# DEVELOPMENT AND ECONOMIC EVALUATION OF A PATIENT-CENTERED CARE MODEL FOR CHILDREN WITH DUCHENNE'S OR BECKER'S MUSCULAR DYSTROPHY IN NORTH INDIA

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# DECLARATION

I, hereby declared that the presented work in the thesis entitled **"Development and economic evaluation of a patient-centered care model for children with Duchenne's or Becker's Muscular Dystrophy in North India"** in fulfilment of degree of **Doctor of Philosophy (Ph. D.)** is outcome of research work carried out by me under the supervision of Dr. Pooja Kansra, working as Professor & COD Economics, in the Mittal School of Business of Lovely Professional University, Punjab, India. In keeping with general practice of reporting scientific observations, due acknowledgements have been made whenever work described here has been based on findings of other investigator. This work has not been submitted in part or full to any other University or Institute for the award of any degree.

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# CERTIFICATE

This is to certify that the work reported in the Ph. D. thesis entitled **"Development and Economic Evaluation of a Patient-Centered Care Model for Children with Duchenne's or Becker's Muscular Dystrophy in North India"** submitted in fulfilment of the requirement for the reward of a degree of **Doctor of Philosophy (Ph.D.)** in Mittal School of Business, is a research work carried out by Titiksha Sirari, 41800848, is a bonafide record of his/her original work carried out under my supervision and that no part of the thesis has been submitted for any other degree, diploma or equivalent course.

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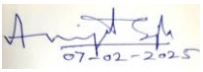

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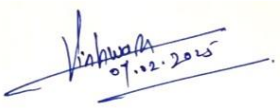

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# ABSTRACT

Duchenne Muscular Dystrophy (DMD) is a genetic disorder caused by a mutation in the X-linked recessive gene, predominantly affecting males. DMD leads to irreversible damage to skeletal muscles, resulting in their weakness, loss of ambulation, respiratory problems and cardiomyopathy. Affected boys become non-ambulatory and become wheelchair-bound by the age of 11 to 13 years. Their life expectancy is restricted to late teens or early twenties. Evolution in the knowledge related to DMD over the years has changed the natural history of the disease. Early diagnosis and interventions have prolonged survival. Does early diagnosis and interventions prolong survival in resource-limited settings like India? The disease requires a multi-disciplinary team approach for comprehensive management that encompasses diagnostic services, physical therapy, orthotics, respiratory therapy, corrective surgeries e.g orthopaedic.

In late stages of disease ventilation support and palliative therapy is also required. The absence of such comprehensive care is directly linked to compromised quality of life for both patients and their families, exacerbating the substantial financial burden imposed by the disease. The disease has substantial direct and indirect care costs. While studies from Europe and the United States shed light on the cost of illness (COI) associated with the disease, there is a notable gap in research from low- and middle-income countries (LMICs). The patient-centered care model has proven effective in managing various chronic illnesses, reducing care costs and yielding better health outcomes. Therefore, there is a felt need to develop an effective patient-centered care model tailored in LMICs to improve the QoL of affected families.

The study aims to estimate the economic burden on families associated with the disease. The study also intended to understand the various socioeconomic factors that are directly or indirectly associated with the COI/economic burden. Subsequently, the study sought to develop a patient-centered care model (intervention) to empower affected families in coping with the disease, ultimately reducing the financial burden. It finally aimed to compare the intervention's impact with routine care on the quality

of life (QOL) of both caregivers and patients, as well as the economic burden, after a 6-month period.

A quasi-experimental design study was considered to answer the study objectives. The study was conducted in two phases, each spanning a 6-month period. Phase one involved observing patients receiving routine hospital care, while phase two implemented the intervention. The rationale behind developing the Patient-Centric Care (PCC) model for DMD patients was to provide patients and their parents/caregivers with essential basic information, enabling them to actively engage in obtaining comprehensive care, particularly in resource-limited settings. The developed intervention was termed the comprehensive DMD-telecare model. This includes teleconsultation as one of its key components to reduce patient physical visits Teleconsultation would be especially beneficial for non-ambulatory patients. The study is based on three major outcomes, the health-related-quality-of-life (HrQoL) of the patient, the QoL of caregivers, and the economic burden of disease on the families.

During the recruitment phase of the first phase, patients were interviewed using a pretested semi-structured schedule adapted from prior studies conducted in alike settings. This interview aimed to gather information on sociodemographic characteristics and quality of life. Subsequently, after treatment initiation, patients were contacted via telephone, WhatsApp and other internet mediums to assess the economic burden. Data on out-of-pocket (OOP) expenditure incurred for the treatment of DMD were collected during these follow-up interactions. As the study evolved WhatsApp came to be a very innovative idea regarding data collection on a real-time basis and this reduced recall bias substantially. This real-time connection with the patient's caregiver also facilitated the implementation of patient-centric care - the intervention. Socioeconomic status was assessed using OP Agrawal scale. health-related quality of life (HRQoL) data were collected from both patients and proxy caregivers at baseline, following phase 1 (routine care), and subsequently after phase 2 (intervention care). The disease-specific QoL tool, PedsQL TM 3.0 Neuromuscular Module, was utilized to assess HRQoL. Additionally, the EuroQoL 5-dimension questionnaire (EQ-5D 5L) was employed to measure QoL specifically in

children affected by DMD. The QoL of caregivers was assessed using the self-administered WHOQOL-BREF tool.

The socioeconomic status, education level, patient's age, and proximity to healthcare facilities are key predictors of the economic-burden over families. The majority of enrolled children were from upper-middle socioeconomic backgrounds, with nuclear families. Most participants were from Punjab, Haryana, and Chandigarh. Caregivers were predominantly mothers, primarily from rural areas with up to matriculation education. Notably, most children were in the late non-ambulatory disease stage (Stage III). Frequent falls and difficulty in walking were common initial symptoms, with nearly half of the parents lacking prior knowledge about DMD. Utilization patterns of care revealed increased rehabilitation service usage during the intervention, particularly in late-stage healthcare needs.

Teleconsultation significantly reduced physical visits, though some patients did not participate due to various reasons like declining ambulation or lack of interest. The pathway to care showed an average of 2.9 treatment agencies consulted before arriving at the study site, with longer pathways observed in advanced disease stages.

Financially, the study revealed a significant annual mean direct expenditure, primarily stemming from direct medical costs, with indirect costs primarily associated with unpaid caregiving. was significantly positively associated with the family's socioeconomic status, the education level of the primary caregiver, and the distance of their residence from the health facility. Conversely, the direct cost of illness was negatively associated with the overall quality of life of the primary caregiver. Whereas indirect costs were linked to caregiver's educational status and urban residence. In joint families, where multiple caregivers are involved, productivity loss is further amplified. Catastrophic health expenditure (CHE) prevalence was notably high, especially among patients traveling from distant areas and those with late symptom onset. Distress financing risk was elevated for patients with delayed symptom recognition and those experiencing CHE. However, disease severity, insurance coverage, and socioeconomic status did not alter CHE or distress financing risk.

The intervention, designed through stakeholder discussions, aimed to reduce the financial burden on families dealing with the disease. Pre and post-intervention (PCC model) evaluations assessed the program's effectiveness in achieving this goal. Children's mean age remained stable throughout the study, with an increase in non-ambulatory patients. The intervention notably reduced total direct costs by about 10 percent, indicating decreased medical resource utilization and associated expenses. Direct medical expenditure constituted a significant portion of the total cost of illness, primarily driven by costs related to procedures, user fees, and rehabilitation services. The intervention led to a significant reduction in direct medical expenditure, primarily attributed to the teleconsultation component of the PCC model. Conversely, the total indirect cost increased in the intervention phase due to heightened economic burden from unpaid caregiving. However, caregiver empowerment was associated with improved caregiving quality and increased hours spent on caregiving. Whereas, disease severity continued to increase over time, negatively impacting patients' physical health. The patient-centred intervention did not significantly affect the health-related-quality-of-life (HrQoL) of children affected with the disease but positively influenced the overall quality-of-life (QoL) of caregivers. The intervention demonstrated promising outcomes in reducing direct medical costs and enhancing caregiver empowerment and QoL, underscoring the importance of tailored intervention in managing chronic conditions like DMD. However, the study lacks a controlled design and doesn't cover the late-stage disease, potentially limiting its generalizability and completeness in understanding the economic burden.

The increasing prevalence and prolonged survival of DMD patients are expected to increase the economic burden on families and healthcare systems. DMD imposes both financial strain and diminished quality of life on patients and caregivers. Patient engagement, particularly through patient-centered care interventions, holds promise in lessening this burden by empowering patients to take ownership of their health. The study demonstrates a significant reduction in direct medical expenditure during the intervention phase, primarily attributed to teleconsultation, which mitigated travel costs. However, indirect costs increased due to unpaid caregiving, although it didn't directly impact household expenditure. The interventions also improved

caregivers' QoL and prevented the decline in patient’s QoL. The study emphasizes the effectiveness of telemedicine in reducing costs and advocating for its integration into healthcare models, especially during the COVID-19 era, to improve disease management and follow-up care. Furthermore, tailored interventions targeting lower socioeconomic status can further reducing the financial burden and enhance caregivers' quality of life.

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Want to begin with “I believe in you…”

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***Titiksha Sirari***

***Dated:***

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| S.No | ABBREVIATIONS | FULL FORM |
|---|---|---|
| 1 | AFO | Ankle-Foot Orthosis |
| 2 | BMD | Becker Muscular Dystrophy |
| 3 | BWS | Best-Worst Scaling |
| 4 | CCWG | Care Considerations Working Group |
| 5 | CDC | Centers for Disease Control and Prevention |
| 6 | CHE | Catastrophic Heath Expenditure |
| 7 | CK | Creatine Kinase |
| 8 | CEA | Cost-Effective-Analysis |
| 9 | CUA | Cost-Utility-Analysis |
| 10 | CMA | Cost-Minimizing-Analysis |
| 11 | CBA | Cost-Benefit-Analysis |
| 12 | CI | Confidence Interval |
| 13 | COI | Cost Of Illness |
| 14 | PCP | Patient Care Pathways |
|  | DMD | Duchenne Muscular Dystrophy |
| 15 | DVT | Deep Vein Thrombosis |
| 16 | DEXA | Dual X-ray Absorptiometry) |
| 17 | EQ-5D | EuroQol- 5 Dimension |
| 18 | FCC | Family Centered Care |
| 19 | FVC | Forced Vital Capacity |
| 20 | GDP | Gross Domestic Product |
| 21 | HRQOL | Health-Related Quality Of Life |
| 22 | HIV | Human Immunodeficiency Virus |
| 23 | INR | Indian National Rupee |
| 24 | ICF | International Classification Of Functioning |
| 25 | IHC | Immune Histo Chemistry |
| 26 | ICRC | Institutes Collaborative Research Committee |
| 27 | IR | Intervention Room |

| | | |
|---|---|---|
| 28 | KFO | Knee-Foot-Orthosis |
| 29 | LMIC | Low and Low Middle Income Countries |
| 30 | LPU | Lovely Professional University |
| 31 | MMT | Manual Muscle Test |
| 32 | MD | Muscular Dystrophies |
| 33 | MLPA | Multiplex Ligation-dependent Probe Amplification |
| 34 | MID | Minimum Important Difference |
| 35 | NGS | Next-Generation Sequencing |
| 36 | NHLBI | National Heart, Lung, and Blood Institute |
| 37 | NHP | National Health Policy |
| 38 | NEP | National Education Policy |
| 39 | OPD | Out Patient Department |
| 40 | OOP | Out-of-Pocket |
| 41 | PCR | Polymerase Chain Reaction |
| 42 | PCC | Patient-Centered Care |
| 43 | PPMD | Parent Project Muscular Dystrophy |
| 44 | PEI | Patient Engagement Index |
| 45 | PGIMER | Post Graduate Institute Of Medical Education And Research |
| 46 | QALY | Quality Adjusted life years |
| 47 | SF | Section Form |
| 48 | SDG | Sustainable Development Goal |
| 49 | SES | Socio Economic Status |
| 50 | SD | Standard Deviation |
| 51 | SPSS | Statistical Package for the Social Sciences |
| 52 | SMA | Spinal Muscular Atrophy |
| 53 | SOC | Standard Operating Care |
| 54 | SOP | Standard Operating Protocol |
| 55 | TREAT-NMD | Treat Neuromuscular Diseases |
| 56 | UDID | Unique Disability Identity |

| | | |
|---|---|---|
| 57 | VAS | Visual Analog Scale |
| 58 | WTP | Willingness To Pay |
| 59 | WB | Western Blot |
| 60 | WTP | Willingness-to-pay |
| 61 | WHOQOL-BREF | World Health Organization Quality of Life-BREF |

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# CHAPTER I
# INTRODUCTION

## 1.1. Muscular Dystrophies (MD)

Muscular Dystrophies (MD) are conditions with progressive weakness of the "skeletal muscles" due to their degeneration This leads to different levels of functional disabilities. (Suthar & Sankhyan, 2018). These dystrophies are genetic in origin and differ according to the clinical features, pattern extent of the weakness of muscles, age when first appeared, amount of progression with time and type of inheritance. Some MDs are seen in infancy or early childhood, while others do not appear until middle age or later (Darras, 2023). There are more than 30 MDs and Duchenne Muscular Dystrophy (DMD) is the most common form of muscular dystrophy in childhood (Younger, 2023). DMD is a chronic, lifelong disease; the affected children become non-ambulatory at the age of 13 and die by their late teens or early twenties. Becker MD is like DMD, but is less severe with preserved ambulation till adulthood (Centers et al., 2009).

## 1.2 Duchenne and Becker Muscular Dystrophy

Duchenne muscular dystrophy (DMD) is a rare and progressive muscular disease, primarily affecting boys. It is the most common muscular dystrophy of childhood, with an incidence of approximately 1 in 3,500 to 6,000 male live births. This condition is caused by mutations in the DMD gene, which encodes the protein dystrophin, essential for muscle fiber strength and stability. The absence of functional dystrophin leads to muscle degeneration and weakness, beginning in early childhood (Emery, 1991). There is no study on the prevalence or incidence of DMD in India. However, a study from southern India involving 275 genetically confirmed patients with DMD was conducted to investigate the natural course of motor milestones (Singh et al., 2018).

Dystrophin gene mutations which are located on the Xp21 chromosome cause Duchenne and Becker Muscular Dystrophy. The consequence of a dystrophin gene mutation is the production of a small, truncated and lesser amount of dystrophin protein. Dystrophin is an essential protein component of the muscle cell membrane (Nicholson et al., 1993). The absence of functional dystrophin protein causes

premature muscle degeneration, resulting in irreversible progressive damage to skeletal muscles. This results in weakness of the muscles, loss of ambulation, breathing issues and cardiomyopathy. No definitive treatment is currently available for this irreversible disease (Bushby et al., 2010a). Its management mostly revolves around a multidisciplinary approach. These management strategies mainly focus on symptomatic management of various manifestations, primary and secondary complications. This may include physical therapy, orthopedic appliances used for support, respiratory therapy, corrective orthopaedic surgery and speech therapy (Bushby et al., 2010b). By the age of 3 to 5 years, boys afflicted with this condition encounter challenges in rising from a seated position, engaging in running or ascending stairs. This is primarily attributed to calf hypertrophy and proximal muscle weakness in the lower extremities. Independent ambulation becomes unfeasible for these children by the age of 9.5 years (Suthar & Sankhyan, 2018). Looking at adolescents and young individuals with DMD, have shown that their quality-of-life (QOL) is compromised. This is due to physical constraints and limited social involvement (Pangalila et al., 2015). Improving and maintaining higher functional abilities via devices could potentially elevate their QOL by enabling more active participation in social activities (Narayanaswami et al., 2015).

The studies on cost of illness from various countries have suggested that the economic burden due to the disease is substantial and these costs increase as the condition worsens (Landfeldt, 2016). For instance, in 2013, medical costs ranged from 4,420 Euro (€) for stage 1 patients to 68,968 Euro (€) for stage 5 patients, a big difference. These studies help us see how costs change and vary in different places. Research over the last three decades in this field has led to rapid strides regarding the understanding of the natural history of DMD and how it impacts our QoL and finances (Ryder et al., 2017).

### 1.3 Management of Disease

Managing any chronic disease aims to prevent illness and enhance quality of life. Making care widely accessible can involve reducing costs, expanding insurance coverage and promoting telehealth (Velasco et al., 2019). These steps improve

healthcare availability, benefiting more individuals. Since there's no definitive medical treatment for DMD, the focus is on mitigating the effects of the dystrophin protein's absence. This approach forms the primary basis of DMD care (Pane et al., 2014). Corticosteroids and immunosuppressants are utilized to decelerate muscle degeneration while antibiotics are employed to combat respiratory infections. (Birnkrant et al., 2010). The use of systemic steroids was introduced as standard care in 1990 and there is sufficient data to show improvement in disease outcomes by slowing down the progression of the disease and extending survival (Matthews et al., 2016).

A study involving numerous clinically confirmed DMD patients across multiple countries assessed the impact of glucocorticoids on key clinical outcomes. Patients on corticosteroids-maintained ambulation for a longer period required less scoliosis surgery or assisted ventilation (Bertoni, 2008). However, steroids did not appear to offer clear cardiac muscle protection at age 20. In patients aged 20 and older, steroids demonstrated some statistically significant cardioprotective effects (Birnkrant, et al., 2016). Assistive technology along with occupational therapy are also beneficial. A few complicated cases of Duchenne may require pacemakers to address cardiac abnormalities and assisted ventilation to manage respiratory muscle weakness. (Birnkrant, et al., 2018).

In individuals with Duchenne Muscular Dystrophy, the use of glucocorticoid treatment is linked to a lowered risk of losing meaningful mobility, upper limb milestones and even death over their lifespan. This treatment directly contributes to an improved quality of life through medical care (Henricson et al., 2013). However, this improvement comes with a price as the financial burden on families. Additionally, it is uncertain how much the quality of life actually improves solely due to medical care (Mitterer et al., 2021). Duchenne kids need a continuum of care, a comprehensive and coordinated approach throughout their life span. This involves integrating various stages of care seamlessly to provide continuous support and treatment (McDonald et al., 2018).

In the era of corticosteroid treatment, a systemic review was conducted to revisit the present natural history of the disease. The key clinical milestones were summarized according to age. The key clinical milestones studied were loss of ambulation, scoliosis, ventilation, cardiomyopathy and mortality. The clinical outcome of DMD patients on corticosteroids in various healthcare environments and involving a significant number of older patients have shown that there is a definitive gain in walking ability, reduction in scoliosis surgery and ventilation requirements, and somewhat lesser heart issues. A significant proportion, approximately 16 percentage, survive up to 20 years with this treatment. Further studies involving adult patients show that survival after 20 years is assessable. By the age of 30, around 44.2 percentage to 56.8 percentage of individuals experience mortality rates (Szabo et al., 2021).

Corticosteroids have even prolonged survival. Natural history studies indicate that losing the ability to walk usually occurs in the early teens, while the requirement for ventilation and heart problems arises in the late teens (Wasilewska et al., 2020). Mortality arises in the third or fourth decade of life. Differences in rates could be due to factors like study design, treatments and mutations. Thus, we have gone ahead to mitigate the impact of the disease and extend the time available for affected patients (Rodger et al., 2012). Most of the clinical outcome measures describing the natural course of illness are taken from European countries or North American studies. The health status of any region not only depends upon the healthcare delivery system but also upon various other factors like social, economic and political diversity (Chongsuvivatwong et al., 2011).

Hence, it is important to consider southern Asia to have an insight into the natural course of the disease as the region contributes to about one-fourth of the world population (worldBank, 2022). Contrary to the studies from Europe and Northern America the kids become non-ambulatory by the age of 10 to 11 and survival is also restricted to 15 years(about 98 percentage of the cohort). Although DMD is a more homogeneous disorder, symptoms are not affected by phenotypic or genotypic variants; but the outcome is affected by the pattern of healthcare (Singh et al., 2018).

Extended survival leads to significant life transitions, needs newer guidelines and newer concept to care. A fresh perspective on health acknowledges the need for strategies beyond biomedicine (Tulchinsky & Varavikova, 2014). The health field concept highlights biology, lifestyle, environment, and healthcare as key determinants of health. This emphasizes public health's importance and forms the foundation for health promotion (Tulchinsky, 2018).

The paradigm of patient care has advanced into a multidisciplinary framework. It no longer focuses solely on addressing the primary symptoms of the disease (Roberti & Tassinari, 2015). The contemporary objective encompasses the optimization of functional independence, enhancement of quality of life and augmentation of overall longevity for individuals affected by DMD (Uttley et al., 2018). Achieving these objectives requires coordinated and multidisciplinary care. In 2010, the DMD Care Considerations Working Group (CCWG) identified eleven key components of care, which include: (1) diagnosis, (2) neuromuscular management, (3) rehabilitation, (4) gastrointestinal and nutritional management, (5) respiratory management, (6) cardiac management, (7) orthopedic and surgical care, (8) psychosocial management, (9) primary care and emergency care, (10) endocrine management (covering growth, puberty, adrenal insufficiency, and bone health), and (11) care transitions across the lifespan (Bushby et al., 2010b).

The primary management approach for the disease still revolves around physiotherapy and glucocorticoids, which play a crucial role in delaying disease progression milestones. In addition, DMD-CCWG committee has provided the basic framework for the management of the disease. It has also raised certain issues related to the support and care required. The committee has highlighted the need for educating the patient and caregivers for optimal care. Their societal inclusion, autonomy and smooth transition of care require appropriate interdisciplinary coordination (Bushby et al., 2010a; Birnkrant et al., 2018a; Birnkrant et al., 2018b; Birnkrant et al., 2018c).

### 1.4 Impact of New Care Guidelines

Individuals diagnosed with DMD should get ahead a successful transition into adulthood, encompassing various aspects such as education, employment, healthcare, and social needs. A smooth transition from adolescence to adulthood is essential for ensuring their overall well-being and quality of life. This specifically refers to the transition of health services from pediatric to adult (Wasilewska et al., 2020). The transition period is described as lasting from 16 to 18 years of age Initially, due to a lack of appropriate treatment, most of the patients could not make it up to 18 years of age. So a transition of care was never an issue in the past (Rodger et al., 2012).

There is no effective treatment available for DMD so far. Treatment approaches and methodologies have evolved, particularly with the emergence of gene therapy and gene-editing tools. Despite these advancements, their clinical impact remains limited thus far. (Hotta, 2015). With improved medical care, there is an improvement in the as well as the survival rate of DMD patients. Thus, there is a felt need for proper planning for the transition process (Wasilewska et al., 2020).

Considering DMD as a lifelong chronic disease that progresses with the advancement of age, the major challenge is to provide continuous, effective therapy and coordinated care from multiple disciplines which can be the mainstay of the much-needed high-quality medical care by DMD patients. (Rothman & Wagner, 2003; Ramli & Taher, 2008). In addition, patient engagement is also needed to reduce the cost of treatment. For taking an active role in one's health; the patient must have the knowledge, skill, and confidence to manage his chronic condition as well as perform health-promoting behaviors. However, studies have shown that the management of various chronic diseases may yield improved clinical outcomes with patient engagement (Coulter, 2012). A clinical trial was conducted in Taiwan on 148 asthma patients, who were engaged in an educational program for self-care for 6 months as an intervention. Controls were given the usual treatment. It was found that lung function improved significantly in the intervention arm (Huang et al., 2009). Another study from Ireland conducted over 64 patients with bronchiectasis found that when patients were engaged in in-person, group-based patient self-management for 8

weeks, no significant improvement was seen in lung function; however, patients receiving the intervention were satisfied with the care and learned new self-management techniques (Lavery et al., 2011).

### 1.5 Paradigm Shift in Care, from a "Newtonian Worldview" to a Holistic View

The world is experiencing a shift from a mechanistic or Newtonian perspective to a holistic one. This new approach emphasizes human values, creativity, and evolution in problem-solving (Samet, 2011). This represents a significant shift, given that the reductionistic approach still largely influences the scientific community. This perspective impacts how we understand human health. The biomedical or reductionistic view treats patients as isolated issues, but its main limitation is the failure to recognize that effective disease management requires a comprehensive approach that includes social, economic, sanitary, environmental, and political factors (Morin, 1992; Collins, 1995).

An inclusive paradigm comprises two key components: self-empowerment of the patient, involving the acquisition of skills and control over resources to enable autonomy in decision-making; and holism, which entails a comprehensive understanding of health that acknowledges the interconnectedness of spiritual, physical, emotional, and social factors influencing wellbeing. (World Health Organization, 2006; Angulo & Losada, 2015). Thus, empowering patients means making them take ownership of their own health care. This can be accomplished by discussing treatment options with the patient or caregivers and assisting them in selecting the best management strategies, settings, and needs (Sirari et al., 2023).

Involving patients in taking ownership of their own healthcare is called patient engagement (Coulter, 2012). Patient education is an essential component of this engagement behaviour. Patients need to be informed about the choices available, clinicians and insurance, coordinate communications among providers and manage complex treatments on their own. Studies have shown that patient-provider communication increases health outcomes (Gruman et al., 2010; Jenerette & Mayer, 2016).

### 1.6. Patient and Caregiver Ownership

Patient engagement is one of the tools for handling chronic diseases. Both patient engagement and chronic disease management need to be very much patient centeric (Clancy, 2011). The health care model is used to empower patients, thereby creating ownership and defining their role in the disease management in patient- and family-centered care (Vahdat et al., 2014). This encourages active collaboration and participation in decision-making by patients, their families and healthcare providers in designing and managing a customized and comprehensive plan for care. This concept is not new; however, due to the multidimensional complexities, the roles are not well defined for both clinicians and patients (McAllister et al., 2012; Jenerette & Mayer, 2016). The common elements through which this health model can operate are (Reid et al.,2005; Suter, Oelke et al., 2009):

- The healthcare system's mission, vision, values, leadership, and quality-improvement are aligned with patient-centered goals.
- Care is coordinated, collaborative and accessible. The required care is provided at the required time and in the required place.
- Care not only focuses on physical comfort but also the emotional well-being.
- Patient and family penchants, morals, cultural traditions and socio-economic status are valued.
- Patients and their families are an integral part of the care team and has a crucial role in decisions at both the levels i.e patient and system.
- The company of family members in the health care setting is encouraged and also facilitated.
- Information is shared with the family member wholly and promptly to ensure their participation in making informed decisions.

The Patient-Centered Care improves not only individual health outcomes but also collectively at the population level. Healthcare delivery systems are benefited from better satisfaction scores among patients and their families. It enhances the credibility of providers among healthcare consumers. This leads to improved productivity among clinicians and ancillary staff (Hibbard, 2003; Hibbard et al.,

2013b). Strategic planning in healthcare supply chain management is crucial (Vactor, 2012). Emphasis on interactive customer relationships and collaborative communication enhances healthcare operations' effectiveness and support. It also works for system in better resource allocation, reduces expenses, and increases financial margins throughout the continuum of care (Bauman et al., 2003). A quasi-experimental design study from Pennsylvnia, studied the impact of family-centered care in multidisciplinary rounds on health outcome of pediatric patients and thus attaining the satisfaction level of hospital staff as compared with conventional rounds. The intervention (model) of conducting in-patient hospital rounds that emphasizes teamwork and empowers hospital staff. The patient and family are the central points of the entire procedure. Staff satisfaction and the family's ability to participate in their care are significant in using this model (Rosen et al., 2009). A framework of patient engagement works at two different levels. Firstly, at patient care by the providers where patients get information about a condition and make their decision regarding preferences for treatment. Secondly providers reach out to the patients, taking their experience into account and making an advisory to fill the gaps at the first level. Policy making is at 3rd level, where patients and communities are involved (Carman et al., 2013).

The primary goal of patient-centered care is to provide "car*e that is respectful of and responsive to individual patient preferences, needs, and values and ensures that patient values guide all clinical decisions*"(Hare, 2018). The long-term sustainability of this healthcare model is necessary to combat chronic disease; one of the essential components of this model is patient self-care (Luxford et al., 2011). Patient involvement is affected by mainly 5 major factors: Related to patient (demographic characteristics of patients), related to illness (severity of illness), related to healthcare professional (knowledge and beliefs of healthcare professionals), related to healthcare setting (primary or secondary care) and in relation to task (whether the clinical abilities of clinicians are major challenges to required patient safety) (Davis et al., 2007). A patient-centered care model ensures patient engagement, which is related to better health outcomes and reduced costs. The model has been proven to reduce

adverse events, reduce malpractice claims, ensure better employee retention, shorten hospital stays, and lower hospital costs per patient (Charmel & Frampton, 2008).

### 1.7 Patient Engagement and Cost-Cutting

A comprehensive study, using the micro-costing method revealed the substantial economic burden of Duchenne and Becker Muscular Dystrophy. The study, encompassing 363 patients, demonstrated annual costs of up to $78,913 for DMD and $39,060 for BMD. The economic impact primarily originated from informal care, indirect costs due to reduced productivity and direct medical expenses like rehabilitation services and medical aids, all closely linked to disease progression. This research highlights the correlation between disease progression, declining health-related-quality-of-life and the escalating economic burden associated with complications (Katz et al., 2014). Similar findings were reported in a European study involving 770 patients. The study unveiled that the annual direct cost of illness per patient varied within the range of $23,920 to $54,270. This cost was significantly higher, ranging from 7 to 16 times, than the per-capita health expenditure in these countries. Indirect and informal care expenses constituted over 60 percentage of the total cost, with a household burden of $58,440 to $71,900 (Landfeldt et al., 2014). These findings underscore the significance of considering the economic implications of rare diseases in health policy assessments, intervention programs, novel therapies and future research initiatives (Angelis et al., 2015). The direct costs of DMD patients in the US, drawing from medical claims of commercially insured individuals with DMD. The resource used and the entire medical costs of DMD were extensive and increased with age (Thayer et al., 2017).

Although it is difficult to quantify life in monetary terms, policymakers still make assumptions about society's willingness-to-pay (WTP) for a life or a life-year in deciding the allocation of public funds (Thavorncharoensap et al., 2013). However, studies focusing on the cost of DMD have mainly estimated the direct costs of the illness (Ryder et al., 2017). In addition, human costs are estimated in terms of indirect costs, valued at the loss in production to society following the "Human Capital Approach". However, this understates the actual societal loss (Trovato, 2020). The

average annual mortality cost associated with DMD was calculated based on the mean total life-years that patients would have had if they were not affected by the disease. These figures were determined to be 3313, 4470, 16,105 and 3564 for Italy, Germany, the United States and the United Kingdom, respectively. The estimated average mortality cost in millions was €248 for Italy, €335 for Germany, €1,208 for the United States and €267 for the UK (Landfeldt et al., 2017).

These findings highlighted the considerable financial impact. This data plays a crucial role in shaping health economic policies, resource allocation and further research endeavors (Drummond & McGuire, 2001). In the 21st century, economic evaluation is playing an increasing role in the allocation of healthcare resources. Proper allocation of resources is crucial for sustainable development in any institution, particularly in healthcare. The cost of illness studies offers valuable insights for policymakers at both micro and macro levels. The inform decisions regarding the cost-effectiveness of new interventions and help allocate funds for further research. Additionally, these studies aid in prioritizing diseases for preventive interventions, ensuring that resources are utilized effectively to address the most pressing healthcare needs. (Zaleski, 2008; Drummond et al., 2015).

The substantial economic burden associated with rare diseases poses a significant obstacle in disease management, especially in countries with limited health risk pooling mechanisms. This, in turn, results in a catastrophic financial burden on affected families (Prinja et al., 2019; Adachi et al., 2023). In situations where definitive care is either absent or prohibitively expensive, families may choose not to pursue even available symptomatic treatments. Therefore, reduction of disease-related expenditures should be prioritized (National Policy for Rare Diseases, 2021). There is existing evidence that provides insights into cost-saving strategies for long-term, lifelong diseases.

The concept of patient engagement is derived from the marketing conceptualization of consumer engagement, which facilitates consumer satisfaction (Hibbard, 2003). Patient engagement plays a crucial role in healthcare delivery systems, often leading to reduced expenditures and improved health outcomes. The

studies consistently highlight the significant impact of patient engagement on cutting healthcare costs. Actively engaged patients demonstrated substantial cost savings compared to those less engaged, highlighting the crucial role of patient involvement in shaping effective and efficient healthcare delivery systems (Hibbard et al., 2013). While many studies have explored the impact of patient engagement on health outcomes, there remains a gap in understanding its effects on healthcare costs. Although research has shown that tailored interventions to increase patient involvement in their disease management can lead to improved health outcomes, fewer studies have specifically examined the associated costs (Hibbard & Greene, 2013). Patient engagement is directly linked to improved health outcomes and a better quality of life (Marzban et al., 2022).

### 1.8 Patient Engagement and Optimization of Quality-of-Life (QoL)

As medical care improves, individuals with DMD are experiencing longer lives, reaching their third decade. This extended lifespan means more years dealing with functional limitations and caregiving. The previous studies have shown mixed results regarding the quality of life (QoL) among DMD patients (Landfeldt et al., 2018). The capacity to participate in daily activities significantly influences health outcomes, spanning mobility, personal care, education, recreation, spirituality, community engagement and social interactions. Physical disability notably diminishes participation, thereby impacting quality of life (Wei et al., 2017). However, studies have indicated that while physical participation is lower in DMD patients, other participation levels remain similar (Uttley et al., 2018).

A study comparing life activity participation and perceived QoL in younger & adolescent boys with DMD against healthy peers revealed noteworthy findings. While perceived QoL exhibited no variance between the DMD group and controls, older adolescents with DMD displayed significantly lower physical, social, and overall QoL scores. This approach using QoL as an outcome measure, provided insights beyond traditional physical outcomes. Individuals with DMD showed diminished involvement in physical activities, lower physical and social scores and compromised school-related scores. Moreover, their total quality of life (QoL) scores was

significantly lower when compared to unaffected boys. Emotional scores remained unaffected, even among older boys. While QoL scores were lower in DMD patients compared to unaffected peers, they remained relatively consistent over time within the DMD cohort, despite disease progression (Bendixen et al., 2012). In a Taiwanese study, adolescents and young individuals with DMD were evaluated to understand their QoL. HrQOL was assessed using the Short-Form-36(SF-36), while global QoL was measured using the World Health Organization Quality of Life (WHOQOL-BREF). The study also examined the relationship between functional status and QoL. The results demonstrated that these patients experienced poor HrQOL and global QoL. Among the attributes assessed using SF-36, physical function was the most affected (90.7 percentage), while mental health was the least impacted (7.8 percentage). Although differences in physical activity were noted across age groups, other health attributes showed no significant variations. The domain that saw the greatest impact on WHOQOL-BREF scores was physical health. Both adolescents and young men faced significant challenges in the social domain of QoL (Lue, Chen, & Lu, 2017). Additionally, it was observed that measured HrQOL, using the Pediatric-Quality-of-Life-Inventory (PedsQL). Often involving parent-proxy scores for reporting their child's QoL (Ryder et al., 2017).

An exploration was conducted to comprehend life themes in individuals with DMD. Approximately 45 published articles were examined, leading to the creation of a thematic framework encompassing four key life themes: firstly physical; secondly psychological; thirdly social and fourthly well-being. DMD entails a multifaceted quality-of-life construct that traditional QoL tools struggle to fully capture. The review also shed light on how the disease adversely affects caregivers' quality of life (Uttley et al., 2018). The caregivers of individuals with DMD experience significant mental and physical burdens, which impact their daily activities and work lives. This often results in impaired HrQoL, pain, stress, depression, sleep quality, sexual dysfunction, family function and self-esteem issues (Landfeldt et al., 2018). Most of the time, primary caregivers are mothers and the perceived HrQoL of their sons significantly influence their own health-related-QoL and emotional suffering. Consequently, the physical and emotional well-being of mothers impacts their

participation in social activities, which are essential for managing stress among caregiving parents. The screening of mothers for anxiety and depression is recommended so that proper intervention could be taken to improve psychosocial functioning among these families. DMD is an incurable chronic disease that places a financial, social and psychological strain on families (Jackson et al., 2021).

In a large multicentric study, around 770 caregivers were surveyed. Most caregivers were parents and from the middle class. The average EQ-5D utility-index ranged between 0.85 and 0.77 in ambulatory patients and 0.88 to 0.57 in caregivers' ratings of patients' health and mental status. About 70 percentage of caregivers reported anxiety and depression and the annual household expenditure was increased (Landfeldt et al., 2016). In the health sector, patients engagement in their own health enhances their level of satisfaction and leads to better health outcomes (Graffigna et al., 2014). In a study from Hong Kong, patient engagement levels were correlated with HrQoL; 686 patients were enrolled in the general outpatient clinic. The patient engagement index (PEI) was used as a reliable and valid instrument to evaluate their PE level. HrQoL was assessed using the EQ-5D. The results indicated that HrQoL was directly related to the level of patient engagement. This association, however, could be influenced by physical, social and psychological factors (Xu et al., 2019). The patient-centered care models enhance patients' engagement in their treatment more specifically for chronic life-long disease. A study involving six hundred and fifteen diabetic patients receiving patient-centered care (PCC) found a correlation between improved physical and mental components of quality of life (QoL), enhanced diabetes self-care management, and better medication adherence (Williams et al., 2016).

### 1.9 Role of Stakeholders in DMD Patient Care as Partners (Patient-Centered Care Model)

Managing lifelong diseases like DMD necessitates a multidisciplinary approach, focusing on care transition, building enduring patient relationships, integrating expertise through teamwork, and maintaining thorough documentation (Bushby et al., 2010b). Each team member's role, from diverse disciplines, must be

clearly defined to facilitate training in the Patient-Centered Care (PCC) approach (Wigert & Wikström, 2014). Tailored treatment and support based on individual needs are vital (Rosen et al., 2009). However, further research is needed to integrate PCC into rare diseases and implement PCC education for team members effectively.

Patient and caregiver inn DMD, the patient is usually a young child, might not fully comprehend their diagnosis while the caregiver experiences various emotional phases (Schwartz et al., 2021). After the diagnosis, caregivers initially feel shocked, finding it hard to believe. This denial mode sometimes leads them to seek opinions from various forms of available options, ranging from medical, traditional healers, faith healers etc. This contributes to financial burdens even before the actual disease management begins and delaying the start of standard care (Fujino et al., 2016). They transition from shock and disbelief to a phase of distress and sadness as they come to terms with the reality of the diagnosis. Over time, they begin to cope with the challenges presented by DMD, finding ways to manage and support the patient. This emotional journey, from shock to acceptance, is often more intense for caregivers, who bear the responsibility of understanding and supporting the DMD patient's needs (Forman, 2020). To bridge this emotional gap and minimize the financial burden, it's essential to provide proper counseling and educational sessions to caregivers and patients during their initial hospital visits. This support can help in gaining the confidence of caregiver thereby to make caregivers understand the disease, manage their emotional responses and access appropriate care sooner, ultimately improving the overall well-being of both the patient and the caregiver (Espinosa et al., 2021).

Family members often make sacrifices to support their loved ones, sometimes increasing their working hours or relying on state benefits to cover extra costs. Poor family relationships can hinder chronic disease management and create emotional challenges. Some families, however, grow closer as they work together to support each other (Read et al., 2011).The financial burden is one of the major challenges faced by families. The expenses related to treatment, transportation for appointments, hiring caregivers and modifying the home environment can strain family finances, leading to stress and worry (Golics et al., 2013).

Caring for a family member with a long-term disease often leaves little time for leisure activities, leading to burnout among family members (Mohandas et al., 2021). While the emotional journey can be tough, families play a vital role in providing care, emotional support and navigating the challenges of chronic diseases. Society contributes by providing essential support, creating an accommodating environment and advocating for improved understanding and services. Such support aids affected individuals in accessing necessary resources and services, while an inclusive environment ensures equal participation and opportunities. Advocacy initiatives by society raise awareness, drive policy changes and enhance the well-being of individuals living with disabilities caused by the disease (Iryna & Franz, 2009).

The government's crucial role in managing chronic rare diseases is driven by political will and commitment. In India, the Ministry of Health and Family Welfare's recognition of the challenges posed by rare diseases is evident in the National Health Policy 2017 (The National Health Policy, 2017). In accordance with the National Health Policy 2017, the "National Rare Disease Policy 2021" was crafted to address rare diseases. The policy promotes cost-effective treatment research, local drug production and early detection through screening initiatives. Whereas eight centers of excellence are designated for specialized rare disease care, highlighting the government's commitment to quality management. To gather precise data, a nationwide rare disease registry is established within these centers, reinforcing the government's resolve to tackle rare disease challenges (National Policy for Rare Diseases, 2021). Researchers are essential stakeholders alongside patients, advocacy groups and others. They conduct basic research and clinical trials. Ataluren is conditionally approved in the EU for some patients with Duchenne Muscular Dystrophy. Exon skipping treatments are being developed, with applications for marketing authorizations filed. This reflects ongoing efforts to make advancements in managing the disease (Straub et al., 2016).

## 1.10 National Rare Disease Policy 2021

DMD is a rare, severe and chronic neuromuscular disease, demanding long-term care. The families grapple with emotional and financial hardships due to its debilitating nature (Birnkrant et al., 2018c). The public health approach to rare diseases is scattered across the population, it is not about identifying the risk factors and abolishing them. The factors associated with rare diseases are usually innate or congenital and therefore irremovable (Patsos, 2001). Major challenges in public health regarding rare diseases are: 1) Difficult early diagnosis 2) exact single-case definition is not available. 3) international classification of disease codes for rare diseases is not available; 4) underlying molecular and physiological mechanisms are unknown. 5) treatment is complex and drugs are expensive. 6) no definite standard of care for treatment and rehabilitation, as much of the research work is already ongoing 7) very few longitudinal data available 8) screening strategies are not much efficient (Ayme et al., 2008) and most of the registries and databases are limited. Despite of these challenges, there are reasons quite valid reasons to have a public health approach to address rare diseases. Collectively, rare diseases affect substantial populations. They can severely affect caregiver's lives; economic impact is often substantial for patients, their families and society in general. The actual burden of rare diseases is still unclear. The public health approach may not seem applicable for orphan diseases. The comprehensive public health approach to manage conditions related to these diseases can be considered. The potential benefits of this approach include: 1) it reduces the impact of disease on the patients, their relatives, caregivers and society in general. 2) better management of disease ensures smooth transition between pediatric and adult care. 3) improved health, QoL and life expectancy. This enhances patient's participation in their communities, workplaces and society in general (Valdez et al., 2016).

The Ministry of Health and Family Welfare of Government of India also felt the need to address the issues related to rare diseases, which was reflected in the National Health Policy 2017 of India. The Government of India (GOI) has approved a separate "National Rare Disease Policy 2021," which recommends research on

indigenous and local production of medicines to achieve low-cost treatments for rare diseases. The policy also emphasizes screening and early detection for rare diseases. Financial support is proposed for certain rare diseases classified in the Group A list under the "Rashtriya Arogya Nidhi and Pradhan Mantri Jan Arogya Yojana". Additionally, eight tertiary care centers of excellence for managing rare diseases have been identified. These centers will establish a hospital-based national registry of rare diseases to ascertain the exact burden of rare diseases in the country (The National Health Policy, 2017; National Policy for Rare Diseases, 2021).

Considering a global paradigm shift in patient care, from doctor or health-caregiver-centric to a patient-centric, holistic approach for the management of chronic diseases and the strong political will to manage rare diseases in India, this study was contemplated. Actually, no other study has touched upon these aspects of DMD in India. Thus, core component of providing primary care in chronic life-long disease is to ensure *QoL* through *patient-centered care (PCC)* and in a *cost-effective manner (National Policy for Rare Diseases, 2021).* This study sought to focus on the QOL gained by the children and caregivers after adopting patient-centered care that reduces the financial burden on the families over a time period.

### 1.11 Rationale of the Study

Evolution in the knowledge related to the disease over the years has made early diagnosis, early interventions then prolongated of survival possible. With the prolonged survival and lifelong disease, it is important to the patient must be getting most of the recommended services at the doorstep. The chronic disease like DMD requires regular follow-up, multidisciplinary management along with persistent caregiving. The impact of disease is both in financial form as well as affecting the QoL. The primary care is to provide first-line care and to maintain the quality of life in the patients living with DMD. The disease is not only chronic and debilitating disease but also a rare disease, increased burden is foreseen in the future due to advancement of diagnostic techniques and increase coverage of medical care. Resultant prolonged survival and the increasing prevalence of disease are likely to

have a huge economic burden over the families as well as health care system. DMD is associated with a substantial economic burden.

The projected economic burden of DMD across different countries highlights the substantial impact of the disease on both individuals and society. Indirect and informal care costs, ranging from 18 percent to 43 percent of total costs, underscore the hidden financial toll on families and caregivers. The total societal burden was estimated at between $80,120 and $120,910 per patient and annum and increased markedly with disease progression. The corresponding household burden was estimated at between $58,440 and $71,900 (Landfeldt et al., 2014). These figures illuminate the challenges families face in managing the financial implications of DMD. The economic context of the rare disease is a very important input to evaluate any health policy or intervention programme related to new treatments, financial support schemes to families (Straub et al., 2016).

The patient engagement is one of the tools for handling chronic diseases. DMD is a chronic disease, like any other rare disease. The PCC healthcare model aims to empower patients, enabling them to take ownership and defining their role in disease management within a framework of patient/ family-centered care (Coulter, 2012). This ensures active partnership and participation in decision-making between patients, their families and the providers to plan a customized comprehensive strategy for care. The roles of this model are not very clear either to the clinician or the patients due to the multidimensional which complexities in case of rare diseases ( Hibbard & Greene, 2013).

There is the political will to address care and treatment related to rare diseases. The Indian government, in alignment with the National Health Policy 2017, recognizes the significance of addressing rare diseases. This policy emphasizes patient-centered care to improve the quality-of-life for individuals dealing with chronic, long-term illnesses (The National Health Policy, 2017). Additionally, renowned institutions like the American and Indian Academy of Pediatrics stress the importance of patient and family-centered care, particularly in pediatric healthcare (The Medical Home, 2002).

The present study intends to address by developing a model that takes into account both the consumer (the patient) and the service provider (the healthcare team) to improve the health outcome in the form of improved QoL for both the patient and the caregiver. To best of our knowledge both of these components of public health are not being explored for DMD in India. This study is designed to explore the QoL in patient and their caregivers giving them PCC aimed to reduce the financial burden on the families, followed for a period of time, to evolve a replicable and feasible model of patient-centered individualized care for DMD patients in India. In this era of COVID-19, teleconsultation is the mainstay of access to medical consultation for many chronic diseases. Teleconsultation is the key feature of the present study and is the most appropriate not just for this specific disease but for any other chronic disease.

Furthermore, present study aimed to furnish data on the economic burden of DMD, a rare disease, which remains relatively underexplored in Low- and Middle-Income Countries (LMICs). Assessing the economic burden of a disease holds significant importance in economic evaluations of diverse interventions. These studies provide the foundation for informed decisions regarding the "cost-effectiveness" of new interventions/management and help allocate resources for research and prioritize diseases for preventive measures.

The current study aimed to assess the economic burden on families of boys with DMD receiving care at a government hospital. It also sought to develop a patient-centered care model to enhance the capacity of families to manage the disease and alleviate financial stress. The study further compared the impact of this intervention with routine care on Quality of Life (QOL) for both caregivers and patients and economic burden after six months. The specific objectives of the study are as follows:

1. To estimate the economic burden_among families of boys with Duchenne Muscular Dystrophy seeking care at a government hospital.

2. To develop and validate a Patient-Centered Care model for building the capacity of the affected families to cope with the disease thus reducing the financial burden on families.

3. To compare the impact of two patient management strategies, i.e., Patient centred care model (intervention) as compared to routine treatment for boys (5-15 yrs) with Duchenne Muscular Dystrophy on Quality Of Life (QOL) (caregiver and patient), economic burden after 6-months of intervention.

### 1.12 Chapter Scheme

This research study is structured into six chapters to provide a comprehensive exploration of the subject matter. Chapter I introduces Duchenne Muscular Dystrophy (DMD), delves into the associated economic burdens and introduces the concept of patient-centered care. In Chapter II, an extensive literature review is presented, covering various aspects of Duchenne and Becker Muscular Dystrophy, including the cost of illness (COI), the Patient-Centered Care (PCC) model and Quality of Life (QoL). This chapter also thoroughly examines research gaps within the existing literature. Chapter III elucidates the research methodology, encompassing research instruments, analytical tools and techniques employed for data analysis. Chapter IV explores socio-economic determinants, clinical factors related to the economic burden and the quality of life experienced by both patients and caregivers. Additionally, this chapter investigates the impact of PCC interventions on the economic-burden and QoL. Chapter V presents the research findings, providing a comparative analysis with relevant studies. This chapter also critically assesses the strengths and limitations of the current study and concludes with a summary and conclusion. Finally, in Chapter VI, policy recommendations derived from the research findings are outlined, offering valuable insights for policy implications and future directions.

# CHAPTER 2

# LITERATURE REVIEW

This chapter provides a comprehensive overview of Duchenne and Becker's Muscular Dystrophies related dimensions to health economics. The chapter emphasizes the significance of health economics in evaluating healthcare models and explores research on costing, exploring diverse methodologies for estimating the cost of illness and underscores the significance of indirect costs and productivity loss in health economics. The economic burden encompassing both direct and indirect factors and their determinants has been discussed. Furthermore, the chapter reviews studies related to QoL, an intangible facet of health, discussing QoL types, measurement tools and interpretation. It highlights research efforts focused on various challenges and coping mechanisms for the management of rare diseases. Further, it talks about patient engagement as patient-centered care model to improve health outcomes and reduce the cost of treatment in chronic long-term diseases. It also examines patient engagement evaluations across different settings and diseases, assessing the strengths and weaknesses of the model. The literature reviewed supports the patient-centered care model as an effective approach for the long-term management of chronic diseases. Finally, the chapter identifies opportunities for future research, offering insights for researchers to contribute to existing knowledge and advance the field.

## 2.1 Economic Evaluation

The science that studies resource allocation to produce optimal health in society is known as health economics (Walley & Haycox, 1997). This facilitates economic theories, methods and concepts in the health sector. Economic evaluation is a crucial component of health economics, gaining prominence in healthcare resource allocation and shaping public satisfaction with the healthcare delivery system (Dang et al., 2016; Singh et al., 2022). The traditional theoretical basis of economic evaluation rests on the welfare analysis. Proper resource allocation is paramount for any institution to achieve sustainable development, particularly in the realm of healthcare. The sustainability further more important when the disease of chronic in nature (lifelong) (Drummond & McGuire, 2001).

Health care industry is one of the most fast-growing industries in India with a growth rate of 17 percentage from 2011 to 2020. The citizens are now expecting more

from the industry, better standard of treatment care at affordable costs. On the other hand, Word Health Organization (WHO) is still categorizing India, where citizens bear “catastrophic costs” cost for their health care(Sriram & Albadrani, 2022). Any new intervention or technology in medicine needs to go through evaluation whether it is providing health benefits within the premise of available resource settings(Kobelt, 2013).

The resources for any program are always finite. In most circumstances, choice of strategies and activities is guided by precedence. In some others, it is guided by a situational analysis which includes the epidemiological profile of the area i.e. burden of problem, effectiveness of the intervention etc. (Dang et al., 2016). The economic evaluation is one such tool to assist in such decision making. It may be defined as ‘the comparative analysis of alternative courses of action in terms of both their costs and consequences’(Drummond et al., 2015). The economic evaluation basically sets out to answer two main questions: first, is linked to the concept of **allocative efficiency** i.e. is a health procedure worth doing compared with other things that can be done with the same resources. Secondly, it addresses the issue of **technical efficiency** i.e. Is it beneficial that the health care resources should be spent in this way, rather than in any other way. This involves addressing issues which are more local in nature and which involve operational decisions such as how do to implement a program (Palmer & Torgerson, 1999).

Most of the times costs are evaluated in monetary-terms to directly compare the available options that allows for a clear evaluation of their respective strengths and weaknesses. A problem in determining the values of the cost items as for many of them, there is no straight forward method for valuation. For example, patient time, that is sacrificed in seeking treatment, is difficult to price. The real societal costs of services that are availed, like physician's time, are not always clear, as price rates for these services can be of poor approximation (Drummond & McGuire, 2001). The most basic methods for economic evaluation includes: cost-minimization, cost-effectiveness-analysis, cost-utility-analysis and cost-benefit-analysis (Kobelt, 2013).

## 2.2 Costing Studies

Costing in healthcare means figuring out the value of everything needed for a medical treatment, like materials, medicine, doctor and patient time and more. This helps us see if the benefits of the treatment outweigh the costs (Teisberg et al., 2020). Health Care Utilization involves tracking or describing the utilization of services by individuals for preventing, treating health problems, maintaining well-being, or gaining health-related information (Andersen, 2008) The associated cost or expenditure involved in the management of disease represents the economic burden of the disease (Diehr et al., 1999).

### 2.2.1 Healthcare Utilization

The statement that healthcare utilization in India is skewed towards the private sector for curative care and towards the public sector for preventive services is generally accurate (Kalita et al., 2023). However, this pattern does not necessarily hold true for rare diseases and there are several reasons for this discrepancy, especially if talk about LIMCs (Schulenburg & Frank, 2015). Most rare diseases (RDs) are intricate, disabling and potentially life-threatening conditions with a genetic basis (Valdez et al., 2016). Despite the substantial health challenges and limited treatments faced by RD patients, their comprehensive impact on healthcare remains unclear (Opazo et al., 2021). Assessing the overall cost and utilization within healthcare systems is a direct approach to understanding the disease burden of RDs (Grosse et al., 2016).

The comprehensive healthcare utilization by pediatric or adult populations with rare diseases (RDs) remains inadequately documented, even in high-income or upper-middle-income countries. Emerging evidence suggests that RDs might exert a disproportionate and substantial impact on healthcare, surpassing the prevalence of RD patients (Schulenburg & Frank, 2015). Pediatric and adult discharges associated with rare diseases (RDs) were significantly higher than the discharges with common chronic (CCs) diagnoses. Thus, health-care utilization was significantly high compared to other chronic diseases. These expenditures contribute almost half of the US national healthcare expenditure (Opazo et al., 2021). A recent study examining

healthcare utilization in pediatric patients total charges for suspected genetic diseases, many of which are rare, accounted for 46 percentage ($57 billion) of the "national bill" for pediatric patients in 2012 (Gonzaludo et al., 2019). Treating patients with rare diseases is often both expensive and challenging. Many of these conditions lack effective therapies, leading to an inequitable pattern of utilization (Schulenburg & Frank, 2015). Medical utilization and expenditures of rare disease patients has shown a trends in inequity in South Korea. The horizontal inequity index (HI) of patients with rare diseases has shown an increase in inpatient utilization and expenditures in a pro-rich state. However, health equity studies for other diseases in South Korea indicated that low-income patients tended to use medical services more than high-income patients due to their greater healthcare needs (Kang & Choi, 2023). In China approximately 61.4 percentage of rare disease patients sought medical consultation from general tertiary medical centers. However, Out-of-pocket expenses constituted 32.9 percentage of the total treatment cost, slightly exceeding the national average. The influence of urban residency on medical costs and utilization was apparent, highlighting the necessity for equitable health insurance coverage considering both common and rare diseases. Thus it is important to consider critical illness insurance options to make comprehensive healthcare coverage certain (Min, Zhang et al., 2019). Rare disease is not rare, considering among the largest population in the world. To attain universal healthcare coverage (UHC), it is imperative to include provisions for rare diseases (RDs) as part of the Sustainable Development Goals, ensuring that the RD population receives quality healthcare without financial hardship (Chung et al., 2023). Strengthening primary health care (PHC) in low- and middle-income countries has been proven to enhance access and health outcomes (Buendia et al., 2022). A review about how PHC impacts patients with rare diseases, considering perspectives from various stakeholders. This review recommends strengthening the primary healthcare system has positive effects on rare disease patients and enhances overall healthcare performance. However, attempts to dismantle this system should be carefully assessed and educational programs promoting its benefits are crucial for public awareness. Encouraging courses and workshops for healthcare professionals, emphasizing multidisciplinary teams, can further improve the system (Ferreira et al., 2023).

The economic burden linked to DMD is considerable, primarily focusing on healthcare utilization, caregiving and diverse resource requirements for individuals affected by the condition (Teoh et al., 2016). The estimates of medical care utilization and expenditures for individuals under 30 with muscular dystrophies in the United States reveal significant financial implications. Privately insured individuals with muscular dystrophy incur approximately $18,930 in incremental annual expenditures, with medical expenses averaging 10 to 20 times higher than those without the condition. Particularly, individuals aged 15 to 19 years exhibit the highest number of inpatient admissions attributed to respiratory infections and cardiac complications.

The socio-economic differences between sub-populations revealed possible explanations for disparities in care. The study emphasizes the public health concern associated with the condition and equitable distribution of health care for UHC (Andrews et al., 2014). A better understanding of service utilization and families' perceptions can help identify and address potential gaps and barriers in caring for individuals with DMD, informing public health policies to support these families (Ferreira et al., 2023). The potential barriers to successful healthcare utilization in include the absence of tailored informational resources and education to support their transition from adolescence to adulthood within the healthcare system. Future studies should investigate the underlying reasons for these barriers and develop optimal transition programs tailored for males with DMD (Paramsothy et al., 2018). In a retrospective population-based study on healthcare utilization, including males under 30 years of age in 2017, the first-year post-diagnosis healthcare utilization included a mean of 0.48 hospitalizations (with a length of stay of 9.37 days), 3.96 general practitioner visits, 28.52 specialist visits and 20.14 ambulatory care visits. The mean direct costs amounted to $18,868 CAD in the first year post-diagnosis (Chen et al., 2022). Inequalities in utilization of healthcare services among patients with muscular dystrophy were observed (Chung et al., 2023). An analysis of health insurance and administrative data revealed disparities in healthcare utilization among different racial groups. Blacks demonstrated lower primary care, specialist care use, therapy and overall utilization compared to whites and other races. However, they had higher rates of hospitalization and emergency treatment. The most significant gap was observed in

outpatient services, with blacks utilizing these services 50 percentage less than whites and 70 percentage less than others. These findings highlight persistent racial disparities in healthcare utilization, even among individuals with access to the same health benefits. Further research is needed to explore additional factors contributing to these disparities, such as resource awareness, health knowledge, or access barriers like transportation (Ozturk et al., 2014). Living with rare diseases imposes substantial burdens on patients, encompassing health, psychosocial, occupational and financial challenges (Ouyang et al., 2011). The financial burden involves both direct (medical and nonmedical) and indirect costs, ranging from hospitalizations and medications to rehabilitation care and support services. The annual costs for certain rare diseases can extend into millions, placing considerable strain on individuals and their caregivers (Opazo et al., 2021).

### 2.2.2 Cost of Illness of Rare Diseases

The cost-of-illness (COI) approach is a comprehensive method to measure the societal impact of a disease. It considers various factors such as disease incidence, prevalence, effects on life expectancy, morbidity and QoL. This approach assesses both direct and indirect financial burdens at different levels from society to individuals and families, providing a wide-ranging perspective on the disease's overall impact. It is often referred to as the economic burden of the illness. (Jo, 2014). In the book elementary economic evaluation in health care descried COI studies are descriptive, these itemize, price and sum all the costs related to a particular problem and finally gives an idea of economic burden(Jefferson et al., 2000). The most tedious is to estimate the intangible costs. A good assessment of an intervention needs to examine all costs direct as well as consequential irrespective of who are the people bears them or where they are occurring in society(Drummond & McGuire, 2001; Drummond et al., 2015).

However, Rice (2000) described cost of illness (COI) studies and their importance. COI can be classified into two major categories: (1) core costs are direct resultant of the illness and (2) cost is related to the costs that includes non-health costs. There are different perspectives to conduct COI studies: societal, healthcare

system, third-party players, industries, the government, patients and their families (Russell et al., 1996). Every perspective has its own different set of cost items does each perspective finally leads to a different kind of result for the same disease. The choice of perspective for any study depends upon the research question and among which group the researcher wants to estimate the economic burden (Afroz et al., 2018). The societal perspective covers the maximum components of cost and most of the time this COI is preferred but if the focus is to see the impact of illness on the patient and their families, we can use participant and family perspectives to have an insight at a family level. This cost includes out-of-pocket expenditures which are losses or household production losses (Rice, 2000). Understanding the economic dimensions and burden of disease is pivotal in securing funding for drug development initiatives targeting rare diseases. Most studies on rare diseases primarily took the societal perspective into account, with a significant portion also considering the third-party payer perspective, which represents health insurance or public healthcare. However, none of these studies included the perspective of patients or their families (Bastida et al., 2017).

Conventionally, cost is divided into three major domains direct cost indirect cost and intangible cost. Intangible costs are difficult to measure and also there are related controversies, so they are often not considered in COI studies (Yousefi et al., 2014). Breakdown of the types of costs that are typically considered to understand the economic impact of a disease, involves considering various costs. These costs are categorized as direct (medical & non-medical expenses), indirect (productivity loss) and intangible cost (pain and reduced QoL). Additionally, preventive measures, research and development and public health initiatives play a role in managing the overall economic burden of the disease on society(Hu & Wagner, 2000).

In a comprehensive review of COI studies across 42 rare diseases in 25 countries, a notable portion focused on DMD. The predominant research approach involved prevalence-based estimation with cross-sectional designs and a bottom-up methodology. Data collection relied heavily on questionnaires administered to patients or caregivers (67 percent), although databases or registries were also used in 48 percent of the studies. The inclusion of costs related to lost productivity, non-

medical expenses, and informal care costs was observed in 68 percentage, 60 percentage, and 43 percentage of the studies, respectively. The majority of studies on rare diseases were conducted in European countries, with the USA being the second most common location for research (Pérez et al., 2021). The COI of rare diseases (RDs) in Hong Kong, where approximately one in 67 individuals live with one or more rare diseases, was estimated to be HK$1,594,339,530 (approximately US$204,402,504). This constitutes 4.3 percentage of all inpatient costs during the period of 2015-2016 (Chiu et al., 2018). In 2020, a cross-sectional study estimated the total societal cost of rare diseases (RDs) at $484,256 per patient per year, with significant variation (SD $730,736; range $2,920–$6,161,275). The pediatric RD patients had notably higher annual costs ($840,908; SD $954,250) compared to patients from adult age group ($324,126; SD $534,329) ($p < 0.001$) (Chung et al., 2023).

The cost of illness for DMD in 2012 varied annually from €7,657 in Hungary to €58,704 in France. Non-healthcare expenses made up 64-89 percent of the total, with informal care being a significant factor (Bastida et al., 2017). Annual healthcare costs averaged $10,046 AUD per individual, significantly surpassing average health expenditures in Australia. The total mean cost was $46,700 (median $32,300), with healthcare expenses making up 22 percent of this amount (Teoh et al., 2016). A multicentric (cross-sectional) study across Germany, Italy, the UK, and the US estimated the average annual direct cost of illness per patient between $23,920 and $54,270 (2012 international dollars), 7 to 16 times the average per-capita health expenditure. Indirect costs, making up 71 percent of total costs, were significant. The total societal burden ranged from $80,120 to $120,910 per patient annually, increasing with disease progression, while the household burden ranged from $58,440 to $71,900 (Landfeldt et al., 2014).

**Table: 2.2.1 Cost Profile of the Reviewed Studies Globally**

| Author | Publication Year | Cost of Individual/Household per year |
|---|---|---|
| Larkindale et al. | 2014 | In a multicentric cross-sectional study design, German, Italian, United Kingdom and United States. The mean per-patient annual-direct-cost of illness was estimated to range $23,920 and $54,270 (in 2012 international dollars). The total societal burden per patient year ranged from $80,120 to $120,910 and increased significantly with disease progression. The household burden was estimated to range from $58,440 to $71,900 per annum. |
| Katz et al. | 2014 | Annual burden of DMD €78,913, while BMD's total costs were €39,060 in Germany. For DMD, the total mean annual direct medical costs were €19,346, and the direct non-medical costs were €30,884. For BMD, the total mean annual direct medical costs were €5,140, and the direct non-medical costs were €12,471. |
| Teoh et al. | 2016 | Annual mean healthcare-costs are $10,046 Australian dollars per affected individual. The mean total cost was $46,700 with healthcare costs contributing 22% of the total costs |
| Thayer et al. | 2017 | Direct annual cost for DMD (incl. hospital, test, monitoring etc.) was $23,005 in a study from United States. |
| Bastida et al. | 2017 | Estimated as annual cost per person (DMD) from €7657 in Hungary to €58,704 in France. |
| Conway et al. | 2022 | Direct annual cost (incl. hospital, monitoring, test etc.) $174,701 in Midwestern states of US. |

### 2.2.3 Direct Cost

The direct costs include the medical treatment/ management related to the disease and the related complications (Afroz et al., 2018). Direct cost is further divided into two types: Direct health related costs including hospitalization, outpatient services charges/user charge, ancillary services, overhead allocation for fixed costs of utility items, variable costs of utility items, training in new procedures, monitoring costs, diagnostic costs, consumable supplies, personal time, equipment, cost of drugs, devices provided by the household, cost of rehabilitation services, cost related to training and education of patients and their caregivers example self-care training life support skill training (Shankar et al., 2020). The direct non-health related costs including transportation to appointments, boarding logging, and caregiving for housekeeping or childcare, which aren't directly medical but are part of the overall cost of healthcare. The direct costs are much higher for chronic disease than acute diseases or communicable diseases (Jo, 2014).

In 2020, the average annual cost of rare diseases (RDs) in Hong Kong was estimated at HK$441,951 (US$56,660) per patient. This included 38.9 percent for direct healthcare costs and 61.1 percent for direct non-healthcare and indirect costs. The total economic impact of RDs in Hong Kong was projected to exceed HK$53 billion (US$6.8 billion) for the year. Despite RD patients making up only 1.5 percent of the population, their direct healthcare costs represented 11.4 percent of Hong Kong's total healthcare expenditure (Chung et al., 2023).

The estimated annual per-patient costs for DMD in US in 2014 were $50,952. When considering the entire population, the national costs amounted to $787 million for DMD. Medical costs were predominantly drove by outpatient care, whereas nonmedical expenses were affected by the necessity to adapt housing for the patient and the expenses associated with caregiving. Income loss was significantly associated to the level of care demanded by the patient (Larkindale et al., 2014). A study from Germany findings revealed that the estimated annual burden of DMD, including direct medical & non-medical, informal care and indirect costs was collectively €78,913, while BMD's total costs were €39,060. The total mean annual direct medical costs were €19,346, and the direct non-medical costs were

€30,884. For BMD the total mean annual direct medical costs were €5,140, and the direct non-medical costs were €12,471.(Katz et al., 2014). The study conducted in the US assessed the direct costs associated with a rare disease, Duchenne Muscular Dystrophy (DMD), in a commercially insured population from 2000 to 2009. Comparing DMD patients (n = 75) to controls (n = 750) in a 1:10 ratio, matched by age, gender, and region, the results revealed a 10-fold increase in healthcare costs for DMD patients compared to controls ($23,005 vs. $2,277, P < 0.001). Particularly, costs were significantly higher for DMD patients aged 14-29 years ($40,132 vs. $2,746, P < 0.001), highlighting the substantial and age-dependent economic impact of DMD on healthcare resources in the United States (Thayer et al., 2017). In another study examining direct costs based on claims in 2018, per-patient cumulative costs from ages 5 to 25 were projected. The estimation considered a diagnosis at age 5, independent ambulation until age 11, and survival until age 25. The 20-year per-patient cumulative cost was $174,701 with prednisone ($2.3 million with deflazacort). Additionally, expected out-of-pocket expenses associated with events and medications were $12,643 ($29,194). The study suggested that standardized monitoring of disease progression and treatments may contribute to reducing the overall costs of illness (Conway et al., 2022). However, Ryder et al. (2019) conducted a systemic review of the burden, epidemiology, costs and treatment of DMD. The study was aimed to look much it costs to have DMD and how it affects a person's quality of life. The review concluded that the economic cost of DMD climbs dramatically with disease progression was rising as much as 5.-7 fold from the early ambulatory phase to the non-ambulatory phase (Ryder et al., 2017). The annual direct cost-of-illness per-patient for DMD in the year 2012 in international dollars varied across different countries: Germany: $42,360, Italy: $23,920, United Kingdom: $54,160 and United States: $54,270(Landfeldt et al., 2014).

#### 2.2.3.1 Out-of-Pocket (OOP) Expenditure

The out-of-pocket payments (OOPs) as “direct payments made by individuals to health care providers at the time-of-service use”. OOPs refer to direct payments made by individuals for healthcare services, excluding prepayments through taxes or insurance premiums (WHO, 2023). However, unregulated healthcare expenses pose a significant barrier

to essential medical care, leading to high out-of-pocket payments that risk financial security. In resource-limited settings where healthcare resources are scarce, patient fees become crucial for healthcare workers' income (Nundoochan et al., 2019). However, a challenge arises when these fees take precedence over providing necessary care, potentially hindering programs aimed at assisting those who cannot afford healthcare. This situation complicates efforts to ensure affordable care for all, emphasizing the importance of controlling healthcare costs (Mosadeghrad, 2014). Indicators like catastrophic health expenditure (CHE) and impoverishing health expenditure (IHE) gauge the impact of out-of-pocket payments on households' financial well-being, providing insights into a health system's financial protection performance (Prinja et al., 2019; Sriram & Albadrani, 2022).

Currently, research on rare diseases (RDs) tends to focus on assessing direct medical costs from the viewpoint of the healthcare system (Schulenburg & Frank, 2015; Teoh et al., 2016; Walker et al., 2017; Chiu et al., 2018; Opazo et al., 2021). Consequently, these estimates fail to capture out-of-pocket (OOP) expenditures, which represent additional healthcare costs that is not roofed by the health system. OOP expenses may lead patients to make choices between healthcare and other essential needs (Chung et al., 2023). The financial risk can be assessed using two indicators: household expenditure and out-of-pocket health-expenditure (Chauhan et al., 2018). The catastrophic health expenditure (CHE) is a certified measure for monitoring financial protection within the sustainable development goals. It identifies households where out-of-pocket health expenditure exceeds available resources (Prinja et al., 2019). CHE identifies the households that are drawn into poverty due to this massive out-of-pocket health expenditure. According to Chauhan et al. (2018) the cost of manage head and neck cancer in tertiary care center in Northern India. Using the COI approach, information on out-of-pocket (OOP) expenditure from patients. The average amount spent on different therapies range from ₹12,575 (USD206) to ₹65,257 (USD 1069). The study had limitations in assessing indirect costs such as lost income. It only accounted for the time spent away from work during hospital treatment. For chronic illnesses like cancer, where sickness duration is significant, lost wages could constitute a substantial part of overall expenses. Additionally, analysis did not include productivity loss from premature death, another aspect that could impact the overall economic assessment (Chauhan et al.,

2018). In a limited number of studies addressing financial challenges due to OOP health expenditure related to RDs, two indicators CHE and impoverishing health expenditure (IHE), were assessed. The CHE identified households with OOP health expenditure surpassing 10 percent, 25 percent, and 40 percentage of total household income, indicating "catastrophically" huge spending with respect to available resources (Wagstaff et al., 2018a). The IHE identified households that are pushed down below the poverty line due to OOP health spending, utilizing both the $1.9 and $3.1 per day poverty lines. These indicators helped assess the proportion of patients at risk of financial hardship within the cohort (Wagstaff et al., 2018b).

### 2.2.4 Indirect Cost or Caregiver Burden

The informal caregivers, comprising family, friends, and neighbors, provide uncompensated care, known as informal care. This involves various costs, including expenses for home adjustments and durable goods (Wimo et al., 2002). However, quantifying the value of time invested by informal caregivers is challenging, as it requires translating time into monetary terms for cost-effectiveness analysis. This valuation involves considering the time spent and its monetary value, encompassing paid work, unpaid work, and leisure time(Smith & Wright, 1994). In addition to direct expenses, patients and their families also face indirect costs, such as lost productivity due to work disruptions for both the patient and caregivers(Prinja et al., 2019). Indirect costs include productivity loss or productivity cost lost due to morbidity, mortality, impairment, job abstinence. It also includes forgone leisure time spent by families and visitors attending patients. Indirect costs form “a part of the social welfare losses due to diseases, while the remaining welfare losses are represented by the losses in healthy time resulting from pain, suffering and grief caused by diseases” (Rice et al., 1985).

A study from Hong Kong assessed the indirect cost associated with a patient's rare disease (RD) condition was assessed by considering labor productivity losses for both the patient and informal caregivers, as well as the consumption of healthcare services. The human capital approach, which evaluates time away from work according to wage levels, was utilized to gauge productivity losses. The resulting annual indirect cost was valued at $

103,535 per patient. However, caregivers' productivity losses contributing significantly at 67.7 percent. Among patients receiving informal care support, 68.9 percent of unpaid caregivers experienced employment impact (81.8 percent for pediatric patients, 59.4 percent for adults). The annual cost of forced un-employment or retirement and absenteeism in unpaid caregivers was estimated at $43,538 and $26,517 per patient, respectively. The employment was disrupted in 67.6 percentage of the 102 adult patients employed fulltime or parttime before the RD diagnosis, resulting in a annual total “productivity loss” of $ 46,550 per adult patient (Chung et al., 2023).

In 2012 per-patient annual informal care cost of illness for DMD exhibited notable variations among different countries. Specifically, Germany recorded the highest cost at $18,530, followed by the United Kingdom with $14,340, Italy at $13,160 and the United States at $13,370. These costs encompass the expenses associated with informal care provided by family members, friends, or neighbors. Additionally indirect cost of illness due to production loss, reflecting the economic impact of work disruptions, also differed across nations. Germany experienced the highest indirect cost at $20,770, followed by the United States with $21,550, the United Kingdom at $18,700 and Italy with $18,220. These findings highlight the economic burden of DMD, in view of both direct informal-care and the indirect costs were associated with productivity loss (Landfeldt et al., 2014). The parents of DMD patients tended to quit or reduce employment around stage II of the disease, while many BMD patients were in stage II/IV when parents stopped working. On average, parents missed 14.5 working days yearly due to their son's disease, with 60 percentage of DMD parents feeling career limitations and 49 percentage earning less due to their son's illness. Although gross salaries were similar in 2013 (DMD: €24,168; BMD: €27,414), the progression of the disease led to decreased earnings. Indirect costs, including absenteeism and reduced working time, were significantly higher for DMD (€7,220) compared to BMD (€2,527) in 2013. Moreover, over half of DMD parents and 23 percentage of BMD parents developed medical problems due to their son's disease, leading to increased medical treatment consumption. The study emphasizes how the physical and mental well-being of parents correlates with the severity of their son's impairment, impacting their caregiving, work, and incurring indirect costs (Katz et al., 2014).

Landfeldt et al. (2017) estimated the mortality cost of the DMD. The study had the objective of estimating the mortality cost of DMD. Each patient who would have remained alive was allocated a lost life-year and subsequently mean total number of life-years lost due to DMD was calculated, specific to each country. To calculate the mean mortality cost the estimated mean number of life-years lost was multiplied by a societal WTP for a life-year. Across the studied countries, estimated mean total national annual mortality cost of DMD ranged from €248 million to €1208 million, constituting between 50 percent and 62 percent of our previous estimates of the total burden of illness. The cost associated with excess mortality was found to be the largest cost component of the disease burden, followed by costs associated with impaired quality of life. The present study or related studies may contribute to the acceleration of payer negotiations, such as the pricing and reimbursement of new therapies(Landfeldt et al., 2017)

Information regarding the economic burden of disease is very important for the best use of health care resources especially in limited-resource settings. Chronic diseases places a significant economic burden, particularly on patients in low and lower-middle income countries. There is a significant opportunity to contribute methodologically rigorous research to enhance the accuracy of cost estimation and enable better comparisons between studies (Afroz et al., 2018).

The financial impacts of RDs needs a comprehensive evaluation is crucial as RDs are often chronically devastating, necessitating care for life-time from formal or informal caregivers. Evaluating socio-economic costs from multiple perspectives (different societal levels ) provides a holistic understanding of RD impacts on various stakeholders and financial hardships (Pérez et al., 2021). Currently, economic cost of DMD is not known in LMIC. However, several studies have evaluated the economic burden in European or Western settings. These studies have shown the severe financial impact of disease over society. The micro-costing method was used to examine the economic burden of Duchenne and Becker's Muscular dystrophy (Katz et al., 2014).

## 2.3 Quality of Life

Traditionally the impact of health care has been measured in terms of its effects on mortality, since health is much more than merely being alive, its effect on morbidity is increasingly being considered(Braveman & Gottlieb, 2014). Quality of Life (QoL) is a measure of an individual's overall well-being, encompassing physical, emotional and social aspects and is influenced by their perception of various important life factors, whether health-related or social (Tully et al., 2019).

The two-basic burden related to DMD are emotional and economical apart from the health-related events directly effects the quality of life(Uttley et al., 2018). Despite parents highlighting the significance of quality-of-life challenges for children with DMD, there's limited information on their actual QoL (Webb, 2005). The progressive decline in strength and loss of the ability to engage in activities with peers can result in social exclusion and isolation, potentially shaping how boys with DMD perceive their QoL (Bothwell et al., 2002).

Psychological challenges affecting family members of individuals with DMD include the unpredictable progression of the disease, societal stigma, maternal guilt regarding genetic transmission and intrusion by extended family (Kerr & Haas, 2014). Brothers may experience jealousy due to the attention given to the affected sibling, while sisters might adopt an overprotective maternal attitude. These factors contribute to the complex emotional dynamics within the family (Buchanan et al., 1979). Therefore, it is crucial to assess the stress levels and coping abilities of family members, as these factors can directly influence the quality of life for both the family and the patient (Read et al., 2011).

### 2.3.1 Health-Related Quality of Life in Patients with DMD

In lieu of no cure for DMD, clinical management focuses on symptom treatment, slowing progression and optimizing patients' Health-Related Quality of Life (HRQoL) (Powell & Carlton). HRQoL is defined as the health-related aspects of quality of life, capturing the influence of the disease and its treatment on disability, daily functioning and the individual's ability to lead a fulfilling life based on perceived health (Powell et al., 2020). In

the literature, the terms QoL and HRQoL are often used interchangeably, despite having distinct definitions. The World Health Organization Quality of Life Group defines QoL as 'individuals' perception of their position in life, considering cultural and value systems and in relation to their goals, expectations, standards and concerns. ("The World Health Organization Quality of Life Assessment (WHOQOL) 1995). HRQoL, on the other hand, narrows this concept, concentrating specifically on how illness and treatments affect a person's life (Civita et al., 2005). HRQoL excludes dimensions of life, like environmental quality and political stability, that are beyond the influence of healthcare interventions (Guyatt et al., 1993). It specifically focuses on aspects directly impacted by health and medical treatments.

The impact of the disease on quality of life for both patients and caregivers was evaluated using a range of generic and disease-specific measures. The Pediatric Quality of Life Inventory (PedsQL) was the most commonly used tool, featured in five studies. Other instruments included SF-36, WHOQOL-BREF, Life Satisfaction Index for Adolescents (LSI-A), Hospital Anxiety and Depression Scale (HADS) (0-21), Children Health Questionnaire (CHQ), KIDSCREEN-52 and Fatigue Severity Score (0-5) (Ryder et al., 2017).

In a study encompassing patient-caregiver pairs from Germany, Italy, UK, and the USA, HRQOL was evaluated using the Health Utilities Index questionnaire (HUI) and the Pediatric Quality of Life Inventory (PedsQL). Despite the majority of caregivers (>84 percent) perceiving their patients as being in good health, irrespective of ambulatory class, the mean patient utility significantly decreased from 0.75 in early ambulatory males to 0.15 in the most severely affected patients. Similarly, patient PedsQL scores (0-100) declined from 80 to 57 across different ambulatory classes. This indicates a marked reduction in HRQOL in DMD patients compared to the general population, with significant associations observed with disease progression (Landfeldt et al., 2016). In comparison to healthy children, both the physical and psychosocial QoL for boys with DMD are notably lower. According to self-reported psychosocial QoL, it tends to be higher in older boys (13–18 years) than in younger boys (8–12 years) and is not associated with the use of mobility aids (Uzark et al., 2012). The agreement between parents and children in reporting (QoL) was generally fair to poor. This

suggests that using proxy reports from parents may not accurately reflect the child's own perception and experience of QoL (Wei et al., 2017).

The PedsQL correlates with the level of impairment at baseline, but not changes over 12 months. Additional studies comparing different assessment tools are necessary to gain a better understanding of the intricate relationship between HRQOL and functional performances (Messina et al., 2016). In DMD patients under non-invasive ventilation (NIV), both respiratory impairment and sleep quality independently predicted poor QoL. The significance of sleep quality in DMD is sometimes overlooked, but it should be carefully addressed to enhance overall QoL (Crescimanno et al., 2019). Additionally, longitudinal studies have an advantage of monitoring changes in HRQOL over time can be done, particularly in the context of a progressive disease (Wei et al., 2016). The previous literature regarding the QoL in DMD patients has remained inconsistent, very few studies reported reduced QoL while others concluded no difference between the QoL of children with DMD and healthy children (Wei et al., 2017). The study conducted by Malcolm Kohler et al. (2005) concluded that QoL in DMD children is not correlated with the amount of disability they are having neither with requirement of any kind of intervention. This is quite contradictory that children with severe disabilities had a very good quality of life (Kohler et al., 2005).

The EuroQOL five dimensions questionnaire (EQ-5D) is a widely used tool for assessing HRQOL by measuring various aspects of well-being, including physical, emotional and social functioning (Gusi et al., 2010). This tool is holistic and utility score, expressing Quality Adjusted Life Years (QALY), often used in cost-effectiveness analyses for evidence-based decision-making. Consequently, EQ-5D serves as a valuable tool for health outcomes studies and economic analyses(Yang et al., 2019). The HRQOL for adult patients from Hungary and France was evaluated using country-specific tariffs to calculate EQ-5D utility scores (Carr-Hill, 1992). The assessment revealed an EQ-5D Visual Analog Scale (VAS) score of 50.5 and an EQ-5D index score of 0.24 for these patients. These scores provide insights into the perceived health status and overall well-being of adult patients in Hungary and France, offering a quantitative measure of their qualityof life based on the EQ-5D instrument (Bastida et al., 2017). Use EQ-5D to assess health status in Duchenne Muscular Dystrophy is suitable for wholistic measure of QoL and more comparable across different

diseases. While other measures may be more tailored to specific DMD outcomes, the results from EQ-5D have value that aids in deriving a utility index, contributing to QALY calculations and facilitates cost-effective analysis detection (Crossnohere et al., 2021)

### 2.3.2 QoL in Caregivers with Patients with DMD (Caregiver Burden of DMD)

The caregiver burden is defined as the extent to which caregivers perceive that providing care has negatively affected their emotional, social, financial, physical and spiritual well-being (Fau et al., 1986). The measurement of QoL may vary based on the perspective. The parent proxy-reports become important when a child is too young or unable to self-report QoL. However, differences between self and proxy reporting are recognized across various health conditions, including DMD (Powell et al., 2020). The parents of children with disabilities often report lower HRQoL for their children compared to the children's self-reported assessment (Capitello et al., 2016). The carer's perspectives both for their child and their own QoL, are crucial for gaining a comprehensive understanding of the underlying themes related to QoL (Marques et al., 2013). The previous studies to explore the QoL of caregivers of individuals with DMD. have categorized identified themes of QoL into four domains: i. physical; ii. psychological; iii. social; and iv. well-being. The findings highlights the need for considering the broader impact on the QoL of both caregivers and the wider family of individuals with DMD (Uttley et al., 2018). A study indicates that adults with DMD in Western Europe exhibit higher HRQoL scores compared to those in Eastern European countries (Steffensen et al., 2015). The provision of formal care to patients and their families differs across countries (Cavazza et al.). Notably, a study conducted in India observed that participating caregivers were predominantly male, potentially influenced by cultural disparities in research participation (Thomas et al., 2014).

There is heterogeneity in the QoL of caregivers and it is not consistent across various geographical regions. However, positive associations were identified with patient age, ambulatory status and/or ventilatory support, impacting informal care hours, physical and cognitive challenges, household cost burden with caregivers burden( Katz et al., 2014; Landfeldt et al., 2016). While Baiardini et al (2011) found that family strain was not influenced by disease progression (Baiardini et al., 2011). However, a strong association

between anxiety and depression in caregivers with patient's health and mental status, but not with ambulatory status (Landfeldt et al., 2016). According to Reid (2001) discovered that family stress correlated with psychosocial adjustment and intellectual function of the patient, rather than socio-demographic variables like age, education and wheelchair use (Reid & Renwick, 2001).

**Table: 2.3.1 QoL of DMD patients and their caregivers, Reviewed Studies Globally**

| Author | Publication Year | Tool Used | Study Area | Main Finding(s) |
|---|---|---|---|---|
| Kohler et al. | 2005 | Short-Form 36 (0-100) | Zurich, Switzerland | Non-ventilated patients and Ventilated patients<br>Short-Form 36 physical function scores were massively reduced in both groups (1 ± 2, and 0 ± 0, respectively)<br>Physical component summary (35 ± 6 and 31 ± 6) |
| Bendixen et al. | 2012 | PedsQL 3.0<br>CAPE | Florida, US | Perceived QoL was markedly diminished in children with DMD relative to unaffected controls, except in the emotional domain. |
| Katz et al. | 2014 | PedsQL | Germany | PedsQL: DMD: 56; BMD: 76<br>caregivers, 29% stopped working, and 38% reduced their working hours to care for their sons. 58% diagnosis of sleep problems, 88% experienced back pain, depressed: 32% :,faced anxiety attacks: 31%. Informal care: 9 hrs/day. |
| Pangalila et al. | 2015 | Short-Form 36 (0-100) | Spierziekten Nederland | Fatigue impacts overall qualityof life and physical health/environment, while anxiety is linked to the psychological domain of "quality of life". |
| Moura et al. | 2015 | WHOQOL-BREF and the ZBI (Zarit Caregiver Burden Interview) | Brazil | Mean WHOQOL score: 14.0<br>Mean ZBI score: 26.<br>Caregiver burden was positively associated with patient age. |

| | | | | |
|---|---|---|---|---|
| Landfeldt et al. | 2016 | The EQ-5D-3L; VAS; the SF-12<br>Health Survey and the ZB | Germany, Italy, UK & USA | Mean EQ-5D-3L utility 0.81, mean VAS score 0.74,<br>mean SF-12 Mental Health Component Summary Score 44 and mean ZBI score 29. Half of all caregivers reported being moderately or extremely anxious or depressed |
| Lue et al. | 2016 | HrQoL: Short Form 36 (0-100)<br>Global QoL: WHOQOL-BREF | Taiwan | Adolescents with DMD experience poor QoL and HRQoL, with notable impact on social activities. Functional performance is correlated with QoL in this context. |
| Landfeldt et, al. | 2016 | Health Utilities Index Questionnaire (HUI) Score ( 0-1)<br>Pediatric QualityofLife Inventory (PedsQL) Score 0-100 | Germany, Italy, UK & USA | HUI: 0.75 in early ambulatory males to 0.15 in the most severely affected<br>PedsQL: 80 ambulatory<br>57 most severely affected |
| Messina et al. | 2016 | PedsQL™ Neuromuscular Module (NMM) | Italy | PedsQL (patient baseline Vs 12 month): 81.4 (±12.8) Vs 81.2 (±13.3) (p=0.77)<br>PedsQL (Parents Proxy baseline Vs 12 month): 75.8 (±15.6) Vs 71.3 (±16.6) (p=0.00) |
| Cavazza et al. | 2016 | EQ-5D-3L<br>ZBI | Bulgaria, France, Germany, Hungary, Italy, Sweden & UK | Mean EQ-5D-3L utility; mean VAS score, mean ZBI score, mean hours of informal care/weekly:<br>Bulgaria: 0.51, 0.56, not reported, and 74;<br>France: 0.66, 0.60, 15 and 65;<br>Germany: 0.69, 0.69, 35 and 45;<br>Hungary: 0.69, 0.78, not reported and 55<br>Italy: 0.78, 0.78, 25 and 59<br>Sweden 0.61, 0.68, 33 and 62 |

| | | | | |
|---|---|---|---|---|
| | | | | UK: 0.72, 0.82, 31 |
| Wei et al | 2017 | PedsQL V.0 4<br>Quality of My Life (QoML)<br>(Global health) | Canada | PedsQL (Patient: 58.3; Parent Proxy: 51.9)<br>QoML (Patient: 78.7; Parent Proxy: 70.0)<br>Quality of Life (QoL) and Health-Related "Quality of Life" (HRQoL) are connected yet distinct concepts, as reported by both children with Duchenne Muscular Dystrophy (DMD) and their parents. |
| Bastida et al. | 2017 | EuroQol 5-domain (EQ-5D) | Spain | EQ-5D VAS score and EQ-5D index scores for patient 50.5 and 0.24 and EQ-5D VAS and EQ-5D index scores for caregivers were 74.7 and 0.71 |
| Crossnohere et al. | 2021 | EuroQol 5-domain (EQ-5D) | USA | EQ-5D index was higher in ambulatory than non-ambulatory patients (0.60 v. 0.30) |

## 2.4 Challenges in the Management of Rare or Orphan Diseases

Rare diseases (RDs) pose a global health-care challenge, impacting an estimated 400 to 700 million individuals worldwide (Rodwell & Aymé, 2015). Thus rare diseases are not that rare with over 10,000 recognized rare diseases, they are typically severe, limiting QoL, and potentially life-threatening (Stoller, 2018; Wakap et al., 2020). It is important to understand the challenges that limits management of disease, so as to develop a coping mechanism adressing those challenges.

### 2.4.1 Challenges in Service Utilization for Rare diseases (Duchenne Muscular Dystrophy

1. **Ignorance**

a) **Limited expertise:** Rare diseases often require specialized knowledge and expertise for diagnosis (genetic testing and advanced diagnostic tools) and treatment (Adachi et al., 2023). Public healthcare facilities in India significantly lacks the specialized medical professionals or resources needed to effectively manage rare diseases (Ferreira et al., 2023).

b) **Lack of awareness:** Many healthcare providers may not be familiar with them. This lack of awareness can lead to misdiagnoses or delayed diagnoses (Stoller, 2018).

2. **High treatment cost:** Healthcare is too costly, limited or even unavailable and often lack well-established treatments or medications due to their rarity and the limited research and development efforts focused on them (Pérez et al., 2021).
3. **Unmet medical needs:** Major definitive treatment available for rare diseases, healthcare utilization may be primarily focused on diagnostic services and even these services can be challenging and time-consuming for patients and their families (Schulenburg & Frank, 2015). There is demand for increased efforts in early disease diagnosis, including awareness campaigns, screening programs and education for healthcare providers at the community level to identify rare diseases as early as possible (Valdez et al., 2016).
4. **Genetic/ runs in families:** People conceal the disease as it is often associated with disabilities and life threatening (Liang et al., 2023). This stigma leads to major barrier in the prenatal diagnosis and prevention of spread disease (Zhu et al., 2017).
5. **Difficult to follow-up:** Limited accessibility to diagnosis and treatment often leads to loss to follow-up (Esquivel-Sada & Nguyen, 2018).

6. **Multidisciplinary (co-ordination difficulties among different disciplines):** Most of the rare diseases are chronic, lifelong and usually affecting multiple organ-systems. The management approach is multidisciplinary and long-term. Comprehensive approach to rare disease management that includes not only diagnostic services but also specialized care, rehabilitation and long-term support for patients (Bushby et al., 2010). A model of care that tailors healthcare services to the individual needs of patients with chronic diseases, emphasizing better outcomes and improved "quality of life" (Wigert & Wikström, 2014). Patients and their families with rare diseases often have a strong desire for treatments that can improve their quality oflife and potentially extend lifespan. While definitive cures may not always be available, various supportive and medical interventions can be crucial in managing rare diseases effectively (Sirari et al., 2023).

### 2.4.2 Strategies to Manage the Challenges

a. **Geographical Accessibility:** Improved geographical access to healthcare services in remote areas is crucial for rare disease patients, reducing loss to follow-up and promoting timely, equitable care (Gilkey et al., 2022). This ensures that individuals in underserved regions receive necessary healthcare interventions, ultimately improving their overall health outcomes ( Valdez et al., 2016; Kalita et al., 2023). Despite geographical challenges, addressing rare diseases in underserved areas remains essential due to the nation's limited specialists in this field (Ferreira et al., 2023). Efforts to expand healthcare access and specialist training are crucial for better rare disease management (Kalita et al., 2023).
b. **Teleconsultation services:** Incorporation of teleconsultation into the healthcare system, making quality healthcare services more accessible, especially for patients facing geographical barriers ( Valdez et al., 2016; Gilkey et al., 2022).
c. **Research Funds:** Given the absence of definitive treatments for many rare diseases, there's a critical demand for increased research funding. These funding are essential to gain a better understanding of the prevalence and characteristics of these conditions, paving the way for improved treatments and management approaches (Stoller, 2018).
d. **Capacity Building:** Demand for educational outreach sessions aimed at building the capacity of peripheral healthcare services, including healthcare providers and community healthcare workers, to enhance their ability in detecting and managing

the rare diseases effectively (Opazo et al., 2021). Patients and their families may find it challenging to access appropriate care and if any treatment or management options are available, they may indeed be more likely to be found within the public healthcare system or through specialized government programs or initiatives. This deviation from the typical healthcare utilization pattern, where private healthcare is often preferred for curative care, underscores the unique challenges and circumstances associated with rare diseases in India and globally.

In India "Ministry of Health and Family Welfare" also felt the need to address the issues related to rare diseases, which was reflected in the National Health Policy 2017 of India. Government of India approved a separate "National Policy for Rare Diseases (NPRD) 2021,". The policy's fifth key principle is Patient Centered Care (PCC) and QoL**.** (*The National Health Policy-2017*, 2017). Long-term follow-up is crucial for chronic rare diseases, but data on treatment outcomes are often lacking. High treatment costs contribute to economic strain, especially in resource-constrained settings like India. The condition DMD is classified as Group 3 diseases in NPRD 21. The "Indian Council of Medical Research" (ICMR) introduced a national registry for RDs involving centers across the country. Centres of Excellence (COEs) are designated tertiary hospitals for rare disease diagnosis, prevention and treatment. COEs develop protocols for early diagnosis, care coordination and improving the quality of life. Tertiary prevention focuses on providing better care and rehabilitation for patients with advanced rare diseases. These centers are also engage in education, training, screening, diagnostics, research and treatment. The policy recommends a focus on indigenous research and producing medicines at local level, research work related to low-cost treatment for rare diseases, screening and early detection for rare diseases (*National Policy for Rare Diseases*, 2021).

The GOI is offering financial aid through the Rastriya Arogya Nidhi scheme. Under this, up to Rs. 20 lakhs of support will be provided by the Central Government for one-time treatment of rare diseases listed in Group 1. This assistance is not confined to Below Poverty Line (BPL) families; around 40 percent of the population eligible under the Pradhan Mantri Jan Arogya Yojana norms can benefit from this aid. The treatment will take place exclusively in government tertiary hospitals (Access health international, 2016).

## 2.5 Patient Empowerment

The entire world is witnessing a new paradigm shift to resolve any problem from mechanistic or Newtonian worldview to holistic view that includes human values and creativity. However, the reductionistic approach which isolates health issues, still dominates the scientific fraternity (Morin, 1992). An inclusive paradigm consists of two key elements: patient self-empowerment and holism. Self-empowerment involves developing skills, having control over resources, making autonomous decisions and taking ownership of one's own health (Rothman & Wagner, 2003; Wasilewska et al., 2020).

### 2.5.1 Patient or Consumer Engagement

Patient engagement in healthcare is seen as a game-changing "blockbuster drug" of this century, capable of achieving the "triple aim" of better health outcomes, improved patient care, and reduced costs (Clancy, 2011). The concept of patient engagement is borrowed from marketing, where customers are the center of attention and their satisfaction and engagement are crucial (Graffigna et al., 2014).

Creating personalized interventions is crucial to engage patients continuously in managing their own health (Coulter, 2012). Patients and caregivers should have access to information about their condition, treatment choices, healthcare resources, support networks and relevant policies. This knowledge empowers individuals to make informed health decisions and play an active role in their well-being (Gruman et al., 2010). Using healthcare models that enhance this empowerment can make care more cost-effective, especially for those with long-term conditions. This approach is highly needed in today's healthcare landscape (Chen et al., 2016). Patient engagement can be achieved via patient-centered care model (Tosto et al., 2023).

### 2.5.2 Patient-Centred Care (PCC)

As the name suggests patient are at the center of the PCC frame work (Wigert & Wikstrom, 2014). Patient-centred care increases adherence to management protocols and thereby reducing morbidity and improving QoL for patients (Navon et al., 2006). The outcome can only be optimized when patients are actively participating in their management and taking responsibility for their own health (Bauman et al., 2003). The

PCC practices are linked to good communication, shared decision-making and patient education. Healthcare providers acknowledge PCC's significance, especially for long-term illnesses (Robinson et al., 2008). The three simple routine steps in practicing PCC model are: Firstly, establishing partnership with patients; secondly shared decision making with the patients and lastly documentation. Documentation not only gives information but also helps in keeping transparency and continuity between the patient and health care provider (Ekman et al., 2011).

A qualitative study explored that committed senior leadership, clear communication between provider and the patient, active engagement of patients and families, their feedbacks and capacity building of staff were the key facilitators in delivering patient-centered care (Luxford et al., 2011). The major bottleneck in delivering PCC is the organizational cultural transmission from 'provider-focus' to 'patient focus'(Bertakis & Azari, 2011b). To have an efficient healthcare delivery system, it's important to explore strategies e.g PCC that can enhance capacity and effectively achieve these objectives in various cultural and economic settings (Berwick et al., 2008). A randomized controlled trial was conducted to assess the efficiency of patient-centered care in primary healthcare usage and costs. In the study randomly one group received patient-centered care for less than a year and the other for more than a year. The group with less patient-centered care had annual charges of $1,435, while the longer-duration group had charges of $948, which was 51.37 percent less. Patient-centered care was associated with lower healthcare service use and reduced annual charges (Bertakis & Azari, 2011a, 2011b).

Planning the health care delivery system via PCC model involve: carefully listening each child and his/her family, honoring ethnicity, cultural socio-economic status and their experiences. Ensuring flexibility in policies, measures and provider's way conducting work, to form tailor-made services can be provided as per the demand, cultural and beliefs individually (Chen et al., 2016). Sharing of information that is comprehensive and unbiased with the patients and their families encourage their participation in making an informed decision (Bauman et al., 2003). Providing formal or informal peer-to-peer support for the child /family, identifying and establishing strengths of individual children and their families is the first step towards PCC. Later empowering them to realize their own strengths, shaping the confidence and their participation in choosing and deciding about their own health care plan (Karazivan et al., 2015). The care

is a teamwork approach where families and healthcare workers work together. This approach leads to improved health status and more efficient care, with fewer tests and referrals (McCormack et al., 2011). While the principles of Patient-Centered Care (PCC) exist in medical literature, there's limited literature on how to put these principles into practice (Kumar & Singh, 2017). A patient-centered care hospital's infrastructure encourages family collaboration through a home-like environment that not only meets the needs of the patient, but also meets the needs of family members (Chen et al., 2016).

PCC model definitely improves the health outcomes, still it is important to evaluate how much it costs to the system. In a randomized controlled trial, the cost-effectiveness of routine outpatient department (OPD) care versus an online model for follow-up treatment of patients with psoriasis over 24 weeks was evaluated. While no significant difference was observed in mean change in Dermatology Life Quality Index (DLQI) scores or mean improvement in quality-adjusted life expectancy between the two groups, the cost of follow-up care was 1.7 times lower for patients in the intervention group. Therefore, patient-centered care was found to be cost-saving while maintaining similar effectiveness to standard care. (Parsi et al.,2012). Considering the enhancement in Quality of life at reduced costs is the main stay of health care delivery system presently (Izard et al., 2014). This needs to include efforts to enhance consumer knowledge, skills, or power to become partners in their care ironically these are the major bottle necks. The education and support benefits have been achieved in a variety of ways, ranging from pamphlets to one-on-one teaching sessions, and involving social support groups (Luxford et al., 2011). In the realm of the internet, assessing the capacity of computer systems to provide patients with information, decision support, and connections to experts and other patients is crucial. (Gómez et al., 2016). A study was conducted to assess the improvement in quality of life, reduce health-risk behaviors and more efficient use of medical services when given home access on computer among HIV-positive patients. This study was randomized control trial and results shown better quality of life in users and reported to spend less time in abulatory visits making more phone calls to providers and experiencing fewer and shorter hospitalizations (Gustafson et al., 1999). These outcome of PCC underscores that policymakers must turn their attention to such areas and beyond. Health information technology (HIT) to formulate a coordinated and earnest national health policy in support of PCC. These particular health policies will help health professionals in acquiring and maintaining skills related to PCC. The policy must also

encourage administration to promote a principles of patient-centeredness (Epstein et al., 2010).

### 2.6 Research Gap

Over the years, our understanding of Duchenne Muscular Dystrophy (DMD) has significantly improved, leading to earlier detection, timely treatment, and longer life expectancy for patients. However, as DMD is a rare and lifelong illness, ensuring patients have easy access to necessary services remains a challenge. With advancements in diagnostic methods and broader medical care coverage, the burden of this disease is expected to rise, placing a significant economic strain on families and the healthcare system. While studies like Landfeldt et al. (2014) have assessed the overall cost of illness (COI) in DMD across developed countries with well-established patient registries, similar comprehensive economic assessments are lacking in developing nations. This gap in research limits our understanding of the economic impact in resource-constrained settings, where healthcare systems face unique challenges. Conducting such studies is crucial for informing health policies, designing intervention programs, and developing targeted financial support initiatives tailored to the needs of DMD patients in diverse contexts.

Ministry of Health and Family Welfare, Government of India also felt the need to address the issues related to rare diseases, which was reflected in the National Health Policy 2017. As stated in policy field of rare diseases is complex and heterogeneous. Research on rare diseases is challenging due to their limited patient pool, making clinical understanding incomplete. Long-term follow-up is vital, but data on treatment outcomes are scarce. Available drugs are often expensive, straining resources. In India, only a fraction of potential rare diseases have been identified, mainly involving disorders like muscular dystrophies and spinal muscular atrophy. These diseases place a significant economic burden, especially in resource-limited settings. The policy emphasizes the need for research in this area. The lack of available literature is so insignificant that there hasn't been any economic study conducted on DMD or any rare muscular disease in our country. This highlights the urgency of exploring these areas to better understand and address the challenges posed by these conditions, shared and used.

In the past decade, cost information on health from India has become more available. The studies conducted across different Indian states have generated data on healthcare costs in public (including primary, secondary, and tertiary levels) and private

hospitals. A consistent finding in these studies is the significant variation in healthcare costs within the same sectors and levels of healthcare facilities, both within states and across states in India. Even the out-of-pocket expenditure estimated in a large sample size like National Sample Survey Office (NSSO) variability is much. This cost variation is significant for policy-making and robust research. The cost heterogeneity, due to different payment rates holds significance for both policy-making and conducting robust research. This underlines the need for DMD research on economic burden (family perspective) in public tertiary care settings.

The National Health Policy 2017 emphasizes patient-centered care as the primary approach for managing chronic long-term diseases, aiming to enhance the quality of life. In line with the "Prevention & Control of Rare Diseases" policy of 2021, centers of excellence were tasked with creating standard operating protocols. These protocols are expected to be designed in a manner to improve early diagnosis, enhance care coordination, and ultimately enhance the quality of life for patients with rare diseases across different levels of healthcare. Regarding tertiary prevention, it focuses improved care and rehab for advanced-stage rare disease patients. It involves offering top support to those with various rare disorders, even those with no specific treatment. This enhances the “quality of life” for patients and families. It covers developmental assessment, early stimulation, therapy, aids, and vital emotional and psychological support. The current approach in the management of disease is co-centric to center of excellence areas only where the approach is doctor-driven (top-down approach). There is no published evidence that reflects how PCC model (bottom-up approach) works on reducing financial impact does enhance the QoL. The gap lies in the lack of research on how the **Patient-Centered Care (PCC) model**, which adopts a bottom-up approach, impacts the quality of life (QoL) and financial burden for patients with rare diseases. While the current management predominantly relies on a doctor-driven, top-down approach within centers of excellence, there is no published evidence exploring the effectiveness of PCC in addressing these critical aspects, highlighting the need for studies in this area.

When investigating the QoL in progressive diseases, using cross-sectional data and analyzing each stage (stratification) separately can introduce additional variability into the data. To gain a more comprehensive understanding of how QoL changes with disease progression, it's advantageous to collect longitudinal data. This involves tracking the same individuals over time. By doing so, it can be observed how QoL evolves as the

disease advances. Thus, longitudinal studies are required to address the dynamics of QoL in progressive diseases, as it captures individual changes and trends over time and reduces the variations caused by looking at different stages separately. Most studies investigating QoL in progressive diseases rely on cross-sectional designs, which analyze different disease stages separately and may introduce variability. Our study addresses this gap by adopting a longitudinal approach, tracking the same individuals over time to capture dynamic changes in QoL as the disease progresses and provide a more comprehensive understanding.

On the basis of above facts, it was observed that, both aspects of public health are currently unexplored for DMD in India. This study is intended to explore the QoL of both the patients and caregivers using a Patient-Centered Care (PCC) approach. The aim is to reduce the financial strain on families by following them over time. The goal is to develop a workable and realistic model of individualized care centered around patients, which can be replicated for DMD patients in India. The objective of this study was to assess the financial strain experienced by families of boys with DMD who receive care at a government hospital. The subsequent aim was to create a Patient-Centered Care model (intervention) to empower affected families in handling the disease and thereby lessening the financial burden. Finally, the impact of the intervention was compared to routine care, appraising the changes in QOL for both caregivers and patients.

# CHAPTER III

# RESEARCH METHODOLOGY

A comprehensive understanding of the research methods and data analysis is a prerequisite for research design. This chapter explains the research methodology used to provide a detailed description of study population, sample design, sample size and statistical tools for data analysis. The present chapter is divided into three sections. Section I of this chapter highlights research design and provides detailed information about research topic, objectives, hypothesis, sample design and sample size estimation. Section II is about the intervention program developed during the study period its components and its administration. Section III the discusses research instrument, validity and reliability. Section IV provides a detailed description of statistical tools for data analysis. However, limitations of the present study have been mentioned in section V.

## SECTION I

## 3.1 RESEARCH DESIGN

The current study aimed to assess the economic burden beared by the families of boys with DMD seeking care at a government hospital. Subsequently, it sought to develop a Patient-Centered Care model (intervention) for building the capacity of the affected families to cope with the disease thus reducing the financial burden on families. Then, it finally aimed to compare the impact of intervention with routine care on Quality of Life (QOL) (caregiver and patient), economic burden and cost-effectiveness after 6 months of intervention. The study aimed to develop and evaluate the impact of a multi-pronged intervention package for children having Duchenne Muscular and Becker's Muscular Dystrophies

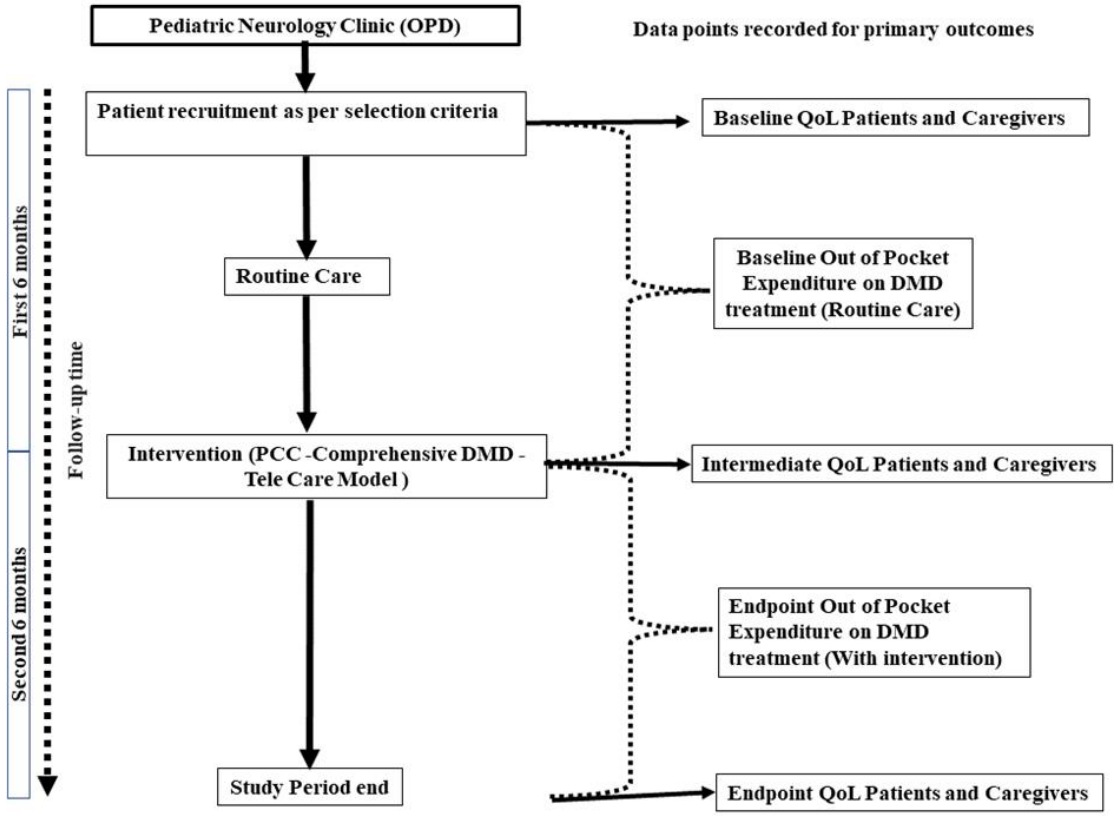
### 3.1.1 Study Design

It was a prospective non-randomized intervention clinical trial. The study design was quasi-experimental and conducted in two phases longitudinally. This is important to note that within the scope of this study, it was difficult to achieve a controlled experimental design. There was no randomization, and the same group of subjects were compared before and after the intervention. The experimental condition could be systematically different across the time frame. Considering this limitation quasi-experimental design was chosen for the study that was conducted in natural setting (Frederickson, 1990).

**Phase I:** Phase one was observational part of the study. The participants (Both old and new OPD patients) were enrolled prospectively from the pediatric Neurology follow-up clinic, Advanced Pediatric Center, Postgraduate Institute of Medical education and Research (PGIMER), Chandigarh. Patients were interviewed at the time of recruitment after receiving their consent to participate in the study for demographic details, socio-economic status, clinical information, information on financial burden. The state of their (patient and caregiver) quality of life was also assessed using the previously validated questionnaires discussed in detail in section II of this chapter. After recruitment in the study, patients were followed for the next 6 months. The routine treatment was given during this period and in routine patients visit the health facility at every 3 months (minimum three visits in 6 month). Patient's families were contacted telephonically or through WhatsApp and other electronic means of communication for the data collection on the economic burden. Initially, it was planned monthly telephonically but, with the use of WhatsApp it became real-time contact with the patients. It helped in reducing the recall bias. After the completion of 6 months patient's parents were contacted and were reminded to visit the health facility for their routine follow-up visit. During this visit, patients were enrolled into the intervention phase (Phase 2- Intervention Phase).

During phase 1, intervention care (Patient-Centered Care) model was developed. The rationale for developing the care model is to provide basic information about disease and treatment options to the patient and their parents/caregivers. Thus, they could participate effectively in the process of obtaining comprehensive care, in a resource-limited setting. The intervention involved, information about the disease, an instructional booklet, a workbook for compliance, and teleconsultation with a neurologist. The rehabilitation team was also a part of this study. The sessions were designed to facilitate caregivers in decision-making by active participation. The set of interventions are collectively named as "Comprehensive DMD-Telecare Model.

**Figure: 3.1.1 Quasi- Experimental Design: Non-Randomized Observational Clinical Trial Study Design**




**Phase 2** was the interventional part of the study, which starts once patient has completed phase 1. During this period, patients were given a set of interventions that were developed during the Phase 1. After a systematic review of the literature and clinical experience, the intervention was designed. Intervention is discussed under the section "intervention development." The patients were followed prospectively for the next 6 months (figure: 3.1.1). Initially, it was planned monthly telephonically; but, with the use of WhatsApp it became real-time contact with the patients. It helped in reducing the recall bias. The details of the follow-up schedule during these phases are given in tables 3.3.1 and 3.3.2.

### 3.1.2 Sample Design

The patients were enrolled prospectively in a consecutive manner to a non-randomized intervention clinical trial study. Enrollment was from the Pediatric Neurology Clinic of Advanced Pediatric Center PGIMER Chandigarh. This institute caters to approximately seven north Indian states which include Haryana, Punjab, Jammu and

Kashmir, Himachal Pradesh, Uttrakhand, the north-western part of Uttar Pradesh, Bihar and Chandigarh.

DMD is a rare disease with paucity in resource-limited settings. The sample selection in the trial/study in a manner similar to epidemiological studies is not possible. For such conditions, usually, convenience sampling is taken. The used method is non-probability hospital-based sampling. After discussing the difficulties with experts in this field and thus the sample size was calculated. The primary outcome measure is the change in Heath related Quality of life (HrQoL) scores of patients. The change in QOL was measured in the observational phase 0-6 months and compared with the change in QOL in the intervention phase 6-12 months. A clinically meaningful difference based on MID (Minimum Important Difference) from HrQoL was taken as 0.1 (Jayadevappa, et al., 2017), taking the standard deviation SD as 0.2 (Bastida et al., 2017), and an allocation ratio of 1:1, the estimated sample size of the was 63 patient in each phase to achieve a level of significance of 95 percent confidence interval (CI) at the power of 80 percent. Accounting for a 10 percent drop-out rate, the total sample size was 70 patients in each phase.

### 3.1.3 Selection Process

During the course of enrollment observing the increased attrition rate as anticipated during sample size calculation and available enrollment time, more participants than the required sample size of the project were enrolled. This discrepancy between observed and anticipated attrition was due to two different time zones of sample size calculation (pre COVID era) and sample collection (post COVID era). The flow of patient numbers and post of the event in the study is depicted in methodology as per STROB (strengthening the reporting of observational studies in epidemiology) guidelines in fig 3.1.2. For phase one, descriptive study, the study population comprised 100 children with Duchenne Muscular Dystrophy and their caregivers. Only 66 participants completed the time period of both the phases.

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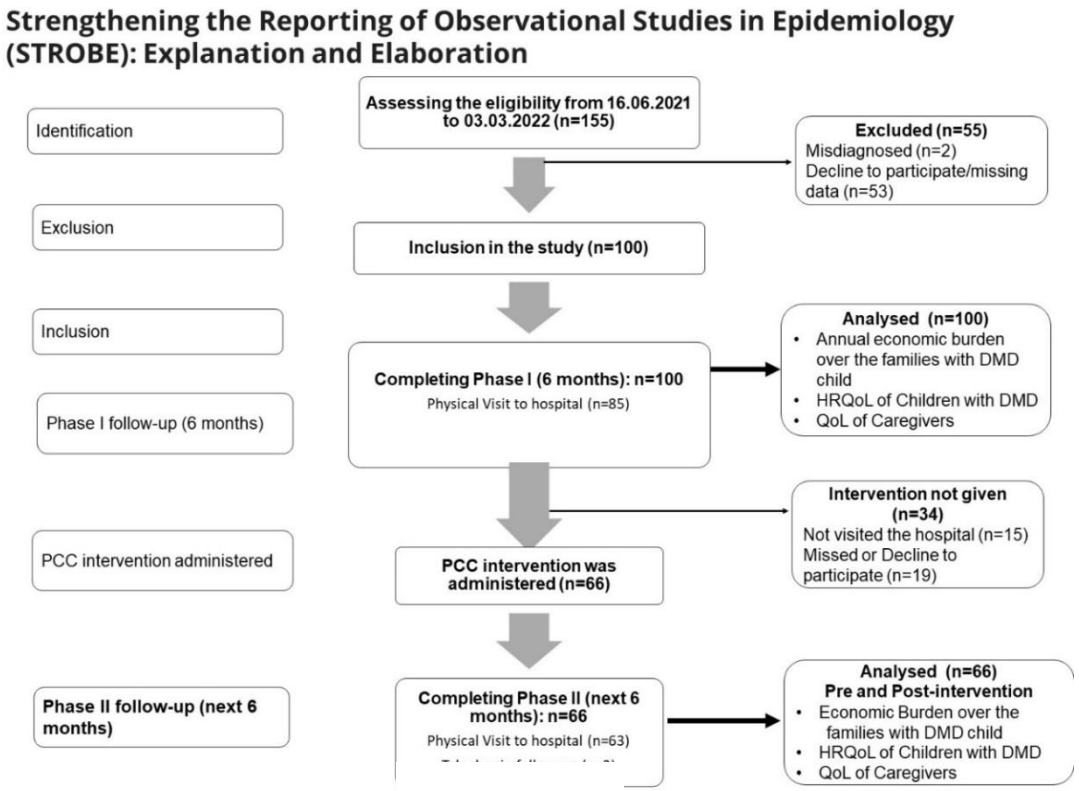
**Figure: 3.1.2 Flow diagram of Non-Randomized Observational Clinical Trial Study**




### 3.1.4 Study Period

After the clearance from the institutional ethics committee, recruitment was initiated in June 2021. The trial was registered under the "Clinical Trials Registry – India" (CTRI), and recruitment was started on the 16$^{th}$ June 2021 and completed on the 31$^{st}$ of December 2021. The study was expected to be of 1 year (6 months for each phase). Thus, expected finish date of this study was December 2022. It was finished well in time. Although the initial preparatory part of the study was too long because of COVID-19, it took about one and a half years to take ethical clearance from the different institutes involved, Institute ethics committee PGIMER, Institute ethics committee LPU and institutes collaborative research committee (ICRC) PGIMER.

### 3.1.5 Study Population

Families/caregivers with 5–15-year-old boys with Duchenne or Beckers MD and their caregivers taking consultation at the Pediatric Neurology Unit of the Department of Pediatrics at Advanced Pediatric Center PGIMER Chandigarh. (Government Tertiary Care Center).

**Caregiver:** The persons who looked after the patients in the hospital or at home (parents/ family members). This excluded hospital staff and paid workers hired by the family.

### 3.1.6 Subject Selection Criteria

Boys with DMD presenting to the Pediatric Neurology Clinic were screened for enrollment in the study. The selection of subjects was done by a pediatric neurologist. Those who fulfilled the inclusion and exclusion criteria were included in the study after taking informed consent. All relevant medical and non-medical conditions were taken into consideration before deciding whether this protocol was suitable for a particular subject.

***Inclusion Criteria:***

(1) Families with affected boys between 5-15 years of age with clinical symptoms suggestive of DMD and increased creatine kinase (CK) levels (CK>1500IU)

(2) Laboratory confirmation: Genetic confirmation by Multiplex Ligation-Dependent Probe Amplification (MLPA)/ Polymerase Chain Reaction (PCR)/ Next-Generation Sequencing (NGS)/SANGER or Muscle biopsy (showing absence of dystrophin expression genetically).

(3) Caregivers of the affected boys have smartphones and internet

***Exclusion Criteria***

1. Subjects/families who were unwilling to participate or unable to comply with the protocol study procedures or visits
2. Consent or assent not given.

### 3.1.7 Ethics Approvals:

The clearance was obtained from the ethics committee of PGIMER and Lovely Professional University (LPU) for the thesis completion. In LPU ethical committee also document that primary ethical responsibility lies where the human trial is conducted. Thereafter, one more clearance was taken before recruitment that is from Institute's collaborative research committee (ICRC) PGIMER. As per ICRC guidelines, the ownership of the data lies with PGIMER. After getting ethical clearance from all the ethics committees involved. The trial was registered under the CTRI, and only after

registration of the trial under CTRI recruitment was started on the 16th of June 2021. Written informed consent was obtained from the parents of all the respondents as well as the concerned authorities. All data is still kept confidential.

**Trial Registration:**

The study protocol was registered with the Clinical Trials Registry India (CTRI/2021/06/034274) and protocol is published with International Registered Report Identifier (IRRID): "PRR1-10.2196/42491."

## SECTION II

## 3.2 INTERVENTION PROGRAMME

The study was conducted prospectively in two phases. Phase one was the observational part of the study and routine care was given. Phase 2 was the interventional part of the study. The intervention was developed during phase 1 later during Phase 2 Intervention was given.

### 3.2.1 Routine Care

The boys suspected of DMD were clinically diagnosed and consulted in the outpatient clinic (OPD) of the Neurology Unit of the Institute. After clinical investigations, a treatment, management plan for the patient care is designed. The patients were managed with multidisciplinary care with the involvement of different departments and health- care professionals. For patient visits different subspecialists as and when required. The requirement of physiotherapy, orthosis and several visits to specialty care units increase hospital visits, confusion and even loss-to-follow-up.

The components of routine management are as follows:

1. Confirm diagnosis: Clinical examination, investigations, genetic confirmation or muscle biopsy.
2. Family counselling
3. Baseline assessment of motor attributes.
4. Baseline cardiac, respiratory functional assessment, spine, bone health, endocrine evaluation.
5. Oral supplementals of calcium and vitamin D (if indicated)

6. Night-time AFO (Ankle-foot orthosis)
7. Advice as physical therapy and stretches
8. Vaccination (immunization).
9. Start steroids as indicated as per standard guidelines.
10. Dietary advice
11. Advice on play and leisure
12. Three monthly clinical check-ups for disease progression and steroid side effects.
13. Prenatal diagnosis and carrier screening.

### 3.2.2 PCC: Comprehensive DMD-Telecare Model

The intervention was developed during Phase 1 after an intense review of the literature and discussion with the specialists from the neurology unit, rehabilitation unit, community medicine and public health. Advice from the Medical photography department of PGIMER was also considered to formulate the intervention program.

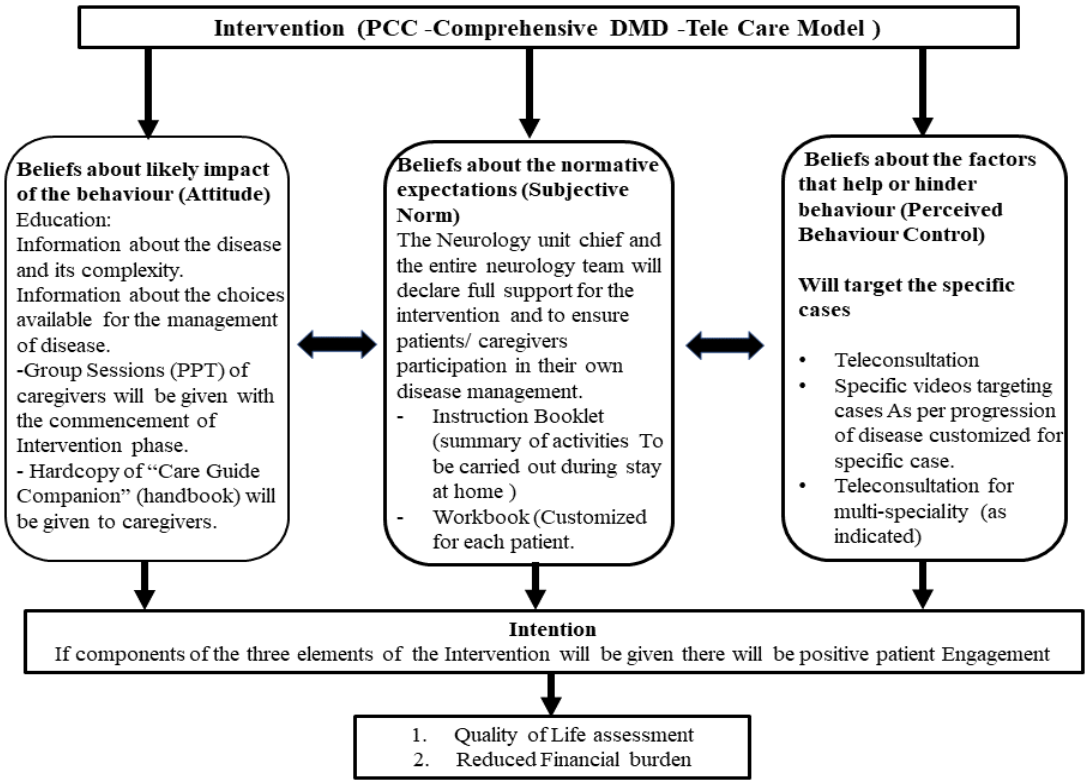
**Rationale of Patient-Centered Care (PCC) Model**

The rationale of developing the Patient Centric Care model in patients of Duchenne Muscular Dystrophy was to provide basic information to the patient and their parents/caregivers so that they can effectively participate in the process of obtaining comprehensive care, in a resource-limited setting. This model is designed to create awareness among the patient and caregivers regarding the disease, it's course of illness and the treatment options available and to facilitate the require behaviour change regarding managing the disease not only pharmacologically but also by improving QoL. Following a systematic literature review, the intervention was developed based on the Theory of planned behavior, with tailoring for each stage of the study (Ajzen, 1991). The conceptual model of Patient-Centered Care (PCC) is illustrated in figure 3.2.1 Recognizing the challenges faced by patients and caregivers, particularly in low- and middle-income countries (LMICs), a telecare component was integrated into the model.

The protocol material for the 'Comprehensive DMD -Tele Care Model' was circulated among 1) Pediatrician/ Pediatric Physiotherapist working with Ped-Neurology Clinic, 2) School of Public Health Experts 3) Cases. A preliminary version of this package was developed. That was circulated among specialists for consensus validity. It was modified as per the feedback received. Thereafter, it was pilot tested among 5

affected children and their caregivers with a quick follow-up of 30 days. The feedback was also gathered from the caregivers and was considered to further modify the intervention package. The purpose of the intervention was to reduce the patient's multiple visits. The families were called for physical follow-up only if indicated and instructed by the neurologist within 6 months. Such visits were recorded and the purpose of these visits was also recorded.

**Figure 3.2.1 Intervention Frame Work**




The patient-centered care model was a Comprehensive DMD -Tele Care Model the components of the model are as follows:

1. ***Informational about the disease:*** Sessions using powerpoint presentation and the "Care Guide Companion" booklet were provided. These were to generate awareness and information about the treatment option available for the disease. The care guide companion (hard copy and digital) was designed in such a way that it helped patients or their caregivers in the decision-making process on their own after the acquisition of knowledge in a comprehensible manner. This information booklet answered the questions of caregivers who visit the health facility and are totally unaware of the disease that their child was having. it was

one of the interventions in the package. components of the book were: 1) basic information related to disease, 2) management of disease, 3) role of the care provider team, 4) support groups to help DMD people

2. ***Instructions and checklist based intervention and follow-up:*** Instructions were given to the patients and their caregivers to make them understand all the activities that were to be carried out once the subject enters phase 2 in the study. Audio-videos and images of instruction of stretches/ physiotherapy, and physical activity were to be provided to the family.
3. ***Compliance diary:*** The patient was to comply with the prescribed procedures and medications. A customized daily planner was given to each patient in the intervention phase to increase his adherence to the prescribed management. This planner included periodic weight, height, blood pressure, daily medication (steroid/ACE inhibitors/Calcium/Vitamin D) and stretching exercise, a check-list for dietary intake, some clinical evaluation that was to be done at home (e.g: Gower's time, 10 MWT, vignos score, etc. (Annexures)
4. ***Rehabilitation kit:*** This kit is comprised of orthotic devices like AFO (Ankle-foot orthosis) or KFO (Knee-orthosis), a wooden wedge, Dietary counselling/checklist, and a guidebook for a local doctor. The researcher ensured that all the components of the kit were provided to the patients.
5. **Tele-Consultation:** Three-monthly video teleconsultation was to be given to the patient by clinicians for providing diagnostic or therapeutic advice through electronic means. The patient did not have to visit the clinic physically. This was to be done through:

a) Review Videos: Videos that patients were to share with the care team regarding their present state of illness. These videos were the recordings of the physical activities that the patient can perform at home and share with the team to evaluate.

b) Customized Videos: As per the clinical requirement additional customized videos were to be shared with patients to address the need.

**6. Multi-specialty Teleconsultation (as indicated)**

Not all subspecialists were to be needed at all ages or stages, but they were to be accessible if necessary. (As indicated)

### 3.2.3 Administration of Intervention Package

The junior residents/ senior residents/faculties of the neurology unit of pediatrics department updated the inclusion and exclusion criteria regarding the protocol and were requested to refer children living with DMD to the intervention room (IR). A referral system was established for this. The researcher recruited the eligible cases on Thursdays and Fridays every week. Once a patient with DMD comes to the pediatric neurology outdoor specialty clinic, he was referred by the doctor to the researcher's room (room no. 5109, 5A, APC) after a clinical workup and necessary investigation/prescription. Once patient completed first 6 months after enrolment patient was given information regarding the disease both with the help of hardcopy of the care guide companion and the digital presentation along with a power point presentation. Usually, it lasted for about 20-30 minutes, but few were very long about 45 minutes to an hour (caregivers with many doubts).

The participants in this phase were encouraged to comply with the instructions about various components of the intervention package. Follow up were done (both virtually and physically) and participants were encouraged to continue with the program. Patients were contacted during the follow-up period through telecommunication and were given multi-speciality teleconsultation that includes: Physiotherapy, orthotic advice, Neurologist, and Dietician. Reminders were given to patients who do not turn up on scheduled follow-up visits at the end of 6 months (intervention end-point).

### 3.2.4. Schedule of Events and Study Parameters

The types and timing of data recorded during phases 1 and 2 are in tables 2 & 3. X= scheduled activity in the timeline.

***Table: 3.2.1 Schedule of Events in Phase 1 (Routine treatment)***

| **Study Period** | **Enrolment in Phase 1** | **Follow -up Weeks** | | | | | |
|---|---|---|---|---|---|---|---|
| **Study Week** | 0 | 4 | 8 | 12 | 16 | 20 | 24 |
| **Visit Window (days)** | 0 | | | ±14 | | | ±7 |
| **Visit Number** | 1 | | | 2 | | | 3 |
| Informed consent | X | | | | | | |
| Physical/clinical examination Physical Visit | X | | | X | | | X |
| Physiotherapy Therapy consultation | X | | | X | | | X |
| Genetic counselling routine | X | | | X | | | X |
| Diet counselling | X | | | X | | | X |
| Information on expenditure | X | X | X | X | X | X | X |
| WHOQOL-BREF for parents | X | | | | | | X |
| EQ-5D for patient | X | | | | | | X |
| 10-meter walk/run test | X | | | | | | X |

In the routine care phase (Phase 1), activities conducted at the health facility during patient visits were documented. These activities are listed in table 3.2.1, and informed consent was obtained from participants during this phase.

***Table: 3.2.2 Schedule of Events in Phase 2 (Intervention Phase)***

| **Study Period** | **Phase 2 (Physical visit)** | **Follow -up Weeks** | | | | | |
|---|---|---|---|---|---|---|---|
| **Study Week** | 24 | 28 | 32 | 36 | 40 | 44 | 48 |
| **Visit Window (days)** | ±7 | | | ±14 | | | ±7 |
| **Visit Number** | 1 | | | 2 | | | 3 |
| Informational about the disease (Hardcopy +Digital) | X | | | | | | |
| Physical/clinical examination | X | | | | | | |
| Physiotherapy Therapy exercise home-program 30 min session (Intervention specific) | X | | | | | | |
| Genetic counselling | X | | | | | | |
| Diet counselling | X | | | | | | |
| Instructional booklet | X | | | | | | |
| Workbook | X | X | X | X | X | X | X |
| Orthotics | X | | | X | | | X |
| Tele-Consultation | X | | | X | | | X |
| Multi-speciality teleconsultation | X | X | X | X | X | X | X |
| Information on expenditure telephonic contact) | X | X | X | X | X | X | X |
| WHOQOL-BREF for parents | X | | | X | | | X |
| EQ-5D for patient/Proxy | X | | | | | | X |
| 10-meter walk/run test | X | | | X | | | X |

During the intervention phase (Phase 2), activities are detailed in table 3.2.2, comprising two physical visits and one tele or remote visit. Various counseling sessions are conducted at the onset of the intervention phase, including physiotherapy exercise home-program sessions lasting 30 minutes (specific to the intervention), genetic counseling, and diet counseling.

## SECTION III

## 3.3 RESEARCH INSTRUMENT

The interview schedules used were both structured and open-ended. The sociodemographic data and OOP expenditure (economic burden) (Annexure I & 2). The EQ-5D EuroQoL 5-dimension questionnaire was to be used to measure QoL in children living with DMD Annexure IV and V. QoL of parents/caregivers will be measured using WHOQOL-BREF i.e Annexure VI.

### 3.3.1. OP Agrawal Scale of Socio-Economic Status

The socio-economic status was assessed using OP Agrawal scale. The OP Agrawal scale of socio-economic status is a 22-item questionnaire that measures the socio-economic status designed for a family in India. The questionnaire covers various aspects of the family's income, education, occupation, assets, social participation, and living conditions. Each item has a different weightage and scoring range from 3 to 9. The total score of the questionnaire can range from 22 to 198. The family's socio-economic status is divided into six categories: very high (more than 76), high (61-75), medium high (46-60), medium low (31-45), weak (16-30), and very weak (less than or equal to 15). The questionnaire is already validated and reliability assessed in Indian Setting. The scale is designed to work for both urban and rural settings in India (Aggarwal et al., 2005). For better understanding and analysis the data were transformed into three categories: High (Very high and high i.e. more than 75); Medium (medium high i.e. 46-60) and Lower (medium low, poor and very poor i.e. less than or equal to 45).

### 3.3.2 Economic Burden

The data on economic burden included information on:

a. Household consumption expenditure (at baseline)
b. Patient-level expenses (out of pocket expenditure) incurred on diagnosis and treatment related to DMD, and coping mechanisms for dealing with the same (During Phase I and Phase II (6 month each).
c. Informal care cost (productivity loss) patients' caregivers were interviewed regarding the time spent daily on the affected child for caregiving during the entire phase. The parents were also interviewed regarding the long-term absenteeism due to BMD/DMD (in days). Caregivers were also asked regarding

their change in job or inability to perform their job due to involvement in caregiving for their affected child due to DMD/BMD.

The patients were interviewed using a pre-tested semi-structured schedule, which was adapted from prior studies conducted in alike settings (NSSO, 2006; Prinja et al., 2016; Prinja et al., 2018). The "cost of illness" approach was followed to assess OOP expenditure, which classifies the same into direct and indirect expenses(Rice, 2000).

At the time of participant recruitment in the first phase, baseline data was collected on their social demographic characteristics and quality of life. Further, after the initiation of the treatment, these patients were again contacted telephonically and through WhatsApp or other internet mediums to assess the financial burden by collecting data on out-of-pocket (OOP) expenditure incurred on the treatment of DMD. The data collection with the help of WhatsApp has evolved as a very innovative idea during the study. This heled in real time contact with the participant and reduced recall bias substantially. The data of economic burden was collected during the entire study period and this real-time connection with the patient's caregiver also facilitated the implementation of patient-centric care - the intervention.

The expenses on the consumables, diagnosis, drugs, user fee, hospitalization or procedure fee were taken into consideration as direct health care expenditure. The direct costs included the medical treatment/management related to the disease and the related complications. The diagnostic costs and routine care costs are also taken into direct costing consideration. To get direct COI for Duchenne “costs of hospitalization”, “drug treatment, and rehabilitation services e.g: physiotherapy or occupational therapy were used. The expenditure on boarding, loading, food and transportation were considered as direct non-healthcare expenditures. The income loss or wage loss by the patient or/and accompanying caregivers during the period of assessment was taken as an indirect expenditure.

### 3.3.3 Quality of Life of Patients and Caregivers

#### 3.3.3.1 EuroQoL 5-Dimension questionnaire and Visual analogue (EQ 5D-3L & VAS)

The health-related quality of life of both patient and proxy from the caregiver was gathered at the baseline, then after phase 1 and second phase 2. The quality of Life of

caregivers will also be recorded at main three data points (baseline, phase 1-end, and phase 2-end). Thus, for measuring the quality of life for caregivers and patients there were three data points. The EQoL 5-D questionnaire (EQ-5D 5L) was used to measure QoL in children living with DMD. QoL of parents/caregivers was measured using the "World Health Organization Quality of Life Instrument" (WHOQOL-BREF).

HrQoL of patients was expressed in terms EQ-5D-5L QoL tool. This tool has EQ-5D descriptive system and the EQ visual analogue scale (EQ VAS). The EQ-5D-5L descriptive system has 5 dimensions, i.e., mobility, self-care, usual activities, pain/discomfort and anxiety/depression. Each of these dimensions has a response in the form of 5 levels of perceived problem: no problems (level 1), slight problems (level 2), moderate problems (level 3), severe problems (level 4) and extreme problems (level 5). Thus, a unique health state is defined by combining 1 level from each of the 5 dimensions. The EQ VAS records the patient's self-rated health on a vertical visual analogue scale, where the endpoints are labeled as "The best health you can imagine" and "The worst health you can imagine". The analysis for creating Qol utility scores based on the EQ-5D-5L tool was done as recommended in its user guide. Each of the patient's conditions or state at a particular time period will be referred to in terms of a 5-digit code. For example, state 11111 implied no problems in any of the 5 dimensions, while state 55555 indicated extreme discomfort in each of the dimensions. Each of these states was further converted into a single index utility value, based on the recently generated tariff value set for the Indian population ( Goni et al., 2017; Jyani et al., 2020; Jyani et al., 2022).

#### 3.3.3.2 Pediatric Quality of Life Inventory Neuromuscular Module

HRQoL was also calculated using the Disease-specific QoL tool, PedsQL TM (Pediatric Quality of Life Inventory TM) 3.0 Neuromuscular Module. The scale score ranges from 0 to 100, where higher scores reflect better quality of life (HrQoL). This scale has 25 items and comprises 3 domains: neuromuscular disease (17 items related to the disease and symptoms), communication (3 items related to the patient's ability to communicate with health care providers and others about illness), and family resources (5 items related to family financial and social support systems).

#### 3.3.3.3 World Health Organization Quality of Life-BREF (WHOQOL-BREF)

A self-administered, WHOQOL-BREF tool was used to assess the QoL of caregivers. This tool has 26 items, describing 4 domains of an individual's QoL i.e., physical health (7 items), psychological health (6 items), social relationships (3 items) and environmental health (8 items). It also contains items for general QoL and overall health (2 items). Response to each item is recorded in 5 levels as scored from 1 to 5. The raw scores were converted into transformed scores ranging from 4-20. Later, these transformed scores are again transformed into main domain scores ranging from 0-100 scale. Higher scores mean better QoL(Sathvik et al., 2008).

## SECTION IV

## 3.4 STATISTICAL TOOLS AND DATA ANALYSIS

### 3.4.1 Research Hypothesis

Based on literature review, to study the objectives three different hypotheses have been formulated based on the proposed model. Three major outcomes of the study are the Health-related quality of life (HrQoL) of the patient, the quality of life (QoL) of caregivers, and the economic burden of disease on the families. The Hypotheses were framed based on the above-given objectives.

**$H_{01}$:** The Patient centered care model does not impact the economic burden on families with children living with Duchenne Muscular Dystrophy.

**$H_{02}$:** The patient centred care model does not impact the overall Quality of life of caregivers of children living with DMD.

**$H_{03}$:** The patient centred care model does not impact the health-related quality of life of children living with DMD.

### 3.4.2 Economic Burden

The economic burden and QoL data were analyzed in the similar manner for both the phases and later parameters/ outcome variables were compared to justify the hypothesis. Initially, primary data was entered in Epi Info™ 7, Division of Health Informatics & Surveillance, Center for Surveillance. Data were analysed using both Epi Info 7 and SPSS Ver 23.

The Mzean OOP expenditure sustained during the assessment period on different modes of treatment was calculated. The OOP expenditure was reported in Indian currency- INR and indirect expenditure was estimated for the follow-up treatment for both phases. Human capital approach was used in assessing the wage loss of caregivers (Lensberg et al., 2013). To address the cost of unpaid caregiving formula in the previous study from Germany was modified for the Indian setting (Katz et al., 2014).

### 3.4.1 The calculation of Indirect Cost for unpaid caregiving during Phase I & Phase II in the Indian Scenario

1. Loss of productivity due to unpaid caregiving: **LP $_{\text{(unpaid caregiving):}}$ (Un-paid $s_{LT}$ + Unpaid $s_{ST}$) X $S_{gdp}$**

i. Loss of productivity for non-working unpaid caregivers Phase I/Phase II = **LP $_{\text{(unpaid caregiving*)}}$**

ii. Unpaid $s_{LT}$ = Long-term caregiving for DMD during Phase I/Phase II (in days)

(Parents with no wage loss taking leave for the care of their child due to BMD/DMD)

iii. Unpaid $s_{ST}$ = Short-term caregiving for BMD/DMD during Phase I/Phase II (in hrs converted into days)

(Parents and all caregivers providing daily care (in hours) to their child living with BMD/DMD)

iv. $S_{gdp}$ = GDP (per capita gross domestic product converted into per day)

unpaid caregiving* is caregiving by a working parent during non-working hours (caregiver's leisure time)

**The formula for the cost of absenteeism and changes in the working situation**

(Productivity loss of caregiver who is not able to work due to their involvement in caregiving)

2. Loss of productivity working caregivers **(Phase** I/Phase II) = **LP $_{\text{(absenteeism**)}}$ + LP $_{\text{(changes)}}$**

i. Loss of productivity due to absenteeism = ($Abs_{LT}$ + $Ab_S$ ) X Sd

ii. Abs $_{LT}$ = Long-term absenteeism due to child's BMD/DMD during Phase I/Phase II (in

days)

iii. $Ab_S$ = Short-term absenteeism- absent in hours due to child's BMD/DMD during Phase I & II (in hours converted into days)

iv. Absenteeism**: Wage loss of caregivers due to absenteeism due to child's BMD/DMD

Sd = actually gross salary per day

3.Loss of productivity due to change in a work situation: $LP_{change} = S_{lost} X t + D_{lost} X S_{di}$

i. Change (loss) in wages of caregivers due to their child living with DMD/BMD.

ii. $S_{lost}$: Loss of gross salary loss per day caused by changes in working hours due to child's disease

iii. t: number of days with reduced salary due to changes in working hours during Phase I&II/ in 2021 and 2022

iv. $D_{lost}$ = number of working days lost due to child's disease during Phase I& II/ in 2021 and 2022

v. $S_{di}$ = actually gross salary per day, inflation-adjusted

Per capita GDP India (31 March 2022) = 2321.104

The productivity loss is taken as a loss in the gross domestic product (GDP) per capita per hour.

The financial risk was assessed in terms of catastrophic health expenditure and distress financing. Expenditure on DMD treatment which exceeded the threshold of 40 percent of non-food household consumption expenditure was considered catastrophic health care expenditure ( Reid et al., 2005; Serra et al., 2011). Money borrowed by the households with or without interest or selling of assets (cattle, house, land etc.) to cater the expenditure were classified as distress financing ( Huffman et al., 2011; Prinja et al., 2016; Chauhan et.al, 2016). Multiple logistic regression was used to assess the risk of catastrophic health expenditure and financial distress, considering factors such as age, income, treatment type, insurance, location, and disease stage at diagnosis. Catastrophic expenditure in DMD is unlikely due to the lack of definitive care; however, sensitivity analysis was performed to evaluate its prevalence at different thresholds (20 percent, 30 percent, 50 percent).

### 3.4.3 General Statistical Considerations

The subject listings were created for each CRF module, summary tables for continuous variables contain the following statistics: n, mean, standard deviation, standard error, 95 percent confidence intervals (CIs) on the mean, median, minimum, and maximum. The summary tables for categorical variables will include N (population), n (subgroup), percentage, and 95 percent CIs on the percentage. Where applicable, the summary data (mean, standard error) were presented in graphical form against the time to visit. Unless otherwise specified, all inferential analyses were 2-sided at the alpha (α) 0.05 level of significance. The number of subjects screened and enrolled, and the number of subjects completing all planned study assessments were summarized by treatment. The subjects who discontinue the study drug or are removed from the study prematurely were to be summarized by reason for discontinuation and treatment.

#### 3.4.3.1 Mean

Mean (X) is a fundamental concept of statistics and mathematics. In statistics, it is a measure of central tendency of a probability distribution. "Mean is estimated by taking the sum of all observations in a data set divided by the total number of observations in a given data set"

$$\overline{X} = \frac{\sum X}{N}$$

Where,

$\sum X$ = Summation of all observations.

N = Number of observations.

#### 3.4.3.2 Median

1. The median in general is described as the middle value arranged in a sorted, descending or ascending, list of numbers. Occasionally median can be more descriptive for a data set than the mean value. "For odd number of observations, the middle number is the median value and if there is an even number of observations, the median is the mean of the two middle values"

    1. For an odd number of observations:

Median=Value at position ((n+1) ÷ 2)

2. For an even number of observations:

Median = {Value at position (n/2) + Value at position (n/2+1)} ÷ 2

These positions refer to the sorted list of values.

Where,
N = Total number of observations.

### 3.4.3.3 Chi- Square ($\chi 2$)

"*Chi-square ($\chi 2$) statistic is a technique which measures the difference between the observed and expected frequencies of the outcomes of a set of events*" (Kumptala et al., 2913). The chi-square is performed to check whether two variables are associated to each other or not. It is useful for studying the difference in categorical variables, particularly when study variables are nominal in nature (*viz.* region, age groups etc.).

$$\chi 2 = \sum \frac{(Oi - Ei)^2}{Ei}$$

Where,

O*i* = Observed frequency.
E*i* = Expected frequency.

### 3.4.3.4 Logistic Regression

This technique is performed when the dependent variable is nominal or categorical
in nature. According to Field (2009), "*determines the impact of multiple independent variables presented simultaneously to predict membership of one or other of the two dependent variable categories*".

In logistic regression, a logistic transformation of the odds serves as depending variable:

$$\text{Log (odds)} = \text{Logit (P)} = \text{In} \left(\frac{p}{1-p}\right) \qquad \text{……..1}$$

Taking above dependent variable and adding a regression equation for

independent variable, logistic regression is:

Logit (P) = $\beta_0 + \beta_1X_1 + \beta_2X_2 + \beta_3X_3 \ldots\ldots\ldots\ldots \beta_nX_n$ ……2

Equation

$$p = \log\left(\frac{p}{1-p}\right) = \beta_0 + \beta_1X_1 + \beta_2X_2 + \beta_3X_3 \ldots\ldots\ldots\ldots \beta_nX_n \quad \ldots\ldots 3$$

Where

P= Probability of a case is in a particular category.

$\beta_0$ is the intercept.

$\beta_1, \beta_2, \ldots\ldots\beta_n$ are the coefficients for the independent or predictor variables $X_1, X_2, ..X_n$

e is the base of the natural logarithm.

### 3.4.3.5 Multiple Linear Regression

The common multilinear regression equation used was:

$y = \beta0 + \beta1x1 + \beta2x2 + \beta3x3 + \beta4x4 + \ldots\ldots\ldots\ldots\ldots.. + \beta nxn$

Where,

y : total cost of illness or total direct cost of illness or total indirect cost of illness or indirect cost related to changes in work or absenteeism due to illness.

x1, x2, x3….. xn : are the predictors listed above

β0 : The intercept 0 is the value of the outcome y when all predictor variables are zero.

β1, β2, β3…….. βn are regression coefficient obtained from the regression model.

The challenge in comparing regression coefficients, as they may be in different units, is akin to comparing "apples and oranges." The standardized coefficients address this by expressing them in a common set of statistically reasonable units, enabling more meaningful comparisons. The standardized coefficient obtained from the regression equation. Standardized coefficients enable researchers to compare the relative impact of various explanatory variables in a path model by adjusting for standard deviations. This facilitates comparison despite differences in units of measurement. The coefficient of $R^2$ is defined as the multiple correlation coefficient. The f-statistic was derived using the residual mean square from the multiple regression.

## SECTION V

## 3.5 LIMITATIONS

1. The study had certain limitations in terms of its design. Enrollment in the study were hospital based and most of the patients visiting the health facility are ambulatory within the age group of 5 to 12 years. Thus, economic burden assessed in the study was based on ambulatory DMD patients visiting the health facility. The studies in the past have shown that the maximum financial burden is in non-ambulatory stage of disease (Stage V). This group was not covered in the study.
2. The study design was not working in a controlled fashion, with no randomization, a single group is compared pre- and post-intervention. The experimental conditions may be systematically different across a time frame.
3. However, no special medical cost was captured for the subset of stage 5 disease in which basic life support is needed. Therefore, cost was similar to the lesser advanced stages of diseases. The usual medical course that is given universally to all the patience of DMD. Where, as none of the patients was followed till later stages of DMD where more expensive management is required e.g: external respiratory management (invasive for non-invasive devices). DMD patients in Indian are rarely followed up to the stage of severe morbidity as they are confined to their homes and do not seek any medical care.
4. The study reported that follow-up was challenging as with other progressive neuromuscular disorders without definitive treatment. Most patients hailed from far flung places and villages from where regular commute was daunting. Most parents/caregivers in this study preferred only physiotherapy or alternative medicine because of personal choices, local beliefs, cultural biases, an undue apprehension regarding adverse effects of medications, and the lack of efficacy of most medicines utilized in treating this disease.
5. A significant limitation of telecare, particularly in non-urban areas, is the lack of access to smartphones and reliable internet connectivity among many individuals. These technological barriers make it challenging for patients and caregivers in rural or underserved regions to benefit from telemedicine services. Without adequate access to smartphones or digital literacy, it becomes difficult for these populations to connect with healthcare providers, schedule consultations, or utilize

telecare platforms effectively. This digital divide exacerbates healthcare disparities, leaving a substantial portion of the population without access to critical medical services, especially for managing chronic or rare diseases that require regular follow-ups and specialized care.

# CHAPTER IV

# ECONOMIC BURDEN AND DETERMINANTS OF DUCHENNE & BECKER'S MUSCULAR DYSTROPHY

The DMD affects skeletal muscles causing a severe degree of disability in later childhood(Bushby et al., 2010). The condition is incurably reducing life expectancy to the late teens or early twenties(Suthar & Sankhyan, 2018). This study aims to understand the economic burden and socio-economic determinants of the quality of life of patients suffering from "Duchenne Muscular Dystrophy" (DMD)and their caregivers. The current study was aimed to estimate the economic/financial burden by analyzing three key components:

1. Direct Medical Costs,
2. Direct Non-Medical Costs
3. Indirect Cost

The study included only the burden on the families due to the disease. This cost was collected in the form of out-of-pocket expenditure by the family in the management of the disease. There is no coverage for health costs available for DMD in the present study. The first objective of the study was to estimate the "economic burden" on families with a child living with DMD or BMD. Most of the caregiver respondents of the MD represented the clinical severity stages II and III (88 percent). There was only one patient from stage V.

Whereas, only 8 percent of patients were "poor free beneficiaries" exempt from user fees and charges for various investigations in government settings. But this benefit is only restricted to government settings. However, if the poor free beneficiary sought consultations or investigations outside of government settings, they were still required to bear these expenses out of pocket. This placed a direct financial burden on families, which was not covered by any existing support mechanisms. Majority rare diseases like Duchenne has significant unmet needs due to the scarcity of available and effective treatments, combined by limited research efforts in this field. Most of the socio-economic studies conducted are confined to COI studies (Angelis et al., 2015). Socio-economic determinants of disease are seldom explored. The socio-economic status measures the social and economic position based on income, education and occupation. In a developing nation like India, with limited resources and supportive care for these patients widely dependent on their socio-economic status. Caregiver burden could be any physical, emotional, social or financial stress of providing care to a person with a chronic disease

(Mielck et al., 2014). Low SES groups seem to be faced with a double burden: increased levels of health impairments and, lower levels of valuated HRQL once health is impaired.

### 4.1 Socio-demographic Characteristics of Study Population

The study population comprised pairs of children and caregivers/parents with Duchenne Muscular Dystrophy and their caregivers. As DMD is a rare X-linked recessive disease majority of the affected population is males. The age of the study subjects at the time of enrolment ranged between 5 to 13 years with a mean age of 8.32 years (S.D ± 2.0) Statistical distribution is normal as normality test for age the is not significant p=0.177 (Shapiro-Wilk test of normalcy). Most studies evaluating economic burden in DMD patients use secondary databases with larger samples (363 to 770), mainly from Europe (Germany, Italy, United Kingdom) and the US, with mean age around 12 years(Landfeldt et al., 2014; Katz et al., 2014). Fewer OPD-based studies with smaller samples exist, e.g., Portugal (46 pairs mean age of 8 years in ambulatory and 21 years in non-ambulatory patients) (Labisa et al., 2022) and Brazil (27 patients). A study from South India included 275 participants with a mean age of 8.1 years (Singh et al., 2018). The present study is OPD-based with a smaller sample size, and most patients are ambulatory. These characters resemble with other Indian or OPD based studies with younger cohort with smaller sample.

#### 4.1.1 Socio-economic Status of Respondents

While 77.6 percent of the caregivers were educated more than in high school only 8.2 percent of them did not receive any formal education. None of them reported their income below the present poverty line. O.P. Aggarwal classification system was used to have an insight socio-economic status of the families having boys living with DMD. In total 22 individual questions (Items) were asked, including occupation, education, monthly family income, household assets etc. These variables were assessed to have a comprehensive knowledge of the socio-economic components of society. Only 7 percent of the families were from higher socio-economic status while 51 percent and 39 percent of families belonged to the upper-middle class and lower-middle class respectively according to the classification used (table 4.1.1**)**. There is a paucity of studies that specifically mention socio-economic status (SES) as a determinant of economic burden (EB) or healthcare utilization in the context of healthcare.

**Table 4.1.1 Socio-demographic determinates of the economic burden of DMD and BMD over the families.**

| ***Baseline characteristic*** | **Clinical Severity** | | | | | **Overall** |
|---|---|---|---|---|---|---|
| | **I** | **II** | **III** | **IV** | **V** | |
| ***Age in Years Mean (n±SD)*** | | | | | | |
| *At Enrolment* | 5.0±0.0 | 6.9±1.8 | 9.1±1.6 | 9.1±2.1 | 11.0± N.A | 8.32 ±2.0 |
| ***SES Status*** | n (%) | n (%) | n (%) | n (%) | n (%) | n (%) |
| *High* | 1(75.0) | 3 (9.4) | 1(1.8) | 2 (25.0) | 0 (0) | 7(7) |
| *Upper middle* | 0 (0) | 15 (46.9) | 31(55.4) | 5 (62.5) | 0 (0) | 51(51) |
| *Lower Middle* | 2(75.0) | 12 (37.5) | 23 (41.1) | 1(12.5) | 1 (100) | 39(39) |
| *Poor* | 0 (0) | 2 (6.3) | 1(1.8) | 0 (0) | 0 (0) | 3(3) |
| *Total* | 3(100) | 32 (100) | 56 (100) | 8 (100) | 1 (100) | 100(100) |
| ***Education of Caregiver*** | | | | | | |
| *At least Graduate* | 1(33.3) | 9 (28.1) | 9 (16.1) | 5 (62.5) | 0(100) | 24 (24) |
| *Matriculation* | 1(33.3) | 12(37.5) | 33 (58.9) | 0(100) | 0 (100) | 46 (46) |
| *No matriculation* | 1 (33.3) | 11(34.4) | 14 (25.0) | 3 (37.5) | 1(100) | 30 (30) |
| *Total* | 3 (100) | 32 (100) | 56 (100) | 8(100) | 1(100) | 100 (100) |
| ***Type of Family*** | | | | | | |
| *Nuclear* | 1(25.0) | 17(53.1) | 31(55.4) | 5 (62.5) | 0 (0) | 54 (54) |
| *Joint* | 2 (75.0) | 15(46.9) | 25(44.6) | 3(37.5) | 1 (100) | 46(46) |
| *Total* | 3(100) | 32 (100) | 56 (100) | 8 (100) | 1 (100) | 100(100) |
| ***Locality*** | | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| *Urban* | 0 (0) | 9 (28.1) | 18 (32.1) | 1(12.5) | 0 (100) | 28 (28) |
| *Rural* | 3 (100) | 23 (71.9) | 38 (67.9) | 7 (87.5) | 1 (100) | 72 (72) |
| *Total* | 3 (100) | 32 (100) | 56 (100) | 8 (100) | 1(100) | 100 (100) |
| ***Geographical Distribution*** | | | | | | |
| *Chandigarh (Tricity)* | 0 (0) | 2 (6.3) | 7 (12.5) | 0 (0) | 0 (0) | 9 (9) |
| *Punjab* | 1 (33.3) | 11 (34.4) | 17(30.4) | 3(37.5) | 0 (0) | 32 (32) |
| *Haryana* | 0 (0) | 6(18.7) | 10(17.9) | 1(12.5) | 0 (0) | 17(17) |
| *Eastern states* | 0 (0) | 5(15.6) | 6(10.7) | 0 (0) | 0 (0) | 11(11) |
| *UK* | 0 (0) | 1(3.1) | 2(3.6) | 0(0) | 1(100.0) | 4(4) |
| *HP* | 2 (66.7) | 4(12.5) | 10(17.9) | 1(12.5) | 0 (0) | 17(17) |
| *J & K other farther* | 0 (0) | 3(9.4) | 4 (7.1) | 3(37.5) | 0 (0) | 10(10) |
| *Total* | 3(100) | 32 (100) | 56 (100) | 8 (100) | 1 (100) | 100(100) |
| ***Respondent at Baseline*** | | | | | | |
| *Mother* | 1 (25.0) | 20 (58.8) | 32 (60.4) | 5 (62.5) | 0 (0.0) | 58 (58) |
| *Father* | 2 (50.0) | 11 (32.4) | 21 (39.6) | 3 (37.5) | 1 (50.0) | 38 (38) |
| *Others* | 1 (25.0) | 3 (8.8) | 0 (0.0) | 0 (0.0) | 1 (50.0) | 4(4) |
| *Total* | 4 (100) | 34 (100) | 53 (100) | 8 (100) | 2 (100) | 100(100) |

***Source:*** *Authors calculation established on primary data.*

The above information provides an overview of the distribution and characteristics of the participants in the study, including their socio-economic status, family structure, geographical origin, primary caregivers, and the duration of the intervention. (table 4.1.1)

***Socio-demographic Profile:*** The majority of the participants belonged to the upper-middle socio-economic status (SES), comprising 51 percent of the total participants. The

least number of participants belonged to the poor SES, accounting for only 3 percent of the total.

***Family Structure*:** A significant portion of the children in the study came from nuclear families, constituting 54 percent of the total.

***Geographical distribution:*** The participants were from various regions, with the highest percentage coming from Punjab (32 percent). Other significant contributors included Haryana and Himachal Pradesh, both at 17 percent. Eastern states represented 11 percent of the participants. Jammu and Kashmir accounted for 10 percent, while Chandigarh had 9 percent. The smallest representation was from Uttarakhand, with only 4 percent of the participants.

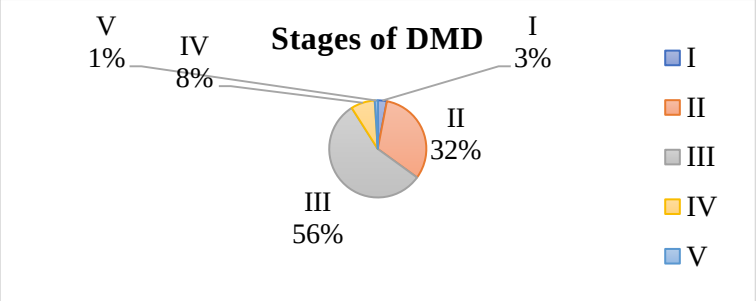
***Primary Caregiver:*** At the baseline, most of the respondents were mothers, comprising 58 percent of the total. Mostly from rural areas (72 percent) and educated up till matriculation (46 percent). In previous studies, caregivers were predominantly parents, with mothers being the primary caregivers in most cases ( Katz et al., 2014; Labisa et al., 2022).

### 4.2 Clinical determinates of the economic burden of Duchenne Muscular Dystrophy

***Clinical stages of disease:*** Most of the children enrolled in the study were in the late non-ambulatory disease stage (Stage III), comprising 56 percent of the total participants. Remarkably, only one child was in the late non-ambulatory stage at the time of enrollment. In contrast, registry-based enrollment tends to have a more uniform distribution of children across various stages of the disease. However, there were still relatively fewer patients enrolled in the late non-ambulatory stage (Landfeldt et al., 2014; Shehata et al., 2023).

***Caregiver Awareness Regarding Symptoms:*** A substantial portion of caregivers (40 percent) reported noticing a problem with their child during the early non-ambulatory disease stage. Conversely, approximately 45 percent of caregivers observed the signs late, after their child had reached 5 years of age.

**Figure 4.2.1 Distribution of Study Population with Clinical Severity**




***Source:*** *Authors calculation established on primary data.*

This data sheds light on when caregivers became aware of the initial symptoms, indicating that a significant portion detected the issue during the early stages, while a substantial percentage noticed it later in their child's development.

**Table 4.2.1 Clinical determinates of the economic burden of Duchenne over the families.**

| ***Baseline characteristic*** | **Clinical Severity** | | | | | **Total (100)** |
|---|---|---|---|---|---|---|
| | **I** | **II** | **III** | **IV** | **V** | |
| ***Type of Disease(p=0.784)*** | | | | | | N (%) |
| DMD | 3 (100) | 29 (90.6) | 49 (87.5) | 8 (100) | 1 (100) | 90 (90) |
| BMD Intermediate | 0(0) | 3 (9.4) | 7 (12.5) | 0 (0) | 0 (0) | 10(10) |
| ***Family History (p=0.913)*** | | | | | | |
| Present | 0 (0) | 5 (15.6) | 10 (17.9) | 1 (12.5) | 0 (0) | 16 (16) |
| Not Present | 3 (100) | 27 (84.4) | 46 (82.1) | 7 (87.5) | 1 (100) | 84 (84) |
| ***Affected Brother (p=0.969)*** | | | | | | |
| Yes | 0 (0) | 3 (9.4) | 6 (10.7) | 1(12.5) | 0 (0) | 10 (10) |
| No | 3 (100) | 29 (90.6) | 50 (89.3) | 7 (87.5) | 1 (100) | 90 (90) |
| ***Age when noticed the symptoms (p=0.003)*** | | | | | | |
| Up to 5 Years | 3 (100) | 25 (78.1) | 22 (39.3) | 4 (50) | 1 (100) | 55 (55) |

| | | | | | | |
|---|---|---|---|---|---|---|
| After 5 years | 0 (0) | 7 (21.9) | 34 (60.7) | 4(50) | 0(0) | 45 (45) |
| ***Symptom First Noticed (p=0.477)*** | | | | | | |
| Difficulty in walking | 1 (50.0) | 7 (24.1) | 11 (19.6) | 3 (13.0) | 1 (4.3) | 23 (24) |
| Inability to climb stairs | 0 (0) | 3 (10.3) | 5 (8.9) | 0 (0) | 0 (0) | 8 (8.3) |
| Difficulty in getting up from the floor | 1 (50.0) | 9 (31.0) | 14 (25.0) | 2 (25.0) | 0 (0) | 26 (27.1) |
| Pain in muscles/limbs | 0 (0) | 0 (0) | 1 (1.8) | 0 (0) | 0 (0) | 1 (1.0) |
| Frequent falls | 0 (0) | 7 (24.1) | 23 (41.1) | 1 (12.5) | 0 (0) | 31(32.3) |
| Muscle enlargement | 0 (0) | 3 (10.3) | 2 (3.6) | 1 (12.5) | 0 (0) | 6 (6.3) |
| Loss of ambulation | 0 (0) | 0 (0) | 0 (0) | 1 (12.5) | 0 (0) | 1(1.0) |
| ***Who noticed first (p=0.594)*** | | | | | | |
| Parents | 1 (33.3) | 16 (50.0) | 33 (58.9) | 3 (37.5) | 1 (100) | 54 (54) |
| Other family members | 1 (33.3) | 7 (21.9) | 15 (26.8) | 2 (25.0) | 0 (0) | 25 (25) |
| School | 0 (0) | 2 (6.3) | 6 (10.7) | 2 (25.0) | 0 (0) | 10 (10) |
| Neighbourhood | 0 (0) | 2 (6.3) | 1 (1.8) | 0 (0) | 0 (0) | 3 (3) |
| accidental finding or others | 1 (33.3) | 5 (15.6) | 1 (1.8) | 1 (12.5) | 0 (0) | 8 (8) |
| ***Knowledge about the disease (p=0.98)*** | | | | | | |
| Yes | 1 (33.3) | 11 (34.4) | 30 (53.6) | 5 (62.5) | 0 (0) | 47 (47) |
| No | 2 (66.7) | 21 (65.6) | 26 (46.4) | 3 (37.5) | 1(100) | 53 (53) |

***Source:*** *Authors calculation established on primary data.*

Table 4.2.1 provides insights into the clinical determinants affecting the economic burden on families dealing with Duchenne Muscular Dystrophy (DMD). The majority of

patients (90 percent) suffered from DMD, while 10 percent had intermediate BMD. A family history of DMD was absent in most cases (84 percent), although 10 percent of patients had an affected brother. The symptoms were noticed in 55 percent of children up to 5 years of age, and there was a significant association with disease severity. The children diagnosed at a later age tended to be more adversely affected. Frequent falls were the most common initial symptom (31 percent), followed by difficulty in walking (24 percent), difficulty getting up from the floor (27.1 percent), and the inability to climb stairs (8.3 percent). In most cases, parents (54 percent) or other family members (25 percent) were the ones who initially detected the symptoms, and 8 cases were accidental findings. It's notable that a significant proportion (47 percent) of parents had no prior knowledge about DMD. These clinical determinants shed light on the varied ways in which DMD affects patients and their families from the initial symptoms to the role of family members in recognizing and addressing the condition. Additionally, the lack of awareness about DMD among some parents underscores the importance of education and support in managing the disease.

### 4.3 Healthcare Utilization Pattern of the Study Population

The healthcare utilization was high in patients after getting the intervention, although the utilization pattern also changed with the implementation of the intervention. Over all utilization of care improved after intervention maximum utilization was seen with Physiotherapy/ Rehabilitation Care. One of the most noticeable differences observed during the intervention was the utilization of rehabilitation services, such as physiotherapy and respiratory management, which increased significantly. The genetic consultations saw a remarkable increase, nearly doubling the count of consultations compared to routine care in phase I. As a notable impact, 11 mothers from the study population sought and received further prenatal diagnostic consultation and care at the tertiary care center after genetic consultations. It's important to note that the study did not capture the utilization of healthcare services for prenatal diagnosis and management, as this aspect was not directly related to the cost of illness of the disease under investigation. The utilization pattern for nutritional counseling also followed a similar trend, with increased usage.

In the second phase of the study, fewer physical visits were recorded due to the introduction of teleconsultation by a neurologist as part of the intervention. This

teleconsultation approach significantly reduced the physical and economic burdens on patients, particularly benefiting the approximately 40 percent of patients residing in distant provinces. Utilization patterns of care during phase I, Phase II and Non-participation in the intervention are shown table 4.3.1

**Table 4.3.1 Utilization pattern of health resources by Dystrophinopathy Patients**

| ***Healthcare Services*** | ***Utilization during phase 1 (routine care) (N)*** | ***Utilization during phase 2 (intervention) (N)*** | ***Utilization not opted for intervention (care) (N)*** |
|---|---|---|---|
| Neuro OPD | 195 | 129 | 68 |
| Cardiac OPD | 15 | 11 | 4 |
| Ophthalmic Care | 5 | 6 | 1 |
| Physiotherapy/ Rehabilitation Care | 139 | 194 | 59 |
| Genetic consultation | 8 | 17 | 8 |
| Physiotherapy related to Respiratory Care | 5 | 42 | 6 |
| Nutritional counselling | 12 | 24 | 5 |
| Inpatient treatment | 1 | 0 | 1 |
| Other superspecialist consultation | 45 | 62 | 59 |

***Source:*** *Authors calculation established on primary data.*

***Note:*** *Multiple Responses.*

Approximately 33 percent of the study population did not participate in the developed intervention, and this was attributed to various reasons. These reasons included instances of lost follow-up due to a decline in ambulation, a lack of interest in the intervention due to the unavailability of definitive treatment, or migration to distant locations far from the healthcare facility. Consequently, patients facing these challenges discontinued their participation in the study prematurely. The utilization pattern during this period indicated that very few healthcare services were accessed, as illustrated in table 4.3.1 Some patients made just one visit to the health center, typically to the neurology outpatient department (OPD). For those who did not enter Phase 2 of the study, the total mean period of follow-up was 172.1 days. This data underscores the challenges

faced by a significant portion of the study population, leading to their discontinuation from the study and limited engagement with healthcare services during this period.

In a study conducted in Germany, it was found that 85 percent of Duchenne Muscular Dystrophy (DMD) patients and over half of Becker Muscular Dystrophy (BMD) patients expressed the need for medical aids. DMD patients became reliant on wheelchairs at a median age of 15, while BMD patients reached this stage at a median age of 40, indicating the progressive nature of the disease. As the disease severity increased, there was a corresponding increase in the need for direct medical care (Katz et al., 2014). Analysis of healthcare service utilization across different countries revealed varying patterns. In Germany, the most utilized services were non-medical community services, such as home assistance and transportation, followed by aids, devices, and home investments. In Italy and the United Kingdom, non-medical community services were also highly utilized, along with services provided by physiotherapists or occupational therapists. In the United States, aids, devices, and home investments were the most utilized services, followed by non-medical community services (Landfeldt et al., 2014). The lack of supportive services for rare diseases like Duchenne Muscular Dystrophy (DMD) is not unique to India. Similar findings were observed in a study from Egypt, where only five patients were in the non-ambulatory stage, comparable to the single patient identified in this study cohort. Additionally, the cost for medical aids and devices constituted only 3.5 percent of the total cost in the Egyptian study, indicating a limited utilization of these essential resources (Shehata et al., 2023).

### 4.3.1 Healthcare / Patient Care Pathway of DMD and BMD Patients:

The patient care pathways (CPW) play a crucial role in structuring the care process, particularly within the framework of Patient-Centered Care (PCC) approaches. Failing to develop and comprehend CPWs can result in diagnostic delays and unequal access to available treatments. This burden of complex care coordination often falls upon patients and their families, leading to significant personal, professional, and social challenges that can adversely affect their "quality of life."

In the present study, the pathway to care was examined, revealing that parents consulted an average of 2.9±1.4 treatment agencies before arriving at the study site. While, number of agencies consulted increased with the severity of the disease (with a median of 4 in stage IV), no significant difference was observed. Notably, some patients had to navigate

up to 8 different agencies in their pursuit of care. These findings emphasize the need for streamlined and efficient patient care pathways to improve the overall quality and accessibility of healthcare services.

In this study, the longer the pathway to care, the more time it took for patients to complete it. In the present study, the meantime consumed to traverse this pathway was about 12.5 months with a maximum span of 84 months (7.0 years). Notably, the patient with the longest journey hailed from Jammu and Kashmir, highlighting the impact of distance on access to specialized care (speciality care center). The number of healthcare agencies consulted was linked to the time spent in seeking care, but there was no correlation with the disease's severity. Notably, roughly 80 percent of patients received a correct diagnosis by the age of 8, a critical point as it falls within the period when the child is still ambulatory according to the disease's natural progression. This emphasizes the importance of early diagnosis and intervention for improved patient outcomes. (table 4.3.2).

**Table 4.3.2 Number of Agencies consulted and the time consumed in traversing the pathway to PGIMER with the severity of disease**

| ***Variables*** | ***Clinical Severity*** | | | | | ***Total*** | ***p value*** |
|---|---|---|---|---|---|---|---|
| | **I** | **II** | **III** | **IV** | **V** | | |
| Number of agencies consulted Mean ±SD | 3.0±1.0 | 2.8±1.1 | 3.0±1.5 | 2.9±1.2 | 3.0 | 2.9±1.4 | 0.72 |
| Time taken for pathway to care (months) Mean ±SD | 10.7±11.7 | 11.8±18.3 | 13.0±15.6 | 9.7±12.1 | 36.0 | 12.5±16.1 | 0.65 |
| Time to last follow-up (months) Mean ±SD | 5.3±4.9 | 4.5±3.6 | 4.5±4.2 | 5.7±6.1 | 24.0 | 4.8±4.6 | 0.01 |

***Source:*** *Authors calculation established on primary data.*

The mean time to last follow-up to the health facility was 4.8 months ranging from 0 to 24 months the data is not normally distributed. The scheduled follow-up time-period is of 3 months, the lag is uniformly present in all the stages of illness. The maximum delay was found in the early non-ambulatory (IV Stage) and late non-ambulatory stages. Non-ambulation makes travelling very difficult so physical follow-ups to the OPDs are very difficult and delayed. There is significant delay in last follow ups with the advancement of disease.

### 4.3.2 Challenges Faced by Patients in the Healthcare Facility

Even after patients reach a premier institute like PGI (Post Graduate Institute), several underlying challenges persist. A significant majority of patients (93 percent) encountered difficulties related to traveling when visiting PGI. This indicates that travel logistics can be a substantial obstacle for many patients seeking care. The same proportion of parents of patients (93 percent) reported challenges in understanding doctors' instructions in a single interaction, often they have to ask them to repeat the explanation. An effective communication can be crucial for treatment adherence and patient understanding.

However, three-fourths of the patients expressed dissatisfaction with frequent visits to PGI, with transportation being the primary hindrance. This suggests that the burden of repeated visits, coupled with transportation issues, impacts patient access to care. Surprisingly, only 39 percent of patients were informed about the availability of prenatal diagnostic options for genetic diseases. This highlights the need for improved patient education and awareness regarding available diagnostic tools.

**Table 4.3.3 Challenges in access to the Healthcare facility as reported at the time of enrolment**

| ***Challenges*** | **Clinical Severity** | | | | | **Total** |
|---|---|---|---|---|---|---|
| | **I** | **II** | **III** | **IV** | **V** | |
| | n (%) | n (%) | n (%) | n (%) | n (%) | n (%) |
| *Problems faced visiting PGI travelling* | 3 (100) | 29 (90.6) | 52 (92.9) | 8 (100) | 1 (100) | 93 (93) |
| *Problems faced due to frequent visits* | 2 (66.7) | 20 (62.5) | 43 (76.8) | 5 (62.5) | 1 (100) | 71 (71) |
| *Work-time loss in each visit* | 2 (66.7) | 22 (68.8) | 43 (76.8) | 6 (75.0) | 1 (100) | 74 (74) |
| *Transportation* | 2 (66.7) | 20 (62.5) | 43 (76.8) | 6 (75.0) | 1 (100) | 72 (72) |
| *waiting period* | 2 (66.7) | 19 (59.4) | 44 (78.6) | 6 (75.0) | 1 (100) | 72 (72) |
| *Difficult access to diagnostic facilities* | 2 (66.7) | 19 (59.4) | 44 (78.6) | 6 (75.0) | 1 (100) | 71 (71) |

| | | | | | | |
|---|---|---|---|---|---|---|
| *Asking doctors to repeat the instruction to have a clear understanding* | 3 (100) | 29 (90.6) | 52 (92.9) | 8 (100) | 1 (100) | 93 (93) |
| *satisfaction with the doctor's explanations* | 3 (100) | 25 (78.1) | 40 (71.4) | 6 (75.0) | 1 (100) | 75 (75) |
| *Unaware of prenatal diagnosis* | 1(33.3) | 12 (37.5) | 19 (33.9) | 6 (75.0) | 1 (100) | 39 (39) |

Table 4.3.3 depicts the challenges in access to the healthcare facility. A significant proportion of patients (74 percent) reported experiencing a loss of work time, indicating the economic impact of their medical visits. Transportation difficulties, were prevalent, with 72 percent of patients facing challenges in getting to healthcare facilities. Long waiting periods were a concern for 72 percent of patients, suggesting a need for improved efficiency in healthcare delivery. Accessing diagnostic facilities posed difficulties for 71 percent of patients, potentially impacting timely diagnosis and treatment. Issues arising from frequent visits were reported by 71 percent of patients, reflecting the burden of repeated medical appointments. Addressing these multifaceted challenges is essential to ensure a smoother healthcare experience and better outcomes for patients dealing with these issues.

### 4.4 Estimation of the Total Cost of Illness

**Annual Direct Expenditure:** The annual mean direct expenditure was estimated at 56.6 (ranging from 11.9 to 289.3) x ₹1000, indicating the financial burden borne by families dealing with Duchenne Muscular Dystrophy (DMD) or Becker Muscular Dystrophy (BMD). This direct cost accounted for approximately 52 percent of the total annual cost of illness. The most substantial cost component contributing to the per-person annual total cost of illness in routine care was the total direct expenditure, accounting for 51.7 percent. Notably, a substantial portion of this expenditure is allocated to direct medical management, encompassing expenses related to medicines, vaccines, diagnostics, surgical procedures, and other medical interventions. Most of the direct expenditure was driven by direct medical costs, making up 30.6 percent of the total COI. Comparatively, in Germany, the situation differs. The maximum expenditure for total direct medical costs of illness in Germany accounts for just 25 percent of the total COI. In Portugal this expenditure was 19 percent of total COI, as compared to the present cohort this ratio

(30.61) is quite less. In this context, a major proportion of direct medical costs is attributed to expenses related to medical aids ( Katz et al., 2014; Labisa et al., 2022).

This discrepancy in cost distribution between this study and Germany highlights the variation in healthcare expenditure patterns and highlights the relatively higher direct medical management costs faced by families dealing with Duchenne Muscular Dystrophy (DMD) in the Indian setting. Additionally, these costs are directly borne by the family as out-of-pocket (OOP) expenses, raising the likelihood of financial risk (Chiu et al., 2018). In contrast the OOP expenditures that were non-reimbursed payments were 6.9 percent in Germany, 7.3 percent in Italy, 5.0 percent in the UK and 7.0 percent in the US of total annual cost of illness (Landfeldt et al., 2014).

### 4.4.1 Direct Medical Expenditure

The direct medical expenditure specifically amounted to 33.5 x ₹ 1000, with a range from 5.6 to 157.3 x ₹ 1000. This category is about 31 percent of the total annual cost of illness in families providing routine care for DMD or BMD. That the basic treatment for BMD and DMD was found to be quite similar, resulting in comparable costs of illness. Approximately 45 percent of the direct medical cost was attributed to the expenses associated with medicines, indicating a substantial portion of the financial burden in this category. Additionally, rehabilitation constituted a significant chunk of the out-of-pocket expenditure by caregivers, accounting for approximately 27.7 percent (table 4.4.1). These findings highlight that families dealing with DMD or BMD face substantial financial challenges, especially when it comes to paying for medical treatments and rehabilitation.

The expenditure on lab and diagnostics was observed to be the least, primarily because many parents utilized the lab and diagnostic facilities provided by PGIMER. These government facilities offer services at highly subsidized rates. However, due to challenges like difficult access and long waiting periods, some parents opt for private facilities, resulting in increased out-of-pocket expenses. Direct medical costs remained relatively consistent across all clinical stages of DMD. It's important to note that there's limited data on the cost of illness in stage 5 as the study included only one patient from this category. This lack of data is due to the fact that most patients in this stage do not seek consultation often because they lack the necessary support systems. Traveling in

resource-limited public settings, which are not disability-friendly is incredibly challenging for them. Consequently, their quality of life is severely affected due to the absence of the medical and social support required for their basic well-being.

The cost of rehabilitation services represented a significant portion of the direct medical burden, accounting for 8.47 percent of the total cost of illness (COI). It's important to note that even though there were nine children who were wheelchair-bound, with an average age of 9.2 years, none of them had access to a wheelchair at the time of enrollment. Remarkably, only one out of these nine children received a wheelchair during the second phase of the study. This highlights that the costs associated with aids, such as wheelchairs, which are reflected in direct medical costs, were relatively low compared to the actual needs and demands of these patients.

Furthermore, no special medical costs were captured for stage 5, which requires basic life support. The study mainly focused on the standard medical care provided universally to all patients with DMD, potentially leaving out specific needs associated with more advanced stages of the disease. Major components that acted as drivers in costing were aids supporting mobility such as wheelchairs which, represented not only the most frequently used aids but also the most expensive devices (inclusive of other home care tools) that were not captured in the study. It's important to note that these costs related to mobility aids were not specifically captured in the study, highlighting their potential impact on the overall financial burden faced by families managing DMD or BMD. These findings highlight the critical importance of accessible healthcare and support systems in addressing the unique needs and challenges faced by patients in different stages of DMD. Additionally, they underscore the need for comprehensive healthcare services to provide essential aids and support, particularly in the later stages of the disease, to improve patients' quality of life.

### 4.4.2 Estimation of the Direct Non-Medical Cost of Illness

The direct medical costs specifically pertain to medical treatments and services. In contrast, total direct costs encompass both medical and non-medical costs associated with the disease. The costs include charges for personal assistance at school and work, travel, boarding & lodging, food, and informal expenses over traditional healers, over different religious rituals, and other informal payments during visits to health facilities. For instance, it is customary that gifts be given to the head of the household while visiting

someone's household in India. The gift could be in cash or kind. Whenever a patient makes a visit to the health center and stays at a relative's place, they spent some money on gifts (Chauhan et al., 2018). Apart from it, there are other miscellaneous informal costs like money spent on vehicle parking charges, heiring a wheelchair, etc. Although there was no significant difference in direct medical costs across the various stages of the disease the study found a significant increase in non-medical costs as the disease severity advanced. This increase was particularly pronounced when transitioning from DMD stage IV to stage V, as indicated by the ANOVA test (F=3.1). It's important to note that the direct non-medical expenditure in stage 1 was 38.6 (ranging from 11.0 to 64.7) x ₹ 1000, which was higher than the expenditure in stages 2 and 3. Frequent visits to healthcare institutions are crucial for accurately diagnosing conditions like DMD. These visits involve various assessments and tests. However, these repeated visits can contribute to the overall economic burden on patients and their families, especially when considering factors like travel, lodging, and related expenses. Patients in stage 1, typically those recently diagnosed with DMD, had frequent health facility visits primarily for diagnosis and medication initiation. These visits often required baseline workup procedures such as immunizations for pneumococcal and varicella, tuberculin tests, baseline ECHO, and more.

The major cost drivers in direct non-medical costs were food, travelling, boarding, and lodging for obtaining medical care. Specifically, the combined cost of travel, boarding, and lodging constituted about 14.3 percent of the total annual cost of illness. In stage 4, the mean cost of travel, boarding, and lodging was 38.8 x ₹ 1000, nearly three times that of stage 3. This cost increased significantly with the severity of the disease, as confirmed by the ANOVA test (F=5.1). The progression of the disease made movement increasingly difficult for patients, leading to longer durations of stay during medical visits. This posed challenges in using public transport systems, further increasing both transportation and lodging costs.

Whereas**,** one component of non-medical direct cost is "informal cost". In Indian culture, it's customary to give gifts, which can be in cash or kind, when visiting someone's home. The patients in this study incurred such informal costs when they stayed at a relative's place while visiting the health center. These expenses, including gifts and miscellaneous informal costs like car parking and obtaining a wheelchair, were

categorized under informal costs. House modifications made as per the child's needs were also included in this category, although only three caregivers incurred such costs.

Furthermore, expenditure on food increased as the severity of the disease advanced. The cost in stage IV was 9.5 x ₹ 1000, nearly three times higher than in stage III (3.4 x ₹ 1000). However, costs were not compared with stage 5 as there was only one patient in this group. This increasing trend in food expenditure was statistically significant with disease severity, as confirmed by the ANOVA test (F=2.8). These findings highlight how the economic burden associated with DMD is not solely driven by medical costs but also significantly impacted by non-medical costs, particularly as the disease progresses to more advanced stages. In this study, no one incurred non-medical expenses, such as house modifications, charges for personal assistance at school and work, or miscellaneous costs like artificial nutrition and alternative therapies. Informal care cost, as well-trained nurses are available in the community for the palliative care of Muscular dystrophy patients in few developed nations(Katz et al., 2014). In a study from Egypt only 1.9 percent of the total annual cost of illness was spent on home modifications related to child's illness (Shehata et al., 2023).

The direct non-medical cost in this study from Germany was less than 14 percent excluding informal care which was 27 percent. Thus, this 30.6 percent of total COI is directly on families as a financial burden. DMD is not covered under any government scheme for common people of India only few Government employees get reimbursements but none of study subject was beneficiary. The proportion of expenditure on account of direct non-medical expenditure was 21.1 percent in present study. This expenditure is 50 percent more than the expenditure found in the study from Germany. However, in the study cohort none of the patients incurred any expenditure for house or automobile modification. On the contrary none of the nine non-ambulatory patients of the present cohort have a access to even simple non-customized wheelchair. The Government supported provision for wheelchairs is grossly lacking with regard to coverage, design, production, and supply. Only 10 percent of the global disabled population requiring wheelchairs are provisioned with a wheelchair. By far, the greatest hurdle in the use of available assistive technology in patients with disability in low- and middle-income countries is an unsurmountable stigma associated with disability. (Guidelines on the Provision of Manual Wheelchairs in Less Resourced Settings., 2008; Barbareschi et al., 2021; Gupta et al., 2021).

**Table 4.4.1 Mean Annual Cost of Duchenne Muscular Dystrophy in ₹ x 1000 (min-max) on the Families with Routine Care (Phase I)**

| Variables | Mean | Ratio of total COI [%] | Clinical Severity | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | | I | II | III | IV | V | P- value |
| Drugs (Medicines +Vaccine) | 15.2 (2.0-58.4) | 13.93 | 15.4 (6.9-28.0) | 16.2 (4.0-58.4) | 14.8 (2.0-54.6) | 14.8 (6.4-29.6) | 11.7 | 0.399 |
| Diagnostic (Lab tests+ ECHO+Xray+DEXA) | 2.1 (0-17.0) | 1.93 | 2.0 (0-4.7) | 2.6 (0-17.0) | 2.0 (0-9.2) | 1.3 (0-3.9) | 1.5 | 0.583 |
| Procedure orthotic User fee | 6.9 (0-60.9) | 6.28 | 5.1 (3.9-6.2) | 5.1 (0.07-15.7) | 8.2 (0.01-60.9) | 6.3 (0.01-10.8) | 30.4 | 0.343 |
| Costs of rehabilitation services | 9.3 (0-104.3) | 8.47 | 31.5 (0-70.2) | 5.2 (0-44.3) | 10.0 (0-104.3) | 13.3 (0-48.1) | 0 | 0.254 |
| Direct medical expenditure | 33.5 (5.6-157.3) | 30.61 | 54.0 (12.1-106.8) | 29.0 (9.1-105.5) | 35.0 (5.6-157.3) | 35.7 (11.0-70.4) | 13.2 | 0.573 |
| Costs for “personal assistance” at school | 1.6 (0-60.8) | 1.45 | - | 0.4 (0-12.2) | 2.6 (0-60.8) | - | - | 0.655 |
| Travel_Boarding/Lodging | 15.6 (0.2-167.0) | 14.30 | 32.2 (8.8-61.7) | 11.8 (0.2-36.7) | 13.8 (0.3-51.9) | 38.8 (2.7-167.0) | 3.6 | 0.003 |
| Food and other costs | 3.8 (0-34.1) | 3.44 | 5.2 (1.3-12.2) | 2.8 (0-10.6) | 3.4 (0-34.1) | 9.5 (0-29.3) | 1.5 | 0.045 |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| Direct Informal (traditional healing) | 2.1 (0-46.0) | 1.93 | 1.2 (0.9-2.0) | 0.9 (0-5.2) | 2.6 (0-46.0) | 3.9 (0-19.5) | 912 | 0.459 |
| Direct non-medical expenditure in INR | 23.1 (0.6-195.9) | 21.11 | 38.6 (11.0-64.7) | 16.0 (0.7-47.3) | 22.5 (0.6-132.0) | 52.2 (2.9-195.9) | 6.1 | 0.01 |
| Total direct expenditure | 56.6 (11.9-289.3) | 51.72 | 92.7 (52.5-171.5) | 45.0 (11.9-125.1) | 57.5 (12.4-289.3) | 87.9 (13.9-218.0) | 43.3 | 0.324 |
| $LP_{(unpaid\ caregiving)}$ | 33.5 (7.3-88.2) | 30.6 | 28.2 (18.5-44.1) | 31.7 (7.3-66.1) | 33.5 (7.3-88.2) | 39.4 (26.3-53.9) | 58.8 | 0.424 |
| $LP_{(absenteeism)} + LP_{(changes)}$ | 19.3 (0.0-311.6) | 17.7 | 3.5 (0.0-7.0) | 21.2 (0.0-278.2) | 21.5 (0.0-311.6) | 4.5 (0.0-31.7) | 1.8 | 0.918 |
| Total indirect COI | 52.8 (14.7-352.9) | 48.28 | 31.7 (18.6-47.6) | 52.9 (14.7-322.3) | 55.0 (14.7-352.9) | 44.0 (26.3-68.5) | 62.1 | 0.844 |
| Total annual COI | 109.4 (27.4 – 534.2) | 100 | 124.4 (71.0-200.5) | 97.9 (27.4-370.0) | 112.5 (34.0-534.2) | 131.8 (81.7-244.4) | 78.0 | 0.735 |

***Source:*** *Authors calculation established on primary data*

The major source of direct non-medical costs in this study from Germany was informal care cost which accounted for 27 percent of the total COI. The study found that informal care costs emerged as the most significant driver of direct resource consumption, particularly in severe dystrophinopathies. These costs also exerted a notable influence on the overall healthcare burden. Informal care which involved family members and friends providing medical services, was assessed as the time spent by non-working caregivers effectively resulting in a loss of their leisure time. These costs were considered as the genuine expenses associated with caregiving, measured by comparing them to the customary payment for formal care services. Consequently, the study categorized informal care costs as part of the direct costs (Katz et al., 2014).

The cost of informal care has been classified as care provided by unpaid caregivers as part of the indirect cost category. There are two main reasons for this classification. Firstly, the caregivers in the study typically lack the extensive training or medical education that formal caregivers possess, unlike the unpaid caregivers in a home setting. Secondly, non-availability of the price of a close market substitute for formal caregivers specifically trained to care for patients with DMD or other chronic pediatric illnesses in the study context (table 4.4.1). The leisure time of caregivers sacrificed in caregiving as they were engaged in caregiving activities, which primarily involve self-care for daily routines of the patients. This leisure time sacrificed is captured as a loss of productivity due to willingness to pay and as such, it is categorized as part of the indirect cost of illness (COI). This approach aims to capture the economic impact of informal caregiving on caregivers' productivity in the present study

### 4.4.3 Estimation of the Annual Indirect Cost of Illness

The indirect cost is the productivity loss due to the illness. This loss stems from a reduction in a person's ability to work due to the disease. It can manifest as absenteeism from work or a change in one's work situation. Sometimes, it's related to spending leisure time on caregiving responsibilities. Caregiver’s "willingness to pay" is essentially how much leisure time they're willing to sacrifice for caregiving to avoid undesirable outcomes. It reflects the value they place on caregiving responsibilities in the face of challenges posed by the illness

(Steigenberger et al., 2022). Willingness to pay is a way to measure the indirect cost resulting from the loss of productivity due to the illness of the patient or the caregiver's ability to work. Salaried caregivers typically receive paid leave from their employers, which is beneficial for both employee productivity and the employer. These leaves are crucial for parents too, as spending time with their children offers numerous benefits("Annual leave: Why you should use it up, and how to make the most of it" 2022).

When a caregiver takes leave to care for their child, they're essentially sacrificing their leisure time, resulting in a loss of productivity. This is known as long-term unpaid caregiving. Other family members also play a role in caring for the affected child, termed as short-term unpaid caregiving. The total hours of short-term caregiving were converted into days. To calculate the indirect cost, both long-term and short-term unpaid caregiving days were added together and multiplied by the Gross Domestic Product (GDP) per capita per day in India. Considering the productivity of leisure time to be the Gross Domestic Product (GDP) of India.

Besides from caregivers willingness to pay, indirect costs also consider the loss of working capacity due to illness, "lost working time," and "loss of productivity. Over study captured the indirect cost in two sections: a) calculating caregivers' willingness to pay for their unpaid care and b) calculating the lost working time of parents or caregivers who had to quit or reduce their employment due to their child's illness. It's important to note that the working time of patients wasn't captured as none of them were in the working age group("Human Capital Approach Human capital approach," 2008). The study has not captured the working time of patients as none of the patients is in the working age group.

**a) Estimation of Indirect Costs caused by "Willingness to Pay"**

Around 33 percent of the caregivers in the study were employed and were able to take paid leaves granted by their employers. There was no wage loss during their hospital visits and also so whenever they took leave for long-term caregiving for their DMD/BMD son. Additionally, approximately 46 percent of the families in the study were joint families. In such families, the responsibility of caregiving is not solely shouldered by the parents but is

often shared among various family members. Apart from their parents their grandparents, uncles, aunties, cousins, and other family members. In this study, the primary caregivers who visited the health facility for consultations were not always the child's parents. Approximately 35 percent of households involved grandparents in caregiving, alongside the parents. In about 8 percent of households, uncles and cousins played a role in caregiving. In 7 percent of cases, elder siblings were responsible for caregiving.

This diversity in caregiving arrangements highlights the wide spectrum of caregiving givers in India, where the responsibility for caring for a child with DMD/BMD extends beyond just the immediate family. Despite the importance for human development, care work often goes unrecognized. This is partly because, being unpaid, it is not reflected in economic indicators such as GDP. But valuing unpaid care work would highlight women's contributions in households and communities and draw attention to their material conditions and well-being, with a possible implication for policymaking. In India unpaid care is estimated at 39 percent of GDP (Human Development Report, 2015).

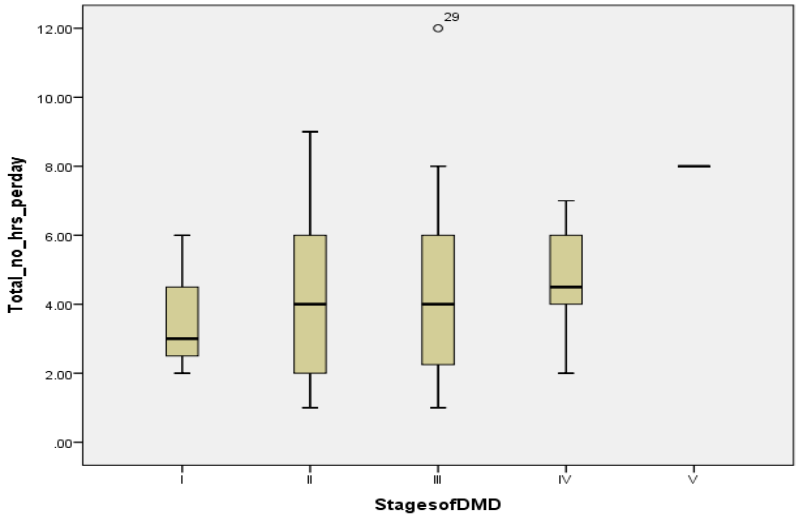
The hours of caregiving were later converted into days and combining both long-term and short-term unpaid caregiving to get a total count of caregiving days. This helped us understand the overall time and effort invested by unpaid caregivers. The study valued the productivity of leisure time using India's Gross Domestic Product (GDP). To calculate the economic impact, the total number of unpaid caregiving days were multiplied by the per capita GDP per day. This approach helped us quantify the loss of productivity in monetary terms. To prevent overestimation of time, primary caregivers were asked about the average number of hours in a day provisioned exclusively to informal care, discounting working hours (if working) and/or leisure time as in an earlier study from Portugal (Labisa et al., 2022).

Informal care in Western setup is categorized in direct cost and it was found to be the major deciding factor for resource consumption mainly in severe disease and had a notable influence on the total healthcare burden. However, in this particular study informal care given by unpaid caregivers is captured as indirect cost of illness as this is caregiver's willingness to pay their leisure time in caregiving leading to loss of productivity. In another

study from Portugal, a proxy good method was used to value the informal costs. Here the average wage paid to the patient care giver was compared to the close market substitute according to the Portuguese neuromuscular patients association (APN) and was used to estimate the effect of indirect wage loss. This is the same method that was adopted by the study from Germany discussed above. In the study from Germany, informal care was about 27 percent of the total COI and informal care cost for ambulatory patients in Portugal was estimated to be 50 percent of the total COI. The productivity loss due to informal caregiving is about 30 percent of the total cost of illness (table 4.4.1). This is the most important cost-driver of the indirect cost of illness. This cost increased with the severity of the disease but the finding was not clinically significant. The same pattern was found in the German study where the cost of care increased with a variety of clinical phenotypes whereas informal care cost was less for the non-ambulatory patients (42 percent of total COI) in the study from Portugal.

As different studies have adopted different assumptions to calculate the informal care cost so it is difficult to compare the data. The mean number of hours dedicated to informal care would be a better entity for comparison purposes. This informal care must have two subdivisions paid informal care and unpaid informal care. Thus, paid informal care will be projected in the direct cost of illness while unpaid informal care will be reflected as a loss of productivity (willingness to pay) as an indirect cost. The mean number of hours per day was 9.4 ±10.9, 6.1 ± 2.0 and 4.3±2.1 for German, Portuguese, and the present study respectively. Unpaid caregiving in the study is almost half of the German study and 30 percent less than the Portuguese study although the findings are not significant. The mean number of hours spent in care efforts per day increases with the severity of the disease in the present study. However, this finding is not statistically significant as most of the study population was ambulatory (91 percent). The same pattern was seen in the study from Germany and Portugal. In the present study total mean hours per day in caregiving were 4.3 ±2.1 hrs and 31 percent of the children were getting care for 6 hours or more. Although the mean number of hours spent in care efforts per day increased with the severity of the disease the findings were not statistically significant. (table: 4.3.2)

**Figure: 4.4.1 Time (hours) spent in caregiving per day with the severity of the disease**




***Source:*** *Authors calculation established on primary data*

Another component of the indirect cost of illness is loss of productivity due to absenteeism and change in working conditions leading to reduced wage. This proportion was 17.7 percent of total COI and comparable to the cost incurred in the study from Portugal (14 percent) while this is almost double the cost in Germany (9 percent). These outcome measures showed that the loss of productivity of caregivers due to DMD is quite high in India. Present cohort has none of the patients in the economically productivity group (pediatric age group) so it is difficult to comment upon the loss of productivity due to the illness of the patients. Therefore, only productivity loss considered was related to parents missing work or changes in caregivers' work schedules. However, it is evident from the data that caregivers in India have been overburdened with caregiving as compared to European countries.

The overall indirect cost includes loss of productivity due to absenteeism and change in working conditions for both patients and parents in Germany 36 percent of total COI which is much higher than the study from Portugal which is 22 percent in the ambulatory phase and in this study, it was 17.7 percent In the study, the entire burden is borne by parents and this enforces the need to explore ways to reduce the burden on caregivers thereby reducing the indirect cost of illness. The same correlation was seen in the study from the US

where there was a direct correlation between the loss of income and the amount of care needed by the patient(Larkindale et al., 2014). The mean annual cost for unpaid caregiving was ₹33.5 (7.3-88.2) x 1000. The cost of caregiving persistently increased with the "severity of the disease" still there was no significant difference noticed. This cost is almost 30 percent of the total COI equal to the "total direct medical cost".

b) **Estimation of Indirect Costs Caused due to Changed Working Capacity**

In the present cohort, 24 percent of the parents either graduated or have a higher university education. However, around 41 percent of the caregivers have daily waged Laboure's or have a petty business. For this group, taking leave from their jobs for hospital visits or to care for their child resulted in wage loss, affecting their income. Approximately 67 percent of caregivers experienced wage loss while caring for their child or visiting the hospital for consultations. During Phase I, caregivers spent an average of 6.6 days getting medical consultations. The mean annual loss of productivity due to absenteeism with routine care amounted to ₹ 3,365. Alarmingly, around 9 percent of mothers in this study group had to quit their jobs in order to provide proper care for their sons. The mean annual "loss of productivity" due to the loss of work was even higher, reaching ₹ 11,597 with routine care.

### 4.5. Determinant of Economic Burden of DMD

To understand the key factors influencing the cost of treatment, it's vital to study various elements that can impact the treatment expenses. These factors include disease severity, disease type, barriers to accessing care, knowledge about care, as well as other sociodemographic and clinical factors.

#### 4.5.1 Association of Health Expenditure with Clinical Severity of Disease

When analyzing the influencing factors of direct and indirect costs from various perspectives (patients, caregivers, and families), Katz et al., found significantly higher mean costs for Duchenne Muscular Dystrophy (DMD) compared to this study ($p < 0.001$). Notably, in the most severe clinical stage, the direct cost of illness (Direct COI) for DMD was highest,

nearly three times higher than that for Becker Muscular Dystrophy (BMD).(Landfeldt et al., 2014; Katz et al., 2014).

However, when compared mean costs between BMD and DMD, the total annual cost of illness (COI) with the intervention was higher for DMD, along with informal annual payments. Specifically, the total COI for DMD was found to be 8 times higher than that for BMD in the cohort. These variations may be attributed to differences in the study populations, healthcare systems and methodologies employed in the two studies.

Health care utilization and expenditures for children and young adults with muscular dystrophy in a privately insured population was studied by Ouyang et. al. (2008). In the study average medical expenditures was noticed to be 10 to 20 times higher in individuals with muscle dystrophy when compared to individuals without muscular dystrophy. The highest number of inpatient admissions related to respiratory infections and cardiac complications was seen in individuals aged 15 to 19 years. The findings underscore the need for appropriate treatment options for individuals with muscular dystrophy as they age (Ouyang et.al., 2008). There are several discrepancies comparing the present results to those of previous studies, since different national healthcare systems or different types of muscular dystrophies with inhomogeneous patient cohorts were analyzed which do not allow a close comparison (Ouyang et al., 2008; Larkindale et al., 2014).

### 4.5.2 Association of Health Expenditure with Family Structure

The indirect cost of caregiving by unpaid caregivers annually without intervention is higher in joint families as compared to nuclear families. The loss of productivity in caregiving by unpaid caregivers in the Indian scenario where people still live in joint families with their grandparents, uncles and aunts appears to be more but this loss of productivity is shared by all the family members. Thus, the actual productivity loss per person is quite less in joint families. Kumar, et al (2013) restored the connection between family structure and child health. The descriptive statistics revealed that children from non-nuclear families exhibited better nutritional status and higher immunization coverage in comparison to children in nuclear families. These findings suggest a potential association between family

structure and child health outcomes (Kumar & Ram, 2013). When compared between loss of work as a challenge to health facility visits, those caregivers who found the loss of their professional work as a barrier to visiting the health facility were spending less on direct treatment costs. Indirect costs caused by parents due to loss of work/absenteeism were just 6 percent in Germany as compared to this study (17.7 percent). Hence, the loss of work is the major bottleneck to visiting health facilities.

**Table 4.5.1 Parameters Affecting the Economic Burden ₹ x 1000(min-max) of DMD/BMD**

| **Family Structure** | **Mean (Nuclear)** | **Mean (Joint)** | **t stats** | **p-value** |
|---|---|---|---|---|
| Indirect Cost Caregiving Phase1 annual | 42.5 (14.7-251.2) | 64.9 (14.7-352.9) | -1.84 | 0.07 |
| **Work loss major challenge in PGI Visits** | No | Yes | | |
| Direct Cost on Diagnostic Phase1@annual | 3.0 (0.0-12.0) | 1.8 (0.0-17.0) | 1.99 | 0.049 |
| Direct Cost on Informal Payment in Phase1@annual | 4.0 (0.0-46.0) | 1.4 (0.0-23.3) | 2.02 | 0.046 |

***Source:*** *Authors calculation established on primary data*

Interestingly, study found that joint families incurred higher expenses in indirect caregiving compared to nuclear families. Moreover, when comparing the challenge of work loss during visits to PGI, statistically significant differences ($p<0.05$) were observed in areas: Direct cost for diagnosis in phase-I and direct cost for annual informal payments in phase 1 for food. In all these cases, there was a noticeable increase in expenditure associated with the work loss challenge during PGI visits. Additionally, the expenditure for annual indirect costs for caregiving was higher when prenatal diagnosis was not explained and this difference was statistically significant ($p<0.05$). These findings highlight how various factors, including family structure, work loss challenges, and the provision of information about prenatal diagnosis, can significantly influence the overall cost of treatment.

**Table 4.5.2 Annual economic burden in routine care (Phase I) vs States (Mean ₹ per 1000 (min-max)**

| Vs State | CHD (Tricity) | PB | HR | Eastern states | UK | HP | J&K others | p-value |
|---|---|---|---|---|---|---|---|---|
| DC Drugs | 11.7 (3.8-21.0) | 13.0 (2.0-33.8) | 17.0 (4.0-46.2) | 11.0 (4.4-15.2) | 28.4 (11.7-33.8) | 17.0 (4.4-54.7) | 19.0 (10.4-29.6) | 0.045 |
| DC Travel Boarding Lodging | 1.8 (0.2-4.7) | 11.9 (3.5-41.0) | 8.2 (1.8-31.1) | 14.4 (1.6-28.4) | 20.0 (3.7-51.5) | 20.7 (23.7-61.7) | 43.0 (5.6-167.0) | 0.00 |
| DC FOOD informal other payment | 0.7 (0.0-1.9) | 2.8 (0.0-11.2) | 2.0 (0.0-9.5) | 1.0 (0.0-2.2) | 1.8 (1.1-2.8) | 5.7 (0.0-34.1) | 12.9 (0.0-29.3) | 0.00 |
| Direct non-medical expenditure | 9.3 (0.5-65.5) | 18.9 (4.6-71.6) | 11.1 (2.4-38.2) | 17.0 (24.1-30.6) | 31.7 (6.1-77.7) | 30.7 (4.1-132.0) | 59.4 (10.1-195.9) | 0.00 |
| Total Direct Cost | 39.3 (12.4-207.9) | 49.1 (13.1-160.0) | 45.0 (11.9-131.2) | 35.2 (21.5-53.0) | 81.1 (19.3-125.1) | 73.2 (14.6-289.3) | 101.3 (32.3-218.0) | 0.008 |

CHD Tricity: Chandigarh, Sahibzada Ajit Singh Nagar, Panchkula; PB: Punjab; HR: Haryana; UK: Uttarakhand;
HP: Himachal Pradesh; J&K: Jammu and Kashmir

***Source:*** *Authors calculation established on primary data*

These findings highlight significant variations in expenditure patterns based on the place of residence, with different regions in India experiencing varying costs associated with the care and treatment of Duchenne patients.

### 4.5.3 Association of Health Expenditure with Socio-economic Status and determinants

The annual total direct cost of illness and indirect cost of illness with routine care cost were directly associated with the "socio-economic status" of the families in the cohort, when

binary correlation was assessed. This association was significant with P=0.006 and P=0.013 respectively. In India, public hospitals are often preferred by poor-illiterate with no medical insurance as it provides affordable medical services although high indirect cost due to loss of productivity and access barriers was a downside. But this is not the case with DMD as the site is one of the premier institutes of medical education and research in India so Caregivers from all social and economic groups visited the facility equally. People with high opportunity costs and higher social backgrounds were more serious about treatment. However, few caregivers with higher education mainly postgraduates wished not to seek any drug treatment for their sons as they were aware of the side effects of steroids and have knowledge that no definite treatment is available (Patra & Bandyopadhyay, 2020). The determinants of economic burden on the families identified are travelling distance from the main health care facility, socio-economic status of the family, and type of disease (Duchenne/Becker's or intermediate).

### 4.5.4 Multiple Linear Regression (MLR)

A multiple linear regression (MLR) was conducted to estimate the impact of exposure while accounting for the influence of other variables, thereby controlling for potential confounding factors. Multiple linear regression was employed to assess the influence of different predictors on the total cost of illness, total direct cost of illness and total indirect cost of illness. The predictors used were SES, child age, educational status of main caregiver, family structure, locality (urban/rural), distance of provinces to the health facility, main caregiver, parent’s knowledge of DMD clinical severity and HrQoL (EQ 5D utility index), Overall QoL caregiver, who noticed first and age when first noticed.

**Direct Cost of Illness**

**Table 4.5.3 Multiple linear regression analysis for determinants of Annual Direct cost of illness (routine care).**

| Model Summary | | | | | | |
|---|---|---|---|---|---|---|
| **Model** | **R** | **$R^2$** | **Adjusted R2** | **SE of the estimate** | **F= MS regression/ MS residual** | **Sig** |
| 1 | 0.776 | 0.601 | 0.546 | 37560.6968 | 10.942 | 0.00 |
| **Coefficients** | | | | | | |
| **Variables** | | | **β** | **Std. Error** | **t** | **value** |
| Constant | | | | 36419.965 | -1.341 | 0.183 |
| SES** | | | 0.730 | 611.989 | 6.499 | 0.000 |
| Age | | | -0.165 | 2346.104 | -1.929 | 0.057 |
| Education Caregiver** | | | 0.270 | 3966.357 | 2.450 | 0.016 |
| Family structure | | | -0.041 | 8418.696 | -0.542 | 0.590 |
| Locality (urban/rural)** | | | -0.193 | 9092.974 | -2.506 | 0.014 |
| parent's knowledge of DMD | | | -0.117 | 7888.912 | -1.653 | 0.102 |
| Distance of provinces** | | | 0.393 | 5148.477 | 5.207 | 0.000 |
| Main caregiver | | | 0.049 | 6904.375 | 0.686 | 0.495 |
| EQ 5d index baseline | | | -0.029 | 12317.870 | -0.364 | 0.717 |
| Overall QoL caregiver** | | | -0.168 | 145.227 | -2.249 | 0.027 |
| Who noticed first | | | 0.086 | 3340.351 | 1.188 | 0.238 |
| Age first noticed | | | 0.065 | 5465.094 | 0.831 | 0.408 |
| Cooks distance statistic (minimum=0.000; maximum=0.292) | | | | | | |
| ** Two-tailed significant values | | | | | | |

***Source:*** *Authors calculation established on primary data.*

After adjusting for confounding factors using multivariate analysis, it was found that the annual direct cost of illness was significantly positively associated with the family's

socioeconomic status, the education level of the primary caregiver, and the distance of their residence from the health facility. Conversely, the direct cost of illness was negatively associated with the overall quality of life of the primary caregiver. Although the child's age is not significantly associated with the cost of illness, the relationship is negative. This suggests that as the child ages or as the disease worsens, the direct cost of illness tends to increase.

Higher socioeconomic status and education levels often result in increased spending on disease management due to access to more resources. Additionally, challenges in accessing healthcare facilities, such as distance or availability, can elevate treatment costs, underscoring the need for equitable healthcare access.

The MLR model for Total Cost of Illness considered predictors such as SES, child age, educational status of main caregiver, locality (urban/rural), distance of provinces to the health facility, main caregiver, parent’s knowledge of DMD clinical severity and HrQoL indexes of children, Overall QoL caregiver, health related QoL of caregiver, who noticed first and age when first noticed to assess their influence. The total cost of illness (COI) for families with a child affected by DMD under routine care is significantly influenced by socio-economic status (SES), residential distance from the health facility, and urban living. COI increases with greater distance from the health facility. Surprisingly, an inverse association with age was observed, possibly due to challenges faced by non-ambulatory patients. Additionally, advanced-stage patients were underrepresented, likely due to mobility constraints.

**Table 4.5.4 Multiple linear regression analysis for determinants of Annual Total cost of illness (routine care).**

| Model Summary | | | | | | |
|---|---|---|---|---|---|---|
| **Model** | **R** | **$R^2$** | **Adjusted R2** | **SE of the estimate** | **F= MS regression/ MS residual** | **Sig** |
| 1 | 0.751 | 0.564 | 0.487 | 79871.2679 | 7.254 | 0.00 |
| **Coefficients** | | | | | | |
| **Variables** | | | **β** | **Std. Error** | **t** | **value** |
| Constant | | | | 109485.497 | 0.920 | 0.360 |
| SES** | | | 0.532 | 1289.419 | 4.602 | 0.000 |
| Age** | | | -0.324 | 5256.274 | -3.477 | 0.001 |
| Education Caregiver | | | -0.036 | 16914.868 | -0.321 | 0.749 |
| Locality (urban/rural)** | | | -0.203 | 18954.237 | -2.530 | 0.013 |
| parent's knowledge of DMD | | | -0.058 | 17009.664 | -0.757 | 0.451 |
| Distance of provinces** | | | 0.223 | 11136.723 | 2.759 | 0.007 |
| EQ 5d index baseline | | | -0.076 | 31453.340 | -0.757 | 0.451 |
| Overall QoL caregiver | | | -0.068 | 361.423 | -0.804 | 0.423 |
| Clinical severity | | | -0.050 | 32373.073 | -0.603 | 0.548 |
| Age first noticed | | | 0.139 | 11565.097 | 1.685 | 0.096 |
| HRQoL of caregiver | | | -0.102 | 258.483 | -1.271 | 0.207 |
| Who noticed first | | | -0.058 | 7275.877 | -0.734 | 0.465 |
| PeadsQoL (Physical Health) | | | -0.037 | 612.830 | -0.380 | 0.705 |
| Main caregiver | | | -0.089 | 14609.565 | -1.175 | 0.243 |
| Affected Brothers | | | -0.145 | 28940.601 | -1.854 | 0.067 |
| ** Two-tailed significant values | | | | | | |
| Cooks distance statistic (minimum=0.000; maximum=0.322) | | | | | | |

***Source:*** *Computer based outputs established on primary data.*

The total cost of illness (COI) for families with a child affected by DMD highlights significant disparities in healthcare access and affordability. Socio-economic status (SES) plays a crucial role, with higher costs burdening wealthier families more, while those from

lower SES backgrounds might struggle with treatment affordability. The increased COI with greater distance from health facilities underscores the challenge of geographical accessibility, particularly in rural or underserved areas. The inverse relationship with age may reflect the compounding difficulties faced by non-ambulatory patients, who encounter barriers to continuous care as their condition worsens. This suggests a need for targeted public health interventions, such as improved transportation services, telemedicine, and financial support, to ensure equitable access to care for all families affected by DMD, regardless of their socio-economic status or location.

A multiple linear regression model for indirect cost of illness considered predictors such as child's age, educational status of main caregiver, family structure (nuclear/joint), distance of provinces to the health facility, main caregiver, parent's knowledge of DMD, and HrQoL indexes of children, Overall QoL caregiver, health related QoL of caregiver, who noticed first and age when first noticed to assess their influence. The indirect cost of illness for families with a child affected by DMD was significantly influenced by the educational status of the primary caregiver. Caregivers in urban areas, with better access to health facilities, often spend more time on caregiving, leading to higher indirect costs from lost productivity. In joint families, where multiple caregivers are involved, productivity loss is further amplified. Families traveling from distant locations incur additional wage loss due to the time spent on healthcare visits. Furthermore, the child's age or disease progression and their health-related quality of life are inversely related to the total indirect cost of illness.

**Table 4.5.5 Multiple Linear Regression Analysis for Determinants of Annual Indirect Cost of illness (routine care).**

| Model Summary | | | | | | |
|---|---|---|---|---|---|---|
| **Model** | **R** | **$R^2$** | **Adjusted R2** | **SE of the estimate** | **F= MS regression/ MS residual** | **Sig** |
| 1 | 0.776 | 0.601 | 0.552 | 40878.66433 | 12.075 | 0.00 |
| **Coefficients** | | | | | | |
| **Variables** | | | **β** | **Std. Error** | **t** | **value** |
| Constant | | | | 28401.324 | 5.262 | 0.000 |
| Age** | | | -0.279 | 2505.000 | -3.451 | 0.001 |
| Education Caregiver** | | | -1.021 | 8890.983 | -8.433 | 0.000 |
| Family structure** | | | 0.266 | 13548.796 | 2.381 | 0.019 |
| Distance of provinces** | | | 1.016 | 9032.645 | 8.044 | 0.000 |
| parent's knowledge of DMD | | | -0.071 | 8633.827 | -1.001 | 0.320 |
| EQ 5d index baseline | | | -0.241 | 13559.068 | -3.043 | 0.003 |
| Age first noticed | | | -0.052 | 13863.249 | -0.459 | 0.647 |
| HRQoL of caregiver | | | -0.059 | 133.399 | -0.780 | 0.437 |
| Overall QoL caregiver | | | -0.020 | 159.421 | -0.268 | 0.789 |
| Who noticed first | | | 0.021 | 3761.875 | 0.279 | 0.781 |
| Main caregiver | | | -0.118 | 7307.264 | -1.714 | 0.090 |
| ** Two-tailed significant values | | | | | | |
| Cooks distance statistic (minimum=0.000; maximum=0.428) | | | | | | |

***Source:*** *Computer based outputs established on primary data.*

*The educational status of the primary caregiver plays a significant role. Those with higher levels of education often engage more deeply in caregiving leading to increased productivity losses. Caregivers in urban areas have better access to health facilities and spend more time managing their child's care, thereby experiencing higher indirect costs from lost productivity. This contrasts with caregivers in rural areas who might face different challenges. Furthermore, in joint families where multiple caregivers are involved in caregiving intensifies productivity loss. Families travelling from distant locations incur*

*additional wage losses and productivity costs due to the time required for healthcare visits, highlighting the impact of access issues. Conversely, a better health-related quality of life in children is associated with lower indirect costs. These factors collectively illustrate how socio-economic, geographical, and personal variables impact the financial burden on families dealing with DMD emphasizes the need for targeted public health strategies to mitigate these indirect costs.*

### 4.6 Financial Risk Protection or Catastrophic Health Expenditure

The World Health Organization (WHO) defines health expenditure as catastrophic when it surpasses 40 percent of a household's remaining income after covering basic subsistence needs. Financial risk is evaluated based on catastrophic health expenditure (CHE) and distress financing. In the context of expenditures on Duchenne Muscular Dystrophy (DMD) treatment, if it exceeds the 40 percent threshold of non-food household consumption expenditure, it is considered catastrophic health care expenditure. Among the recruited patients, 40 percent (n = 100) suffered from catastrophic health expenditure at the 40 percent threshold. The prevalence of catastrophic expenditure changed to 76 percent, 63 percent, and 29 percent when the threshold for catastrophic expenditure was taken as 20, 30, and 50 percents respectively. Forty five percent of the patients reported having faced distress financing. The association of catastrophic health expenditure and distress financing with their various predictors of cost of care such as the age of the patient, progression of the disease, age when noticed the symptoms, socio-economic status of caregiver, knowledge about the disease, educational status of caregiver, provinces from where the patient is traveling to the health facility, locality and family structure are presented in the table 4.6.1

**Table 4.6.1 Association of catastrophic health expenditure and distress financing with various predictors of COI**

| **Predictors of COI** | | **Catastrophic health Expenditure N(%)** | **p value (χ2 test)** | **Distress financing N(%)** | **p value (χ2 test)** |
|---|---|---|---|---|---|
| Age of the patient | <=9 Years | 31 (77.5) | 0.775 | 35 (77.8) | 0.707 |
| | > 9 Years | 9 (22.5) | | 10 (22.2) | |
| Stage of disease | Pre or early ambulatory | 11 (27.5) | 0.34 | 16 (35.5) | 0.99 |
| | Late ambulatory | 24 (60.0) | | 25 (55.6) | |
| | Non-ambulatory | 5 (12.5) | | 4 (8.9) | |
| Age when noticed the symptoms | Up to 5 years of age | 14 (35.0) | 0.001 | 17 (37.8) | 0.002 |
| | After 5 years of age | 26 (65.0) | | 28 (62.2) | |
| Socio-economic status | Upper | 0 (0) | 0.037 | 2 (4.4) | 0.105 |
| | Middle | 19 (47.5) | | 19 (42.2) | |
| | Lower | 21 (52.5) | | 24 (53.3) | |
| Educational status of caregiver | At least graduate | 9 (22.5) | 0.67 | 11 (24.4) | 0.24 |
| | Matriculation | 17 (42.5) | | 17 (37.8) | |
| | At least graduate | 14 (35.0) | | 17 (37.8) | |
| knowledge about the disease | Yes | 26 (65%) | 0.003 | 26 (57.8) | 0.051 |
| | No | 14 (35%) | | 19 (42.2) | |
| Locality | Urban | 9 (22.5) | 0.317 | 9 (20.0) | 0.107 |
| | Rural | 31(77.5) | | 36 (80.0) | |
| Family structure | Nuclear | 21 (52.5) | 0.806 | 22 (48.9) | 0.35 |
| | Joint | 19 (47.5) | | 23 (51.1) | |
| Distances of provinces to the health facility | Nearby Travel | 16 (20.0) | 0.000 | 21 (46.7) | 0.02 |
| | Nearby Difficult Travel | 8 (20.0) | | 9 (20.0) | |
| | Farthest travel | 16 (40.0) | | 15 (33.3) | |
| Catastrophic health expenditure | Yes | NA | NA | 31 (68.9) | 0.00 |
| | No | NA | | 14 (31.1) | |
| Distress financing | Yes | 31 (77.5) | 0.00 | NA | NA |
| | No | **9 (22.5)** | | **NA** | |

**Source:** Authors calculation established on primary data.

The CHE was significantly less among patients belonging to higher socio-economic status as compared to lower socio-economic (χ2 6.6; $p < 0.037$), caregivers ignorant to the outcome of disease (χ2 8.8; $p < 0.003$) and traveling from nearby provinces (χ2

15.2; p < 0.000), and parent who noticed symptoms up to 5 years of age ($\chi 2$ 10.8; p < 0.001). The caregivers ignorant about what is the prognosis of the disease are less liable to have CHE as compared to patients having knowledge about the disease outcomes (OR 0.3; 95% CI 0.125– 0.671), caregivers who noticed the disease after the age of 5 years are 4 time more liable to get into CHE (OR 4.0; 95% CI 1.72– 9.35), Patient who are traveling from farthest places to take health care from PGI are 8.4 time more susceptible to get into CHE (OR 8.0; 95% CI 2.64– 26.7) as compared to people traveling from nearby provinces, Susceptibility to get into CHE is 11 times more in the patients who are going through distress financing ( OR 11.3; 95% CI 4.36 – 29.36). Caregivers who noticed the disease after the age of 5 years are 4 times more liable to get into DF (OR 3.7; 95% CI 1.6–8.4). The patient who are traveling from farthest places to take health care from PGI is 4.4 times more likely susceptible to get into DF (OR 4.4; 95% CI 1.48– 13.1) as compared to people traveling from nearby provinces.

The results at 40% threshold, showed that the odds of catastrophic expenditure was significantly lesser for those were ignorant about the disease (OR: 0.2, 95% CI: 0.1– 0.5, p-value: = 0.002), as compared to caregivers having knowledge about the disease. (table 4.6.2) Secondly, those who were traveling from far-flung areas were significantly 5.6 times more prone to face CHE as compared to patients traveling from nearby areas (OR: 5.6, 95% CI: 1.4 – 23.2, p-value: = 0.02). The odds of CHE were 5.1 times more in patients with CHE (OR 5.1 95% CI: 1.8 – 15.0). The risk of distress financing was higher among patients whose parents noticed the symptoms in later ages (after 5 years). This risk was 2.7 times higher (OR 2.7. 95% CI: 1.0 – 7.5). The odds of distress financing were 7.3 times more in patients with CHE (OR 7.3 95% CI: 2.4 – 22.3). The severity of the disease (progression of disease/age of patient), locality of the household presence of any insurance/subsidy entitlement, and socio-economic status did not alter the risk of suffering from catastrophic expenditure and distress financing.

**Table 4.6.2 Adjusted odds ratios for association of catastrophic health expenditure and distress financing with predictors of COI**

| Predictors of COI | | Catastrophic health Expenditure N (%) | | Distress financing N (%) | |
|---|---|---|---|---|---|
| | | OR (95% CI) | P value | OR (95% CI) | P value |
| Age when noticed the symptoms | Up to 5 years of age | - | | - | |
| | After 5 years of age | 2.2 (0.7- 6.5) | 0.16 | 2.7 (1.0-7.5) | 0.05 |
| Socio-economic Status | Upper | - | | - | |
| | Middle | 0.5 (0.11 – 2.0) | 0.32 | 0.47 (0.06-3.9) | 0.49 |
| | Lower | 1.0 (0.14 – 7.0) | 0.99 | 0.95 (0.1-10.2) | 0.96 |
| Educational status of caregiver | At least graduate | - | | - | |
| | matriculation | 0.6 (0.1 – 3.5) | 0.45 | 0.55 (0.14-2.23) | 0.40 |
| | At least graduate | 0.56 (0.09 – 3.5) | 0.54 | 1.2 (0.29 - 6.18) | 0.93 |
| knowledge about the disease | Yes | - | | - | |
| | No | 0.2 (0.1– 0.5) | 0.002 | 0.68 (0.23-1.9) | 0.46 |
| Distances of provinces to the health facility | Nearby Travel | - | | - | |
| | Nearby Difficult Travel | 1.1 (0.34 – 3.7) | 0.84 | 1.03 (0.29-3.71) | 0.96 |
| | Farthest travel | 5.6 (1.4 – 23.2) | 0.02 | 1.65 (0.42-6.45) | 0.47 |
| Catastrophic health Expenditure | Yes | - | - | 7.3 (2.38-22.3) | 0.001 |
| | No | - | - | - | |

| | | | | | |
|---|---|---|---|---|---|
| Age of Patient | <=9 Years | - | | - | - |
| | > 9 Years | 0.4 (0.09 – 1.6) | 0.12 | - | - |
| Distress Financing | No | - | | - | - |
| | Yes | 5.1 (1.8 – 15.0) | 0.003 | - | - |
| Omnibus Test of Model | | Sig. = 0.000 | | Sig. = 0.000 | |
| Nagelkerke (R2) | | 0.500 | | 0.436 | |
| -2 Log likelihood | | 91.6 | | 99.03 | |

***Source:*** *Authors calculation established on primary data.*

It is worth noting that there is a lack of available studies that assess financial risk in terms of catastrophic expenditure for DMD specifically. However, there is one study from China that examines CHE in the context of seven rare diseases. In the study CHE was 40 percent and prevalence of catastrophic expenditure changed to 76 percent, 63 percent, and 29 percent when the threshold for catastrophic expenditure was taken as 20, 30, and 50 percents, respectively. Forty five percent of the patients reported having faced distress financing. The CHE for DMD in China, calculated at a threshold of 40 percent, was notably lower (only 0.0015) compared to this study (40 percent). It's important to highlight that the study from China did not include an assessment of distress financing. The data used to estimate the percentage of CHE in the China study relied on secondary data from estimated income. In contrast, the primary data was directly collected from individual families in a prospective manner in the present study. This study just estimated the cost incurred on the drugs available for the treatment which is very high from the affordability limit of individuals. So, no consumption no cost. This difference in data collection methods and sources may contribute to variations in the CHE percentages observed between the two studies(Xin et al., 2016).

The multiple logistic regression analysis at a 40 percent threshold for catastrophic expenditure has shown Caregivers who were unaware of the disease had significantly lower odds of facing CHE (OR: 0.2, 95% CI: 0.1–0.5, p-value: 0.002) compared to those with knowledge about the disease. Having a deeper understanding of the disease empowers

individuals to explore various treatment options, especially in the absence of a definitive cure, thus adding to cost.

The significant increase in the likelihood of catastrophic health expenditure (CHE) among patients from remote areas, as shown by odds of 5.6 times higher, underscores the need for teleconsultation services. Teleconsultation can help bridge the geographical gap, providing access to healthcare for those in distant areas and potentially reducing CHE. Patients whose parents noticed symptoms at a later age (after 5 years) had a 2.7 times higher risk of resorting to distress financing. This underscores the importance of early disease detection, emphasizing the need for increased awareness and education among peripheral physicians about the disease.

## 4.7 Government Policies Addressing the Transition of Muscular Dystrophy Patients from Childhood to Adulthood

***Employment:*** The employment situation for DMD (Duchenne Muscular Dystrophy) patients in India is dismal. A notable legal judgment by Honourable MR. JUSTICE BHARGAV D. KARIA on 07/06/2021 shed light on some crucial facts. Under the Persons with Disabilities Act of 1995, Muscular dystrophy was not explicitly listed as a disease resulting in physical disability. However, in the Disability Act of 2016, Muscular dystrophy was included under the category of locomotor disabilities but only if the disability exceeded 50 percent. These legal definitions and classifications have significant implications for DMD patients in terms of accessing disability benefits and support services, as well as their potential for employment opportunities.

***Unique Disability Identification (UDID) cards:*** These cards are issued by the government to individuals with disabilities. UDID cards serve as official documents that provide proof of disability. The primary purpose of UDID cards is to facilitate access to government welfare schemes, benefits, and services that are intended to support people with disabilities. These cards help ensure that individuals with disabilities receive the assistance and accommodations they need to improve their quality of life and participate more fully in society. Numerous pending petitions from individuals suffering from Muscular dystrophy,

seeking the issuance of unique disability identification (UDID) cards, have experienced prolonged delays, resulting in the cards not being issued rendering the beneficiaries from getting access to various government benefits and support services. ("Shivsankar Karunasankar Pandya Vs State of Gujarat on 7 June, 2021," n.d.)

***National Education Policy 2020 (NEP):*** None of the patients from the present study has gone under any kind of vocational training that would help them in fetching gainful employment in the future. In India, the education system faces significant challenges in supporting children with disabilities. Once a child becomes non-ambulatory, parents often cease sending them to school due to transportation difficulties. Most schools lack disability-friendly infrastructure, hindering the inclusion of disabled children. Social support systems for both children and their caregivers are inadequate. Although India's National Education Policy 2020 emphasizes inclusive education, implementation remains limited. Despite the Rights of Persons with Disabilities Act of 2016, which advocates for inclusive schooling, disabled children struggle to progress beyond primary education, with only 9 percent completing secondary education. Access to necessary facilities like ramps and accessible toilets is severely lacking in schools, with less than 40 percent having ramps and only 17 percent having accessible toilets. Additionally, access to electricity, crucial for technological advancements in education, is available in only 59 percent of schools. These barriers underscore the urgent need for improved accessibility and support within the education system to ensure disabled children can access education and opportunities for vocational training and employment in the future.

## 4.8 Conclusion

The socio-economic status of families, education, age (progression of disease) and distance of health care facilities from patients are perceived as primary predictors of economic burden over the families. The present chapter discusses the socio-economic, demographic and clinical characteristics of the study respondents. This chapter also estimates the total annual cost of illness and illustrates various costs associated with the disease. The table 4.1.1 highlighted the socio-demographic determinates of the economic burden of DMD in northern India. The mean age of the children at the time of enrollment was 8.32 ±2.0 years. The majority of the participants belonged to the upper-middle socio-economic status (SES),

comprising 51 percent of the total participants and no one was below poverty line. Regarding the family structure most of the of the children in the study came from nuclear families (54 percent). The participants were from northern states of India, maximum were from Punjab (32 percent). Other significant contributors included Haryana and Himachal Pradesh, both at 17 percent. Eastern states represented 11 percent of the participants. Jammu and Kashmir accounted for 10 percent, while Chandigarh had 9 percent. The smallest representation was from Uttarakhand, with only 4 percent of the participants. Most of the primary caregivers were parents (96 percent) and mothers were mostly involved as primary caregiving (58 percent). Mostly from rural areas (72 percent) and educated up till matriculation (46 percent).

Most of the children enrolled in the study were in the late non-ambulatory disease stage (Stage III), comprising 56 percent of the total participants. Table 4.2.1 provides insights into the clinical determinants affecting the economic burden. The majority of patients (90 percent) suffered from DMD, while 10 percent had intermediate BMD. Only 10 percent of patients had family history, symptoms were noticed in half of children up to 5 years and there was a significant association with disease severity (using $\chi 2$ ; $p=0.003$). Children diagnosed at a later age tended to be more adversely affected. Frequent falls were the most common initial symptom (31 percent), followed by difficulty in walking (24 percent), difficulty getting up from the floor (27.1 percent). It's notable that a significant proportion (47 percent) of parents had no prior knowledge about DMD.

Utilization patterns of care during phase I, Phase II and Non-participation in the intervention are shown table 4.3.1 The study found that while respiratory management for DMD patients primarily involved up to physiotherapy only. However, healthcare needs were unfortunately unmet for many DMD patients due to a lack of knowledge and motivation within the study cohort. Whereas during the intervention, the utilization of rehabilitation services e.g. physiotherapy and respiratory management were increased. Genetic consultations saw a remarkable increase, nearly doubling the count of consultations compared to routine care in phase I. As a notable impact, 11 mothers from the study population sought and received further prenatal diagnostic consultation and care at the tertiary care center after genetic consultations. Similar findings were noticed for nutritional counseling.

Teleconsultation during the intervention phase reduced physical visits considerably. Approximately 33 percent of the study population did not participate in the developed intervention, due to lost follow-up due to a decline in ambulation, a lack of interest in the intervention due to the unavailability of definitive treatment, or migration to distant locations far from the healthcare facility.

Furthermore, pathway to care was examined, revealing that parents consulted an average of 2.9±1.4 treatment agencies before arriving at the study site. While the number of agencies consulted increased with the severity of the disease (with a median of 4 in stage IV). The meantime consumed to traverse this pathway was about 12.5 months with a maximum span of 84 months (7.0 years). Notably, patient with the longest journey Jammu and Kashmir, highlighting the impact of distance on access to specialized care. A significant proportion of patients reported experiencing a loss of work time (74 percent), transportation difficulties (72 percent) of patients facing challenges in getting to healthcare facilities.

Long waiting periods were a concern for 72 percent of patients, accessing diagnostic facilities posed difficulties for 71 percent of patients and frequent visits were reported as a challenge in 71 percent of patients. The economic burden of a disease is measured through cost-of-illness (COI) studies, which quantify the "maximum amount that could potentially be saved or gained if a disease were to be eradicated.(Pérez et al., 2021)". The annual mean direct expenditure was estimated at 109.4 (ranging from 27.4 to 534.2) x ₹ 1000, indicating the financial burden borne by families dealing with DMD or BMD. This direct cost accounted for approximately 52 percent of the total annual cost of illness. Most of the direct expenditure was driven by direct medical costs, making up 30.6 percent of the total COI. This estimate is different from Germany based study the total direct medical costs of illness was only 25 percent of the total COI same is with Portugal (19 percent of total COI), as compared to the present study this ratio (30.61) is quite less. In this context, a major proportion of direct medical costs is attributed to expenses related to medical aids ( Katz et al., 2014; Labisa et al., 2022). The proportion of expenditure on account of direct non-medical expenditure in the study was 21.1 percent. However, in the present cohort none of the patients had expenditure

for house or automobile modification. On the contrary none of the nine non-ambulatory patients in the present cohort had any access to even simple non-customized wheelchair.

Loss of productivity is caused by disease induced deficit in working capacity( Katz et al., 2014; S. Prinja et al., 2016). The mean annual cost for unpaid caregiving was 33.5 (7.3-88.2) x ₹1000. The productivity loss due to informal caregiving is about 30 percent of the total cost of illness and loss of productivity due to absenteeism and change in working conditions leading to reduced wages was 17.7 percent. These outcome measures showed that the loss of productivity of caregivers due to DMD is quite high in India. The disease progression hindered patient mobility, prolonging medical visits and complicating public transport access, thereby elevating transportation and accommodation expenses. Direct non-medical costs primarily comprised food, travel, and lodging, constituting 14.3 percent of total annual illness costs. In stage 4, travel, boarding, and lodging costs averaged 38.8 x ₹1000, almost triple that of stage 3. This cost increased significantly with the severity of the disease, as confirmed by the ANOVA test (F=5.1). The study found that joint families (64.9 x ₹1000) incurred higher expenses in indirect caregiving compared to nuclear families (42.5 x ₹1000). Moreover, caregivers who found work loss not a major challenge in visiting the health facility were significantly spending more (3.0 x ₹1000) in diagnostic care when comparing with who found it more challenging (1.8 x ₹1000) (p=0.049).

The multiple linear regression model identified key socio-economic and demographic determinants of direct cost routine care. The model's significant F-statistic value of 10.942 indicates goodness of fit. After adjusting for confounding factors using multivariate analysis, it was found that the annual direct cost of illness was significantly positively associated with the family's socioeconomic status, the education level of the primary caregiver, and the distance of their residence from the health facility. Although the child's age is not significantly associated with the cost of illness, the relationship is negative. This suggests that as the child ages or as the disease worsens, the direct cost of illness tends to increase. The multiple linear regression models were used to identify predictors after adjusting for confounders for total cost of illness and indirect cost of illness (Caregiving). The F-statistic values were 7.254 and 12.075, respectively, indicating good model fit. The total cost of illness

(COI) for families with a child affected by DMD highlights significant disparities in healthcare access and affordability. Socio-economic status (SES) plays a crucial role, with higher costs burdening wealthier families more, while those from lower SES backgrounds might struggle with treatment affordability. The increased COI with greater distance from health facilities underscores the challenge of geographical accessibility, particularly in rural or underserved areas. Similarly, indirect cost of illness is associated with the educational status of the main caregiver, with caregivers in urban areas spending more time on caregiving. The productivity loss is higher in joint families due to multiple caregivers spending more time on caregiving. Families traveling from distant locations incur additional wage loss due to the time spent on healthcare visits. Furthermore, the child's age or disease progression and their health-related quality of life are inversely related to the total indirect cost of illness.

The out-of-pocket (OOP) health expenditures refer to additional healthcare costs not covered by the health system, which patients or their families must pay themselves( Prinja et al., 2019). These expenses may compel patients to make difficult choices between healthcare and other essentials, leading to an inequitable distribution of health services. The universal healthcare coverage (UHC) policies are crucial to achieving one of the sustainable development goals (SDGs) established by Member States of the United Nations (UN)(Chiu et al., 2018). Among the recruited patients, 40 percent (n = 100) suffered from catastrophic health expenditure at the 40 percent threshold. The prevalence of catastrophic expenditure changed to 76 percent, 63 percent, and 29 percent when the threshold for catastrophic expenditure was taken as 20, 30, and 50 percents respectively. Forty five percent of the patients reported having faced distress financing. The CHE was significantly less among patients belonging to higher socio-economic status as compared to lower socio-economic ($\chi 2$ 6.6; $p < 0.037$), caregivers ignorant to the outcome of disease ($\chi 2$ 8.8; $p < 0.003$), and traveling from nearby provinces ($\chi 2$ 15.2; $p < 0.000$), and parent who noticed symptoms up to 5 years of age ($\chi 2$ 10.8; $p < 0.001$).

Table 4.6.2 exhibits the key predictors of catastrophic expenditure that were identified by using the logit model. Based on the “Omnibus Test of Model”, the value of 0.000 exhibits the model to be significant. Multiple logistic regression at 40 percent

threshold, showed that the odds of catastrophic expenditure was significantly lesser for those were ignorant about the disease (OR: 0.2, 95% CI: 0.1– 0.5, p-value: = 0.002), as compared to caregivers having knowledge about the disease. Secondly, those who were traveling from far-flung areas were significantly 5.6 times more prone to face CHE as compared to patients traveling from nearby areas (OR: 5.6, 95% CI: 1.4 – 23.2, p-value: = 0.02). The odds of CHE were 5.1 times more in patients with CHE (OR 5.1 95% CI: 1.8 – 15.0). The risk of distress financing was higher among patients noticed the symptoms in later ages (after 5 years). This risk was 2.7 times higher (OR 2.7. 95% CI: 1.0 – 7.5). The odds of distress financing were 7.3 times more in patients with CHE (OR 7.3 95% CI: 2.4 – 22.3). The severity of the disease (progression of disease/age of patient), locality of the household presence of any insurance/subsidy entitlement, and socio-economic status did not alter the risk of suffering from catastrophic expenditure and distress financing.

# CHAPTER V

# PATIENT-CENTERED CARE MODEL

# &

# IMPACT ON ECONOMIC BURDEN AND QUALITY OF LIFE

## 5.1 Patient-Centered Care model (Intervention)

The care required in a rare disease like DMD is not the usual medical care. Here, the patient/caregiver-physician relationship grows with time along with evolution in the knowledge of disease and its management strategies (Ayme et al, 2008). As the disease outcomes and their management are changing, the healthcare provider must inform, engage and be an interactive partner in the therapeutic process for better compliance and adherence to standard operating care (SOC). Majorly, treatment-seeking behavior by the patients is limited due to scant knowledge about the disease, lack of appropriate multidisciplinary healthcare and access to the treatment (Takeuchi et al., 2020). The involvement of multiple disciplines is often neglected by the healthcare sector directly impacting health care delivery. In addition to medical management, self-care, as well as caregiving by the patients' parents, is very important. Patient engagement is very critical in the management of chronic rare diseases as the patient takes ownership of his/her health. Patient-centered care is one such intervention that ensures patient involvement. Patient/family engagement improves health outcomes and reduces health expenditure. Patient engagement, health outcomes and health expenditure are dependent upon the geographical location of healthcare delivery system.

During the study period, a novel intervention known as the Patient-Caregiver-Centered (PCC) model was developed. The primary objective of this model was to empower both patients and caregivers by providing them with essential knowledge about the disease and available treatment options. Additionally, it aimed to encourage their active involvement in the management of the disease to achieve comprehensive care, particularly in a resource-limited setting. A crucial component of the PCC model was teleconsultation. This component was specifically designed to address the challenges posed by long-distance travel, which can be especially burdensome for functionally disabled patients. Moreover, it aimed to make medical consultations readily available, a particularly important feature during the COVID-19 era when in-person visits were restricted or limited.

The patient-centered care model was a Comprehensive DMD -Tele Care Model the components of the model are as follows:

| S. No. | Components of Intervention Management |
|---|---|
| 1 | **Informational about the disease:** Sessions using PowerPoint presentation and the "Care Guide Companion" booklet is provided (hard copy and digital). The booklet is designed in such a way that after the acquisition of knowledge in a comprehensible manner it helping children/ caregivers in participating decision-making process of their treatment. Components of the book are: 1) basic information related to disease, 2) management of disease, 3) role of care provider team, 4) support groups to help people living with DMD. (Annexure - VIII) |
| 2 | **Instructions and Checklist** based intervention and follow-up: Instructions is given to the patients and their caregivers to make them understand all the activities that is carried out once subject enters the phase 2 in the study. Audio-videos and images of instruction of stretches/ physiotherapy, physical activity are provided to the family. |
| 3 | **Compliance Diary:** The patient is expected to comply with the prescribed procedures and medications. A customized daily planner is introduced to each patient in the intervention phase to increase their adherence to the institutional set management protocol. This contains periodic monitoring of weight, height, Blood pressure, daily medication (steroid/ACE inhibitors/Calcium/Vitamin D) and also their daily exercise routine. Some clinical evaluation are expected to be performed at home (e.g: Gower's time, vignos score, Brookes scores etc which are monitored by the team telephonically) (Annexure -VII) |
| 4 | **Tele-Consultation:** Three-monthly video teleconsultation is given to the boys by clinicians for providing diagnostic or therapeutic advice through electronic means. The boy does not have to visit the clinic physically. This is done through: |

| | |
|---|---|
| | 1. Review Videos: Videos that patients share with the care-team regarding their present state of illness. These videos are the recordings of the physical activities that the patient can perform at home and share with the team to evaluate.<br>2. Customized Videos: As per the clinical requirement additional customized videos are shared with patients to address the need. |
| 5 | **Multi-specialty teleconsultation**<br>Not all these subspecialists are needed at all ages or stages, but they must be accessible if necessary. **(as indicated)** |
| **A sample of model components is given in the annexures of the thesis** | |

The intervention (PCC model) was developed during the study period. The objective of the model was to empower the patient and caregiver by giving them knowledge about the disease, and treatment options available and encouraging their involvement in taking management of the disease to obtain comprehensive care, in a resource-limited setting. The most important component of the intervention was teleconsultation, this component was targeted to address long-distance travel by functionally disabled patients can also make medical consultations available in covid ERA.

It is noteworthy that the premiere health center involved in this study extends its services to a wide geographic region, covering patients with various types of muscular dystrophy, including DMD, BMD, and Spinal Muscular Atrophy (SMA). This broad coverage spanning seven provinces in Northern India underscores the potential impact and reach of the PCC model in improving the care and well-being of individuals affected by these debilitating conditions.

## 5.2 Economic Burden During Intervention phase (Phase II)

The annual total means direct expenditure with intervention was 44.0 (6.5-305.4) X ₹1000. Direct cost is about 41.16 percent of the total annual cost of illness. The direct medical expenditure in INR is 23.1 x ₹1000, ranging from 2.4-16.5 x ₹1000. Direct medical expenditure is about 22 percent of the total cost of illness per year in families with PCC intervention. Considering the medical cost these were same across the follow up period (with or without intervention). (table 5.3.1) The least expenditure was on orthotics/aids as discussed earlier this is the most common unmet need in the study. Although they were nine children who were wheelchair-bound, but none of them had a wheelchair at the time of enrollment. Only one out of these 9 children got a wheelchair during the second phase of the study. Thus, costs of aid which are reflected in direct medical costs were quite low compared to the demands on needs. The cost of rehabilitation services was the next most cause of direct medical burden (3.64 percent of total COI) during phase 2.

| Table 5.2.1 Mean annual Cost of Duchenne Muscular Dystrophy in ₹ x 1000 (min-max) on the families with PCC intervention (Phase II) | | | | | | | |
|---|---|---|---|---|---|---|---|
| Variables | **Mean** | **Ratio of total COI [%]** | **Clinical Severity** | | | | **p- value** |
| | | | **I** | **II** | **III** | **IV** | |
| Drugs (Medicines +Vaccine) | 14.6 (1.8-126.3) | 13.66 | 17.2 (6.9-36.5) | 10.6 (1.8-32.6) | 17.7 (3.8-126.3) | 11.2 (8.1-14.2) | 0.388 |
| Diagnostic (Lab tests+ ECHO + Xray +DEXA) | 2.5 (0.0-21.5) | 2.36 | 0.8 (0-2.4) | 1.2 (0-6.8) | 2.8 (0-21.3) | 8.2 (0.9-17.0) | 0.006** |
| Procedure_orthotic<br>User fee | 2.1 (0-96.6) | 1.94 | 0.25 (0.03-0.7) | 0.4 (0-1.3) | 3.7 (0-96.6) | 3.6 (0-1.7) | 0.736 |
| Costs of rehabilitation services | 3.9 (0-60.8) | 3.64 | 8.1 (0-24.3) | 1.4 (0-20.9) | 4.4 (0-60.8) | 9.8 (0-28.1) | 0.358 |
| Direct medical expenditure | 23.1 (2.4-16.5) | 21.60 | 26.3 (6.9-37.2) | 13.6 (2.3-35.2) | 28.6 (3.3-16.5) | 29.6 (10.5-51.5) | 0.145 |
| Personal assistance for school and work attendance | 2.6 (0-85.2) | 2.41 | - | - | 50.1 (0-85.2) | - | 0.499 |
| Travel_ Boarding/Lodging | 11.8 (0.4-50.0) | 11.00 | 17.2 (4.0-27.8) | 11.5 (0.7-48.3) | 11.2 (0.38-50.0) | 14.4 (4.0-27.9) | 0.797 |
| Food other costs | 5.0 | 4.70 | 7.9 | 2.9 | 6.4 | 3.9 | 0.869 |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | (0-133.2) | | (0.6-21.0) | (0-19.0) | (0-133.2) | (0.12-10.2) | |
| Informal (traditional healing) | 1.5 (0-8.0) | 1.44 | 2.6 (0.4-5.2) | 11.7 (0-5.9) | 1.4 (0-6.8) | 3.9 (0-8.0) | 0.035 |
| Direct non-medical expenditure | 20.7 (0.4-140.3) | 19.56 | 27.7 (4.9-54.0) | 15.6 (1.3-61.4) | 23.9 (0.4-140.3) | 22.1 (6.3-40.9) | 0.628 |
| Total direct expenditure | 44.0 (6.5-305.4) | 41.16 | 54.0 (39.8-61.2) | 29.2 (8.6-73.2) | 52.5 (6.5-305.4) | 51.7 (43.4-57.8) | 0.01** |
| LP (unpaid caregiving) | 46.3 (14.7-75.4) | 43.3 | 47.0 (25.7-58.8) | 45.3 (14.7-66.1) | 45.1 (14.7-73.5) | 58.9 (44.1-75.4) | - 0.709 |
| LP (absenteeism**) + LP (changes) | 16.6 (0-285.1) | 15.5 | 11.7 (0-2.2) | 22.5 (0-285.1) | 15.1 (0-182.5) | 8.0 (0-34.6) | 0.305 |
| Total indirect COI | 62.9 (18.0-343.9) | 58.84 | 48.2 (27.0-61.0) | 67.8 (20.7-344.0) | 60.2 (18.0-241.3) | 67.0 (56.1-78.7) | 0.433 |
| Total COI | 107.0 (40.6-417.1) | 100 | 102.2 (88.2-117.5) | 96.9 (43.3-417.1) | 112.7 (40.6-339.0) | 118.8 (105.4-136.5) | 0.845 |

** Two-tailed significant values

***Source:*** *Authors calculation established on primary data*

The non-medical costs significantly increased with disease severity with the shooting increase from stage IV and V however no significant difference was found in Phase II during the intervention. There were only 4.5 percent of patients who appointed assistants as informal care at school during both phases. There was almost a 30 percent of reduction in cost for personal assistants as informal care at a school. But this reduction is not statistically significant (table: 5.3.1). There was almost a 12 percent cost reduction in Phase II over travel boarding and lodging and this difference was found to be statistically significant. Tele-consultation is the major cause behind cost reduction over travel boarding and lodging in Phase II. So far costs in Phase II all costs are reduced but the cost of food has increased in phase II by 31 percent (figure 5.3.1). Although this difference is not statistically significant and this rise in cost is very much contradictory to the decreased cost of travel, boarding, and lodging with decreased visits to the main health care centre due to the provision of teleconsultation in the intervention phase. There is a reduction in the informal cost intervention phase, statistically not significant. The direct non-medical expenditure came out to be the same in both phases.

## 5.3 Impact of Economic Burden During Non-Intervention (Phase I) and Intervention Phase (Phase II)

The direct medical expenditure was reduced in the intervention phase by 19.5 percent and this decrease was statistically significant. The teleconsultation component of PCC intervention had a major impact on cost curtailment on procedures, user fees, orthotic devices and the cost of rehabilitation services. The teleconsultation potentially increases healthcare access by making care more affordable for both patients and their families with many chronic diseases. Use of teleconsultation which was promoted during the COVID-19 pandemic, telemedicine has reduced costs, including those related to transportation and missed work, while offering high-quality care and regular follow-ups (Gilkey et al., 2022). The non-medical costs increased significantly with disease severity with a sharp rise between stages 4 and 5 with routine care, but this increase was not seen during the intervention phase as the teleconsultation component was taking care of travel-related costs.

This study is short term study, no two data points have duration more than 365 days. In short-term studies where the economic impact is not much (unless inflation is very high) inflation correction is not done. This data was interpreted in consultation with health economists. The correction of inflation was not suggested by the experts due to the short-term

nature of the study. The consumption of direct medical resources was significantly higher in patients during the routine care phase compared to the intervention phase. Although the data did not show statistical significance, the cost was notably lower during the intervention phase. It's important to note that the non-statistical values may be attributed to the shorter period of follow-up in the intervention phase.

**Table 5.3.1 Economic burden in ₹ 1000 (min-max) during non-intervention (Phase I) and Intervention phase (Phase II) compared at an annual rate**

| Variables | **Phase I** | | Phase II | | **t stats** | P value (paired t-test) |
|---|---|---|---|---|---|---|
| | **Mean** | **Ratio of total COI [%]** | **Mean** | **Ratio of total COI [%]** | | |
| Drugs (Medicines + Vaccine) | 13.3 (2.0-58.4) | 13.93 | 14.6 (1.8-126.3) | 13.66 | 0.134 | 0.894 |
| Diagnostic (Lab tests+ ECHO+ Xray +DEXA) | 2.2 (0-17.0) | 1.93 | 2.5 (0.0-21.5) | 2.36 | 0.066 | 0.948 |
| Procedure orthotic User fee | 5.4 (0-60.9) | 6.28 | 2.1 (0-96.6) | 1.94 | 2.69 | 0.009** |
| Costs of rehabilitation services | 7.7 (0-104.3) | 8.47 | 3.9 (0-60.8) | 3.64 | 1.89 | 0.031* |
| Direct medical expenditure | 28.7 (5.6-157.3) | 30.61 | 23.1 (2.4-16.5) | 21.60 | 2.33 | 0.022** |
| Costs for personal assistance for school | 2.0 (0-60.8) | 1.45 | 2.6 (0-85.2) | 2.41 | -1.00 | 0.321 |
| Travel, Boarding & Lodging | 13.5 (0.2-167.0) | 14.30 | 11.8 (0.4-50.0) | 11.00 | 2.32 | 0.023** |
| Food & other costs | 3.8 | 3.44 | 5.0 | 4.70 | -0.45 | 0.654 |

| | | | | | | |
|---|---|---|---|---|---|---|
| | (0-34.1) | | (0-133.2) | | | |
| Informal (traditional healing) | 2.0 (0-46.0) | 1.93 | 1.5 (0-8.0) | 1.44 | -0.97 | 0.333 |
| Direct non-medical expenditure | 20.9 (0.6-195.9) | 21.11 | 20.9 (0.4-140.3) | 19.56 | 0.844 | 0.402 |
| Total direct expenditure | 55.4 (11.9-289.3) | 49.6 | 44.0 (6.5-305.4) | 41.16 | 1.93 | 0.029* |
| LP (unpaid caregiving) | 33.5 (7.3-88.2) | 30.6 | 46.3 (14.7-75.4) | 43.3 | -7.45 | 0.00** |
| LP (absenteeism**) + LP (changes) | 19.3 (0.0-311.6) | 17.7 | 16.6 (0-285.1) | 15.5 | 1.07 | 0.287 |
| Total indirect COI | 56.3 (14.7-352.9) | 50.4 | 62.9 (18.0-343.9) | 58.84 | -1.31 | 0.194 |
| Total COI | 111.6 (27.4 – 534.2) | 100 | 107.0 (40.6-417.1) | 100 | 0.53 | 0.596 |
| * One-tailed significant values<br>** Two-tailed significant values | | | | | | |

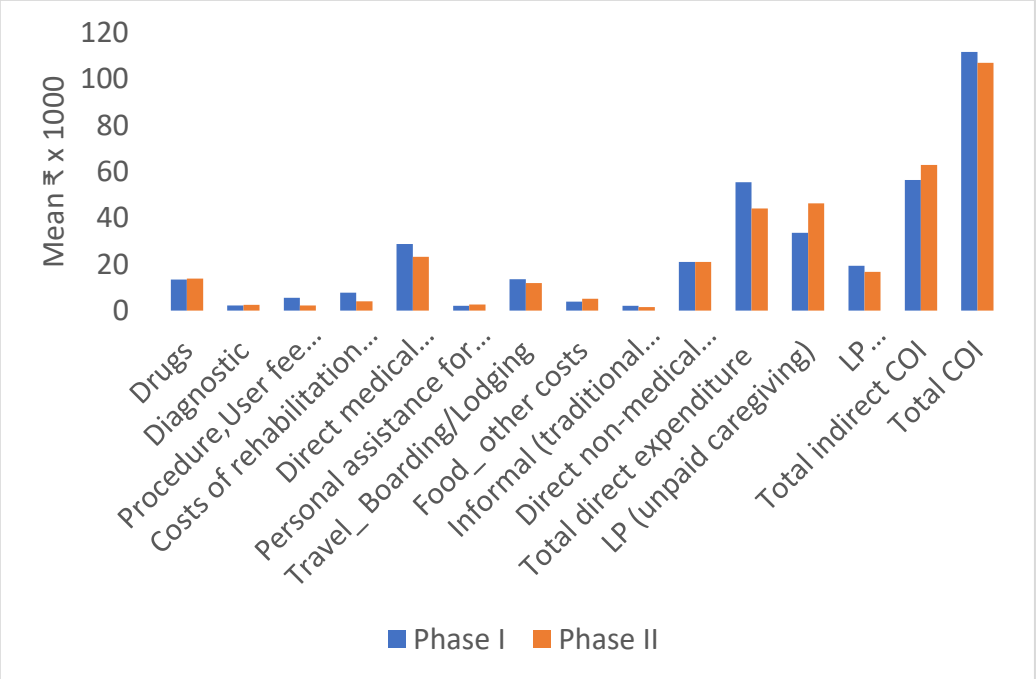
***Source:*** *Authors calculation established on primary data*

The family's or patients' perspective measures COI in the OOP and the productivity lost due to the illness. The third objective of the study was to measure the difference between economic burden and QoL with routine care and with intervention. There was no significant difference in direct medical cost across the different stages of the disease in Phase I, however direct medical expenditure was found to be reduced in the intervention phase by 19.5 percent and this decrease was statistically significant. The major contributor to this reduction of the cost was decreased cost of procedures, user fee, orthotic devices, and cost of rehabilitation

services. The out-of-pocket expenditure on procedures and orthotic devices decreased considerably in Phase 2. Mean expenditure was reduced by almost 60 percent. (paired t-test z=-2.7). Expenditure on rehabilitative services like Physiotherapy also decreased with intervention by almost 50 percent but no statistical significance was found. (paired t-test z = -1.9).

However, contradictory to the direct expenditure (non-medical and medical), the total indirect cost increased in the intervention phase this is subjected to the increased economic burden due to loss of productivity in unpaid caregiving. However, the loss of productivity due to absenteeism and change in the working capacity of the caregiver decreased (6.2 percent) like all other costs in phase II. The burden of the loss of productivity in unpaid caregiving (willingness to pay) increased by 37.9 percent in Phase II. This difference was highly statistically significant and very clearly visible in the comparative bar chart (figure: 5.3.1). The unpaid caregiving cost increased the overall indirect cost of illness in Phase II by 11.7 percent statistically not significant (Paired t Test, z=1.31).

**Figure 5.3.1 Expenditure during non-intervention (Phase I) and Intervention phase (Phase II)**




***Source:*** *Authors calculation established on primary data*

The total COI was reduced by 4 percent although is not statistically significant. So the study cannot reject the null hypothesis, that there is no change in economic burden with

or without intervention. The time spent in caregiving was not associated with the severity of disease. However, it was significantly associated with the empowerment of the caregiver. Training and education about the disease and its management empower the caregiver and increase enhanced caregiving. The mean number of hours spent in unpaid informal care with routine care was 4.4 hours while during the intervention phase, the time duration increased to 6.1 hours. This was reflected in the burden of the loss of productivity in unpaid caregiving (willingness to pay). Economic Burden increased significantly by 37.9 percent in Phase II (intervention phase). Training programs change the behaviour of caregivers, caregivers become more receptive to counsellors and educators for DMD patients and care providers (Ota et al., 2006). The studies have shown that training and knowledge of both caregiver and affected children have a positive effect on managing disease and improving self-care practices at home. It works especially where disease management is complex and has to be continued lifelong (Fukkink & Lont, 2007).

### 5.4 Quality of Life of Children

The socio-economic status is directly associated with QoL of many life-threatening diseases. This study included 100 boys (5-15 years) with DMD and their caregivers at the baseline. Health-related QoL and global QoL were assessed with EQ 5D-3L utility index for children with DMD and World Health Organization "Quality of Life-BREF (WHOQOL-BREF)"(Saxena et al., 1998) Quality of Life among caregivers of children living with DMD. The World Health Organization Quality of Life (WHOQOL-BREF) questionnaire is a scale that is extensively used to carry out assessments and to internationally compare the quality of life". The same is used in the present study. The baseline data of 100 caregivers were collected. WHOQOL-BREF instrument comprises of four domains of QOL namely physical health, psychological, social relationships, and environment.

#### 5.4.1 Health-related Quality of Life Among Affected Children

The quality of life (QoL) of patients to decrease as a disease progresses, and this is often reflected in various health-related quality of life measures like the EQ-5D and the Pediatric Quality of Life Inventory (PedsQL). The EQ-5D-5L offers an advantage in its suitability for estimating quality-adjusted life-years (QALYs) and other applications requiring summarization of EQ-5D-5L profile data into a single numerical value.This simplifies the representation of overall health-related quality of life, making it a valuable tool in various healthcare assessments and research. Summarisation of QoL into a single number

makes it easier to compare different countries, different time-periods, and different diseases (Devlin et al., 2022). However, disease-specific "quality-of-life" measuring tool like PeadsQoL provides better insight into how disease progression affects HrQoL. The quality of life (QoL) among children with DMD presents conflicting findings across studies. While some research indicates a reduced QoL in these children, others find no significant difference between the QoL of children with DMD and that of healthy children. However, it's important to note that the variability in findings could be influenced by the methods used to measure QoL within each study(Kohler et al., 2005; Melo & Moreno, 2007). This possibility gives EQ-5D-5L an advantage over other tools, although the appropriateness of the EQ-5D-3L in rare conditions has not been evaluated. It is important to understand whether this generic measure of HrQoL is comprehensive, relevant and understandable to people with rare conditions. The use of EQ-5D can be used to inform the cost-effectiveness of emerging therapies.

The median age of the boys was 9 years and 91 percent were ambulatory while 9 percent were wheelchair-bound. EQ 5D-3L utility index scores for Health-related quality of life (HrQoL) for children were computed based on the recently generated tariff value set for India (Jyani et al., 2022). The mean EQ 5D utility score at the baseline at the time of recruitment was 0.58 ±0.38. The distribution of the scores is not normality distributed. (Kolmogorov-Smirnova normality test $p < 0.05$). The patient's self-rated health is through a visual analogue scale i.e EQ-VAS. The mean EQ-VAS score is 71.3 ± 20.4. The patient's "self-rated health visual analogue scale score" showed no definite trend. The score was more at the time of the start of the intervention than the baseline but again showed a decline at the end of the intervention (table 5.6.1) EQ 5D utility scores of HrQoL of patients significantly decreased with the progression of the disease.

**Table 5.4.1 Health-related Quality of Life (HrQoL) Assessment for Children Living with DMD and BMD**

| Quality of Life Scales | Mean score ±SD (Overall) | Clinical Severity | | | | | p-value |
|---|---|---|---|---|---|---|---|
| | | I | II | III | IV | V | |
| *EQ 5D-3L utility index for Children* ***Score (0-1)*** | 0.58 ±0.35 | 0.98 ±0.02 | 0.78 ±0.22 | 0.50 ±0.35 | 0.19 ±0.24 | 0.0 | 0.00 |
| *PedsQL™ 3.0 Neuromuscular Module.* ***(Physical Health)*** ***Score (0-100)*** | 77.7 ±17.7 | 99.0 ±1.7 | 84.9 ±11.0 | 74.4 ±19.3 | 68.7 ±13.0 | 41.2 | 0.03 |
| *PedsQL™ 3.0 Neuromuscular Module.* ***(Communication)*** ***Score (0-100)*** | 39.3 ±31.0 | - | 47.6 ± 42.8 | 36.0 ± 26.6 | 41.6 ±27.4 | - | 0.48 |
| *PedsQL™ 3.0 Neuromuscular Module.* ***(Family Resources)*** ***Score (0-100)*** | 56.8 ±33.7 | - | 63.6 ±40.4 | 56.2 ±32.3 | 45.0 ±26.4 | - | 0.52 |

***Source:*** *Authors calculation established on primary data*

### 5.4.2 PedsQL (Pediatric Quality of Life Inventory) Neuromuscular Module

HRQoL was also calculated using the disease-specific QoL tool, PedsQL TM (pediatric quality of life inventory) 3.0 Neuromuscular Module. This scale has 25 items and comprises 3 domains: neuromuscular disease (17 items related to the disease process and associated symptomatology), communication (3 items related to the patient's ability to communicate with health care providers and others about his/her illness), and About Our family resources (5 items related to family financial and social support systems). Items are scaled on a 5-point likert scale from 0 (Never) to 4 (Almost always). The scale used in this study ranges from 0 to 100, where higher scores mean a better quality of life (HrQoL). The mean PeadsQoL-Physical Health, PeadsQoL-communication and PeadsQoL-Family Resources were 77.7±17.7, 39.3 ±31.0 and 56.8 ±33.7 respectively. The pediatric quality of

life inventory is a tool used to assess the QoL of children and adolescents with chronic health conditions. The neuromuscular module specifically focuses on neuromuscular diseases. Like the EQ-5D, PedsQL scores also decrease with disease progression. This indicates that as the disease worsens, young patients with neuromuscular conditions are experiencing a decline in their overall QoL, likely due to the specific challenges and limitations associated with their condition.

Interestingly, quality of life related to communication and family resources remained the same throughout the disease progression. This could suggest that despite the overall decline in health-related QoL, certain aspects of life, such as communication and family support, are relatively stable. This could be due to adaptive strategies or external support systems that help patients maintain these aspects of their QoL (table 5.4.1).

**5.5 Quality of Life among Caregivers**

Instruments used to measure Duchenne carer's quality of life have limited psychometric evidence (Carlton et al., 2022). WHOQOL-BREF tool is a more generic questionnaire analyzing the quality of life in a general context, regardless of how the disease affects the family (Roncada et al., 2015). Thus, it is more comparable like EQ-5D-5L. Health-related quality of life for boys and the overall quality of life of caregivers were assessed at three different points of time to get a better comparison with the progression of disease. It also helped in evaluating interventions that help in improvement in quality of life.

The study assessed the QoL of caregivers using the WHOBREEF scales. The results showed that the average scores for caregivers' overall QoL and overall health-related QoL were 29.7 ± 26.0 and 53.0 ± 34.7, respectively. This comparison indicates that caregivers' overall QoL was significantly worse than their health alone. This suggests that aspects of their lives beyond physical health have been adversely affected and merit further investigation. This suggests that there are other aspects of their lives that have been negatively affected and require further investigation. The WHOBREEF scale examined different domains to assess the caregivers QoL, including physical, psychological, social, and environmental factors (table: 5.5.1). Understanding these different areas can provide valuable insights into the challenges caregivers may be facing and help identify areas where support and intervention may be needed to improve their overall well-being.

However, 69.5 percent of caregivers rated their overall quality of life to be poor or very poor with a similar percentage (68.4 percent) expressing dissatisfaction with their health

at baseline. These findings underscore the significant impact of caregiving on caregivers' well-being. The distribution of domains of quality-of-life scores was significantly different in the normality test run for all the domains of quality-of-life except for Social relationship at baseline and the start of the intervention, environmental at baseline i.e they are not normally distributed.

**Table 5.5.1 Quality of Life (QoL) Assessment for Caregivers of Children Living with DMD & BMD**

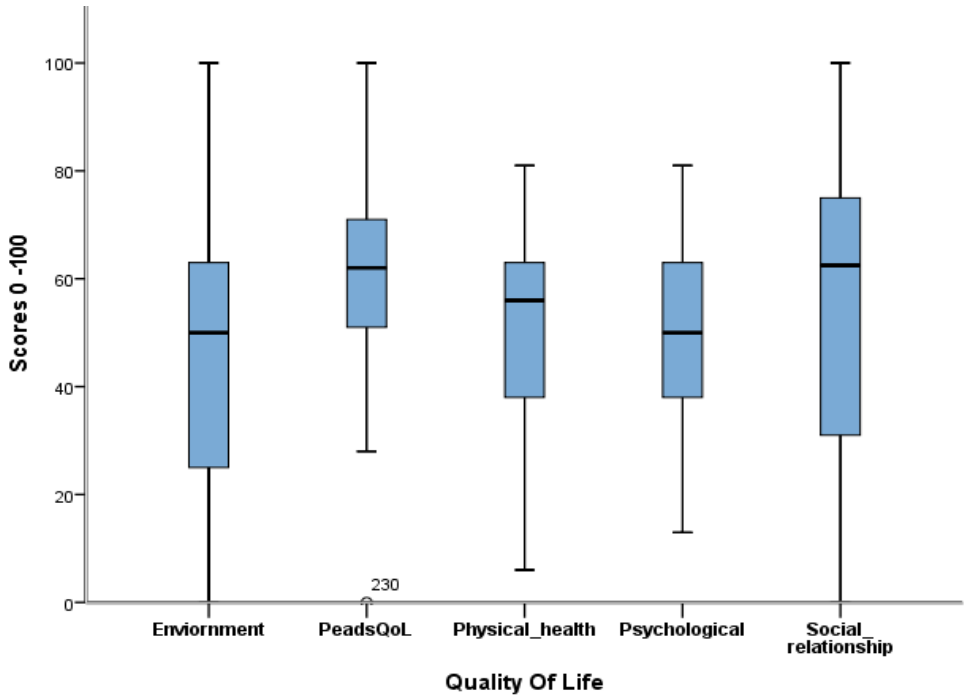
| Quality of Life Scales | Mean score ±SD (Overall) | Clinical Severity | | | | | p value |
|---|---|---|---|---|---|---|---|
| | | I | II | III | IV | V | |
| WHOBREEF Overall quality of life Score (0-100) | 29.7 ±26.0 | 16.7 ±28.9 | 34.4 ±31.6 | 29.0 ±23.2 | 25.0 ±18.9 | 0.0 | 0.503 |
| WHOBREEF Overall health Score (0-100) | 53.0 ±34.7 | 99.7 ±14.4 | 46.9 ±34.0 | 55.3 ±34.7 | 53.1 ±31.1 | 0.0 | 0.11 |
| WHOBREEF Physical Domain Score (0-100) | 50.6 ±14.4 | 46.0 ±19.3 | 51.7 ±12.0 | 49.2 ±15.4 | 49.6 ±13.6 | 31.0 | 0.62 |
| WHOBREEF Psychological Score (0-100) | 49.8 ±16.8 | 50.0 ±27.1 | 54.8 ±14.7 | 46.8 ±16.6 | 54.7 ±14.74 | 19.0 | 0.43 |
| WHOBREEF Social relationship Score (0-100) | 54.7 ±26.9 | 77.0 ±6.9 | 57.0 ±26.0 | 54.0 ±27.5 | 48.5 ±24.4 | 0.0 | 0.16 |
| WHOBREEF Environmental Score (0-100) | 45.6 ±24.7 | 60.7 ±23.9 | 48.2 ±24.7 | 42.5 ±24.3 | 57.2 ±21.6 | 0.0 | 0.83 |

***Source:*** *Authors calculation established on primary data*

The mean scores of QoL for caregivers are almost similar in all four aspects of life, their physical health, psychological health, aspect of their social life and the environmental domain at the baseline. The environmental domain in WHOBREEF depicts how caregivers

think of the health-related facility are available and accessible to them in the surroundings. This aspect of QoL appears to be similar to the other domains, suggesting that caregivers' views on the adequacy and convenience of healthcare facilities align with their perceptions of physical health, psychological well-being, and social aspects of their lives. This finding could have important implications, as it implies that caregivers' overall QoL is relatively uniform across these different dimensions at the baseline. However, it's essential to monitor these aspects over time to understand if any changes occur or if interventions are needed to maintain or improve caregivers' QoL in the environmental domain and other areas of their lives as they continue their caregiving responsibilities.

**Figure: 5.5.1 Quality of life of caregivers in the four aspects of life and Health-related quality of life (Neuromuscular) of Dystrophinopathy patients.**




***Source:*** *Authors calculation established on primary data*

The study provides valuable insights into the QoL of caregivers across different stages of the disease, specifically focusing on physical health, psychological health, social health, and environmental health (table 5.5.1).

**Physical QoL:** At the baseline, caregivers generally reported good physical QoL except in stage V, where it was moderate. However, it is important to note that stage V had only one patient, so it's challenging to draw strong conclusions for this stage. It seems that physical

QoL may be influenced by disease progression, but further data would be needed to confirm this.

**Psychological QoL:** The caregiver's psychological health did not appear to be significantly affected by the progression of the disease, although no single stage showed very good psychological health. Psychological health was lower in stage I, possibly due to the emotional impact of the initial diagnosis.

**Social QoL**: The social health at baseline has shown a decreasing trend with the clinical severity of the disease, although this relationship did not reach statistical significance (ANOVA P=0.16). The caregivers reported excellent social relationships in stage I but declined to moderate in stage IV and very poor in stage V. This suggests that caregiving responsibilities and the demands of the disease may have an impact on caregivers' social well-being.

**Environmental QoL:** The environmental health was consistently reported as good across all stages of the disease. This indicates that caregivers felt that health-related facilities were available and accessible in their surroundings, regardless of disease severity.

The burden on caregivers in DMD is considerable and this burden is directly related to the severity and progression of the disease. Just like the patients, the assessment of HrQoL was conducted at three different data points over a 12-month period. Similarly, the QoL of caregivers was also evaluated at these time intervals. About two third of caregivers rated their overall quality of life to be poor or very poor and were dissatisfied with their health at baseline. A study conducted explored the burden of social and professional support in families of patients with muscular dystrophies (MDs) in Italy. About 77 percent reported a feeling of loss, 74 percent felt sad and 54 percent felt constrained in leisure activities. About 3.2 percent of caregivers always felt depressed. Among caregivers, 63.4 percent never encountered any difficulties when going on Sunday outings, while only a small percentage, 3.6 percent, reported facing economic challenges in this regard(Magliano et al., 2015). About 32 percent of the caregivers in the study always felt depressed, much higher as compared to a study from Italy. Fifty three percent (53 percent) of caregivers always found a great deal of difficulty in getting an opportunity for leisure activities. In the study on caregivers QoL from Italy, 56.6 relatives bore the guilt of transmitting illness to their children. 63.2 percent believed that they have received adequate information from clinicians on how to cope with patient's medical emergencies. Some relatives stated they felt sure they have someone who

would take care of them in case of their own physical illness, and 76.8 percent reported 2 or more trustworthy friends on whom they could rely. Further, 75.7 percent believed their friends would help them in the case of the patient's emergencies. In the present study, 61 percent of respondents believe that they were adequately informed regarding the disease. Scores of QoL of social relationships at baseline for caregivers suggest that moderate social support from relatives and friends was available. The psychological burden was directly related to severity of disease in the Italian study and quality of life of psychological health of caregivers in the present study was moderate and no association with the severity of disease was ascertained (Landfeldt et al., 2016). Only 44 percent of caregivers felt safe in their day-to-day life.

It was found that like any other genetic disease, DMD disease has an impact on several aspects of life, ranging from economic burden, lifelong dependency, and associated social stigma. A cross-sectional study from Southern Karnataka of India assessed the quality of life and mental health condition among caregivers of genetic disease patients using the WHO-BREF scale. The median QoL scores for the physical health, psychological, social relationship, and environmental health scores were 81, 69, 75, and 69 respectively. The psychosocial health and environmental and social relationships were the most affected (Mohandas et al.,2021). Another study from Brazil assessed the QoL of caregivers of children suffering with asthma using the WHOQOL-BREF questionnaire and compared the scores with parents of healthy children. Mean scores for physical, psychological, social, and environmental QoL were 63.03, 63.66, 63.74 and 55.38 respectively for caregivers. Environmental health was impacted the most. Authors elucidated, family factors as key components of understanding the quality of life not only for children with the disease but also for their caregivers. Family factors which were analysed included family resources (material and assistance) as well as family challenges (future perspectives). Such factors are potentially modifiable and can be included in practical interventions aiming in improving the quality of life of family members. The authors also suggested that psychological interventions focused on the recognition and appreciation of care, along with the positive revaluation of stressful situation, can help in coping strategies and improve the parents' quality of life (Roncada et al., 2015).

The median QoL scores for physical health, psychological, social relationship, and environmental health scores were 56, 56, 56 and 50 respectively. These scores suggest that the impact of DMD on the caregiver's QoL is worst. The most affected domain was environmental health. A web-based observational study conducted by Schwartz et al. (2021)

included DMD caregivers, and parents of children with and without DMD stratified by subjects Age Group. There were many Outcomes which included quality of life, resilience, caregiver impact, stressful life events, financial strain, out-of-pocket expenditures, work productivity and unrealized ambitions. Compared to parents without a DMD child, DMD caregivers reported good physical health but bad mental health. Providing caregiving support for DMD teenagers was the most difficult challenging, caregivers had sacrificed their education and professional life to meet demand of caregiving. DMD caregivers have to face certain hidden costs that impact their health and financial well-being, but parents of DMD children of all ages maintain notable resilience and positivity (Schwartz et al., 2021). The present study observed the same pattern was seen, quality of life of physical, psychological, social and environmental health remained almost the same with the advancing stages of disease at baseline. However, at the end of the study after 12 months when again the quality-of-life assessment was conducted for caregivers, it was seen that quality of life in the domains of environment and Physical health have shown significant deterioration with advancing stages although psychological health and social health were unaltered.

### 5.6 Determinant of Quality of Life

EQ 5D utility scores of HrQoL of patients significantly decreased with the progression of the disease. This decline in means course of HrQoL was also noticed at the start of the intervention and the end of the intervention. At all the data points the decline was statistically significant. (table 5.6.1). The maximum quality of life score was 0.98 in stage 1 at baseline the same score was maintained at the start of the intervention and a slight decline was noticed at the end of the intervention (0.96). This further reinforces that with the progression of age, HrQoL declines as the disease progresses. In stages II and III of the disease different trend was noticed, the baseline scores of 0.78 and 0.50 increased to 0.79 and 0.57 respectively by the end of the intervention. The same trend was noticed in stage IV as well at the start of 0.04 but an improvement is seen by the end of the intervention it was raised to 0.11. (table 5.6.1).

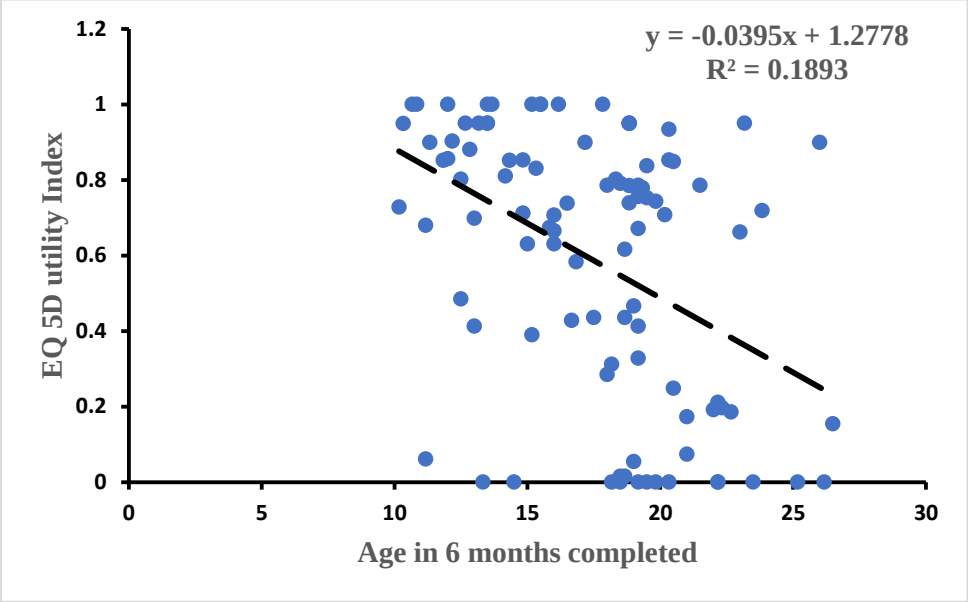
**Table 5.6.1 Health-related quality of life of dystrophinopathy patients with clinical severity of the disease.**

| *Quality of Life* | Clinical Severity | | | | | p-value |
|---|---|---|---|---|---|---|
| | I | II | III | IV | V | |
| | Mean score ±SD | Mean score ±SD | Mean score ±SD | Mean score ±SD | Mean score ±SD | |
| EQ 5D score at baseline | 0.98±0.02 | 0.78±0.22 | 0.50±0.35 | 0.19±0.24 | 0.0 | 0.00 |
| EQ 5D score at "Start of intervention" | 0.98±0.02 | 0.78±0.26 | 0.57±0.34 | 0.04±0.09 | - | 0.007 |
| EQ 5D score at "End of intervention" | 0.96±0.07 | 0.79±0.23 | 0.57±0.31 | 0.11±0.17 | - | 0.047 |

***Source:*** *Authors calculation established on primary data*

HrQoL scores at baseline (EQ 5D utility scores and PedsQoL) were not significantly associated with SES. However, EQ 5D utility scores are significantly and inversely associated with age. Linear regression showed that the EQ 5D utility scores index changed by 18.9 percent with every 6 months change in age (figure 5.6.1). All four domains of QoL in caregivers were directly associated with SES. Linear regression showed that the domains i.e. physical health, psychological, social relationship and environment in caregivers changed by 3.2 percent, 4.8 percent, 7.2 percent and 16.3 percent with every single unit change in SES. All four domains of QoL in caregivers are significantly correlated with each other.

**Figure 5.6.1 : EQ 5D-3L utility index scores directly associated with advancing age**




***Source:*** *Authors calculation established on primary data*

### 5.6.1 Multiple Linear Regression Analysis

A multiple linear regression was conducted to estimate the impact of exposure while accounting for the influence of other variables, thereby controlling for potential confounding factors. Multiple linear regression was employed to assess the influence of different predictors on the QoL of caregivers in all the domains and HrQoL children. The predictors used were SES, educational status of main caregiver, family structure, locality, distance of provinces to the health facility, respondent type, age, parents knowledge of DMD clinical severity and HrQoL (EQ 5D utility index). The common multilinear regression equation used was: $y = \beta0 + \beta1x1 + \beta2x2 + \beta3x3 + \beta4x4 + \ldots\ldots\ldots\ldots\ldots\ldots + \beta nxn$

Whereas, y: total cost of illness or total direct cost of illness or total indirect cost of illness or indirect cost related to changes in work or absenteeism due to illness. x1, x2, x3….. xn : are the predictors listed above β0 : The intercept 0 is the value of the outcome y when all predictor variables are zero.

**HrQoL (EQ 5D index baseline) :** In the study, multiple linear regression analysis was performed to account for the potential confounding effects of various covariates. The covariates that were considered for confounders in the analysis included:

1. **Age at baseline:** This is likely a crucial factor to consider since age can have a significant impact on HrQoL, where illness is progressive.

2. **Family structure:** The family's structure or composition may influence the caregiving dynamics and support available to the child, which in turn can affect HrQoL.
3. **Type of Disease**: The two disease are considered one is DMD and the other is milder form i.e. BMD or intermediate disease.
4. **Family History:** When family history of a disease is present, caregivers often have prior knowledge or awareness of the condition. This prior awareness can indeed contribute to improved HrQoL for the child with the disease.
5. **Family structure:** Whether joint or nuclear, can affect caregiving dynamics and support, potentially influencing the health-related quality of life of children with health conditions.
6. **SES:** SES can significantly impact HrQoL outcomes, as it reflects economic and social factors that influence access to resources, healthcare, and overall well-being.

**Table 5.6.2: Adjusted association of QoL of children and their caregivers with socio-economic factors, clinical severity and distance from health facility in a multilinear regression model**

| Dependent Variables of QoL (N=100) | Predictors *Standardized Coefficients (pValue)* | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | **SES** | **Education Status of main caregiver** | **Family Structure** | **Locality** | **Distances of provinces to the health facility** | **Respondent at Baseline** | **Age in Years at baseline** | **DF** | **Parent's knowledge of DMD** | **Clinical Severity** | **CHE** | **EQ 5D-3L utility index** |
| EQ 5D-3L utility index for Children Score (0-1) | 0.11 (0.41) | 0.1 (0.46) | -0.10 (0.29) | -0.13 (0.17) | **0.20 (0.04)** | -0.01 (0.88) | **-0.42 (0.00)** | - | 0.06 (0.51) | **-0.29 (0.001)** | - | - |
| PedsQL 3.0 Neuromuscular Module. (Physical Health) Score (0-100) | -0.02 (0.88) | -0.06 (0.67) | -0.08 (0.42) | 0.10 (0.32) | **0.20 (0.048)** | -0.06 (0.53) | **-0.45 (0.00)** | - | -0.01 (0.95) | **-0.18 (0.056)** | - | - |
| Overall quality of life Score (0-100) | **0.32 (0.002)** | - | - | -0.01 (0.89) | -0.03 (0.74) | - | - | - | - | 0.13 (0.21) | - | -0.06 (0.55) |
| Overall health Score (0-100) | -0.24 (0.136) | -0.12 (0.426) | 0.176 (0.104) | - | -0.082 (0.479) | - | - | -0.11 (0.31) | - | -0.016 (0.886) | - | 0.046 (0.682) |
| Physical Domain Score (0-100) | **0.184 (0.081)** | - | - | 0.009 (0.931) | -0.043 (0.686) | - | - | - | - | 0.134 (0.226) | - | 0.046 (0.676) |
| Psychological Score (0-100) | **0.31 (0.048)** | -0.05 (0.74) | -0.16 (0.13) | | -0.02 (0.84) | - | - | 0.18(0.09) | - | 0.02 (0.83) | - | 0.09 (0.41) |
| Social relationship Score (0-100) | **0.42 (0.004)** | -0.06 (0.65) | -0.09 (0.37) | 0.15 (0.14) | -0.02 (0.86) | 0.09 (0.32) | -0.04 (0.70) | | 0.11 (0.25) | -0.13 (0.20) | -0.03 (0.79) | 0.13 (0.24) |
| Environmental Score (0-100) | **0.49 (0.00)** | -0.12 (0.36) | -.124 (0.16) | -0.06 (0.49) | -0.15 (0.11) | -0.07 (0.38) | 0.01 (0.91) | - | 0.02 (0.85) | 0.133 (0.12) | 0.03 (0.77) | **0.25 (0.01)** |

SES: Socio-economic status
CHE: Catastrophic health expenditure
DF: Distress Financing

***Source:*** *Authors calculation established on primary data*.

After adjusting for confounders, the statistically meaningful regression equation found that the HrQoL of children was significantly associated with three factors (table: 5.6.2):

1. **Age at baseline:** This suggests that as children grow older, their HrQoL decrease.
2. **Clinical severity:** This suggests that as clinical severity in children increases from ambulation to non-ambulation quality of life worsen.
3. **Distances of provinces to the health facility**: Surprisingly patients residing in far flung areas had better quality of life as compared to nearby places.

**Table 5.6.3 Analysis of variance for the multiple regressions related to QoL of children and their caregivers**

| **Dependent Variables of QoL (N=100)** | **Source of variation** | **Sum of Squares (SS)** | **d.f.** | **Mean Square** | **F= MS regression/ MS residual** |
|---|---|---|---|---|---|
| EQ 5D-3L utility index for Children Score (0-1) | Regression | 4.25 | 9 | 0.473 | 5.08, p= 0.000 |
| | Residual | 8.38 | 90 | 0.093 | |
| | Total | 12.63 | 99 | | |
| PedsQL Neuromuscular Module. (Physical Health) Score (0-100) | Regression | 9045.63 | 9 | 1005.07 | 4.12, p=0.000 |
| | Residual | 21960.02 | 90 | 244.00 | |
| | Total | 31005.65 | 99 | | |
| Overall quality of life Score (0-100) | Regression | 8115.38 | 5 | 1623.07 | 2.586, p=0.031 |
| | Residual | 59003.37 | 94 | 627.69 | |
| | Total | 67118.75 | 99 | | |
| Overall health Score (0-100) | Regression | 5878.22 | 7 | 839.74 | 0.682, p=0.687 |
| | Residual | 113221.78 | 92 | 1230.67 | |
| | Total | 119100.00 | 99 | | |
| Physical Domain Score (0-100) | Regression | 1203.88 | 5 | 240.77 | 1.157, p=0.336 |
| | Residual | 19558.76 | 94 | 208.07 | |
| | Total | 20762.64 | 99 | | |
| Psychological Score (0-100) | Regression | 3461.26 | 7 | 494.47 | 1.862, p=0.085 |
| | Residual | 24426.18 | 92 | 265.50 | |

| | | | | | |
|---|---|---|---|---|---|
| | Total | 27887.44 | 99 | | |
| Social relationship Score (0-100) | Regression | 20766.04 | 11 | 1887.82 | 3.240, p=0.001 |
| | Residual | 51270.96 | 88 | 582.62 | |
| | Total | 72037.00 | 99 | | |
| Environmental Score (0-100) | Regression | 28494.83 | 11 | 2590.44 | 7.14, p=0.000 |
| | Residual | 31936.48 | 88 | 362.91 | |
| | Total | 60431.31 | 99 | | |

***Source:*** *Authors calculation established on primary data*

In contrast to the findings for children's HRQoL , QoL for caregivers, all domains of their QoL demonstrated a significant and direct correlation with socio-economic status and to each other. Environment and social relationship domain of QoL caregivers is positively correlated to EQ 5D utility scores (r=0.22 p=0.028 and r=0.21 p=0.033) but not to PedsQoL health (r=0.17 p=0.086 and r=0.18 p=0.066) of children. While psychological and physical health of caregivers are independent of HRQoL of their children. This implies that the different aspects of caregivers' well-being, such as physical, psychological, social, and environmental factors, were influenced by their socio-economic status.

However, multiple linear regression analysis was performed to account for the potential confounding effects of various covariates. The covariates considered for all sorts of QoL are listed in table 5.6.2 After adjusting for confounders, the statistically meaningful regression equation found that the HrQoL of children was significantly inversely associated with two factors age of child, disease severity and directly associated with distances of provinces to the health facility. Similarly, adjusted overall QoL, psychological, social relationship and environment QoL of caregivers were directly associated with SES. Only environment QoL of caregivers was additionally directly associated with HRQoL of their child. Quality of physical health of caregivers is independent of any such factor. These findings emphasize the multifaceted nature of caregivers' experiences and the need to consider socio-economic factors when assessing and addressing their QoL. Tailored interventions and support programs can be developed to target specific areas of need based on the SES and education levels of caregivers(Hibbard et al., 2013). These results indicate that

age and the type of disease are key factors that should be taken into account when evaluating and supporting the HrQoL of children in the context of the study. It's important to continue monitoring and researching these factors to provide more targeted and effective interventions for improving the well-being of children with different health conditions.

## 5.7 Quality of Life Among Patients and their Caregivers with Non-Intervention (Phase I) and Intervention Phase (Phase II)

The change in the QoL of physical health after the intervention was noticed, and the QoL physical health significantly deteriorated with clinical severity. The reason may be during the intervention which was patient-centric, physiotherapy and Rehab videos were shared with the caregivers reinforcing the importance of regular physiotherapy in maintaining the functional abilities of their child. This reinforcement improves their caregiving skills, nonetheless, patients with severe disease were difficult to manage and require more proactive care. That had made a significant impact on their physical health with the progression of the disease however overall QoL of the physical health of caregivers increased after the intervention.

The EQ 5D utility scores have shown an increasing trend with time but the median remained the same. The rising trend in the scores of HrQoL is an unusual finding as the disease is progressive and has a severe impact on the functional mobility of the patients. The data itself has shown deterioration in the functional status of these patients. The likely cause of kids reporting better quality of life was that they felt more involved in the management of their own disease after an intervention. Patients felt ownership in the management of their own disease and this is the basic objective of the PCC model.

**Table 5.7.1 Comparison of QoL of Children and caregivers during non-intervention (Phase I) and Intervention phase (Phase II)**

| Quality of Life (N=66) | Baseline | Start of intervention | End of intervention | p- value (paired t-test) Baseline and endpoint | p- value (Paired t-test) Start point and end point |
|---|---|---|---|---|---|
| EQ 5D utility score | 72.9±20.1 | 76.4±21.2 | 73.8±21.4 | 0.15 | 0.75 |
| Physical health | 51.4 ±13.9 | 52.3 ±12.8 | 55.1 ±12.0 | 0.024 | 0.021 |
| Psychological health | 51.2 ±16.6 | 51.7 ±17.4 | 53.7 ±17.4 | 0.204 | 0.178 |
| Social health | 54.4 ±24.9 | 56.9 ±26.2 | 55.3 ±20.5 | 0.461 | 0.668 |
| Environmental Health | 48.15 ±22.5 | 48.2 ±22.0 | 55.5 ±13.9 | 0.00 | 0.00 |

***Source:*** *Authors calculation established on primary data*

HrQoL of patient showed no improvement after intervention. This finding is not unusual quite usual as health-related quality of life depends upon severity or progression of disease with time. The, scores had not shown any deterioration as well, this was a positive impact, as over time the quality of life patients has not deteriorated as expected. At baseline about 66 percent of caregivers mentioned / assessed their quality of life to be very poor or poor by the end of the intervention there were only 40 percent mentioned their quality of life to be poor or very poor. The physical health quality of caregivers has improved significantly and the same was true for environmental health. However, quality of psychological health and social relationships remained the same even after intervention. During interactive sessions with the patient and their caregivers at the time of the start of phase 2, caregivers were given a booklet as well as given hands-on training regarding how to deliver home-based Physiotherapy training sessions. The videos of exercise sessions were also shared with the patients mentioning do's and don'ts about the exercise and also about what are the harmful

effects caregivers might go through while taking care/ physiotherapy to their child and how to avoid them. This was reflected in their improved physical health after the intervention. During interactive sessions in Phase 2 caregivers discussed the elements of the family environment and the physical environment which were hindering caregiving. The research team along with caregivers and patients discussed the appropriate solutions that might facilitate a child's health.

### 5.8 Conclusion

The intervention was designed considering the needs of the stakeholders (children, their parents/caregivers and health care providers) by conducting group discussions, which was essential for the success of any intervention. Pre and post-intervention data were used to evaluate the programme effectiveness, on whether this patient-centered care model (intervention) could contribute to reducing the financial burden on families. The children's mean age was 9.4 ± 2.0 years initially, and 9.9 ± 2.1 years at the study's end. Non-ambulatory patients increased from 9 to 16. The intervention notably reduced the total direct cost proportion to about 10 percent, indicating reduced medical resource use and associated costs. This suggests a positive outcome in terms of reducing medical resource consumption and related expenses for patients. However, severity of the disease continued to increase. The annual total mean direct expenditure with intervention was ₹44.0 (6.5-305.4) x 1000. The direct cost accounts for about 41.16 percent of the total annual cost of illness. The direct medical expenditure is approximately ₹23.1 x1000, ranging from ₹2.4-16.5 x 1000. This constitutes about 22 percent of the total cost of illness per year in families with PCC intervention. Orthotics/aids, a common unmet need, had minimal expenditure. The rehabilitation services accounted for the next significant portion of direct medical burden (3.64 percent of total COI) during Phase 2. There's a significant difference in direct medical costs between different disease stages with intervention (ANOVA test P=0.01). This contradicts findings from other studies where both direct medical and non-medical COI increased with disease severity, particularly between DMD stages IV and V (Katz et al., 2014; Landfeldt et al., 2014). Unlike settings offering mobile nursing services and house modifications, unavailable in India, present study highlights food, travel, boarding, and

lodging as major cost drivers. Constituting about 11 percent of the total annual cost of illness. However, this cost didn't vary significantly during the intervention phase, likely due to telemedical consultations in the PCC model. In the intervention phase, caregiving was not found to correlate with disease severity, as depicted in table 5.2.1. However, caregiver empowerment enhances caregiving quality in terms of hours spend in caregiving, as illustrated in table 5.3.1 Enhanced caregiver empowerment corresponded to improved caregiving, particularly among unpaid caregivers. These results highlight the beneficial effects of caregiver empowerment on caregiving quality and, consequently, on the quality of life for both patients and caregivers.

During the intervention phase, there was a significant 19.5 percent reduction in direct medical expenditure, attributed largely to the teleconsultation component of the PCC intervention. This reduction encompassed costs related to procedures, user fees, orthotic devices, and rehabilitation services. The out-of-pocket expenditure on travel, boarding & lodging decreased considerably in the intervention Phase. Mean expenditure was reduced by almost 16 percent. (Paired t-test P=0.023). Teleconsultation particularly emphasized during the COVID-19 pandemic, enhances healthcare accessibility by reducing costs associated with transportation and missed work, while ensuring continuous high-quality care and regular follow-ups(Gilkey et al., 2022). Contradictory to the direct expenditure (non-medical and medical) the total indirect cost increased in the intervention phase this is subjected to the increased economic burden due to loss of productivity in unpaid caregiving. However, there was a notable decrease (6.2 percent) in productivity loss related to absenteeism and change in the working capacity of the caregiver during Phase II. The burden of loss of productivity in unpaid caregiving significantly increased by 37.9 percent in Phase II, contributing to an 11.7 percent rise in the overall indirect cost of illness, although statistically not significant (paired t test, z=1.31). The duration of caregiving was not associated with the disease severity but correlated significantly with caregiver empowerment. Education and training on disease management empower caregivers, enhancing caregiving quality (Ota et al., 2006). Mean hours spent in unpaid care increased from 4.4 to 6.1 during the intervention, reflecting a 37.9 percent economic burden rise. Training programs modify caregiver behavior, caregivers become more receptive to counsellors and educators from DMD specialists. Although this

study lacked sufficient follow-up duration, previous research underscores the positive influence of caregiver training on disease management and home-based self-care practices, especially in lifelong and complex conditions like DMD (Fukkink & Lont, 2007).

The EQ-5D utility index, using adult tariffs, was applied to the Indian pediatric population for the first time. HrQoL scores of children as mean EQ 5D (S.D) utility score and PedsQoL for physical health were 0.58 ±0.38 and 77.7±17.7 respectively. Both were inversely correlated with advancing age and clinical severity of disease. HrQoL of boys has no significant correlation with their socio-economic status. The study evaluated caregivers' quality of life (QoL) using WHOBREEF scales, revealing average scores of 29.7 ± 26.0 for overall QoL and 53.0 ± 34.7 for health-related QoL. The caregivers rated their overall QoL significantly lower than their health alone. WHOBREEF assessed physical, psychological, social, and environmental domains. Around 69.5 percent rated their QoL as poor or very poor, with 68.4 percent dissatisfied with their health initially. Physical QoL remained generally good, while psychological health remained consistently not very good, and social health showed a declining trend with disease severity, though not statistically significant (P=0.16). Environmental health remained consistently reported as good across all disease stages. The environmental and social relationship domains of caregivers' quality of life (QoL) were positively correlated with EQ 5D utility scores (r=0.22, p=0.028 and r=0.21, p=0.033), but not with the Pediatric Quality of Life (PedsQoL) health of children (r=0.17, p=0.086 and r=0.18, p=0.066). However, psychological and physical health of caregivers appeared to be independent of the HrQoL of their children. This suggests that caregivers' well-being is influenced by various factors, including socio-economic status (SES).

A multiple linear regression was utilized to evaluate the impact of exposure while controlling for other variables and potential confounding factors. This analysis assessed the influence of various predictors on caregivers' QoL across different domains and the HrQoL of children. Predictors included socio-economic status (SES), educational status of the main caregiver, family structure, locality, distance to health facilities, respondent type, age, parental knowledge of DMD clinical severity, and HrQoL (EQ 5D utility index). After adjusting for confounders, the regression equation showed a significant inverse association

between the HrQoL of children and their age, disease severity and a direct association with the distance of provinces to the health facility. Additionally, adjusted overall QoL, psychological, social relationship and environmental QoL of caregivers were directly associated with SES. Only the environmental QoL of caregivers was additionally directly associated with the HRQoL of their child. Quality of physical health of caregivers remained independent of these factors.

The intervention had a notable impact on the QoL of physical health with a significant deterioration observed with increasing clinical severity. During the patient-centric intervention, caregivers were provided with physiotherapy and rehab videos, emphasizing the importance of regular physiotherapy in maintaining their child's functional abilities. This reinforcement improved caregivers' skills, but patients with severe disease required more proactive care, which significantly impacted their physical health as the disease progressed. Nonetheless, overall QoL of caregivers' physical health increased after the intervention. The EQ 5D utility scores exhibited an increasing trend over time, although the median remained constant. This finding is somewhat unexpected given the progressive nature of the disease and the observed deterioration in patients' functional mobility. The likely explanation for this trend is that children felt more involved in managing their disease after the intervention. This sense of ownership in disease management aligns with the primary objective of the patient-centered care (PCC) model. The intervention did not impact the economic burden on families with children living with DMD. Similarly, the PCC model did not affect the HrQoL of children living with DMD. However, the PCC model did impact the overall QoL of caregivers of children living with DMD.

# CHAPTER VI

# SUMMARY, CONCLUSION & RECOMMENDATIONS

Duchenne Muscular Dystrophy (DMD) is an X-linked recessive disease with an incidence of 1 in 3500 to 6000 live-male births(Birnkrant et al., 2018). It progresses from an ambulatory to non-ambulatory stage and the child usually gets wheelchair confined by 11-13 years and death occurs in late teens or early twenties(Bello et al., 2016; Suthar & Sankhyan, 2018; Jumah et al., 2019). The progressive and irreversible damage to skeletal muscles results in weakness, loss of ambulation, breathing issues, and cardiomyopathy in DMD patients. DMD requires a multidisciplinary team approach for holistic management. The disease is progressive in nature however, muscle degeneration can be slowed down with the use of corticosteroids (Matthews et al., 2016; Wong et al., 2017; McDonald et al., 2018; Suthar et al., 2021). This is a lifelong chronic condition that cannot be cured, only supportive therapies exist (Bushby et al., 2010).

Several studies conducted in Europe, the United States, and Australia have consistently shown that Duchenne Muscular dystrophy (DMD) imposes a significant economic burden(E. Landfeldt et al., 2014). The mean total annual cost per patient varies widely across countries, ranging from €9000 (2012) (Cavazza et al., 2016) in Bulgaria to €79,000 (2013) in Germany (Katz et al., 2014). Studies have shown that this burden increases markedly with disease progression. Although no such study has been conducted from the South-East Asian region (Landfeldt et al., 2014; Ryder et al., 2017). The substantial economic burden of diseases like DMD can have catastrophic effects on households, particularly in countries where a significant portion of healthcare expenses is paid out-of-pocket (OOP). In India, around 47.1 percent of the total health expenditure was borne by individuals directly through out-of-pocket payments in the fiscal year 2019-20 (National Health Accounts Estimates for India 2019-20). Economic aspects and disease burden knowledge play an important role in raising funding for drug development programs for rare diseases. Most rare disease studies primarily adopted the societal perspective, and a significant portion also considered the third-party payer perspective, representing health insurance or public healthcare. However, none of these studies included the perspective of patients or their families (Bastida et al., 2017).

Due to the progressive nature of the disease the health-related quality of life deteriorates with the severity of the disease. Quality of life encompasses not only physical health but also psychological, social, and overall well-being of affected individuals. Disease burden adversely affects caregivers' quality of life, impacting their daily activities and work lives(Uttley et al., 2018). Caregivers of individuals with DMD experience significant mental and physical burdens, leading to impaired health-related quality of life, sleep quality, family function, depression, pain, stress, sexual dysfunction, and self-esteem issues. There is heterogenesity in the QoL of caregivers, Its not consistent across various geographical regions. Positive associations were identified with patient age, ambulatory status and/or ventilatory support, impacting informal care hours, physical and cognitive challenges, household cost burden with caregivers burden (Landfeldt et al., 2016; Katz et al., 2014). Quality of life serves as a vital measure for assessing the effectiveness of clinical interventions, the quality of healthcare services, and the impact of interventions on patients' well-being. Changes in quality of life reflect patients' satisfaction with their treatment, healthcare models, or delivery systems, providing valuable insights into the overall effectiveness and utility of healthcare interventions and services (Bendixen et al., 2012; Lue et al., 2017; Wei et al., 2017).

Empowering patient with the help of healthcare delivery model that has potential for improving cost-effectiveness of care, especially for people affected by long term conditions is need of an hour (Chen et al., 2016). The patient-centred care increases adherence to management, reduces loss to follow up in the healthcare facility and in turn, reduces morbidity and improves quality of life for patients(Bauman et al., 2003). The healthcare delivery system frequently using patient engagement in reducing expenditure and improving health outcomes (Hibbard & Greene, 2013; Hibbard et al., 2013). The impact of disease is both in financial form as well as affecting the QoL. The primary care is to provide first line care and to maintain the quality of life in the patients living with DMD. Cost of illness (COI) studies are important in informing decision makers when evaluating the value of new treatments. The Patient-Centered Care (PCC) model serves as a primary instrument for empowering patients, ultimately leading to cost reduction and improved health outcomes.

## 6.1 Rationale of the Study

DMD is a chronic, debilitating and rare disease, increased burden is foreseen in the future due to advancement of diagnostic techniques and increase coverage of medical care. Resultant prolonged survival and the increasing prevalence of disease is likely to have a huge economic burden over the families as well as health care system. The DMD is associated with a substantial economic burden. Indirect and informal care costs, ranging from 18 percent to 43 percent of total costs, underscore the hidden financial toll on families and caregivers. The total societal burden was estimated at between $80,120 and $120,910 per patient and annum, and increased markedly with disease progression. The corresponding household burden was estimated at between $58,440 and $71,900 (Landfeldt et al., 2014). These figures highlight the challenges families face in managing the financial implications of DMD. The economic context of the rare disease is a very important input to evaluate any health policy or intervention programme related to new treatments, financial support schemes to families (Straub et al., 2016).

The patient engagement is one of the tools for handling chronic diseases. The PCC healthcare model is used to empower patients, fostering ownership and describing their role in managing the disease within the framework of patient and family-centered care (Coulter, 2012). This ensures active partnership and participation in decision-making between patients, their families and the providers to plan a customized comprehensive strategy for care. The roles of this model are not very clear either to the clinician or the patients due to the multidimensional which complexities in case of rare diseases (Hibbard & Greene, 2013). There is the political will to address care and treatment related to rare diseases. The Indian government in alignment with the National Health Policy 2017, recognizes the significance of addressing rare diseases. This policy emphasizes patient-centered care to enhance the quality of life for individuals dealing with chronic, long-term illnesses (National Health Policy- 2017).

This study is designed to explore the QoL in patient and their caregivers giving them PCC aimed to reduce the financial burden on the families, followed for a period of time, to

evolve a replicable and feasible model of patient-centered individualized care for DMD patients in India. In this era of COVID-19, teleconsultation is the mainstay of access to medical consultation for many chronic diseases. Teleconsultation is the key feature of the study, so this study will be most apt not just for this specific disease but for any other chronic disease. Additionally, the study aimed to provide data on the economic burden of DMD (rare disease), a relatively underexplored area in LMICs.

The current study aims to address a gap by developing a model that considers both the patient and the healthcare team to enhance health outcomes, particularly the Quality of Life (QoL) for both patients and caregivers. This holistic approach, focusing on Patient-Centered Care (PCC), is novel in the context of Duchenne Muscular Dystrophy (DMD) in India. The study seeks to estimate the economic burden on families with DMD patients receiving care at a government hospital, followed by implementing an intervention aimed at reducing this financial strain. Finally, the impact of the intervention on QoL and economic burden were compared to routine care after a 6-month period.

### 6.2 Economic Burden on Families with Children Living with Duchenne

The results of the analysis revealed the annual mean direct expenditure was estimated at ₹109.4 (ranging from ₹27.4 to ₹534.2) x 1000, and it is approximately 52 percent of the total annual cost of illness. Most of the direct expenditure was driven by direct medical costs, making up 30.6 percent of the total COI. In the present cohort, none of the patients incurred expenses for house or automobile modification. Conversely, none of the nine non-ambulatory patients in the cohort had access to even simple non-customized wheelchairs. The loss of productivity is caused by disease induced deficit in working capacity, the mean annual cost for unpaid caregiving was ₹33.5 (7.3-88.2) x 1000. The pathway to care for Duchenne Muscular dystrophy (DMD) diagnosis involves significant costs as caregivers consult multiple healthcare agencies for diagnosis. On average, parents consulted 2.9 treatment agencies before reaching the study site, with no significant difference observed based on disease severity. The lengthy pathway to care, averaging 12.5 months, emphasizes the need

for more efficient patient care pathways. The pathway to care is not captured in the present direct cost of illness due to considerable recall bias.

The mean time to the last follow-up at the health facility was 4.8 months, despite quarterly follow-ups being recommended. ANOVA results (P=0.01) indicate a significant delay in last follow-ups as the disease advances. Direct non-medical costs, mainly including food, travel, and lodging, accounted for 14.3 percent of the total annual illness costs. In stage 4, travel, boarding, and lodging costs averaged ₹38.8 x 1000, nearly three times that of stage 3. This expense significantly increased with disease severity, as evidenced by the ANOVA test (F=5.1). Furthermore, the expenditure on food increased with the advancement of disease severity. For instance, the cost in stage IV was ₹9.5 x 1000, almost three times higher than in stage III (3.4 x 1000 ₹). However, costs were not compared with stage 5 as there was only one patient in this group. This escalating trend in food expenditure was statistically significant with disease severity, as confirmed by the ANOVA test (F=2.8).

The productivity loss due to informal caregiving is about 30₹ of the total cost of illness and loss of productivity due to absenteeism and change in working conditions leading to reduced wages was 17.7 percent. The mean number of hours per day spent in caregiving, reflecting productivity loss due to informal caregiving, was 9.4 ± 10.9 for Germany, 6.1 ± 2.0 for Portugal, and 4.3 ± 2.1 for the present study.

## 6.3 Socio-economic Determinants of Economic Burden with Children Living with Duchenne

The majority of participants belonged to the upper-middle socio-economic status (SES), comprising 51 percent. Most children in the study came from nuclear families (54 percent). Participants primarily from northern states of India, with the highest representation from Punjab (32 percent). Other significant contributors included Haryana and Himachal Pradesh, both at 17 percent. Eastern states represented 11 percent of the participants, with Jammu and Kashmir accounting for 10 percent and Chandigarh for 9 percent. The smallest representation was from Uttarakhand, with only 4 percent of participants. Most primary caregivers were parents (96 percent), with mothers predominantly serving as primary

caregivers (58 percent). The majority hailed from rural areas (72 percent) and had education up to matriculation (46 percent). The challenges reported by patients included work time loss (74 percent), transportation difficulties (72 percent), long waiting times (71 percent), accessing diagnostic facilities (71 percent), and managing frequent visits (71 percent).

The study revealed that joint families incurred higher indirect caregiving expenses (₹64.9 x 1000) compared to nuclear families (₹42.5 x 1000). Additionally, caregivers who perceived work loss as less challenging spent significantly more on diagnostic care (₹3.0 x 1000) compared to those who found it more challenging (₹1.8 x 1000) (p=0.049). The Pearson correlation analysis showed a significant direct association between the annual total direct cost of illness and the socio-economic status (SES) of the caregiver (P=0.006), with a 7.5 percent increase in expenditure for every unit increase in SES. Similarly, the annual indirect cost of illness was directly associated with SES (P=0.013), with a 6.1 percent increase in expenditure for every unit increase in SES, as per the linear regression equation.

The multiple linear regression analysis identified key socio-economic and demographic factors that influence the direct and indirect costs of illness for families with a child affected by Duchenne muscular dystrophy (DMD). The model's goodness of fit was confirmed by significant F-statistics. Higher annual direct costs are significantly associated with the family's socioeconomic status, the education level of the primary caregiver, and the distance from the health facility. As the child ages or the disease worsens, costs tend to rise, although the relationship is not statistically significant. Indirect costs, including caregiving, are linked to the caregiver's education level, with urban caregivers spending more time on care. Joint families and those traveling from distant locations face higher productivity and wage losses. Additionally, as the child ages or the disease progresses, indirect costs increase, inversely impacting their quality of life. These findings highlight disparities in healthcare access and affordability, with socio-economic status and geographical factors playing crucial roles.

Additionally, indirect cost of illness (caregiving) increased with the deterioration of health-related QoL in children with DMD and greater parental awareness of the disease.

Similarly, indirect cost of illness was associated with the educational status of the main caregiver, with urban caregivers spending more time on caregiving.

## 6.4 Catastrophic Healthcare Expenditure of DMD Among Households in Northern India

The out of pocket (OOP) refer to added costs on health not covered by the health system, which patients or families must pay themselves(Prinja et al., 2019). These expenses may compel patients to make difficult choices between healthcare and other essentials, leading to an inequitable distribution of health services. World Health Organization (WHO) defines health expenditure as catastrophic when it surpasses 40 percent of a household's remaining income after covering basic subsistence needs. Among the recruited patients, 40% experienced catastrophic health expenditure, which rose to 76 percent, 63 percent, and 29 percent when the cut off for CHE was set at 20 percent, 30 percent, and 50 percent, respectively. Additionally, 45 percent of the patients experienced distress-financing (DF). The association between CHE and DF was examined in relation to various predictors of the cost of care. Factors such as the patient's age, disease progression, age at symptom onset, socio-economic status of the caregiver, knowledge about the disease, educational status of the caregiver, province of origin, locality, and family structure were considered. Significant associations were observed between CHE and higher socio-economic status ($\chi 2$ 6.6; $p < 0.037$), caregiver ignorance of the disease outcome ($\chi 2$ 8.8; $p < 0.003$), proximity of the patient's province to the healthcare facility ($\chi 2$ 15.2; $p < 0.000$), and symptom onset before the age of 5 ($\chi 2$ 10.8; $p < 0.001$).

The caregivers who lacked knowledge about the disease prognosis were less likely to experience CHE compared to those who were aware of the disease outcomes (OR 0.3; 95% CI 0.125–0.671). Patients whose symptoms were noticed after the age of 5 were four times more likely to face CHE (OR 4.0; 95% CI 1.72–9.35). Those traveling from distant locations to seek healthcare at PGI were 8.4 times more susceptible to CHE (OR 8.0; 95% CI 2.64–26.7) compared to those from nearby provinces. Susceptibility to CHE was 11 times higher in patients facing distress financing (DF) (OR 11.3; 95% CI 4.36–29.36). Similarly, caregivers

whose patients noticed the disease after age 5 were four times more likely to face DF (OR 3.7; 95% CI 1.6–8.4). The patients traveling from distant locations were 4.4 times more likely to experience DF (OR 4.4; 95% CI 1.48–13.1) compared to those from nearby provinces.

In the multiple logistic regression analysis (Omnibus test of model), conducted at a 40 percent threshold for catastrophic expenditure, caregivers who were unaware of the disease had significantly lower odds of facing catastrophic health expenditure (CHE) (OR: 0.2, 95% CI: 0.1–0.5, p-value: 0.002) compared to those with knowledge about the disease. Understanding the disease empowers individuals to explore treatment options, which may contribute to higher costs in the absence of a definitive cure. Additionally, patients from remote areas had a significant increase in the likelihood of experiencing catastrophic health expenditure, with odds 5.6 times higher. This highlights the potential benefit of teleconsultation services in bridging geographical gaps and providing healthcare access to those in distant areas, potentially reducing CHE. Furthermore, patients whose parents noticed symptoms at a later age (after 5 years) had a 2.7 times higher risk of resorting to distress financing. This emphasizes the importance of early disease detection and the need for increased awareness and education among peripheral physicians about the disease.

### 6.5 Impact of Intervention on Economic Burden and Quality of life.

During the study period, intervention known as the Patient-Caregiver-Centered (PCC) model was developed to empower both patients and caregivers by providing them with essential knowledge about the disease and available treatment options. The intervention imparted their active involvement in the management of the disease. A crucial component of the PCC model was teleconsultation to address the challenges posed by long-distance travel, especially for functionally disabled patients. Moreover, it aimed to make medical consultations readily available, which became particularly important during the COVID-19 era when in-person visits were restricted or limited. During interactive sessions in Phase 2, caregivers received hands-on training on delivering home-based physiotherapy sessions, along with educational materials and videos demonstrating exercises and potential risks to avoid. In the intervention phase direct medical expenditure decreased significantly by 19.5 percent (28.7 x ₹ 1000 (routine care) to 23.1 x ₹1000 (intervention care). This decline was

primarily attributed to the teleconsultation component of the PCC intervention. The implementation of teleconsultation effectively mitigated travel-related expenses as travel, boarding & lodging costs were significantly decrease from 13.5 x ₹1000 (routine care) to 11.0 x ₹1000 (intervention care) (paired t test statistics p=0.023). The utilization of direct medical resources was notably higher during routine care compared to the intervention phase, although this difference did not reach statistical significance, possibly due to the shorter follow-up period in the intervention phase.

Contrary to the direct expenditure, total indirect cost increased during the intervention phase, primarily due to economic burden from loss of productivity in unpaid caregiving. However, there was a decrease (6.2 percent) in productivity loss due to absenteeism and changes in caregiver working capacity, similar to other cost reductions observed in Phase II (intervention phase). The burden of productivity loss in unpaid caregiving statistically significantly increased by 37.9 percent in Phase II. Although the increase in unpaid caregiving costs contributed to an 11.7 percent rise in the overall indirect cost of illness in Phase II, this difference was not statistically significant (Paired t Test, z=1.31). Training and education on disease management significantly empowered caregivers, enhancing caregiving quality and positively impacting disease management and self-care practices at home. This effect was particularly pronounced in complex, lifelong disease management scenarios. The mean duration of unpaid informal care during routine care was 4.4 hours, which increased to 6.1 hours during the intervention phase, reflecting the increased burden of productivity loss in unpaid caregiving.

EQ-5D utility index, using adult tariffs, was applied to the Indian pediatric population for the first time. HrQoL scores of children as mean EQ 5D (S.D) utility score and PedsQoL for physical health were 0.58 ±0.38 and 77.7±17.7 respectively. These scores were inversely correlated with advancing age and clinical severity of disease but no correlation with their socio-economic status. Caregivers' QoL, assessed using WHOBREEF scales, showed average scores of 29.7 ± 26.0 for overall QoL and 53.0 ± 34.7 for health-related QoL, with caregivers rating their overall QoL lower than their health alone. Physical QoL remained generally good, while psychological health was consistently moderate, and social health showed a trend

towards decline with disease severity. Environmental health remained consistently good across all stages. Caregivers' QoL domains were positively correlated with EQ 5D scores but not with PedsQoL of children. Linear regression analysis showed changes in SES associated with alterations in caregivers' QoL domains: physical health (3.2 percent), psychological (4.8 percent), social relationship (7.2 percent), and environment (16.3 percent). Multiple linear regression analysis was conducted to address potential confounding effects, considering various covariates listed. After adjustment, the regression equation revealed that the health-related quality of life (HrQoL) of children was significantly inversely associated with two factors: the age of the child and disease severity, while being directly associated with the distance of provinces to the health facility. Similarly, the adjusted overall QoL, psychological, social relationship, and environmental QoL of caregivers were directly associated with SES. Additionally, only the environmental QoL of caregivers was directly associated with the HrQoL of their child. The quality of physical health of caregivers appeared to be independent of these factors.

The HrQoL of patients did not show improvement after the intervention, which is not unusual given that HrQoL often depends on the severity or progression of the disease over time. However, scores did not deteriorate either, which is a positive outcome indicating that the patients' quality of life did not worsen as expected. At baseline, about 66 percent of caregivers rated their quality of life as very poor or poor, but by the end of the intervention, this percentage decreased to 40 percent, indicating an improvement. The physical health quality of caregivers significantly improved, as did their environmental health, although the quality of psychological health and social relationships remained unchanged.

### 6.6 Recommendations

The study has public health implications and can guide the policymakers in formulating policy or guidelines for a more patient-centric program in managing a chronic life-long rare disease not only in India but also in other low- and middle-income countries. This study also provides the most recent description of illness costs and treatment patterns in DMD. There is lack of scientific data on impact of DMD in LMIC.

1. The study indicates a lack of demand for medical and supportive care for advanced stages of disease (stages 4 and 5), primarily because caregivers are unaware of available treatment options. Thus, centres of excellence on rare diseases should launch targeted training and educational programs for caregivers and communities. This approach will help generate the necessary demand for medical resources and supportive care.

2. The study highlights limited access to wheelchairs and rehabilitation in India, exacerbated by inaccessible public spaces and lack of training. To improve disability support, a comprehensive system re-evaluation is needed, emphasizing collaboration among government, NGOs, private organizations, and healthcare providers to create an inclusive environment for all.

3. The study revealed that majority of expenses are attributed to direct medical costs, with the proportion of these costs exceeding those incurred by European countries that benefit from medical coverage funded by their federal governments. Consequently, financial burden on families in India is much higher compared to other countries. To address this disparity, the government should alleviate these costs by supporting medical treatments, periodic diagnostic assessments, and necessary surgeries for all patients, regardless of income.

4. The study has observed that caregivers in India faces a significant caregiving burden. The study has identified that caregivers spend approximately 13 days annually exclusively for visiting healthcare facilities. Many caregivers reported taking time off for health facility visits but did not specify their work absences related to caregiving or health issues arising from their responsibilities. The study suggests to improve healthcare accessibility for such diseases with teleconsultation as a viable solution. The other methods like use of helpline for meeting information needs of patients, their families and caregivers is a viable solution.

5. In addition to teleconsultation, establishing helplines to address the informational needs of patients with rare diseases, their families, and caregivers is identified as an

effective approach. These helplines can assist in scheduling visits to specialized centers and coordinating arrangements for multidisciplinary healthcare management.

6. Another approach to improving healthcare accessibility is by establishing specialized centers at district-level hospitals. This initiative requires highly skilled healthcare professionals; however, the current shortage of specialized manpower presents a significant challenge. This gap can be addressed by training healthcare professionals at the district level through outreach sessions conducted by the eight designated Centers of Excellence across India. Such training programs will enable early disease detection, ensure smooth referral processes, and expand the reach of essential rehabilitation services necessary for managing these conditions effectively.

7. Ayushman Bharat provides insurance for various pediatric and adult conditions requiring hospitalization, it excludes rare diseases like Duchenne Muscular Dystrophy. Similarly, the Niramaya Health Insurance Scheme under the National Trust offers up to ₹1 lakh in coverage for people with disabilities, covering hospitalization, therapies, and surgeries. However, this amount is insufficient for the high costs of treating rare diseases. A proposed scheme with a ₹50 lakh lifetime coverage for rare diseases is under consideration, though its effectiveness is yet to be evaluated. Apart from that it doesn't include supportive management of the disease.

8. The study has shown that integrating teleconsultation into standard care protocols can help effectively to reduce the financial burdens on patients and caregivers. It can enable the caregivers to follow treatment guidelines while lowering direct as well indirect cost of treatment. Although less favored by caregivers of recently non-ambulatory patients, teleconsultation was well-received by previously non-ambulatory and ambulatory patients.

9. The study finds that higher socio-economic status and education levels lead to increased spending on DMD treatment due to better access to resources and understating. However, distance from healthcare facilities can significantly elevate costs, highlighting disparities in access to health facility. These findings underscore

the need for targeted public health interventions, such as patient-centered care and financial support, to ensure equitable access to care for all families affected by DMD.

10. This study provides the baseline data on the economic burden experienced by families with a child having DMD in the Southeast Asian region. Additionally, study can also be referred to generate quality-of-life years from DMD QoL data. This information can then be used to inform economic models of the cost-effectiveness of interventions in DMD.

11. The transition from pediatric to adult care and social involvement is vital in DMD care. Education and employment offer independence and well-being. NEP 2020 emphasizes non-discrimination, accessible infrastructure, accommodations, and support for children with muscular dystrophy. Prioritizing education and employment opportunities can help to foster independence, inclusivity, and enhances the overall quality of life for affected individuals.

12. The Patient-Centered Care (PCC) model, which prioritizes individual needs and integrates with standard operating care, is highly effective in fostering collaboration among patients, caregivers, and healthcare providers. Indian centers of excellence like PGIMER can implement this model to create a single program addressing various rare or chronic diseases in children. This approach would streamline care, improve outcomes, and bolster caregiver support.

### 6.7 Scope of Future Studies

Future research can be carried out to cover the following research areas:

1. To assess the long-term health outcomes and quality of life improvements in Patient-Centred Care for DMD/BMD children.
2. A comprehensive analysis of cost-effectiveness analysis comparing Patient Centered Care with traditional care models.
3. Evaluate the integration and impact of telemedicine and mobile health technologies in Patient-Centered Care.

4. Further research may be necessary to enhance telemedicine's use in areas with limited technological access.
5. To study cultural and socio-economic factors influencing the adoption and effectiveness of Patient-Centered Care model.
6. Examine caregiver’s support and training programs impact on patient outcomes and caregiver well-being.

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## LIST OF PUBLICATIONS

**The following papers have been published or accepted for publication and thereby fulfilling the minimum program requirements as per the UGC.**

| S.No. | Title of paper with author names | Name of journal / conference | Published date | Issn no/ vol no, issue no | Indexing in Scopus/ UGC-CARE list (*please mention*) |
|---|---|---|---|---|---|
| 1 | Development and economic evaluation of a patient-centered care model for children with Duchenne's Muscular Dystrophy: A quasi-experimental study protocol | JMIR Research Protocols | April 28, 2023 | Volume 12, April 28, 2023 https://doi.org/10.2196/42491 | Scopus |
| 2 | Socioeconomic determinants of the quality of life in boys suffering from Duchenne muscular dystrophy and their caregivers. | Indian Journal of Medical Research | 30-01-2025 | IJMR_83_2024 Accepted | Scopus |
| 3 | Deflazacort dose optimization and safety evaluation in Duchenne muscular dystrophy (DOSE): | European journal of paediatric neurology: EJPN: official | May 2022 | Volume 38, May 2022, Pages 77-84, http://dx.doi.org/10.1016/j.ejpn.2 | Scopus |

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|---|---|---|---|---|---|
| | A randomized, double-blind non-inferiority trial | journal of the European Paediatric Neurology Society | | 022.04.004 | |
| 4 | Comparative performance of verbal autopsy methods in identifying causes of adult mortality: A case study in India | Indian Journal of Medical Research | Oct 2021 | Volume 154(4) Pages 631-640 | Scopus |
| 5 | Bone density and bone health alteration in boys with Duchenne muscular dystrophy: a prospective observational study. | Journal of pediatric endocrinology & metabolism | Apr 2021 | Volume 34(5) Pages 573-581 | Scopus |

Protocol

# Development and Economic Evaluation of a Patient-Centered Care Model for Children With Duchenne Muscular Dystrophy: Protocol for a Quasi-Experimental Study

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## Abstract

**Background:** Duchenne muscular dystrophy (DMD) is a rare progressive muscular disease that primarily affects boys. A lack of comprehensive care for patients living with DMD is directly associated with a compromised quality of life (QoL) for those affected and their caregivers. This disease also has a huge economic impact on families as its treatment requires substantial direct, indirect, and informal care costs.

**Objective:** This study presents a protocol developed to evaluate the feasibility and efficacy of a patient-centered care (PCC) model for children with DMD. The care model was designed with the aim to empower families, improve QoL, and reduce economic burden on their families.

**Methods:** This study is planned as a quasi-experimental study that will enroll 70 consecutive families with boys (aged 5-15 years) with DMD visiting a tertiary care center. The study is being conducted in 2 phases (preintervention and postintervention phases, referred to as phase 1 and phase 2, respectively). During phase 1, the patients received routine care. The study is now in phase 2, with the intervention currently being administered. The intervention is based on the PCC model individualized by the intervention team. The model has a comprehensive DMD telecare component that includes teleconsultation as one of its key components to reduce in-person physician visits at the health facility. Teleconsultation is especially beneficial for late-ambulatory and nonambulatory patients. Data on economic burden are being collected for out-of-pocket expenses for both phases during in-person visits via telephone or messaging apps on a monthly basis. QoL data for patients and their primary caregivers are being collected at 3 time points (ie, time of enrollment, end of phase 1, and end of phase 2). Outcome measures are being assessed as changes in economic burden on families and changes in QoL scores.

**Results:** Participant recruitment began in July 2021. The study is ongoing and expected to be completed by March 2023. The findings based on baseline data are expected to be submitted for publication in 2023.

**Conclusions:** This paper outlines a research proposal developed to study the impact of a PCC model for patients with DMD in low- and middle-income countries (LMICs). This study is expected to provide evidence of whether a multicomponent, patient-centric intervention could reduce economic burdens on families and improve their QoL. The results of this study could guide policy makers and health professionals in India and other LMICs to facilitate a comprehensive care program for patients living with

DMD. The economic impact of a rare disease is an important consideration to formulate or evaluate any health policy or intervention related to new treatments and financial support schemes.

**Trial Registration:** Clinical Trials Registry India (ICMR-NIMS) CTRI/2021/06/034274; https://ctri.nic.in/Clinicaltrials/regtrial.php?modid=1&compid=19&EncHid=14398.21116

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**KEYWORDS**

Duchenne muscular dystrophy; patient-centered care; disabilities; quality of life; caregivers; cost of illness; effective; treatment; policy; caregiver; pediatrics; intervention; psychological; disability

## Introduction

### Background

Duchenne muscular dystrophy (DMD) is an X-linked recessive disease with an incidence of 1 in 3500 to 6000 live male births [1]. It progresses from an ambulatory to a nonambulatory stage, with the child usually becoming wheelchair-bound between 11 and 13 years and death occurring in the late teens or early twenties [2-4]. Even with this short span of life, children with DMD face many challenges, as their progression through various transitional phases impacts not only them but also their families and society at large [5].

The progressive, irreversible damage to the skeletal muscles results in weakness, loss of ambulation, breathing issues, and cardiomyopathy. Currently, there is no definitive treatment to halt the disease's progression. Disease management mostly revolves around symptom management and involves a multidisciplinary approach to address various systemic complications [6].

### Course of Disease

Children with DMD usually remain asymptomatic up to 2 years of age and get diagnosed between 4 and 6 years of age. Most children with DMD are limited to assisted ambulation with the help of long leg braces by the age of 10 years. Age of loss of ambulation globally is between 13 and 14 years, whereas in resource-limited settings, it is between 9 and 11 years [4]. There is no study on the prevalence or incidence of DMD in India. However, a study from southern India [7] on 275 genetically confirmed patients with DMD was conducted to study the natural course of motor milestones. In that study, the mean age of onset of symptoms was 3.7 years, and the mean age at presentation was 8.1 years. During the follow-up period of 15 years, 155 out of 275 (56.4%) children had either become wheelchair-bound or bed-bound or died. The affected children were bound to wheelchairs by a mean age of 10.4 years and bed-bound by 11.8 years. Only 7 (2.6%) died during the follow-up period at a mean age of 15.2 years.

DMD requires a multidisciplinary team approach for holistic management. This includes diagnostic services, physical therapy, orthotics, respiratory therapy, corrective orthopedic surgery, ventilation, feeding support, and speech therapy. Muscle degeneration can be slowed down with the use of corticosteroids, while antibiotics may be needed to fight respiratory infections [8-11]. Occupational therapy, assistive devices, and a wheelchair are also beneficial for some children. Some patients may need a pacemaker for cardiac abnormalities and assisted ventilation for respiratory muscle weakness. Patients diagnosed with DMD should expect to make a successful transition to adulthood, which includes education, health care, and social support to transition smoothly from adolescence to adulthood [12,13].

The DMD Care Considerations Working Group identified eleven components of care: (1) diagnosis, (2) neuromuscular management, (3) rehabilitation management, (4) gastrointestinal and nutritional management, (5) respiratory management, (6) cardiac management, (7) orthopedic and surgical management, (8) psychosocial management, (9) primary care and emergency management, (10) endocrine management (including growth, puberty, adrenal insufficiency, and bone health), and (11) transitions of care across the life span. The mainstay of management is still restricted to physiotherapy and glucocorticoids, which help in delaying loss of ambulation, an important milestone of the disease [6,14]. Treatment modalities also have changed, especially with the advent of gene therapy and gene editing tools. However, these new modalities of treatment have not made a significant clinical impact yet [15].

### Empowering Patients and Caregivers

Globally, we are witnessing a new paradigm shift to resolve problems from a mechanistic or Newtonian worldview to a holistic view. This approach involves human values, creativity, and evolution, and it has affected the approach to understanding human health. Nevertheless, the reductionistic approach dominates the imagination of the scientific fraternity [16]. The biomedical or reductionistic approach sees patients as an isolated problem, whereas we need to understand that we need a holistic approach (ie, social, economic, sanitary, environmental, and political commitment) to manage a disease.

An inclusive paradigm has two major components: (1) self-empowerment of the patient and (2) holism. Empowering patients involves developing skills, control over resources, ensuring autonomy in decision-making, and taking ownership of their health [17,18].

Empowering patients through an effective health care delivery model has the potential to improve the cost-effectiveness of care, especially for people affected by long-term conditions [19]. Patient-centered care (PCC) increases adherence to disease management, reduces loss to follow-up in the health care system, and, in turn, reduces morbidity and improves quality of life (QoL) for patients [17].

3rd Annual National Conference of
Epidemiology Foundation of India

# EFICON 2022


CONFERENCE THEME

*Evidence-Based Medicine for Promoting Health in India*

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33. Abstract ID: EFIPAT093

**TITLE:** Socioeconomic determinants of the quality of life in boys suffering from Duchenne muscular dystrophy and their caregivers.

**Presenting Author:** Dr Titiksha Sirari

**Authors:** Titiksha Sirari[3&1], Amarjeet Singh[2], Renu Suthar[1], Shankar Prinja[2], Vishwas Gupta[3] Manisha Malviya,[1], Akashdeep Chauhan,[4] Naveen Sankhyan[1]

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**Introduction:** DMD affects skeletal muscles causing a severe degree of disability in late childhood. The condition is incurable reducing life expectancy to late teens or early twenties. Socioeconomic status (SES) is directly associated with QoL of many life-threatening diseases. However, impact of SES on the quality of life (QOL) of these patients has not been clearly defined. This study was designed to understand socioeconomic determinants of the quality of life in patients suffering from DMD and their caregivers.

**Methods:** This study included 100 boys (5-15 years) with DMD and their caregivers. Health related QoL and global QoL were assessed with EQ 5D-3L utility index for children with DMD and World Health Organization Quality of Life-BREF (WHOQOL-BREF) for caregivers. Association of QoL with family's socioeconomic status was assessed. Spearman correlations and linear regressions were used to investigate relationships between the variables.

**Results:** Median age of the boys was 9 years; 91% were ambulatory while 9% were wheelchair bound. EQ 5D-3L utility index scores for Health-related quality of life (HRQoL) for children were computed based on the recently generated tariff value set for India. The mean EQ 5D utility score was 0.58 ±0.38 and it is directly associated with advancing age that explains the effect of disease progression. However, EQ 5D utility scores are not significantly associated with SES. All four domains of QoL in caregivers were directly associated with SES. Linear regression showed that the domains i.e., physical health, psychological, social relationship and environment in caregivers changed by 3.2%, 4.8%, 7.2 % and 16.3% with every single unit change in SES.

**Conclusion:** SES is an important factor associated with QoL in caregivers of patients with Duchenne muscular dystrophy. Further, optimum interventions and measures in lower SES stats may be needed to improve QoL in caregivers, which is often overlooked.

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# Deflazacort dose optimization and safety evaluation in Duchenne muscular dystrophy (DOSE): A randomized, double-blind non-inferiority trial

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ARTICLE INFO

*Keywords:*
Duchenne muscular dystrophy
Steroids
Deflazacort
Six minute walk distance
Ambulation

ABSTRACT

*Background:* US food and drug administration has recently approved deflazacort for Duchenne muscular dystrophy (DMD) and recommended the dosage of 0.9 mg/kg/d for patients aged ≥5years. However, data assessing the minimal efficacious dose and need of dose-titration based on age or disease severity is limited.
*Objective:* To determine whether deflazacort 0.45 mg/kg/d (proposed lower dosage) is non-inferior to 0.9 mg/kg/d among newly diagnosed patients with DMD.
*Method:* A double-blinded, non-inferiority, randomized trial, conducted between December 2018 and July 2020. Newly diagnosed patient aged 5–15 years with genetic or muscle biopsy confirmed DMD and baseline 6-min walk distance (6MWD) > 150 m were screened. Patients were randomly assigned (1:1), stratified to prespecified subgroups by age (≤7years and >7years), and baseline 6MWD (≤350 m and >350 m), to receive either 0.45 mg/kg/d or 0.9 mg/kg/d regimens. The primary endpoint was the change in 6MWD, from baseline to week-24 of intervention. The trial was powered with a predefined, non-inferiority margin of 30 m. The analyses were by modified intention-to-treat (mITT).
*Result:* A total of 97 patients were enrolled, 40 receiving 0.45 mg/kg/d and 45 receiving 0.9 mg/kg/d deflazacort comprised of mITT population. For primary endpoint analysis the mean (SD) change in 6MWD from baseline to week-24 was 9.7 m (41.5) in deflazacort 0.45 mg/kg/d, and 34.7 m (43.5) for 0.9 mg/kg/d. The mean difference in change in 6MWD across the group was 24.8 m (95% CI 6.7 to 43, p value 0.008). The mean difference in change in 6MWD in the subgroups of boys ≤7 years of age was 21.8 m (95% CI -0.82, 44.5, p = 0.059), with baseline 6MWD of >350 m was 19.9 m (95% CI -2.4, 42.4; p = 0.08). The incidence of combined moderate to severe treatment-related adverse events was significant in the 0.9 mg/kg/d group by week 24 (odds ratio 0.36 [95% CI, 0.14 to 0.89], p = 0.03).
*Discussion:* The efficacy of proposed low dose deflazacort in comparison to the standard dose did not meet the prespecified criteria for non-inferiority. The low dose deflazacort was non-inferior in subgroup of patients with age ≤7 years and baseline 6MWD of >350 m.
*Trial registration:* Clinical Trial Registry-India Identifier: CTRI/2019/02/017388.

## 1. Background

Duchenne muscular dystrophy (DMD) is an X-linked, progressive, fatal, childhood-onset neuromuscular disorder that affects around 1 in 5000 live male births [1]. Despite the availability of novel therapies, corticosteroids remain the mainstay of evidence-based medical treatment [2]. However, uncertainty remains about the best steroid regimen for the newly diagnosed children with DMD [3–6].

Initial randomized studies of prednisone aiming to improve skeletal muscle strength and function in ambulatory children with DMD found that the efficacy curve plateaued at the dosage of 0.75 mg/kg/d; the minimal efficacious dosage of prednisone was 0.3 mg/kg/d [7,8]. Despite good efficacy, 0.75 mg/kg/d prednisone is associated with significant steroid-related adverse events. Subsequent work revealed that

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Quick Response Code:

# Comparative performance of verbal autopsy methods in identifying causes of adult mortality: A case study in India

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Received January 4, 2019

***Background & objectives*: Cause of death assignment from verbal autopsy (VA) questionnaires is conventionally accomplished through physician review. However, since recently, computer softwares have been developed to assign the cause of death. The present study evaluated the performance of computer software in assigning the cause of death from the VA, as compared to physician review.**

***Methods*: VA of 600 adult deaths was conducted using open- and close-ended questionnaires in Nandpur Kalour Block of Punjab, India. Entire VA forms were used by two physicians independently to assign the cause of death using the International Statistical Classification of Diseases and Related Health Problems (ICD)-10 codes. In case of disagreement between them, reconciliation was done, and in cases of persistent disagreements finally, adjudication was done by a third physician. InterVA-4-generated causes from close-ended questionnaires were compared using Kappa statistics with causes assigned by physicians using a questionnaire having both open- and close-ended questions. At the population level, Cause-Specific Mortality Fraction (CSMF) accuracy and *P*-value from McNemar's paired Chi-square were calculated. CSMF accuracy indicates the absolute deviation of a set of proportions of causes of death out of the total number of deaths between the two methods.**

***Results*: The overall agreement between InterVA-4 and physician coding was 'fair' (κ=0.42; 95% confidence interval 0.38, 0.46). CSMF accuracy was found to be 0.71. The differences in proportions from the two methods were statistically different as per McNemar's paired Chi-square test for ischaemic heart diseases, liver cirrhosis and maternal deaths.**

***Interpretation & conclusions*: In comparison to physicians, assignment of causes of death by InterVA- 4 was only 'fair'. Hence, it may be appropriate to continue with physician review as the optimal option available in the current scenario.**

**Key words** Adult mortality - causes of death - computer-coded verbal autopsy methods - InterVA-4 - physician coding of death - verbal autopsy

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# Bone density and bone health alteration in boys with Duchenne Muscular Dystrophy: a prospective observational study

Renu Suthar, B. V. Chaithanya Reddy, Manisha Malviya, Titiksha Sirari, Savita Verma Attri, Ajay Patial, Minni Tageja, Gunjan Didwal, Niranjan K. Khandelwal, Arushi G. Saini, Lokesh Saini, Jitendra K. Sahu, Devi Dayal and Naveen Sankhyan

From the journal Journal of Pediatric Endocrinology and Metabolism
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## Abstract

# LIST OF PRESENTATIONS

| S.No. | Title of paper with author names | Name of journal/conference | Organized by | Date of Conference |
|---|---|---|---|---|
| 1. | Development and economic evaluation of a patient-centered care model for children with Duchenne's Muscular Dystrophy: A quasi-experimental study protocol | JIMS-PHDCCI-KAS XVIIth International Conference on "Business Leadership in VUCA World:Driving Growth Through Effective Integration of People, Technology, and The Environment" **(Best Paper)** | Jagannath International Management School, New Delhi-110019. | 11th & 12th Feb, 2022 |
| 2. | Development of a patient-centred care model for children with Duchenne's Muscular Dystrophy and its economic evaluation: A Single group trial the mixed-method study protocol | International Conference on "Rethinking business: Designing strategies in the age of disruptions" | Mittal School of Business, Lovely Professional University | 28th January, 2022 |
| 3 | Socioeconomic determinants of the quality of life in boys suffering from Duchenne muscular dystrophy and their caregivers | 3rd Annual National Conference of Epidemiology Foundation of India EFICON-2022; **(Merit Certificate)** | Department of Community and Family Medicine, AIIMS Patna. | 4th & 5th of November 2022 |
| 4 | Intellectual Property Right (IPR) workshop | Intellectual Property Right (IPR) workshop | LPU Phagwara; Jalandhar; Punjab. | 29th & 30th of September 2022 |
| 5 | Socioeconomic determinants of the quality of life in boys suffering from Duchenne muscular dystrophy and their caregivers. | NIMHANS International Symposium On Neuromuscular Disorders-2023 | Department of Neurology, National Institute of Mental Health and Neuro Sciences (NIMHANS), Bangalore – 560029. Email: nisnmd2023@gmail.com | 7th – 9th April 2023 |

## Certificates of Paper Presentations

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on

**"Global Digital Transformation: Navigating Critical Technological, Socio-economic and Cultural Shifts to Build Future-Ready Organisations"**

**11th & 12th Feb, 2022**

**CERTIFICATE OF APPRECIATION**

This is to certify that **Dr./Mr./Ms. Titiksha Sirari** has participated with a paper entitled **Development and economic evaluation of a patient-centered care model for children with Duchenne's Muscular Dystrophy: A quasi-experimental study protocol** at the XVIIth International Conference held on 12th Feb, 2022.

**Dr Manjula Shastri**
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This is to certify that the Best Paper Award is presented to **Dr./Mr./Ms. Titiksha Sirari** with a paper entitled **Development and economic evaluation of a patient-centered care model for children with Duchenne's Muscular Dystrophy: A quasi-experimental study protocol** at the XVIIth International Conference held on 12th Feb, 2022.

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# EFICON 2022

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This is to certify that Dr. Titiksha Sirari has presented an oral ✓ paper / e-poster titled Socioeconomic determinants of the quality of life in boys suffering from Duchenne Muscular Dystrophy and their caregivers in the 3rd Annual National Conference of Epidemiology Foundation of India-EFICON 2022 held at AIIMS Patna on 4th – 5th November 2022.

| Prof. (Dr.) Gopal Krushna Pal | Prof. (Dr.)Umesh Kapil | Prof. (Dr.) C. M. Singh | Dr. Pragya Kumar |
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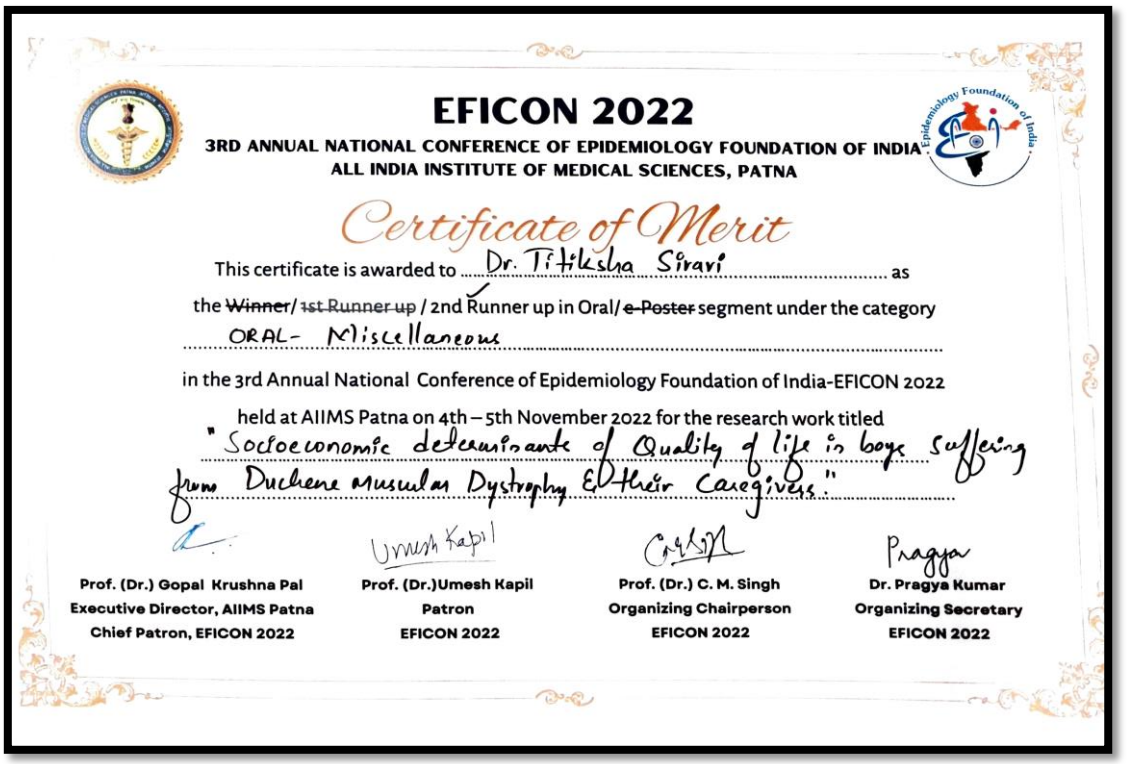

# EFICON 2022

**3RD ANNUAL NATIONAL CONFERENCE OF EPIDEMIOLOGY FOUNDATION OF INDIA**
**ALL INDIA INSTITUTE OF MEDICAL SCIENCES, PATNA**

## Certificate of Merit

This certificate is awarded to Dr. Titiksha Sirari as the ~~Winner~~/ ~~1st Runner up~~ / 2nd ✓ Runner up in Oral/ ~~e-Poster~~ segment under the category ORAL- Miscellaneous in the 3rd Annual National Conference of Epidemiology Foundation of India-EFICON 2022 held at AIIMS Patna on 4th – 5th November 2022 for the research work titled "Socioeconomic determinants of Quality of life in boys suffering from Duchene muscular Dystrophy & their caregivers."

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# Socioeconomic determinants of the quality of life in boys suffering from Duchenne muscular dystrophy and their caregivers

**Titiksha Sirari**[1&3], Amarjeet Singh[2], Renu Suthar[1] , Shankar Prinja[2], Vishwas Gupta[3] Manisha Malviya,[1], Akashdeep Chauhan,[4] Naveen Sankhyan[1]
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## Aim

This study was designed to understand socioeconomic determinants of the quality of life (QoL) in boys with Duchenne muscular dystrophy (DMD) and their caregivers.

## Methods and Results

Sample Size: 100

Sampling Method: Consecutive

Study Site : Pediatric Neurology Unit, Department of Pediatrics, PGIMER, Chandigarh, India

Study Subjects: DMD boys (5-15 years) and their main caregivers

### Assessment of Quality of Life

**Overall QoL (4 domain)**
Physical health
Psychological
Social Relationship
Environment

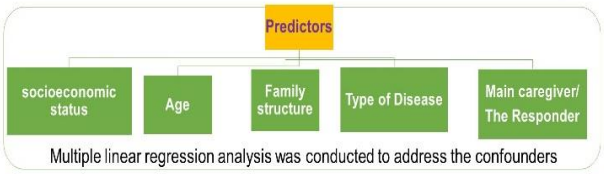
Tool : World Health Organization Quality of Life Scale **(WHOBREEF)**
Mean scores

Caregiver

DMD Boys

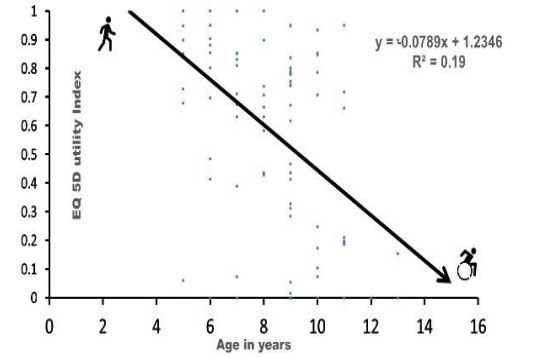
**Health Related QoL**
Tool:
**EuroQol 5-dimensions 5-levels instrument** (EQ-5D-5L)
Mean utility index using India's own Tariff value sets for the First Time

**PedsQL Scores**
Mean scores




Multiple linear regression analysis was conducted to address the confounders

| QoL Scale | Mean ± SD | Scoring Range (Min-Max) |
|---|---|---|
| HR QoL (EQ5D5L) | 0.58±0.35 | 0-1 |
| HR QoL (PedsQL ) | 62.5±15.9 | 0-100 |
| Physical health caregiver (WHOBREEF) | 51.4±13.9 | 0-100 |
| Psychological caregiver (WHOBREEF) | 51.2±16.6 | 0-100 |
| Social Relationship caregiver (WHOBREEF) | 54.4±24.9 | 0-100 |
| Environment caregiver WHOBREEF) | 48.15±22.5 | 0-100 |




EQ 5D-3L utility index scores directly associated with advancing age: Linear regression

- HRQoL of in boys was only associated with age and type of disease and not the socioeconomic status of families.
- There was a 19% deterioration in HRQoL in the boys living with DMD with every unit increase in age.
- Adjusted environmental, Social relationships, and Psychological QoL of caregivers were directly associated with socioeconomic status.
- Every unit gain in the socioeconomic status score, environmental, social relationship, and psychological there is a gain in scores of QoL in caregivers by 1.1, 0.86, and 0.4 units respectively.

## Results

100 families enrolled median age 9 years (IQR: 7.0-9.0)

9% - wheelchair-bound

15% family history


Geographical Distribution:
Chandigarh, Punjab and Haryana: 58%
Uttarakhand & Himachal Pradesh 21%
Eastern Uttar Pradesh and Bihar: 11%
Other distant states: 10%

## Conclusions and Recommendations

HRQoL in boys is directly associated with the age/ progression of disease and not with socioeconomic status of their families.

Thus, in **children QoL can only be addressed by the new therapeutic interventions**

Socioeconomic Status is directly associated with QoL in caregivers of boys with Duchenne muscular dystrophy

Thus, we **need healthcare delivery models that can address socioeconomic inequality**

Studies determining interventions and measures in lower Socioeconomic Status may be needed to improve QoL in caregivers

**Patient-Centric Care Model** is one such healthcare delivery model that reduces health costs and improves health outcomes.

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# GLOSSARY

| | | |
|---|---|---|
| 1 | Adrenal Insufficiency | A rare disorder in which the adrenal glands do not make enough of certain hormones. |
| 2 | Atelectasis | *A complete or partial collapse of the entire lung or area (lobe) of the lung* |
| 3 | Cardiomyopathy | A disease of the heart muscle that makes it harder for the heart to pump blood to the rest of the body. |
| 4 | Caregivers | A family member or paid helper who regularly looks after a child or a sick, elderly, or disabled person. |
| 5 | Catastrophic Illness | An acute or prolonged illness usually considered to be life-threatening or with the threat of serious residual disability. |
| 6 | Creatine Kinase | an enzyme that's found in your skeletal muscle, heart muscle and brain. |
| 7 | Dyspnoea | Difficult, painful breathing or shortness of breath. |
| 8 | Dystrophy | A group of diseases that cause progressive weakness and loss of muscle mass |
| 9 | Economic Burden | A term used to describe problems a patient has related to the cost of medical care. |
| 10 | Genetics | The study of genes and heredity. |
| 11 | Gower's Manoeuvre | The child assumes the hands-and-knees position and then climbs to a stand by "walking" his hands progressively up his shins, knees, and thighs. |

| | | |
|---|---|---|
| 12 | Homogeneous Disorder | Conditions which have the same root cause for all patients in a given group. Usually genetic condition. |
| 13 | Intervention. | A treatment, procedure, or other action taken to prevent or treat disease, or improve health in other ways. |
| 14 | Loss Of Ambulation | Not able to walk around, Bed ridden |
| 15 | Mortality | the number of deaths in one period of time or in one place |
| 16 | Multidisciplinary | A term used to describe a treatment planning approach or team that includes a number of doctors and other health care professionals who are experts in different specialties (disciplines). |
| 17 | Multiplex Ligation-Dependent Probe Amplification (MLPA) | A technique by which up to 45 different sequences can be targeted in a single, semiquantitative polymerase chain reaction (PCR)-based experiment. The sequences detected can be small enabling analysis of fragmented DNA. |
| 18 | Muscle Biopsy | A procedure used to diagnose diseases involving muscle tissue |
| 19 | Mutation | Any change in the DNA sequence of a cell. Mutations may be caused by mistakes during cell division, or they may be caused by exposure to DNA-damaging agents in the environment. |
| 20 | Pneumonia | An infection that inflames the air sacs in one or both lungs. The air sacs may fill with fluid or pus (purulent material), causing cough with phlegm or pus, fever, chills, and difficulty breathing. |
| 21 | Polymerase Chain | A laboratory method used to make many copies of a |

| | | |
|---|---|---|
| | Reaction | specific piece of DNA from a sample that contains very tiny amounts of that DNA |
| 22 | Scoliosis | A condition marked by a side-to-side curve of the backbone. The curve is usually shaped like an S or a C |
| 23 | Tele-Consultation | A synchronous or asynchronous consultation using information and communication technology to omit geographical and functional distance. |
| 24 | Ventilator | A machine used to help a patient breathe. Also called respirator. |

# Appendix I

## General Information/ Socio-economic and Demographic Profile

**C.R. No……..** **NDC No…….** **Study enrollment:** ……

**Name** **Date of birth** **Date of enrollment**

**Categories of Muscular Dystrophy**

DMD: Non-ambulatory | Intermédiate: Non ambulatory | BMD: Ambulatory | Isolated cardiomyopathy

Stages of DMD

Pre-symptomatic (stage I)
Delayed walking or Asymptomatic
Early-Non ambulatory (Stage IV)

Early-Ambulatory (Stage II)

Late-Ambulatory (Stage III)

Late-Non ambulatory (Stage V)

**Contact details of Patient**

Address: State:

Phone Number Smart Phone: Yes No
Alternate contact number Internet availability: Yes No

Name of Respondent/Caregiver ……

Relation with Patient: (Mother/Father/other …… (specify)

Family History: Present/ Not Present; Number of Children affected ……

**Details of Caregivers[§] /Family profile**

| Name | Age/Sex | Relationship with Patient | Education | Occupation | Income @month | Involved in caregiving |
|---|---|---|---|---|---|---|
| | | | | | | |
| | | | | | | |
| | | | | | | |
| | | | | | | |
| | | | | | | |
| | | | | | | |

**Development and economic evaluation of a patient-centered care model for children with Duchenne's Muscular Dystrophy in North India *(Interview schedule LPU &/co PGIMER)***

## Socioeconomic Status of the Family

| Domain | Score | Domain | Score |
|---|---|---|---|
| **1. Monthly per capita income from all sources (in INR)** | | **4. Family possessions (presence of each item given below will carry a score of '1')** | |
| >50000 | 7 | Refrigerator/ TV/Radio/ Transistor/Music system/ AC/ Washing Machine/ Telephone/ Mobile/ Credit card/ Sanitary latrine/ Any newspaper subscribed throughout the month | 10 |
| 20000-49999 | 6 | **5. Type of house** | |
| 10000-19999 | 5 | Own house with 3-4 rooms | **7** |
| 5000-9999 | 4 | Own house with 3-4 rooms/ Rented/Govt. house with 5 or more rooms | 6 |
| 2500-4999 | 3 | Own house with 1-2 rooms/ Rented/Govt. house with 3-4 rooms | 5 |
| 1000-2499 | 2 | Rented/Govt. house with 1-2 rooms | 4 |
| <1000 | 1 | Own jhuggi | 3 |
| **2. Education of either husband or wife who is more educated among them** | | Rented jhuggi | 2 |
| Professional qualification with technical degrees or diplomas e.g. Doctor, Engineer, CA, MBA, etc. | 7 | No place to live, pavement, mobile cart | 1 |
| Postgraduation (non-technical also PhD) | 6 | **6. Possession of a vehicle or equivalent** | |
| Graduation | 5 | 2 or more cars/Tractors/Trucks | 4 |
| 10th class pass but <Graduation | 4 | 1 Car /Tractor/Truck | 3 |
| Primary pass but <10th class | 3 | 1 or more scooter(s)/Bullock cart (s) | 2 |
| <Primary but attended school for at least one year | 2 | 1 or more cycles (not baby cycle) | 1 |
| Just literate but no schooling | 1 | None of the above | 0 |
| Illiterate | 0 | **7. No. of earning members in the family (Nuclear/Joint)** | |
| **3.Occupation of husband, otherwise wife.** | | 3 or more members earning and income pooled | 3 |
| Service in central/State/Public undertakings or Owner of a company employing >20 persons or self-employed professional viz Doctors, CAs, Engineer | 5 | 2 or both husband and wife earning | 2 |
| Service in Private sector or independent business employing 2-20 persons | 4 | Only 1 family member earning | 1 |
| Service at shops, home, transport, own | 3 | No earning member | 0 |

| | | | |
|---|---|---|---|
| cultivation of land | | | |
| Self employed e.g. shops, Rehdies or petty business with income >5000 | 2 | | |
| Self employed with income <5000 (labourer, house wife) | 1 | | |
| None of the family member is | 0 | | |

| **Domain** | **Score** | **Domain** | **Score** |
|---|---|---|---|
| **8.Number of children head of the family has/had** | | **14.Members of family gone abroad in last three years (official or personal)** | |
| 0-1 | 5 | Whole family | 3 |
| 2 | 4 | Only husband and wife | 2 |
| 3 | 3 | Only 1 family member | 1 |
| 4 | 2 | None | 0 |
| 5 | 1 | **15.Possession of agricultural land for cultivation** | |
| ≥6 | 0 | Own agricultural land >100 acres | 5 |
| **9.Facility of some essentials in the family** | | Own agricultural land 51-100 acres | 4 |
| Both tap water supply and electricity | 2 | Own agricultural land 21-50 acres | 3 |
| Only one of above two is present | 1 | Own agricultural land 6-20 acres | 2 |
| None is present | 0 | Own agricultural lan18d 1-5 acres | 1 |
| **10.Education of children (in relation to head of the family)- Exclude under 5 children for this item** | | No agricultural land | 0 |
| All children going/ever gone to school/college | 3 | **16.Possession of non-agricultural land/land for housing or other type of land** | |
| >50% children ever gone/going to school/college | 2 | Own non-agricultural land/land for housing>1000 Sq Yards | 3 |
| ≤ 50% children ever gone/going to school/college | 1 | Own non-agricultural land/land for housing501-1000 Sq Yards | 2 |
| No child ever gone/going to school/college | 0 | | |
| **11.Employment of a domestic servant at home** | | Own non-agricultural land/land for housing 25-500 Sq Yards | 1 |
| Employed >2 full time servants on salary for domestic work | 4 | | |
| Employed only 1 full time servant on salary for domestic work | 3 | Own non-agricultural land/land for housing <25 Sq Yards or Does not own non-agricultural land/land for housing at all | 0 |
| Employed > 3 part time servants on salary for domestic work | 2 | | |
| Employed 1-2 part time servants on salary for domestic work | 1 | **17.Presence of milch cattles in the family for business or non-business purposes** | |
| Employed no servants for domestic work | 0 | | |
| **12.Type of locality the family is residing** | | Own 4 or more milch cattle | 3 |

| | | | |
|---|---|---|---|
| Living in urban locality | 5 | Own 1-3 milch cattles | 2 |
| Living in rural locality | 4 | Own 1 milch cattle | 1 |
| Living in resettlement colony | 3 | Does not own any milch cattle | 0 |
| Living in slums/jhuggis | 2 | **18. Presence of non milch cattle or pet animals in the family** | |
| No fixed living and mobile | 1 | | |
| **13.Caste of the family** | | Own 2 or more | 2 |
| Upper caste | 4 | Own 1 | 1 |
| Other backward class (OBC) | 3 | | |
| Dalits | 2 | | |
| Tribals | 1 | | |

| **Domain** | **Score** | **Domain** | **Score** |
|---|---|---|---|
| **19. Besides the house in which the family is living, the family owns other house or shop or shed etc. of any size whether given on rent or not** | | **21. Parental support in the form of non-movable property** | |
| Own 3 or more | 3 | >50 acres of agricultural land -OR -a house/plot >1000 sq yards -OR - Both | 4 |
| Own 2 or more | 2 | 21-50 acres of agricultural land -OR -a house/plot 501-1000 sq yards -OR -Both | 3 |
| Own 1 | 1 | 1-21 acres of agricultural land -OR -a house/plot 100-500 sq yards -OR - Both | 2 |
| Does not own any | 0 | No agricultural land - BUT - a house/plot 25-100 sq yards | 1 |
| **20. Positions held (besides the positions as employee) by any one member in the family** | | No parental property | 0 |
| Holding position of 3 or more official or non-official organizations viz. president/ chairman/Secretary/Treasurer etc. | 4 | **22. Total amount of income tax paid by the family (include all the earnign members IT)** | |
| Holding position of 1-2 or more official or non-official organizations viz. president/ chairman/Secretary/Treasurer etc. | 3 | >10 lacs | 7 |
| | | 1-10 lacs | 6 |
| | | >50000 but <1 lac | 5 |
| Holding position as member only of executive or other committees of official or non-official organizations. | 2 | >20000 - <50000 | 4 |
| | | >10000-<20000 | 3 |
| | | >5000-<10000 | 2 |
| | | <5000 | 1 |
| Does not hold any such position | 1 | Nil | 0 |
| | | | |
| | | | |

**Development and economic evaluation of a patient-centered care model for children with Duchenne's Muscular Dystrophy in North India *(Interview schedule LPU &/co PGIMER)***

| TOTAL SCORE OFTHIS FAMILY | |
|---|---|
| **Social Status** | **Score** |
| **Upper High** | **≥76** |
| **High** | **61-75** |
| **Upper middle** | **46-60** |
| **Lower Middle** | **31-45** |
| **Poor** | **16-30** |
| **Very Poor or Below Poverty Line** | **≤15** |

## Caregiver Interview Schedule

Q1. When did you first notice that your child is having problem?

| < 3 years | 3 to 5 years | 5 to 8 years | >8years |
|---|---|---|---|

Q.2 What did you first noticed first (mark the symptoms in increasing order)

| **Physical symptom** | **order** | **Age** | **Physical symptom** | **order** | **Age** |
|---|---|---|---|---|---|
| Difficulty in walking | | | Pain in muscles/limbs | | |
| Toe walking | | | Frequent falls | | |
| Difficulty in running | | | Muscle enlargement | | |
| Inability to climb stairs | | | Loss of ambulation | | |
| Difficulty in getting up from floor | | | Others | | |

Q.3. Finally, who told you that your child is having some sort of disease? Doctor=1, Nurse=2, Internet=3, others (Specify)

Q4. What all do you know about DMD? (correct/ incorrect)

| **Why-**<br><br>**What happens (S/S)**<br><br>Prognosis | Predictions<br><br>Any other kind of information related to the disease. |
|---|---|

| **Natural course of disease** | **Clear understanding** | **Not clear understanding** |
|---|---|---|
| Delayed Milestone | | |
| Symptoms | | |
| Progression of disease | | |
| Loss of ambulation or wheel chair bound | | |
| Bead bound | | |
| Pre-Mature Death | | |

**Health Utilization Pattern**

**A) Pathway to care** Q5.1What treatment agencies you consulted so far, your child?

**Development and economic evaluation of a patient-centered care model for children with Duchenne's Muscular Dystrophy in North India *(Interview schedule LPU &/co PGIMER)***

| **Agencies** | **Name of the agency** | **When consulted** | **Referred by** | **Money spent** | **Response** |
|---|---|---|---|---|---|
| Agency 1 (A1) | | | | | |
| Agency 2 (A2) | | | | | |
| Agency 3 (A3) | | | | | |
| Agency 4 (A4) | | | | | |
| Agency 5 (A5) | | | | | |
| Agency 6 (A6) | | | | | |

| **B) Q.6 Problems faced after coming to PGI** | |
|---|---|
| Number of visits made :…… | Money spent in investigations |
| Money spent in transportation …… | Work-time loss in each visit |

| **Problem Faced** | **Yes=2, No=1 (if Yes Specify)** |
|---|---|
| Transport | |
| During registration till getting consultation. **(waiting period)** | |
| In sample collection, its transportation, report collection **(running around)** | |
| Other investigations (conducted in same building or separate) | |
| Any kind of fear or apprehension | |

| | |
|---|---|
| Q7. Were reports of investigations explained to you/family by the concerned doctor? Yes/No | |
| Q8. Were you Satisfied with the explanations given to you regarding the investigations conducted? | |
| Satisfactory =2 | Unsatisfactory=1 |
| If Unsatisfactory specify: | |
| **C) Follow up visits at PGI** | |

Q9. Time since last follow up visit: …….

Q10 What problem faced during follow up visit?

**D) Home Care (Instructions for Home)**

Q12. What Instructions have been given to you for care at home

| **Regarding** | **Instructions given (Yes=2, No=1)** | **Comprehension (Yes=2, No=1)** | **Compliance (Yes=2, No=1)** |
|---|---|---|---|
| Medications (name/ doses) | | | |
| Diet (low fat/salt restrictions) | | | |
| Orthotics: night splints, AFO, KAFO | | | |
| Support devices: wheel chairs, frame, Assisted Cough technique | | | |
| Home Modifications | | | |
| Physiotherapy | | | |
| Anyother | | | |

Q13. Were you informed regarding availability of prenatal diagnosis? (Yes=2, No=1)

Q.14. Did you face any problem in understanding the instructions for the first time? (Yes=2, No=1)

Q.15. Did you ever have to repeat the questions to doctor/nurse to clear your understanding about any given instructions? (Yes=2, No=1)

Q.16. Please specify the instruction in which u faced problem to follow?

Q.17. Were you informed regarding the difficulties you may face in management of disease and various ways to cope them? Yes/No

Q17.1. If Yes, Please specify.

**Anything else**

## Appendix II

**CONSUMPTION EXPENDITURE (Phase 1)**

| How much does your family spend per month on following items: | Expense | | |
|---|---|---|---|
| | 7 days | 30 days | 365 days |
| i. Food: ration (Cereals, pulses, edible oil, bread etc.), Fruits and vegetables, Milk, Milk products, Beverages etc | | | |
| ii. Education (Books, newspaper, fees) | | | |
| iii. Health | | | |
| iv. Bills (Electricity, telephone, water) | | | |
| v. Conveyance, fuel | | | |
| vi. Rents | | | |
| vii. Clothing, Footwear, bedding, curtains etc | | | |
| viii. Entertainment (Cable, cinema, sports, recreation & hobbies) | | | |
| ix. Personal effects (Watch, mobile phone, spectacles, toiletries, jewellery) | | | |
| x. Consumer services (Domestic help, cook, sweeper, barber, tailor, priest, beautician) | | | |
| xi. Pan, Tobacco, alcohol or any other intoxicants | | | |
| xii. Miscellaneous (household appliances, furniture, crockery, animals, or any family function) | | | |
| xiii. Total Monthly Expenditure (to be calculated from above) | | | |

**OUT OF POCKET EXPENDITURE**

| **Source of expenditure** | **Out of Pocket Expenditure** | | | | | | |
|---|---|---|---|---|---|---|---|
| | **V_ 1** | **Tele- FU** | **Tele-FU** | **V_2** | **Tele-FU** | **Tele-FU** | **V_3** |
| Travelling cost | | | | | | | |
| Medicines | | | | | | | |
| Physiotherapy | | | | | | | |
| Genetic consultation | | | | | | | |
| Lab tests/ Diagnostics PGIMER | | | | | | | |
| Lab tests/ Diagnostics Private | | | | | | | |
| Procedure/Surgery | | | | | | | |
| Orthotics: AFO, KAFO, hand splints | | | | | | | |
| Support devices: wheel chairs, frame, PF enhancement devices, Assisted Cough technique | | | | | | | |
| Home Modifications | | | | | | | |
| User fees/Hospital charges (including bed charges) | | | | | | | |
| Informal payment | | | | | | | |
| Boarding/Lodging | | | | | | | |
| Food | | | | | | | |
| Other | | | | | | | |
| Total | | | | | | | |

| **Source of finance** | | | | | | | |
|---|---|---|---|---|---|---|---|
| **Source of finance** | **Amount** | | | | | | |
| | **Visit 1** | **Tele-follow up** | **Tele-follow up** | **Visit 2** | **Tele-follow up** | **Tele-follow up** | **Visit 3** |
| Salary/Savings | | | | | | | |
| Selling of assets | | | | | | | |
| Borrowed from relatives/friends | | | | | | | |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| without interest | | | | | | | |
| Borrowed with interest | | | | | | | |
| **Health insurance (Yes/No)** | | | | | | | |
| 1_ Private Insurance | | | | | | | |
| 2_ Government employee | | | | | | | |
| 2_Disability allowance | | | | | | | |
| 3_Reimbursements (other) | | | | | | | |
| 4_ NRHM scheme | | | | | | | |
| 5_ BPL free/poor free | | | | | | | |
| **Visit 1** | **Visit at the time of enrollment** | | | | | | |
| **Tele-follow up** | Monthly tele-follow ups. | | | | | | |
| **Visit 2** | **At the time of Follow up 1 (3 months)** | | | | | | |
| **Visit 3** | **At the time of Follow up 2 (6 months) end of phase 1** | | | | | | |

***Data Collection Tool for Indirect expenditure in caregiving (Caregiver Details):***

1. What would you have being doing otherwise if your child would have not being taking him for visit? (Multiple response allowed)

Time spent (in hours) on: **(During Visit)**

| | Caregiver 1 (1 day) | Caregiver 2 (1 day) |
|---|---|---|
| Household activities | | |
| Childcare | | |
| Professional work | | |
| Voluntary work | | |
| Leisure activities | | |
| Attending School/University | | |
| Seeking work | | |
| Social work | | |
| Physical workout | | |
| Other (specify) | | |

2. Did other people take over and perform your usual household tasks during you are involved in care of affected child or **during hospital visits**? If yes, fill the appropriate option, there can be more than one answer

| Household activities | Yes/No/NA | Paid/Unpaid | No. of hours |
|---|---|---|---|
| Childcare | | | |
| Professional work | | | |
| Voluntary work | | | |

| | | | |
|---|---|---|---|
| Leisure activities | | | |
| Attending School/University | | | |
| Seeking work | | | |
| Social work | | | |
| Physical workout | | | |
| Other (specify) | | | |

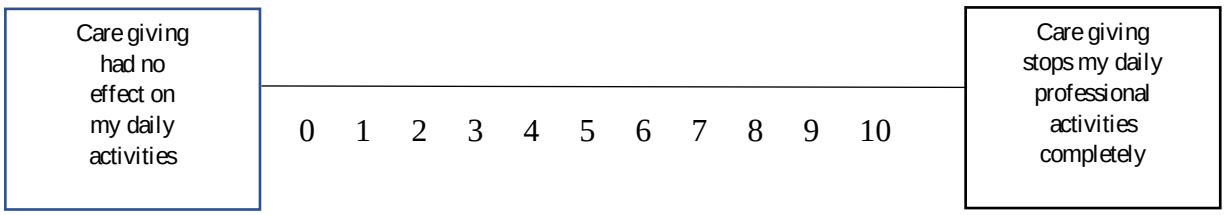
**Caregivers at home:**

| | Caregiver 1 | Caregiver 2 | Caregiver 3 |
|---|---|---|---|
| Relation with patient | | | |
| Address | | | |
| Contact No. | | | |
| Employment status (Yes/No) | | | |
| Nature of employment (Give codes as mentioned in the end of tool) | | | |
| Monthly Gross Income of Caregiver (In Rs) | | | |
| **Time spent daily (hours) on:** | | | |
| Household activities | | | |
| Hours forgone due to care-giving | | | |
| Alternative {1=Yes(paid), 2=Yes (unpaid), 3=No, 4=NA} | | | |
| No. of hours (alternative) | | | |
| Payment to alternative paid worker (In Rs) | | | |
| **Childcare for other Siblings** | | | |
| Hours forgone due to care-giving | | | |
| Alternative {1=Yes(paid), 2=Yes (unpaid), 3=No, 4=NA} | | | |
| No. of hours (alternative) | | | |
| Payment to alternative paid worker (In Rs) | | | |
| **Professional work** | | | |
| Hours forgone due to care-giving | | | |
| Alternative {1=Yes(paid), 2=Yes (unpaid), 3=No, 4=NA} | | | |
| No. of hours (alternative) | | | |
| Payment to alternative paid worker (In Rs) | | | |
| **Voluntary work** | | | |
| Hours forgone due to care-giving | | | |
| Alternative {1=Yes(paid), 2=Yes (unpaid), 3=No, 4=NA} | | | |
| No. of hours (alternative) | | | |
| Payment to alternative paid worker | | | |

| | | | |
|---|---|---|---|
| (In Rs) | | | |
| **Leisure activities** | | | |
| Hours forgone due to care-giving | | | |
| **Attending School/university** | | | |
| Hours forgone due to care-giving | | | |
| **Seeking work** | | | |
| Hours forgone due to care-giving | | | |
| **Social work** | | | |
| Hours forgone due to care-giving | | | |
| Alternative {1=Yes(paid), 2=Yes (unpaid), 3=No, 4=NA} | | | |
| No. of hours (alternative) | | | |
| **Physical workout** | | | |
| Hours forgone due to care-giving | | | |
| **Other (specify)** | | | |
| Hours forgone due to care-giving | | | |
| Alternative {1=Yes(paid), 2=Yes (unpaid), 3=No, 4=NA} | | | |
| No. of hours (alternative) | | | |

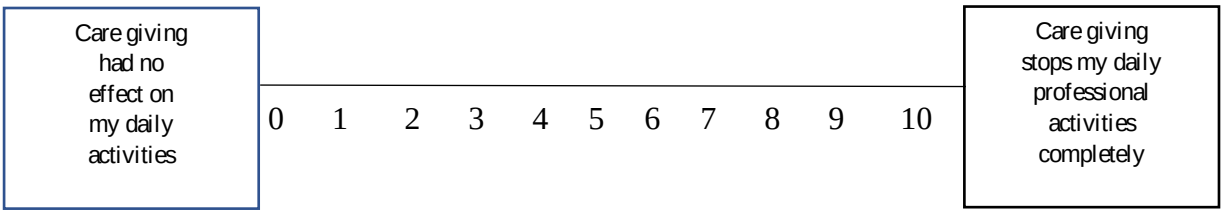
| | | | | |
|---|---|---|---|---|
| * *Alternative Worker;* | Yes (Paid) =1, | Yes (Unpaid) =2, | No=3, | Not Applicable (NA) =4 |

1. Due to care-giving was your performance at work affected:




(Encircle a number)

2. Due to care-giving was your performance on non-professional activities like household
chores, shopping, exercising, studies etc. affected.




(Encircle a number)

**EQ-5D-5L** Date: ……. Individual/Proxy ……

Under each heading, please tick the ONE box that best describes your health TODAY.

**MOBILITY**

I have no problems in walking about ❑
I have slight problems in walking about ❑
I have moderate problems in walking about ❑
I have severe problems in walking about ❑
I am unable to walk about ❑

**SELF-CARE**

I have no problems washing or dressing myself ❑
I have slight problems washing or dressing myself ❑
I have moderate problems washing or dressing myself ❑
I have severe problems washing or dressing myself ❑
I am unable to wash or dress myself ❑

**USUAL ACTIVITIES** *(e.g. work, study, housework, family or leisure activities)*

I have no problems doing my usual activities ❑
I have slight problems doing my usual activities ❑
I have moderate problems doing my usual activities ❑
I have severe problems doing my usual activities ❑
I am unable to do my usual activities ❑

**PAIN / DISCOMFORT**

I have no pain or discomfort ❑
I have slight pain or discomfort ❑
I have moderate pain or discomfort ❑
I have severe pain or discomfort ❑
I have extreme pain or discomfort ❑

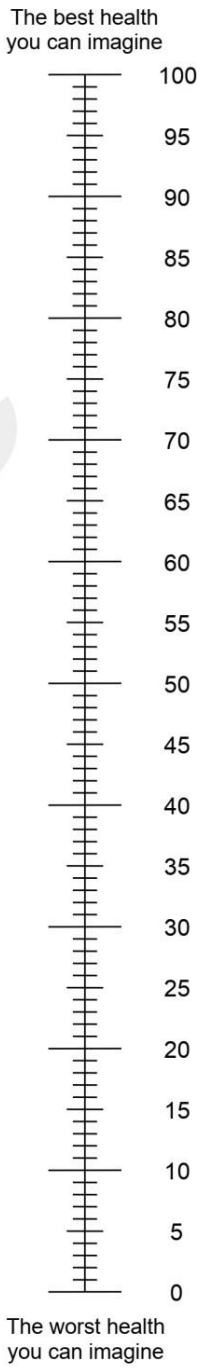
**ANXIETY / DEPRESSION**

I am not anxious or depressed ❑
I am slightly anxious or depressed ❑
I am moderately anxious or depressed ❑
I am severely anxious or depressed ❑
I am extremely anxious or depressed ❑

**EQ-5D-5L** Date: ……. Individual/Proxy ……

- We would like to know how good or bad your health is TODAY.
- This scale is numbered from 0 to 100.
- 100 means the best health you can imagine.
  0 means the worst health you can imagine.
- Mark an X on the scale to indicate how your health is TODAY.
- Now, please write the number you marked on the scale in the box below.

YOUR HEALTH TODAY = ☐



## QUALITY OF LIFE (WHOQOL) -BREF

The following questions ask how you feel about your quality of life, health, or other areas of
your life. I will read out each question to you, along with the response options. **Please choose**
**the answer that appears most appropriate.** If you are unsure about which response to give
to a question, the first response you think of is often the best one.
Please keep in mind your standards, hopes, pleasures and concerns. We ask that you think
about your life **in the last four weeks.**

| | | Very poor | Poor | Neither poor nor good | Good | Very good |
|---|---|---|---|---|---|---|
| **1.** | **How would you rate your quality of life?** | **1** | **2** | **3** | **4** | **5** |
| **2.** | **How satisfied are you with your health?** | **Very dissatisfied** | **Dissatisfied** | **Neither satisfied nor dissatisfied** | **Satisfied** | **Very satisfied** |
| | | **1** | **2** | **3** | **4** | **5** |
| | | **Not at all** | **A little** | **A moderate amount** | **Very much** | **An extreme** |
| **3.** | **To what extent do you feel that physical pain prevents you from doing what you need to do?** | **5** | **4** | **3** | **2** | **1** |
| **4.** | **How much do you need any medical treatment to function in your daily life?** | **5** | **4** | **3** | **2** | **1** |
| **5.** | **How much do you enjoy life?** | **1** | **2** | **3** | **4** | **5** |
| **6.** | **To what extent do you feel your life** | **1** | **2** | **3** | **4** | **5** |

| | | | | | | |
|---|---|---|---|---|---|---|
| | **to be meaningful?** | | | | | |
| **7.** | **How well are you able to concentrate?** | **1** | **2** | **3** | **4** | **5** |
| **8.** | **How safe do you feel in your daily life?** | **1** | **2** | **3** | **4** | **5** |
| **9.** | **How healthy is your physical environment ?** | **1** | **2** | **3** | **4** | **5** |
| | | **Not at all** | **A little** | **Moderately** | **Mostly** | **Completely** |
| **10.** | **Do you have enough energy for everyday life?** | **1** | **2** | **3** | **4** | **5** |
| **11.** | **Are you able to accept your bodily appearance?** | **1** | **2** | **3** | **4** | **5** |
| **12.** | **Have you enough money to meet your needs?** | **1** | **2** | **3** | **4** | **5** |
| **13.** | **How available to you is the information that you need in your day-to-day life?** | **1** | **2** | **3** | **4** | **5** |
| **14.** | **To what extent do you have the opportunity for leisure activities?** | **1** | **2** | **3** | **4** | **5** |
| | | **Very poor** | **Poor** | **Neither poor nor good** | **Good** | **Very good** |

| | | | | | | |
|---|---|---|---|---|---|---|
| **15.** | **How well are you able to get around?** | **1** | **2** | **3** | **4** | **5** |
| | | **Very dissatisfied** | **Dissatisfied** | **Neither satisfied nor dissatisfied** | **Satisfied** | **Very satisfied** |
| **16.** | **How satisfied are you with your sleep?** | **1** | **2** | **3** | **4** | **5** |
| **17.** | **How satisfied are you with your ability to perform your daily living activities?** | **1** | **2** | **3** | **4** | **5** |
| **18.** | **How satisfied are you with your capacity for work?** | **1** | **2** | **3** | **4** | **5** |
| **19.** | **How satisfied are you with yourself?** | **1** | **2** | **3** | **4** | **5** |
| **20.** | **How satisfied are you with your personal relationships?** | **1** | **2** | **3** | **4** | **5** |
| **21.** | **How satisfied are you with your sex life?** | **1** | **2** | **3** | **4** | **5** |
| **22.** | **How satisfied are you with the support you get from your friends?** | **1** | **2** | **3** | **4** | **5** |
| **23.** | **How satisfied are you with the conditions of your living place?** | **1** | **2** | **3** | **4** | **5** |
| **24.** | **How satisfied are you with your access** | **1** | **2** | **3** | **4** | **5** |

| | | | | | | |
|---|---|---|---|---|---|---|
| | **to health services?** | | | | | |
| **25.** | **How satisfied are you with your transport?** | **1** | **2** | **3** | **4** | **5** |
| | | **Never** | **Seldom** | **Quite often** | **Very often** | **Always** |
| **26.** | **How often do you have negative feelings such as blue mood, despair, anxiety, depression?** | **5** | **4** | **3** | **2** | **1** |

***[The following table should be completed after the interview is finished]***

| | | Equations for computing domain scores | | Transformed scores* | |
|---|---|---|---|---|---|
| | | | Raw score | 4-20 | 0-100 |
| 27. | **Domain 1 Physical health** | (6-Q3) + (6-Q4) + Q10 + Q15 + Q16 + Q17 + Q18 | a. = | b: | c: |
| 28. | **Domain 2** Psychological | Q5 + Q6 + Q7 + Q11 + Q19 + (6-Q26) | a. = | b: | c: |
| 29. | **Domain 3** Social relationship | Q20 + Q21 + Q22 | a. = | b: | c: |
| 30. | **Domain 4** Environment | Q8 + Q9 + Q12 + Q13 + Q14 + Q23 + Q24 + Q25 | a. = | b: | c: |

The first transformation method converts scores to range between 4-20, comparable with the WHOQOL-100. The second transformation method converts domain scores to a 0-100 scale.

# Peds QL (Neuromuscular module)

**Child report - Age (8-12)**

| About my neuromuscular disease (problems with…) | Never | Almost never | Sometimes | Often | Almost always |
|---|---|---|---|---|---|
| 1.It is hard to breathe. | 0 | 1 | 2 | 3 | 4 |
| 2.I get sick easily. | 0 | 1 | 2 | 3 | 4 |
| 3.I get sores and/or rashes. | 0 | 1 | 2 | 3 | 4 |
| 4.My legs hurt. | 0 | 1 | 2 | 3 | 4 |
| 5.I feel tired. | 0 | 1 | 2 | 3 | 4 |
| 6.My back feels stiff. | 0 | 1 | 2 | 3 | 4 |
| 7.I wake up tired. | 0 | 1 | 2 | 3 | 4 |
| 8.My hands are weak. | 0 | 1 | 2 | 3 | 4 |
| 9.It is hard to use the bathroom | 0 | 1 | 2 | 3 | 4 |
| 10.It is hard to gain or lose weight when I want to. | 0 | 1 | 2 | 3 | 4 |
| 11.It is hard to use my hands. | 0 | 1 | 2 | 3 | 4 |
| 12.It is hard to swallow food. | 0 | 1 | 2 | 3 | 4 |
| 13.It takes me a long time to bathe or shower. | 0 | 1 | 2 | 3 | 4 |
| 14.I get hurt accidentally. | 0 | 1 | 2 | 3 | 4 |
| 15.I take a long time to eat. | 0 | 1 | 2 | 3 | 4 |
| 16.It is hard to turn myself during the night. | 0 | 1 | 2 | 3 | 4 |
| 17.It is hard for me to go to places with my equipment. | 0 | 1 | 2 | 3 | 4 |

| Communication (problems with…) | Never | Almost never | Sometimes | Often | Almost always |
|---|---|---|---|---|---|
| 1.It is hard for me to tell the doctors and nurses how I feel. | 0 | 1 | 2 | 3 | 4 |
| 2.It is hard for me to ask the doctors and nurses questions. | 0 | 1 | 2 | 3 | 4 |

| | | | | | |
|---|---|---|---|---|---|
| 3.It is hard for me to explain my illness to other people. | 0 | 1 | 2 | 3 | 4 |
| **About our family resources (problems with)** | **Never** | **Almost never** | **Sometimes** | **Often** | **Almost always** |
| 1.It is hard for my family to plan activities like vacations. | 0 | 1 | 2 | 3 | 4 |
| 2.It is hard for my family to get enough rest. | 0 | 1 | 2 | 3 | 4 |
| 3.I think money is a problem in our family. | 0 | 1 | 2 | 3 | 4 |
| 4.I think my family has a lot of problems. | 0 | 1 | 2 | 3 | 4 |
| 5.I do not have the equipment I need. | 0 | 1 | 2 | 3 | 4 |

## Child report – Age 5-7

| **About my neuromuscular disease (problems with…)** | **Not at all** | **Sometimes** | **A lot** |
|---|---|---|---|
| 1.It is hard to breathe. | 0 | 2 | 4 |
| 2.I get sick easily. | 0 | 2 | 4 |
| 3.I get sores and/or rashes. | 0 | 2 | 4 |
| 4.My legs hurt. | 0 | 2 | 4 |
| 5.I feel tired. | 0 | 2 | 4 |
| 6.My back feels stiff. | 0 | 2 | 4 |
| 7.I wake up tired. | 0 | 2 | 4 |
| 8.My hands are weak. | 0 | 2 | 4 |
| 9.It is hard to use the bathroom | 0 | 2 | 4 |
| 10.It is hard to gain or lose weight when I want to. | 0 | 2 | 4 |
| 11.It is hard to use my hands. | 0 | 2 | 4 |
| 12.It is hard to swallow food. | 0 | 2 | 4 |
| 13.It takes me a long time to bathe or shower. | 0 | 2 | 4 |
| 14.I get hurt accidentally. | 0 | 2 | 4 |
| 15.I take a long time to eat. | 0 | 2 | 4 |
| 16.It is hard to turn myself during the night. | 0 | 2 | 4 |
| 17.It is hard for me to go to places with my equipment. | 0 | 2 | 4 |

**Phase II Follow up Sheet(Tele-follow-up) and the Checklist**

Patient Name:

NDC: …….. PCC No ……..

**Appendix VII**

| | | On day of Intervention | F/U 1 | F/U 2 | F/U 3 |
|---|---|---|---|---|---|
| | Date | | | | |
| | Mode of Follow-up (Tele/physical/video) | | | | |
| Examination by Local Physician | Height (Local Physician) | | | | |
| | Height gain (yes/No) | | | | |
| | Weight | | | | |
| | Weight Gain (Yes/No) | | | | |
| | Blood Pressure (In sitting relaxed Position, Rt arm) | | | | |
| | Percentile for hypertension >90$^{th}$ | | | | |
| Functional Status (Reported by Parents) | Frequency of fall Same/decreased/Increased | | | | |
| | Ambulation (Yes/No) | | | | |
| | Gower's Sign time | | | | |
| | 6MWD | | | | |
| | Vignos UL | | | | |
| | Vignos UL | | | | |
| Adverse Event Reported | Back Pain (Yes/No) | | | | |
| | Pain Abdomen (Yes/No) | | | | |
| | Behavioural Issues (aggression/ irritability) | | | | |
| | Any other | | | | |
| Sleep | Time to go to sleep | | | | |
| | Awakening at night | | | | |
| | Day time sleepiness | | | | |
| | Sleep hours | | | | |
| Evaluations in last 6 months | (EYE)For IOP/ Cataract | | | | |
| | (BONE) Fracture: Yes/No | | | | |
| | ECHO (In Last 6 months) | | | | |
| | DEXA (In Last 6 months) | | | | |
| Drugs | Glucocorticoid Therapy On/Off | | | | |
| | ACE inhibitors (yes/no) | | | | |
| | Type of ACE Inhibitors | | | | |
| | Calcium: Yes/ No | | | | |
| | Vitamin D: Yes/No | | | | |
| Night Splint and number of Hours (Yes/No) | | | | | |
| Physiotherapy | Compliance Diary | | | | |

**Appendix VIII**

# Home instructions and compliance diary

**Weekly**

| | | Day 1 | 2 | 3 | 4 | 5 | 6 | 7 |
|---|---|---|---|---|---|---|---|---|
| Drugs:<br>Corticosteroids:Y/N<br>Calcium<br>Vit D<br>Any Other: | | | | | | | | |
| Chest physiotherapy<br>(Breathing Exercises)/<br>Incentive spirometry * (5 repetitions) | Morning | | | | | | | |
| | Evening | | | | | | | |
| Upper Limb Exercises and stretching (30 second each stretch hold on both sides.<br>(5 repetitions) | Morning | | | | | | | |
| | Evening | | | | | | | |
| Lower limb Exercises and stretching (30 second each stretch, hold on both sides.<br>(5 repetitions) | Morning | | | | | | | |
| | Evening | | | | | | | |
| Night Splint<br>Minimum 6 hours/day. | Yes/No | | | | | | | |
| Standing on wooden wedge<br>Minimum 30minutes/day. | Yes/No | | | | | | | |

# ड्यूशेन मस्कुलर डिस्ट्राफी

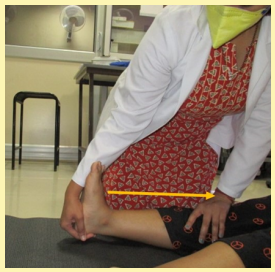

## बेहतर जिंदगी मेरी ज़िम्मेदारी

डॉ तितिक्षा सिराड़ी
डॉ नवीन संख्यान
डॉ रेनू सुथार
डॉ शंकर प्रिंजा
डॉ विश्वास गुप्ता
डॉ मनीषा मालवीय
डॉ अमरजीत सिंह

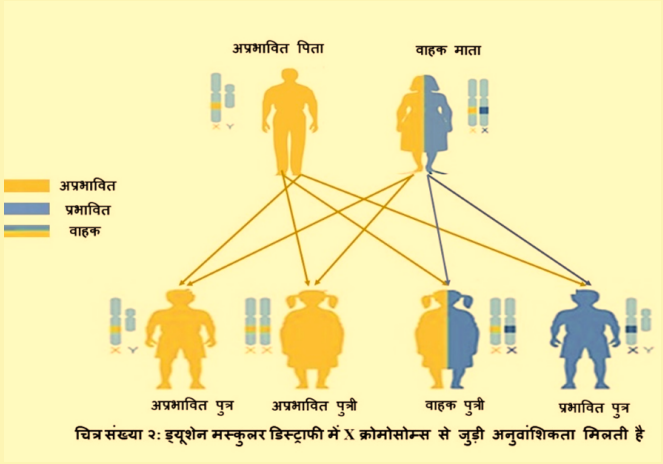
डी.एम.डी मांसपेशियों को प्रभावित करती है और इसके लक्षण केवल लड़कों में देखने को मिलती है

यह डी.एम.डी. की बीमारी हर 3500 लड़कों में से एक को होती है

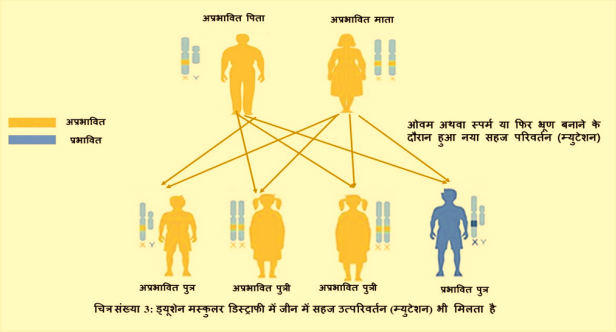
ड्यूशेन में फिजियोथेरेपी विभिन्न कसरतों के माध्यम से लम्बे समय तक अ शारीरिक क्षमता को बनाए रखने में मदद करता है।
मांसपेशियों में खिंचाव करना जोड़ों में संकुचन और विकृति की संभावनाओं को कम करता है।
सहायक उपकरणों (ऑर्थोसिस, कैलिपरस, व्हीलचेयर्स और स्टैंडिंग फ्रेम्स) का इस्तेमाल जीवन की गुणवत्ता को बढ़ता है।
घर में सुरक्षा और गिरने से बचने कुछ संभावित बदलाव के लिए कुछ बदलाव किये जा सकते हैं



गर्भ में पल रहे बच्चे की जेनेटिक जाँच की जासकती है। इससे, जन्म से पहले ही पताचल जाता है की भ्रूण में यह विकार है या नहीं ।
यह जाँच एम्नियोसेंटेसिस और कोरियोनिक विलस सैंपलिंग (सीवीएस)के द्वारा, माँ के गर्भ से नमूने निकल कर किया जा सकती है।
यह बीमारी की रोकथाम में योगदान देता है।



चित्र संख्या २: ड्यूशेन मस्कुलर डिस्ट्राफी में X क्रोमोसोम्स से जुड़ी अनुवांशिकता मिलती है

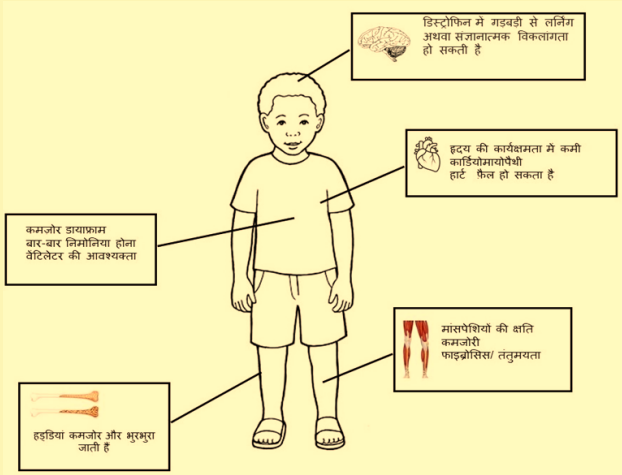


चित्र संख्या 3: ड्यूशेन मस्कुलर डिस्ट्राफी में जीन में सहज उत्परिवर्तन (म्युटेशन) भी मिलता है

डी.एम.डी की बीमारी, जीन में गड़बड़ी होने से होती है, जिसकी वजह से मांसपेशियों में डिस्ट्रोफिन प्रोटीन की कमी हो जाती है, इस कारण यह जल्दी ही क्षतिग्रस्त हो जाती हैं।

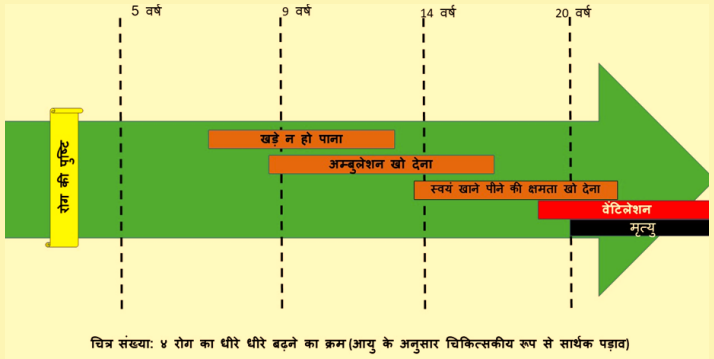
यह जीवन पर्यन्त रहने वाली आनुवंशिक बीमारी (पीढ़ी दर पीढ़ी चलने वाली) है।

जेनेटिक काउंसलिंग के ज़रि, इस बीमारी होने की क्या संभावना होती है? आगे परिवार की योजना कैसे कर सकते हैं? परिवार में इस बीमारी के वाहक की जांच क्या है और इसका क्या महत्व है? समझाते हैं।

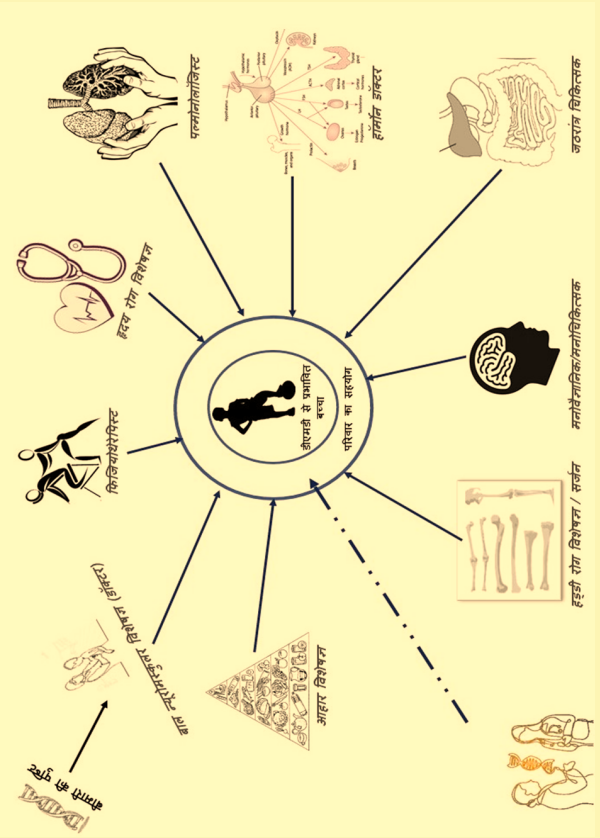



ड्यूचेन होने के मुख्य प्रारंभिक संकेत और लक्षण :

- चलने में समस्या: डीएमडी से प्रभावित बच्चे उनकी उम्र के अन्य बच्चों की तुलना में देरी से चलना शुरू करते हैं।
- जाँच के दौरान क्रैएटिन काइनेज (CK, एक मांसपेशी का प्रोटीन ) का खून में बढ़ा हुआ स्तर मिलने पर , आगे की जाँच की पुष्टि करने के लिए उन्हें न्यूरोमस्कुलर विशेषज्ञ के पास भेजा जाना चाहिए।
- लीवर एन्जाइम का बिना कारण रक्त में बढ़ा हुआ मिलान।
- परिवार में किसी सगे संबंधियों में इस बीमारी की पुष्टि होने से बच्चों में बीमारी होने की संभावनाएं बढ़ जाती हैं।



चित्र संख्या: ४ रोग का धीरे धीरे बढ़ने का क्रम (आयु के अनुसार चिकित्सकीय रूप से सार्थक पड़ाव)




देखभाल के लिए एक टीम की आवश्यकता होती है। इस टीम में न्यूरोलॉजी, कार्डियोलॉजी, पल्मोनोलॉजी, एंडोक्रिनोलॉजी, मनोविज्ञान के विशेषज्ञ और उप -विशेषज्ञ होती है।
टीम के द्वारा उचित समय पर मिलकर, एक समन्वित तरीके से किये गए प्रयास से बालकों की जीवन की गुणवत्ता को बढ़ाया जा सकता है।
बच्चे के माता पिता इस टीम के अभिन्न अंग हैं।
देखभाल करने वाले / परिवार के सदस्य बच्चे की दैनिक गतिविधियों जैसे स्वयं की देखभाल, स्कूली शिक्षा, इधर-उधर जाने में सहायता और भावनात्मक सहायता प्रदान करते हैं। बीमारी के बढ़ने के साथ देखभाल देने के प्रयास में भी वृद्धि हो जाती है, ऐसे में लगातार अपनी स्वस्थ टीम के संपर्क में रहना जरुरी होता हैं।

# बेहतर जिंदगी मेरी ज़िम्मेदारी

# “आशा और विश्वास”

## ड्यूशेन मस्कुलर डिस्ट्राफी

# विषय-सूची

## खंड क: परिचय

ड्यूशेन मस्कुलर डिस्ट्राफी बीमारी मांसपेशियों को प्रभावित करती है और इसके लक्षण केवल लड़कों में देखने को मिलती है। यह पीढ़ियों में चलने वाली बीमारी है। इस से प्रभावित ज्यादातर बच्चे नौ से दस वर्ष की आयु तक आते आते चलने फिरने में असमर्थ हो जाते हैं। यह एक जटिल बीमारी है जो विभिन्न अंगों की मांसपेशियों को प्रभावित करती है। इस बीमारी की पुष्टि होने से लेकर जीवन पर्यन्त बच्चे को लगातार विभिन्न चिकित्सीय परामर्श और अथक देखभाल की ज़रूरत पड़ती है।

अभी तक समाज में इस बीमारी को नजरअंदाज किया जाता रहा है क्योंकि इसका कोई भी प्रभावी इलाज़ नहीं है। परन्तु यह ध्यान देने योग्य बात है कि लोगों में इस बीमारी के बारे में जागरूकता को बढ़ा कर, परिजनों को देखभाल के लिए सजग एवं प्रशिक्षित करके इन बच्चों के जीवन कि गुणवत्ता को बढ़ाया जासकता है।

ड्यूशेन से प्रभावित बच्चे दिमागी स्तर पर अन्य सामान्य बच्चों कि तरह ही होता है। वह अपनी बौद्धिक क्षमता के आधार पर अपनी विक्लांगता से सही डंग से जूझ पाते हैं। मगर, ये तब ही संभव है जब उन्हें अपने आसपास एक सकारात्मक माहौल मिले।

यह पुस्तक परिवार में इस दिशा में एक पहल है। हम आशा करते है कि यह आगे चल कर सामाजिक स्तर पर बदलाव लेन में मददगार सिद्ध होगी। जागरूकता बढ़ाना ही समाज के स्तर पर स्वीकार्यता प्राप्त करने का एकमात्र तरीका है। पुस्तिका, प्रभावित परिवारों के लिए मार्गदर्शक के रूप में सहायक रहेगी।

इस किताब को बीमारी की विभिन्न अवस्थाओं को ध्यान में रखते हुए लिखा गया है। ताकि यह यह पता लग सके कि आप वर्तमान स्थिति में बीमारी के किस पड़ाव पर हैं और भविष्य में आपको क्या कदम उतने की ज़रूरत है। इससे परिवार के साथ-साथ बच्चे भी भविष्य में आने वाली बीमारी की चुनौतियों के लिए अपने आपको तैयार कर सकेंगे।

## खंड ख़: बीमारी  से संबंधित बुनियादी/मूलभूत जानकारी

### 1. ड्यूशेन मस्कुलर डिस्ट्राफी (डी.एम.डी)  क्या है?

यह बीमारी मांसपेशियों को प्रभावित करती है और केवल लड़कों को ही होती है । यह डी.एम.डी.  की बीमारी हर 3500 लड़कों में से एक को होती है। यह बीमारी पीढ़ी दर पीढ़ी चलने वाली है। ये मस्कुलर डिस्ट्रॉफी दो प्रकार होतीं हैं, ड्यूशेन मस्कुलर डिस्ट्राफी (डी.एम.डी) और बेकर्स मस्कुलर डिस्ट्राफी (बीएमडी)। दोनों में से बेकर्स मस्कुलर डिस्ट्राफी कम नुकसानदायक है।

हमें दैनिक कार्य करने के लिए  मांसपेशियों की आवश्यकता होती है।  हम चलने, दौड़ने, सीढ़ियाँ चढ़ने, फर्श से उठकर खड़े होने जैसे विभिन्न कार्यों को करने के लिए मांसपेशियों का उपयोग करते हैं।

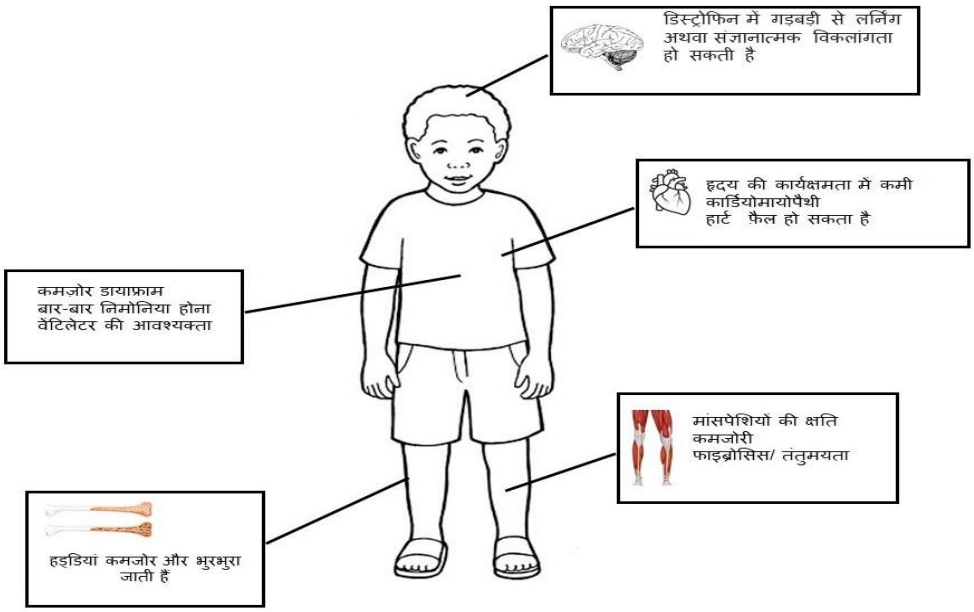
हृदय के सुचारू रूप से काम करने से लेकर सांस लेने तक में मांसपेशियों का इस्तेमाल होता है। ये मांसपेशियां छोटे-छोटे मसल्स फाइबर से बनी होतीं हैं जो प्रोटीन से बने होते हैं। इन प्रोटीनों का निर्माण विभिन्न प्रकार के जींस पर निर्भर करता है।

डी.एम.डी की बीमारी में डिस्ट्रोफिन नाम के जीन में गड़बड़ी हो जाती है, जिसकी वजह से एक बहुत ही महत्वपर्ण प्रोटीन नहीं बन पता है । इस प्रोटीन का नाम है डिस्ट्रोफिन, ये प्रोटीन सभी प्रकार की मांसपेशियां में पाया  है । 'डिस्ट्रोफिन जीन' में गड़बड़ी के से जो फाइबर बनता है वह सामान्य नहीं होता है। इस  फाइबर में डिस्ट्रोफिन प्रोटीन की कमी होती है, इस कारण यह जल्दी ही क्षतिग्रस्त हो जाता है। इस कमी को पूरा करने के लिए शरीर और अधिक मात्रा में मसल फाइबर का निर्माण करता है पर सभी मसल्स फाइबर ख़राब होते हैं ।

डी.एम.डी. एक जीवन पर्यंत रहने वाली बीमारी है। इस बीमारी को क्लिनिकली पांच भागों में बांटा जा सकता है। बीमारी का दुष्प्रभाव उम्र के साथ धीरे धीरे बढ़ता रहता है। उम्र बढ़ने के साथ हाथ पैरों की मांसपेशियां कमजोर पड़ जाती हैं तथा सांस लेने में मदद करने वाली मांसपेशियों की ताकत भी कम हो जाती है। समय के साथ हृदय की मांसपेशियां भी कमजोर पड़ जातीं हैं। हर बच्चे में यह लक्षण उम्र की अलग-अलग पड़ाव पर मिलते हैं फिर भी इस बीमारी को पांच भागों में बांटा गया है। इन मांसपेशियों में कमजोरी के कारण बच्चा देर से चलना व दौड़ना सीखता है। ssउससे फ़र्श पर  से उठकर बैठने में या खड़े होने में दिक्कत आती है। कुछ लड़के अक्सर दौड़ते दौड़ते या चलते-चलते गिर जाते हैं। आमतौर पर, यह कमजोरी तब पकड़ में आती है  जब उनकी शारीरिक क्षमता उनके साथियों से काफ़ी कम रह जाती है। जैसे-जैसे बीमारी बढ़ती है वह हृदय, मस्तिष्क, हड्डियां और पाचन तंत्र की मांसपेशियों को भी अपनी गिरफ़्त में ले लेती है।  डीएमडी / बीएमडी के लगभग ३०% बच्चों में बौद्धिक दोष  (बुद्धिमता की कमि) मिलता है, यह संभवतः केंद्रीय तंत्रिका तंत्र (दिमाग) में असामान्य डिस्ट्रोफिन की वजह से होती है । इस बीमारी में इलाज एवं देखभाल इस बात पर निर्भर करती है कि बीमारी किस स्टेज पर है तथा कौन-कौन से अंग प्रभावित हैं।

उपचार का उद्देश्य है, डी.एम.डी से प्रभवित बच्चे के स्वास्थ्य को उत्तम रखना तथा जहां तक हो सके उसको स्वावलंबी बनाना। बच्चे को एक लम्बा और स्वस्थ जीवन जिनमें सहयोग करना। डी.एम.डी के प्रमाणित चिकित्सीय देखभाल के अंतर्गत फिजियोथेरेपी, स्टेरॉइड्स, हड्डियों की देखभाल, हृदय की देखभाल, हार्मोन, सांस लेने में मदद करने वाली मांसपेशियों की देखभाल, मानसिक एवं व्यावहारिक सहयोग सम्मिलित हैं। सभी प्रकार की चिकित्सीय देखभाल की आवश्यकता एक साथ नहीं पड़ती है। यह रोग की अवस्था और रोग के प्रति व्यक्ति की प्रतिक्रिया पर निर्भर करता है। बीमारी का जल्द से जल्द पता लगना उसकी निदान में बहुत मददगार सिद्ध होता है।

अभी तक हम ये जान गए हैं कि यह जीवन पर्यंत चलने वाली बीमारी है, जो उम्र के साथ बढ़ती जाती है। यह शरीर के विभिन्न अंगों को अलग-अलग समय पर प्रभावित करती है। डी.एम.डी में चिकित्सीय इलाज के साथ-साथ, परिवार के द्वारा की जाने वाली देखभाल की भी बहुत अहम भूमिका होती है। यह बहुत जरूरी है कि परिवार के सदस्यों को चिकित्सीय देखभाल में सम्मिलित किया जाए और परिवार के सदस्यों को भी बच्चे की देखरेख में अपनी भूमिका सुनिश्चित करनी चाहिए। हर बच्चे के लिए उसकी जरूरत के हिसाब से उसकी देखरेख का समुचित इलाज की योजना बनाना आवश्यक है। परिवार, जिसमें डीएमडी से प्रभावित बच्चा है, बीमारी के अनुकूल अपने को ढ़ालना बहुत आवश्यक है।




चित्र १ : डी.एम्.डी. विभिन्न अंगों को प्रभावित करता है।

## 2. यह बीमारी कुछ ही बच्चों को क्यों प्रभावित करती है जबकि दूसरे अप्रभावित रहते हैं ?

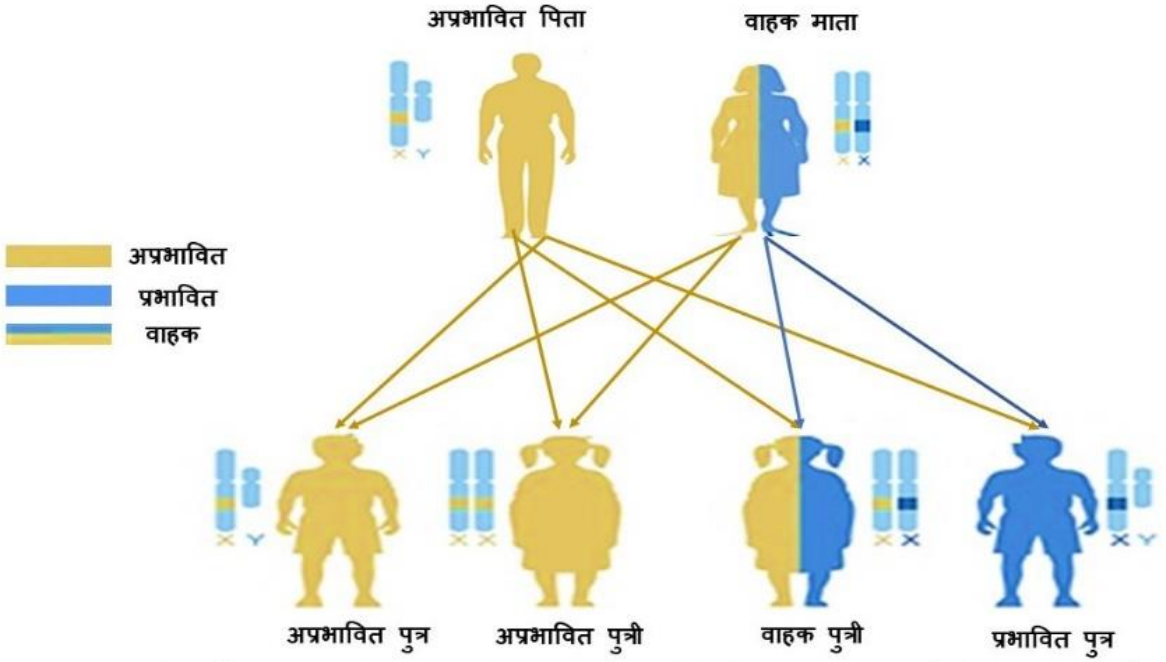
मस्कुलर डिस्ट्रॉफीज नौ प्रकार की होती है। इन में से डी.एम.डी सबसे आमतौर पर मिलती है। जैसे कि पहले भी चर्चा की जा चुकी है की यह एक पीढ़ी दर पीढ़ी चलने वाली बीमारी है। इसका मतलब है कि यह एक आनुवंशिक बीमारी है। अनुवांशिक बीमारियां एक बीमार जीन के पीढ़ियों में चलने के कारण होती हैं।

पहले भी उल्लेख किया गया है कि यह बीमारी उन लोगों में होती है जिनमें डिस्ट्रोफिन नामक जीन में गड़बड़ी हो जाती है। उनकी मांसपेशियों में डिस्ट्रोफिन नामक प्रोटीन दोषपूर्ण होता है। इस प्रोटीन का कार्य मांसपेशियों की कोशिकाओं को स्वस्थ रखना है।अस्वस्थ मांसपेशियां बार-बार टूटती - फूटती रहती हैं, यह बीमारी बढ़ती उम्र के साथ मांसपेशियों को और कमज़ोर करती चली जाती है।

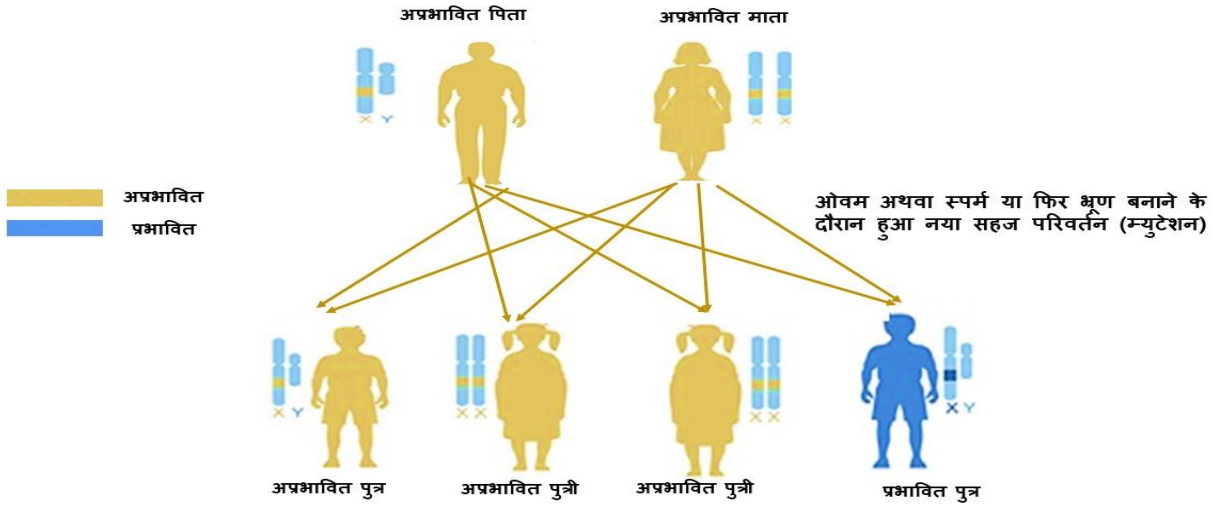
इस बीमारी से सभी बच्चे प्रभावित नहीं होते, यह बीमारी मुख्यतः लड़कों में मिलती है। पुराने ममेरे सगे संबंधियों में इस बीमारी की पुष्टि होने से बच्चों में बीमारी होने की संभावनाएं बढ़ जाती हैं। परंतु यह समस्या परिवार के किसी सदस्य को प्रभावित किये बिना भी आगे की पीढ़ियों में देखी जासकती है। क्यूंकि, जीन परिवार में किसीको भी प्रभावित किये बिना चुप चाप पीढ़ी दर पीढ़ी आगे बढ़ती जाती है इसलिए पहले बच्चे में सही -सही अनुमान लगाना मुश्किल होता है कि उसको यह बीमारी होगी या नहीं। किन्तु, पहले बच्चे में इस जीन की पुष्टि हो जाने पर यह आवश्यक हो जाता है कि हम इस जीन को परिवार में आगे बढ़ने न दें ।

एक महिला में दो एक्स क्रोमोसोम होते हैं तथा पुरुषों में एक एक्स क्रोमोसोम और एक वाई क्रोमोसोम होता है। यह जानना जरूरी है कि यह डिस्ट्रोफिन जीन इंसान के एक्स क्रोमोसोम में पाया जाता है। बीमारी के लक्षण तभी सामने आते हैं जब व्यक्ति के सभी एक्स क्रोमोसोम में दोष होता है । इस जीन की आनुवंशिकता को चित्र संख्या : २ से समझा जा सकता है। एक अप्रभावित पिता और एक वाहक माता के दोनों, लड़के और लड़कियां में यह दोषपूर्ण जीन विरासत में जाने कि बराबर सम्भावना होती है। बेटियाँ जिनमें एक दोषपूर्ण X क्रोमोसोम होता है वे इस जीन को एक पीढ़ी से दो दूसरी पीढ़ी में एक वाहक के रूप में ले जाती हैं, जबकी लड़के/ बेटे ही इस बीमारी से ग्रसित होते हैं। इस प्रकार की आनुवंशिकता जो X क्रोमोसोम से जुड़ी होती है, डिस्ट्रोफिनोपैथीज़ (डी एम् डी अथवा बी एम् डी ) में पाई जाती है। महिला बिनाकिसी बीमारी के लक्षण के भी इस दोषपूर्ण जीन के साथ पूरी जिंदिगी जी सकती है, परन्तु उस महिला के 50% बेटों को यह बीमारी हो सकती है और 50% बेटियां इस बीमारी को दूसरी पीढ़ी में लेजाने केलिए वाहक सिद्ध हो सकतीं हैं। चूँकि यह बीमारी तभी अपने लक्षण दिखाती है जब सभी एक्स क्रोमोसोम में दोषपूर्ण क्रोमोसोम होता है, इसलिए बेटियों / माताओं / बहनों में बीमारी के लक्षण नहीं पाए जाते हैं।

डायस्ट्रोफिन प्रोटीन के उत्पादन के लिए जिम्मेदार जीन आकार में बहुत बड़ा (सबसे बड़ा) होता है। जीन का जबरदस्त बड़ा आकार होने के कारण, इस में सहज उत्परिवर्तन (म्युटेशन) का दर सबसे ज्यादा होता है। डी. एम्. डी. के लगभग एक तिहाई मामलों में इस प्रकार का ही नया म्युटेशन पाया जाता है। इन मामलों में माता-पिता दोनों ही अप्रभावित होते हैं और इनके परिवार में भी किसी को भी इस बीमारी के लक्षण नहीं होते हैं। जीन में सहज उत्परिवर्तन (म्युटेशन) की आनुवंशिकता को चित्र संख्या : ३ से समझा जा सकता है।




चित्र संख्या २: इयूशेन मस्कुलर डिस्ट्राफी में X क्रोमोसोम्स से जुड़ी अनुवांशिकता मिलती है




चित्र संख्या 3: इयूशेन मस्कुलर डिस्ट्राफी में जीन में सहज उत्परिवर्तन (म्युटेशन) भी मिलता है

इस प्रकार, डिस्ट्रोफिनोपैथिस (डीएमडी / बीएमडी) नए या विरासत में मिला उत्परिवर्तनो (म्युटेशन) के कारण हो सकता है।

डीएमडी अथवा बीएमडी उत्परिवर्तन (म्युटेशन) कई प्रकार के होते हैं, पचास प्रतिशत से अधिक के मामलों में विलोपन (जीन का टूट जाना) मिलता है। लगभग 5% मामलों में प्रतिलिपि (जीन के किसी भाग कि प्रतिलिपि) बनजाती है। विलोपन एवं प्रतिलिपि बड़े उत्परिवर्तन हैं जो आसानी से डीएनए डायग्नोस्टिक्स से पकड़ में आ जाते हैं। इनके अलावा अन्य छोटे उत्परिवर्तन भी होते हैं जो इन मायोपैथियों के लिए जिम्मेदार हैं। ये बहुत ही छोटे होते हैं इसलिए इनको बिंदु

(पॉइंट) म्युटेशन कहते हैं, लगभग 20 %-30 % मामले ऐसे होते हैं। रोग के लिए जिम्मेदार म्यूटेशन के प्रकार को जानने की आवश्यकता आनुवांशिक परामर्श लेने में, नैदानिक उपचार और जीनोटाइप पर आधारित नैदानिक दवाई की परख करने में पड़ती है ।(PMID: 26773583).

मस्कुलर डिस्ट्रॉफी से प्रभावित बच्चों में जीन म्युटेशन या बहुरूपता का पता डीएनए डायग्नोस्टिक्स के द्वारा खून के नमूने, एम्नियोटिक द्रव कोशिकाएं एवं कोरियोनिक विलाई के नमूनों में किया जा सकता है। इसके अलावा गर्भ के दौरान भी, गर्भ में पल रहे बच्चे की जेनेटिक जाँच की जासकती है। इससे, जन्म से पहले ही पताचल जाता है की भ्रूण में यह विकार है या नहीं । ताकि, समय रहते उचित इलाज किया जा सके और परिवार में इसके विस्तार को रोका जा सके । यह जाँच एम्नियोसेंटेसिस और कोरियोनिक विलस सैंपलिंग (सीवीएस)के द्वारा, माँ के गर्भ से नमूने निकल कर किया जा सकती है। गर्भ के दौरान जाँच के लिए माँ को गर्भ के १२-१३ हफ्तों के बीच में अस्पताल, जहाँ प्रीनेटल जाँच की सुविधाएं उपलब्ध हैं,संपर्क करने के लिए कहा जाता है।

जिन परिवारों में डी एम डी से प्रभावित बच्चे होते हैं, परिवारवाले हमेशा यह जानने के लिए इच्छुक होते हैं कि उनके बच्चों में यह घातक बीमारी किस कारण से होती है। घरवालों में बीमारी से संबंधित विभिन्न दुविधाएं और मनोवैज्ञानिक दबाव भी होता हैं, जिन्हें संबोधित करने की

जोर दिया गया है। परामर्श में परिवार की वंशावली को बेहतर तरीके से समझने के लिए चर्चा की जाती है ताकि सभी वाहकों की सही सही पहचान की जासके । जिससे ये वाहक अपनी जेनेटिक जाँच करा सकें तथा अपने आने वाली पीढ़ी में इस बीमारी को आगे न बढ़ने दें। परिवार के लिए भविष्य की योजना बनाने के लिए, माता-पिता को प्रसव से पूर्व जाँच की उपलब्धता के बारे में भी बताया जाता है । यह बीमारी की रोकथाम में योगदान देता है।

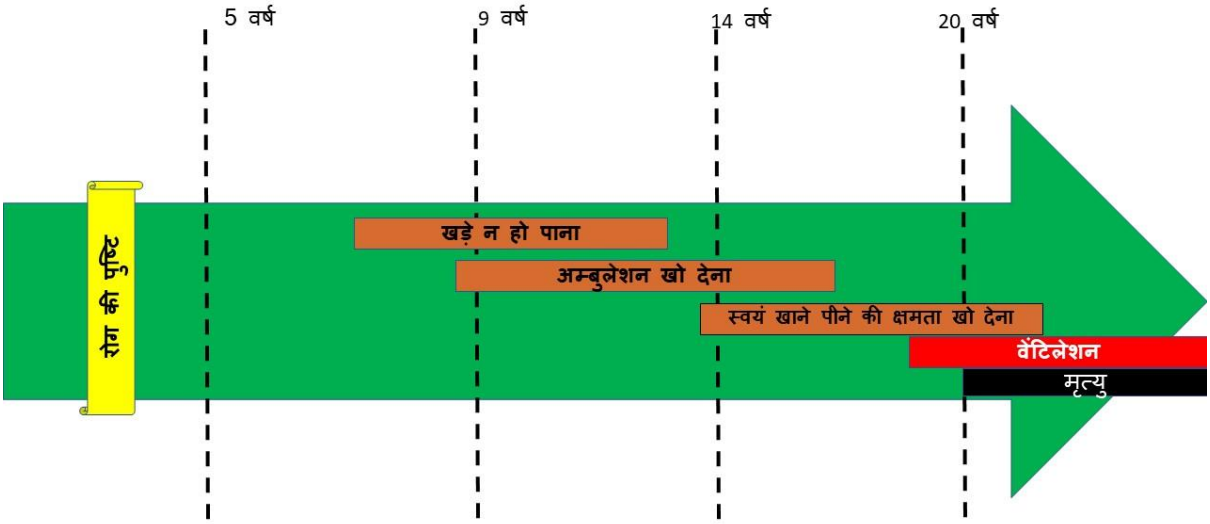
आवश्यकता है। जेनेटिक काउंसलिंग के माध्यम से इन उलझनों को सुलझाया जा सकता है। जेनेटिक काउंसलिंग माता-पिता को इस बीमारी का कारण, बीमारी के प्रकारों एवं मोटे-मोटे तौर पर इसके लक्षणों को समझने में मदद मिलती है। प्रभावित बच्चों के जीवन की गुणवत्ता बनाए रखने के लिए बहु-आयामी देखभाल के महत्व पर भी जोर दिया जाता है।

परिवार में फिर से किसी बच्चे को इस बीमारी होने की क्या संभावना होती है? आगे परिवार की योजना कैसे कर सकते हैं? परिवार में इस बीमारी के वाहक की जांच क्या है और इसका क्या महत्व है? जेनेटिक कॉउंसलिंग में माता पिता को आनुवंशिकीय जाँच को कैसे समझाया जाए? कौन कौन आनुवंशिक परीक्षण उपलब्ध हैं? इन सब प्रश्नों के उत्तर जेनेटिक काउंसलिंग के द्वारा दिए जाते हैं। माता पिता को समझाया जाता है कि कैसे यह बीमारी महिला जीन वाहको द्वारा एक पीढ़ी से दूसरी पीढ़ी में जाती है।

आनुवंशिकता का परामर्श आनुवंशिकीविद् द्वारा दिया जा सकता है, इसके अलावा आपका डॉक्टर / कंसलटेंट या काउंसलर या नर्स भी परामर्श दे सकते हैं। यह बातचीत एक समूह या व्यक्तिगत रूप से हो सकती है। समूहिक बातचीत आमतौर पर आनुवंशिकी परीक्षण से पहले होता है। अनुवांशिकी रिपोर्ट आ जाने के बाद, माता पिता की शंकाओं को व्यक्तिगत तौर पर समझाया जाता है। परिवार में वाहक की जाँच के महत्व पर फिर से

## 3. यह रोग का बच्चों में धीरे धीरे कैसे बढ़ता है ?

ड्यूशेन और बेकर्स मस्कुलर डिस्ट्राफी समय के साथ धीरे धीरे बढ़ाने वाली बीमारी है। रोग के बढ़ने के क्रम और लक्षणों के आधार पर डॉक्टर इस बीमारी को विधिन्न चरणों में चिन्हित करते हैं। ये "चरण" रोग की विशिष्ट समस्याओं की पहचान करने और दिशा निर्देशों पर आधारित देखभाल करने में सहायक होते हैं। कभी-कभी चरणों के लक्षण परस्पर एक-दूसरे से आपस में मिले जुले रहते हैं। फिर भी बीमारी का इन चरणो में विभाजन, निश्चित रूप से उपचार के दौरान आवश्यक एवं व्यापक देखभाल का चयन करने में मदद करते हैं। रोग का धीरे धीरे बढ़ने का क्रम चित्र संख्या ४ से समझा जा सकता है।




**चित्र संख्या: ४ रोग का धीरे धीरे बढ़ने का क्रम (आयु के अनुसार चिकित्सकीय रूप से सार्थक पड़ाव)**

**टिप्पणी: स्टेरॉयड थेरेपी और बेहतर कार्डियक देखभाल के बाद**

ये चरण आपके बच्चे को अस्पताल में मिल रही स्वास्थ्य सेवाओं के साथ-साथ घर पर जो आवश्यक देखभाल मिलनी चाहिए उसका खाका तैयार करने में भी मदद करते हैं। साथ ही आपको बीमारी के अलग-अलग चरणो में अस्पताल की देखभाल करने वाली टीम और आपकी भूमिका की स्पष्ट रूप से जानकारी देने में मदद मिलती हैं ताकि,सही समय पर, सही देखभाल, सही जगह पर प्रदान की जासके।

1. ***प्रथम चरण. बीमारी के लक्षणों से पहले की स्थिति (शिशु अवस्था / बचपन)***

इस अवधि में अधिकांश बच्चों में इस बीमारी का कोई भी लक्षण दिखाई नहीं देता है। अतः ज्यादातर बच्चों में इस दौरान बीमारी का पता नहीं चल पता है। इस चरण में केवल वे ही परिवार परामर्श के लिए सामने आते हैं जिनमें यह समस्या पुरानी पीढ़ियों में देखने को मिलती है। कई बार कुछ अन्य समस्याओं के लिए रक्त परीक्षण के दौरान, यह मर्ज़ परामर्शदाता के पकड़ में आजाता है। इस अवस्था में लक्षण मौजूद भी हो सकते हैं जैसे देरी से चलना या देर से बोलना लेकिन अक्सर किसी का इनकी तरफ़ ध्यान ही नहीं जाता है। इस चरण में किसी भी प्रकार के इलाज की आवश्यकता नहीं पड़ती है। परन्तु, यदि इस चरण में बीमारी होनी की पुष्टि हो जाती है तो मरीज को नियमित जांच के लिए अस्पताल में बुलाया जाता है।

2. ***प्रारंभिक एम्बुलेशन अवस्था (बचपन का चलना)***
प्रारंभिक अम्बुलेटरी स्टेज में लड़के में बीमारी के स्पष्ट लक्षण दिखने शुरू हो जाते हैं। हालांकि ये बहुत ही कम एवं प्रारंभिक संकेत हो सकते हैं :

- बच्चों को फर्श से उठकर खड़े होने में अपने हाथो की मदद लेनी पड़ती है, जैसे कि वह अपने आप पर चढ़ रहे हैं, चित्र संख्या: ५ से आसानी से समझा जा सकता है। इस विशेष तरीका को "गोवेर्स मनुवेर" कहते हैं।
- हमें बच्चों में वेडलिंग वॉक (चाल) भी देखने को मिलती है। जिसमें बच्चा छोटे छोटे कदम लेते हुए अपने शरीर को झुलाते हुए चलता है।
- बच्चा अपने पैरों के पंजों पर चलता है। (टो वॉकिंग)
- पैरों की पिंडलियाँ अपने आकार से ज्यादा बड़ी दिखती हैं। (स्यूडोहाइपरट्रोफी)
- फ़र्श पर लेटे- लेटे सिर या गर्दन को उठाने में कठिनाई महसूस होती है तथा उसको अपने हाथों का सहारा लेना पड़ता है।
- हालांकि बच्चे सीढ़ियों चढ़ लेते हैं, लेकिन चलने के दौरान कठिनाई महसूस करते हैं और बार-बार गिर सकते हैं।
- रक्त में क्रिएटिन कइनेंस का बढ़ा हुआ स्तर, (२०० से अधिक), और लिवर एंजाइम्स के स्तर में बढ़ोतरी (ऐ एस टी या ऐ एल टी) भी ड्यूचेन की तरफ़ इशारा करती है। ऐसी स्थिति में अतिरिक्त जांच की आवश्यकता पड़ती है।

गोवेर्स साइन ड्यूशेन और बेकर्स मस्कुलर डिस्ट्राफी के बच्चों में दिखता है

इसी अवस्था में माता-पिता चिकित्सकीय सलाह लेना शुरू करते हैं। जब जांच के बाद ड्यूचेन का संदेह होता है तो कुछ अतिरिक्त रक्त परीक्षण किए जाते हैं। रोग की पुष्टि, आनुवंशिकता के परीक्षण के बाद ही की जाती है। आनुवंशिकता की जांच डी. एन. ए. में परिवर्तन की पहचान करना है, इसको ही हम जेनेटिक उत्परिवर्तन (म्युटेशन) कहते हैं । इन आनुवांशिक परीक्षणों की व्याख्या करने और इस बात पर चर्चा करने के लिए कि विशेषज्ञ (आनुवांशिक परामर्शदाताओं) की आवश्यकता हो सकती है। चर्चा में यह भी समझाया जाता है कि इस बीमारी का पता चलने पर आपके बच्चे और परिवार पर क्या प्रभाव पड़ सकता है। आनुवांशिकता और इसकी महत्व की चर्चा हम पिछले अध्याय में विस्तार से कर चुके हैं।

डी एम् डी के बच्चों में सीखने की दिक्कत और व्यवहार संबंधी समस्याएं भी अक्सर देखने को मिलती हैं। उनमें से कुछ मस्तिष्क में डिस्ट्रोफिन की कमी के कारण और अन्य शारीरिक सीमाओं की वजह से होती हैं । ये दिक्कतें परिवार वालों के साथ मिल कर काम करने में मुश्किलें उत्पन करती हैं।

इस चरण के इलाज में आमतौर पर प्रारंभिक अवस्था की फिजियोथेरेपी की शुरुआत की जाती है, जो मांसपेशियों को

लचीला बनाए रखता है और जोड़ों की जकड़न को कम करता है। पुनर्वास टीम बच्चों को प्लेटाइम या स्कूल के मध्यांतर के समय उपयुक्त व्यायाम साथ ही साथ अनुकूल शारीरिक शिक्षा पर भी सलाह दे सकती है। इस स्तर में लंबे समय तक मांसपेशियों में खिंचाव प्रदान करने और टखने की गतिशीलता को बनाए रखने के लिए नाइट स्प्लिन्ट्स या "टखने के पैर की अर्थ्रोसिस," या "एएफओ" के इस्तेमाल की सलाह दी जाती है।

स्टेरॉयड थेरेपी की शुरूआत इस चरण में हो जाती है। यह जरुरी है कि इस रोग की पुष्टि करते समय विशेषज्ञ (परामर्शदाता) माता पिता को स्टेरॉयड के इस्तेमाल के बारे में चर्चा करें। इसमें स्टेरॉइड से होने वाले फ़ायदे तथा नुकसानों के बारे में अवगत कराया जाता है।

स्टेरॉयड थेरेपी की शुरूआत करने से पहले निमोनिया और फ्लू से बचाव के लिए पूरा रोगनिरोधी टीकाकरण (प्रोफाइलेक्टिक इम्यूनाइजेशन) आवश्यक है। स्टेरॉयड थेरेपी की शुरुवात में ही शरीर की हड्डियों की गुणवत्ता पर नज़र रखने केलिए एक स्कैन किया जाता है। इस स्कैन को "डेक्सा स्कैन " कहते हैं, यह एक उन्नत क़िस्म का "एक्स रे" है। डेक्सा स्कैन हमें हड्डियों में खनिजों के घनत्व (बोन डैंसिटी) के बारे में जानकारी देता है। डेक्सा स्कैन को नियमित अंतराल पर करते रहना होता है ताकि हड्डियों की सेहत पर नज़र रखी जासके।

हड्डियों को मजबूत रखने के लिए पोषण महत्वपूर्ण है, और विटामिन डी और कैल्शियम युक्त आहार लेने केलिए प्रोत्साहित करना चाहिए। एक पंजीकृत आहार विशेषज्ञ से पोषण संबंधी आवश्यकताओं पर चर्चा करना अत्यंत महत्वपूर्ण है। न्यूरोमस्कुलर बीमारी की नियमित जाँच के दौरान हर ३ महीने में ऊंचाई और वजन पर विशेष निगरानी रखी जानी चाहिए। हृदय की कार्यशीलता की जानने के लिए बीमारी की पुष्टि होते ही ईसीजी (इलेक्ट्रो कार्डियोग्राम) या कार्डिएक एमआरआई या इकोकार्डियोग्राम परिक्षण/ जाँच करानी आवश्यक है।

### 3. *बाद की (लेट) एम्बुलेशन ( जातेहुए बचपन/ किशोर/ युवा वयस्क की) अवस्था*

इस चरण में मांसपेशियों की ताकत में निरंतर कमजोर होने के साथ, चलना बेहद मुश्किल हो जाता है। लड़कों को सीढ़ियों पर चढ़ना तथा फ़र्श से उठना मुश्किल हो जाता है। आखिरकार उनके लिए सहारे के बिना फ़र्श पर से बैठना या फ़र्श पर बैठना असंभव हो जाता है। टखने में संकुचन (कॉन्ट्रैक्टर) हो जाता है, अथार्थ टखनों की गतिशीलता में काफ़ी कमी आजाती है। साथ ही साथ कूल्हे की गतिशीलता में भी कमी आ जाती है।

फिजियोथेरेपी से मांसपेशियों का खिंचाव, व्यक्ति की कार्यक्षमता को बनाए रखने और लम्बे समय तक अपनी चलने फिरने की स्वतंत्रता बनाए रखने में सहायक होती है। जब जोड़ों की जकड़न / संकुचन को केवल फिजियोथेरेपी से संभाला नहीं जा पता तो आर्थोपेडिक विशेषज्ञों के परामर्श की आवश्यक हो सकता है। एक फिजियोथेरेपिस्ट आपके बच्चे की स्ट्रेचिंग अभ्यास की योजना उसके हिसाब से तैयार (कस्टमाइज़) कराता है और व्हीलचेयर जैसे उपकरणों के बारे में अवगत करता है। १० साल की उम्र तक, डीएमडी वाले लड़के चलने के लिए लंबे पैरों के ब्रेसिज़ पर निर्भर हो जाते हैं। ज्यादातर समय वे व्हीलचेयर तक ही सीमित रह जाते हैं। ११ से १३ वर्ष की आयु आते तक वे चलने फिरने की क्षमता खो देते हैं और व्हीलचेयर तक ही सीमित रह जाते हैं।

स्टेरॉयड थेरेपी को निरंतर चलती रहती है, परन्तु साइड-इफेक्ट्स के रोकथाम पर ध्यान देने की आवश्यकता होती है। साल में तीन से चार बार मांसपेशियों के सामर्थ्य की जांच पड़ताल करने की आवश्यकता पड़ती है। वजन पर लगातार नियंत्रण बनाए रखने के लिए निरंतर ध्यान रखा जाना चाहिए और किसी भी चिंता की स्थिति में उचित हस्तक्षेप की आवश्यकता होती है। फ्रैक्चर का खतरा शरीर की गतिशीलता

में कमी के साथ बढ़ता है इसलिए हड्डियों की सेहत की लगातार निगरानी (मॉनीटरिंग) होनी चाहिए। डेक्सा स्कैन हमें हड्डियों में खनिजों के घनत्व (बोन डैंसिटी) के बारे में जानकारी देता है। हड्डियों की सेहत का आंकलन हम रक्त में विटामिन डी की मात्रा तथा रीढ़ की हड्डी (स्पाइनल) एक्स-रे से भी लगा सकती है।

भोजन में विटामिन डी और कैल्शियम के पर्याप्त मात्रा को सुनिश्चित करने के लिए आहार का मूल्यांकन समय-समय पर किया जाना चाहिए। बच्चों में वृद्धि की दर पर निगरानी रखने के लिए बच्चे की ऊँचाई, लंबाई और वजन पर नज़र बनाए रखना बहुत ज़रूरी है।

इस चरण में हृदय की कार्यशीलता पर निगरानी के लिए सालाना ईसीजी (इलेक्ट्रो कार्डियोग्राम) या कार्डिएक एमआरआई या इकोकार्डियोग्राम परिक्षण/ जाँच करानी आवश्यक है। हृदय में किसी भी प्रकार की प्रतिकूल परिवर्तन मिलने पर हृदय रोग विशेषज्ञ कोई भी खास उपचार की सलाह दे सकते हैं।

बच्चों की शैक्षिक आवश्यकताओं को नज़र में रहती हुए मनोवैज्ञानिक और न्यूरोसाइकोलॉजिस्ट उपयुक्त सलाह दे सकते हैं। ये सब उपाय बच्चे को अपने दोस्तों के साथ मिल जुलकर रहने में सहायता प्रदान करते हैं।

4. ***प्रारंभिक गैर-एम्बुलेटरी (जातेहुए बचपन/ किशोर/ युवा वयस्क की) अवस्था***

शुरुआती गैर-एम्बुलेटरी चरण में बच्चों को चलना बहुत मुश्किल रहता है, ब्रेसिज़ के साथ भी। पैरों में लगातार संकुचन के कारण, उनमें से ज्यादातर बच्चे व्हील चेयर पर आ जाते हैं।

वर्त्तमान में प्रतिदिन पैरों की मांसपेशियों के खिंचाव की कसरत / एक्सेर्साइज़ के अलावा फिजियोथेरेपी के जरिये पैरों, कंधे, कोहनी, कलाई और हाथों की उंगलियों की अकड़न को कम करना बहुत ज़रूरी हो जाता है। चलना फिरना बंद हो जाने के बाद रीढ़ की हड्डी का टेढ़ापन (स्कोलियोसिस)पर लगातार नज़र रखना बहुत ज़रूरी हो जाता है। कुछ मामलों में, स्कोलियोसिस कुछ ही महीनों में काफी तेजी से बढ़ सकता है। स्कोलियोसिस और सांस की मांसपेशियों में कमज़ोरी होने के कारण सांस लेनेमें तकलीफ़ होती है। इसलिए फिजियोथेरेपी का मुख्य केंद्र बिंदु बच्चे को सही बैठने का ढंग, हाथों की मांसपेशियों के खिंचाव की कसरत (स्ट्रेचिंग) एवं श्वसनीय व्यायाम सीखना है। पैरों की विकृतियों को बचाने के लिए आर्थोपेडिक उपकरणों की भी आवश्यकता हो सकती है। ये विकृतियां पैरों में दर्द, असहजता एवं उचित जूते ढूढ़ने में बाधक सिद्ध होती है।

इस अवस्था तक आते -आते बच्चे एक लम्बे समय से स्टेरॉयड थेरेपी पर होता है, इसलिए स्टेरॉयड के साइड-इफेक्ट्स की निगरानी एवं रोकथाम पर ध्यान देना अति आवश्यकता हो जाता है। इस चरण में भी स्केलेटल मांसपेशियों की शक्ति एवं कार्यक्षमता तथा वजन को नियंत्रण में रखने के लिए वर्ष में दो बार इनका मूल्यांकन बहुत महत्वपूर्ण है। रीढ़ की हड्डी के फ्रैक्चर को खास तोर से ध्यान में रखते हुए हड्डियों की सेहत की लगातार निगरानी की जानी चाहिए। अब बच्चा खड़ा नहीं हो सकता है, इसलिए बच्चे की ऊंचाई बाँह की लम्बाई ("अलनार लंबाई") तथा निचले पैर ("टिबियल लंबाई") से मापा जा सकता है। भोजन में विटामिन डी और कैल्शियम के पर्याप्त मात्रा को बढ़ाने पर जोर दिया जाना चाहिए। हृदय और श्वास लेने के लिए सहायक मांसपेशियों की काम करने की क्षमता पर नियमित निगरानी रखनी चाहिए। हृदय की कार्यशैली पर निगरानी के लिए सालाना ईसीजी (इलेक्ट्रो कार्डियोग्राम) या कार्डिएक एमआरआई या इकोकार्डियोग्राम परिक्षण/ जाँच करानी है। इस अवस्था में हृदय के आकार में वृद्धि हो सकती है। पल्मोनरी फंक्शन टेस्ट (पी ऍफ़ टी) के माध्यम से श्वसन कार्य की सक्षमता का परीक्षण करना चाहिए। यदि ज़रूरत पड़े तो उचित इलाज भी देना चाहिए।

यह चरण बचपन से किशोरावस्था या किशोरावस्था से युवा वयस्क तक परिवर्तन (ट्रांजीशन) का समय है। ये समय तरुण अवस्था का आगमन है, तो इसकी खास निगरानी की ज़रूरत होती है। इस अवस्था में हार्मोनल बदलाव होना शुरू हो जाते

हैं। यह अच्छा हो अगर आपके लड़के की बाल चिकित्सीय टीम और वयस्क चिकित्सा टीमों में एक सामंजस्य बैठ सके। बच्चे की पढ़ाई, व्यावसायिक दिलचस्पी और चिकित्सा देखभाल प्राप्त करने में उसके चुनाव/ निर्णय को बढ़ावा देना चाहिए। इस उम्र में यह महत्वपूर्ण है कि उसका एक सामाजिक दायरा हो या वह किसी प्रकार के समुदाय का हिस्सा बने। जिनके साथ वह ड्यूचेन के साथ रहने की भविष्य की योजनाओं के बारे में स्वतंत्र रूप से बात कर सकते हैं।

पैलिएटिव केयर (देखभाल) का उद्देश्य है, जीवन की गुणवत्ता का अनुकूलन करना और गंभीर, जटिल बीमारी में लोगों में पीड़ा को कम करना। इस प्रकार की देखभाल महत्वपूर्ण होती है, मुख्यतः यह रोगी एवं उसके परिवार की जीवन की गुणवत्ता को बनाए रखने में सहायक होती है।

### *3. बाद की गैर-एम्बुलेटरी अवस्था (युवा वयस्क/ वयस्क)*

इस अवस्था में निचले अंग के अलावा ऊपरी अंग में भी काफ़ी कमजोरी आ जाती है, उचित आसन में बैठने में अत्यंत कठिनाई होटी है। फिजियोथेरेपिस्ट विभिन्न- विशिष्ट व्यायामों तथा उपकरणों का सुझाव दे सकते हैं जो व्यक्ति को खुद की देखभाल करने में ज़्यादा से ज़्यादा स्वतंत्रता प्रदान करें। ऑक्यूपेशनल थेरेपिस्ट मामूली घरेलू सुधारों के साथ व्यक्ति को स्वयं अपनी देखभाल करने में भी मदद कर सकते हैं (उदाहरण के लिए खाना- पीना, ब्रश करना, दांत धोना, शौचालय की सीट का उपयोग करना अन्य। इस चरण में सहायक - उपकरणों (असिस्टीव- डिवाइसेस) का इस्तेमाल व्यक्ति की स्वतंत्रता को और अधिक सुनिश्चित करता है। ताकि परिवार पर पड़ रहे रोगी की देखभाल का बोझ कम किया जा सके।

इस चरण में भी स्टेरॉयड के साइड-इफेक्ट्स की निगरानी के साथ चलती रहेंगी। श्वसन और ऊपरी अंगों की शक्ति और कार्यों को संरक्षित करने के लिए स्टेरॉयड का आजीवन सेवन करने की सलाह दी जाती है।

साँस लेने की मासपेशियां लगातार कमज़ोर पड़ती जाती हैं, जिसके कारण अपनेआप साँस लेना और अधिक मुश्किल हो जाता है। इस अवस्था में मशीन की सहायता से साँस लेने की सलाह दी जाती है। इसको मैकेनिकल वेंटिलेशन भी कहते है। शरीर की खांसी करने की क्षमता में भी कमी आ जाती है और कभी कभी सहायक उपकरणों (कफ असिस्टीव- डिवाइस) की आवश्यकता हो सकती है। अब श्वसन एवं हृदय की काम करने की क्षमता में लगातार और अधिक गिरावट आजाती है, इसलिए इन दोनों की कार्य की क्षमता की लगातार और बारंबार निगरानी रखना अति आवश्यकता हो जाता है। बोन हेल्थ पर भी निरंतर निगरानी रखने की आवश्यकता होती है। जीवन की गुणवत्ता बनाए रखने में पॉलिटीवे केयर की लगातार अपना काम करती रहती है।

ड्यूचेन के साथ जीवन यापन करने में, किशोरावस्था से वयस्क बनने के क्रम में एक अच्छी योजना बनाने की आवश्यकता होती है। योजना बनाने की शुरुआत १३ से १४ वर्ष की उम्र में कर ली जानी चाहिए। किशोरावस्थाअवस्था से वयस्क बनने की क्रम के बदलाव की योजना इस बात पर निर्भर करती है कि व्यक्ति और उसके परिवार की उसके भविष्य की क्या उम्मीदें और लक्ष्य हैं। भविष्य की इस योजना के मुख्य आधार शिक्षा, रोजगार और समाज में उसकी आवागमन की सहूलियत के आधार पर करना चाहिए। ड्यूचेन के साथ जीवनयापन करना काफ़ी महेंगा है। उसको एक साधारण से जीवन जीने केलिए भी औरों से अधिक संसाधनों की आवश्यकता पड़ती है। अस्पताल की स्वास्थ्य देखभाल टीम (हेल्थ केयर टीम) उपलब्ध संसाधनों का पता लगाने तथा सरकार द्वारा मिलने वाले समर्थन का पता लगाने में भी मदद कराती है। इस स्तर पर दोस्तों के साथ जुड़े रहना और कुछ समुदायों का हिस्सा बने रहना, मानसिक एवं सामाजिक स्थिरता के लिए भी महत्वपूर्ण है।

## 4. क्या यह एक गंभीर बीमारी है? यह एक प्रभावित बच्चे के जीवन पर क्या असर डालती है?

ड्यूशेन की संभावना मात्र से ही मन में कई प्रकार की शंकाएं उत्पन हो जाती हैं। प्रभावित लोगों और उनके परिवार के सदस्यों में चिंता और तनाव हो सकता है।

मरीज के साथ-साथ माता-पिता के दिमाग में कई सवाल आ सकते हैं, जैसे क्या डी.एम.डी. एक खतरनाक बीमारी है? डी.एम.डी. से प्रभावित व्यक्ति कितने वर्ष तक जीवित रह सकता है? स्कूल और सामाजिक स्तर पर यह बीमारी बच्चे के जीवन को कैसे प्रभावित करता है? डी एम् डी वयस्कों के रहन- सहन को कैसे प्रभावित करता है? कौन-कौनसी मांसपेशियाँ अथवा अंग-प्रत्यंग प्रभावित होते हैं? क्या इस बीमारी का कोई निश्चित इलाज है या निकट भविष्य में कोई पूर्ण इलाज आने की सम्भावना है?

डीएमडी से प्रभावित बच्चों में आमतौर पर लक्षण तीन से पांच साल की उम्र तक दिखना शुरू हो जाते हैं परन्तु माता-पिता अक्सर ८ से १० साल में डॉक्टरों की सलाह लेना शुरू करते हैं। बच्चे आमतौर पर १० से १२ वर्ष की आयु तक अपनी चलने फिरने की क्षमता खो देता है और इधर उधर जाने केलिए व्हीलचेयर पर निर्भर हो जाते हैं। अंततः रोगी बिस्तर पर पड़ (बेडबाउंड) जाता है। बीमारी की बाद के चरणों में रोगी को सांस लेने में तकलीफ, हृदय संबंधी समस्या हो जाती हैं। रोगी को अपनी रोज़मर्रा की जरूरतों के लिए दूसरों पर या असिस्टीवे डिवाइसेस (सहयोगी यन्त्रों ) आश्रित रहना पड़ता है। इस प्रकार बीमारी जीवन के विभिन्न दशकों में धीरे धीरे बढ़ती है। बीमारी के इन सभी चरणों की चर्चा पिछले अध्याय में की गई है। इन रोगियों की औसत आयु 17-28 वर्षों के बीच में होती है।

ड्यूशेन मस्कुलर डिस्ट्रॉफी से प्रभावित व्यक्ति का जीवन काल कम होता है और वह निश्चित रूप से जीवन में बाद में चलने फिरने के लिए व्हीलचेयर पर आश्रित हो जाते हैं। परन्तु उम्र के हिसाब से रोग की तीव्रता को पिछले कुछ दशकों में कम किया गया है। यह बहूआयामी इलाज़ और लगातार रोग पर मज़बूत निगरानी रखने की वजह से हुआ है। यद्यपि अभी कुछ दवाइयों के ज़रिये डायस्ट्रोफिन प्रोटीन की बनने की प्रक्रिया को पुनर्स्थापित किया जा चूका है परन्तु ये कुछ ही प्रकार के म्युटेशनों तक ही सीमित है। बीसवीं सदी के अंत तक हमारी चिकित्सा पद्धति इसी बात पे ज़ोर देती थी कि जब तक यह सामान्य रूप से संभव हो, तब तक घर पर रोगी के उपचार करें । हालत बिगड़ने पर ही अस्पतालों का रुख़ करें । इलाज़, बैठने की स्थिति में कुछ आराम प्रदान करना और मृत्यु के सबसे सामान्य कारणों से लड़ने तक ही सीमित था। और अधिकांश समय मरीज सामान्य सामाजिक जीवन नहीं जी पाते थे। आज की चिकित्सा पद्धति रोगियों के दीर्घकालिक इलाज़ और नई- नई पद्धति को इलाज़ की प्रक्रिया में सम्मिलित करने पर ज़ोर देती है। बीमारी के बाद के चरणों में रोगी की जीवन की गुंडवत्ता और जीवन काल को अपनी क्षमता तक बढ़ाना, इस दौर की चिकित्सा पद्धति का प्रमुख़ लक्ष्य है।

बचपन में प्रभावित बच्चा लगभग अपनी आयु वर्ग के किसी भी अन्य बच्चे की तरह ही होता है। वह स्कूल जा सकता है, स्कूल के स्तर पर विभिन्न गतिविधियों में आराम से भाग ले सकता है। परन्तु वह अक्सर गिरत जाता है। वह अपने साथियों की तुलना में धीमी चाल से चलता है और उसके पिंडली की मांसपेशी में अतिपुष्टि और आकार में बड़े दिखाई देते हैं। किंडर गार्डन स्तर (लगभग ४ साल) तक वह दूसरे बच्चों की तरह सामान्य रूप से बढ़ता है। उम्र बढ़ने के साथ वह अन्य लोगों से उन गतिविधियों में बिछड़ जाता है जिनमें शारीरिक क्षमता की अधिक ज़रूरत होती है। जैसे स्कूल में या घर पर उसके लिए सीट या फ़र्श से उठना मुश्किल हो जाता है । फ़र्श पर पड़े सामान उठाना, जूते के फीते बांधना इन सब कामों में भी उसको मुश्किलों का सामना करना पड़ता है । उसके लिए लंबी दूरी तक चलना, सीढ़ी चलना मुश्किल हो जाता है। ५ से ८ साल की उम्र आते आते उसकी चाल सामान्य से थोड़ी अलग़ हो

जाती है। उसको ज़ल्दी-ज़ल्दी थकान महसूस होने लगती है इसलिए उसकी ज्यादातर गतिविधियाँ घर के भीतर तक ही सीमित हो जाती हैं।

किशोरावस्था आते-आते मांसपेशियों की कार्य-क्षमता और अधिक बिगड़ती चली जाती है। मांसपेशियों की यह कमज़ोरी धीरे-धीरे शरीर के ऊपरी भागों में भी आ जाती है। निचले शरीर से जुड़ी गतिविधियों के लिए सहायता की आवश्यकता हो सकती है। किशोरावस्था में चलना असुरक्षित और बोझिल हो जाता है। समय बीतने के साथ बच्चे को सांस लेने में कठिनाई बढ़ती जाती है, हृदय की कार्यक्षमता में कमी और चलनी फिरना के लिए व्हीलचेयर पर निर्भर हो जाता है । वयस्क होते होते उसके लिए रोज़मर्रा की सामान्य लेकिन महत्वपूर्ण गतिविधियां जैसे लेखन, चित्रकारी, गायन और अपनी व्हीलचेयर को यहाँ - वहाँ लेजाना भी मुश्किल हो जाता है । दैनिक गतिविधियों को करने जैसे नहाना-धोना, स्वयं भोजन करने कहीं अधिक समय लगने लग जाता है। कई बरी दैनिक गतिविधियों के लिए उसको सहायता की आवश्यकता पड़ती है। अकेलेपन के कारण कभी-कभी सामाजिक जीवन प्रभावित होता है। व्यक्ति का परिवेश उसकी शिक्षा, आसपास होने वाले सांस्कृतिक आयोजनो, उसकी सामाजिक भागीदारी और साथ ही साथ व्यक्ति का उपचार के विकल्पों के चुनाव पर भी आप प्रभाव डालता है।

उदाहरण के लिए, साँस लेने के लिए वह किस श्वसन उपकरण का चयन करता है, व्यक्तिगत सहायता के लिए क्या वह किसी देखभालकर्ता को पैसे देकर नियुक्त करता है, व्हीलचेयर, कंप्यूटर, स्मार्टफोन का उपयोग करता है और यदि संभव हो तो एक स्वतंत्र आत्मनिर्भर जीवनयापन का चयन हो सकता है। ये सब विकल्प सीधे तौर पर बीमार व्यक्ति और उसके परिवार की जीवन की गणवत्ता को प्रभावित करता है।

रोगियों के लम्बे समय तक इलाज में कई चुनौतियां आती हैं जिनमें से सबसे महत्वपूर्ण है, साँस लेने के लिए बाहर से मशीनी सहायता। बीमारी बढ़ने के साथ साँस लेनेवाली मासपेशियां अत्यधिक कमज़ोर पड़ जाती हैं इसलिए बाहर से मैकेनिकल वेंटिलेशन की आवश्यता पड़ती है। मैकेनिकल वेंटिलेशन एक सरल और आसानी से नियंत्रण किए जासकने वाली तकनीक है, परिष्कृत उपकरणों के उपयोग के बिना भी घर पर प्रारंभिक वेंटिलेशन संभव है। बीमारी के आंखरी चरण में नाक से इनवेसिव वेंटिलेशन की आवश्यकता पड़ती है। डी एम् डी के मरीजों में मृत्यु का प्रमुख कारण श्वांस - प्रणाली की समस्यायें है। परन्तु मैकेनिकल वेंटिलेशन की सहायता से जीवन को लम्बा एवं बेहतर बनाया जा सकता है। वेंटीलेटर के सहयोग पाने वाले मरीज़ों का जीवन कल २५ से २६ साल का होता है। समय के साथ हृदय और श्वसन देखभाल/ इलाज में सुधार आने के कारण डी एम् डी के मरीजों के जीवनकाल में लगातार सुधर आया एक यूरोपियन अध्ययन के मुताबिक़ बिना किसी बाहरी वेंटिलेशन की सहायता के जीवनकाल लगभग १९ वर्ष था जो कि वेंटिलेशन की सहायता से २७ वर्ष तक पहुँच गया। इसके विपरीत बेक्केर्स मस्कुलर डिस्ट्रॉफी के मरीज़ हृदय उचित देखभाल से वयस्क होने तक एक नार्मल ज़िन्दगी जीते हैं, जबकि ठीक इसी तरह की परस्थितियों में एक डी में डी का मरीज़ ३० साल तक जीवित रह सकते हैं ।

संयुक्त राज्य अमरीका के एडम मैकडोनाल्ड, दुनिया के डी एम् डी से प्रभावित सबसे उम्रदर व्यक्तियों में से एक हैं । नीदरलैंड के 54 वर्षीय व्यक्ति भी दुनिया के डी एम् डी से प्रभावित सबसे उम्रदर व्यक्तियों में से एक हैं, जिनके दो भाई थे वे भी ड्यूशेन से प्रभावित थे जो १५ वर्ष और ४१ वर्ष की आयु तक जीवित रहे।

हालांकि कोई इलाज मौजूद नहीं है, फिर भी स्टेरॉइड्स मासपेशियां का टूटना कम करता है और बीमारी की जटिलता को कम करता है। कार्डियोमायोपैथी के बढ़ने के क्रम को भी कम करता है, श्वसन या श्वास संबंधी कठिनाइयों के लिए गैर इनवेसिव और इनवेसिव असिस्टेड वेंटिलेशन कारगर सिद्ध होता है।

इसके अलावा, नई ड्रग थेरेपी तथा जीन थेरेपी विकसित की जा रही हैं। ड्यूशेन का कोई भी एक सीधा सीधा इलाज संभव नहीं है, इस बीमारी का विभिन्न स्तरों पर इलाज करके ही रोकना संभव है।

बहुत कम प्रभावित व्यक्तियों को डीएमडी का उचित इलाज मिलपाता है, यह अस्पताल के दूर स्थित होने की वज़ह और सीमित वित्तीय संसाधनों के कारण होता है।

## खंड ग: बीमारी का इलाज एवं देखभाल

### 1. यह कैसे पता किया जाता है कि बच्चे को यह बीमारी है? (बीमारी का पुष्टीकरण)

एक बार ड्यूशेन का अंदेशा होने पर यह अति आवश्यक हो जाता है कि ज़ल्द से ज़ल्द रोग की पुष्टि की जाए। ताकि उचित इलाज की शुरुआत की जा सके और परिवार को भी इस बीमारी के लिए तैयार करने का यथोचित समय मिल सके। बीमारी की पुष्टि एक न्यूरोमस्कुलर विशेषज्ञ या एक अनुभवी चिकित्सक द्वारा किया जाना चाहिए, जिसके पास न्यूरोमस्कुलर रोगों के इलाज करने का अनुभव हो। न्यूरोमस्कुलर रोगों का विशेषज्ञ ही डी एम् डी के इलाज में कार्यरत टीम का लीडर होता है।

#### *ड्यूशेन होने का अंदेशा कब होता है?*

ड्यूचेन होने के मुख्य प्रारंभिक संकेत और लक्षण :

1. चलने में समस्याः डीएमडी से प्रभावित बच्चे उनकी उम्र के अन्य बच्चों की तुलना में देरी से चलना शुरू करते हैं। उनकी पिंडलियों की मासपेशियाँ बड़े आकार की (अतिपोषित) और कड़ी (टाइट) होती हैं। लड़कों को सीढ़ियों पर चढ़ने में कठिनाई महसूस होती है, वे थोड़ा अज़ीब ढंग से और कभी कभी अपने पैर के पंजों में चलते हैं। वे चलते या दौड़ते समय अक्सर गिर जाते हैं। सबसे अनोखा संकेत जो इन बच्चों में देखने को मिलता है वह है "गोवेर्स साइन",
   उन्हें फर्श से सीधे खड़े होने के लिए अपने शरीर पर हाथों के बल चढ़ना पड़ता है, इस विशेष संकेत के बारे में पिछले खंड में चर्च की जा चुकी है।
2. जाँच के दौरान क्रैएटिन काइनेज (CK, एक मांसपेशी का प्रोटीन ) का खून में बढ़ा हुआ स्तर मिलने पर , आगे की जाँच की पुष्टि करने के लिए उन्हें न्यूरोमस्कुलर विशेषज्ञ के पास भेजा जाना चाहिए। हालांकि क्रैएटिन काइनेज का बढ़ा हुआ स्तर मांसपेशियों की अन्य बिमारियों में भी देखा जा सकता है।
3. लीवर एन्जाइम का बिना कारण रक्त में बढ़ा हुआ मिलान। ये लीवर एन्जाइम हैं : अल्कलाइन अमिनोट्रांसफारसे (ALT), एस्पार्टेट अमिनोट्रांसफारसे (AST), अथवा लेक्टेट डी हाइड्रोजनेज़ (LDL). लेकिन कभी-कभी इन एंजाइमों में वृद्धि से अनावशयक रूप से लीवर की बीमारी की तरफ ध्यान केंद्रित हो जाता है और असल बीमाई की पुष्टि में देरी हो जाती है।
4. अक्सर इन बच्चों में भाषा के विकास में देरी, व्यवहारिक एवं बौद्धिक विकास की समस्याएं देखी जा सकती हैं।
5. परिवार में किसी सगे संबंधियों में इस बीमारी की पुष्टि होने से बच्चों में बीमारी होने की संभावनाएं बढ़ जाती हैं।

#### *रोग की पुष्टि*

डी.एम.डी एक आनुवंशिक बीमारी है और यह डायस्ट्रोफिन प्रोटीन के उत्पादन के लिए जिम्मेदार जीन में आए उत्परिवर्तन (म्युटेशन) की वज़ह से होती है। ये प्रोटीन सभी प्रकार की मांसपेशियों में पाया जाता है । इस बीमारी की पुष्टि जेनेटिक जाँच के द्वारा की जाती है। आनुवंशिकता की जाँच विभिन्न प्रकार के जेनेटिक जांचों के द्वारा, रक्त के नमूने ले के किया जाता है। मसल बायोप्सी, एलेक्ट्रोमोग्राफी जैसे अन्य परीक्षण से भी इस बीमारी का पता चल सकता है पर ये पुष्टिकारी जांचें नहीं हैं।

1. जेनेटिक जाँच

   भले ही अन्य परीक्षणों के द्वारा इस बीमारी का पता लगा लिया जा चूका हो फिर भी जेनेटिक जाँच डी.एम.डी की बीमारी की पुष्टि के लिए ज़रूरी होती है। विभिन्न जेनेटिक परीक्षणों के द्वारा आनुवंशिकता का पता डीएनए

में विस्तृत परिवर्तन (म्युटेशन या बहुरूपता) के माध्यम से लगाया जा सकता है। डायस्ट्रोफिन प्रोटीन के उत्पादन के लिए जिम्मेदार जीन (७८ एक्सॉन ) की जाँच मल्टीप्लेक्स लइगेशन प्रोब एम्पलीफिकेशन (MLPA), पोलीमेराज चेन रिएक्शन (पीसीआर) तथा जीन सीकुएंसिंग की जाती है। जेनेटिक जाँच के द्वारा आनुवंशिकता का पता लगने पर आनुवंशिकीविद् अथवा आपका डॉक्टर / कंसलटेंट या काउंसलर या नर्स , परिणाम काउंसलिंग के ज़रिये आपको समझते हैं। भविष्य में परिवार के लिए योजना बनाने में माता-पिता को प्रसव से पूर्व गर्भ में जेनेटिक जाँच मददगार सिद्ध होती है।

डिस्ट्रोफिन जीन इंसान के एक्स क्रोमोसोम में पाया जाता है, X क्रोमोसोम से जुड़ी आनुवंशिकता को हम पिछले अध्याय में समझ चुके हैं। यह पीढ़ी दर पीढ़ी चलती है और लड़के/ बेटे ही इस बीमारी से ग्रसित होते हैं। चूंकि यह एक बीमारी परिवार में चलती है, इसलिए यह भविष्य के गर्भधारण में भी हो सकती है। जो महिलाऐं डी·एम·डी से प्रभावित लड़के या पुरुष की रिश्तेदार (मौसियां, बहने) होती हैं उनमें वाहक परीक्षण (कर्रिएर टेस्टिंग ) की जेनेटिक टेस्टिंग की सलाह डी जाती है।

***मांसपेशी की बायोप्सी***

जब बच्चे में लक्षण एवं संकेत ड्यूचेन की संभावना जाहिर करते हैं परन्तु आनुवंशिक परीक्षण नकारात्मक होते है, उस परिस्थिति में मांसपेशियों की बायोप्सी की सलाह डी जाती है। मांसपेशी की बायोप्सी एक सर्जिकल प्रक्रिया है जिसमें मांसपेशी का एक छोटा सा टुकड़ा निकला जाता है और लैब में जांच के लिए बेजा जाता है। लगभग एक से दो हफ्ते में जांच का पता चल जाता है। बायोप्सी आमतौर पर क्वाड्रिसेप्स या जांघ की मांसपेशी पर की जाती है। मांसपेशी बायोप्सी से मांसपेशियों की कोशिकाओं में मौजूद डिस्ट्रोफिन की मात्रा के बारे में जानकारी मिल जाती है। यह जांच ड्यूचेन की पुष्टि करने में में सहायक होती है।

इस समय देखभाल का उद्देश्य जल्द से जल्द एक सटीक तौर पर बीमारी की पुष्टि करना होता है । शीघ्र से शीध्र बीमारी का पता चलते ही परिवार के सभी लोगों को ड्यूचेन, आनुवांशिक परामर्श और उपचार योजनाओं के बारे में सूचित किया जा सकता है। इस स्तर पर अस्पताल एवं समाज के द्वारा उचित देखभाल, निरंतर सहयोग और बीमारी से जुड़ी हुई शिक्षा की आवश्यकता होती है।

## 5. इस बीमारी के इलाज के विभिन्न घटक क्या हैं?

### क. न्यूरोमस्कुलर चिकित्सा

ड्यूशेन में शरीर की मांसपेशियों में धीरे- धीरे कमजोरी आती जाती है, इसलिए मरीज़ को एक न्यूरोमस्कुलर विशेषज्ञ के साथ नियमित जांच करते रहनी चाहिए। मांसपेशियों की लगातार बढ़ती कमज़ोरी की जाँच करते रहना बहुत महत्वपूर्ण होता है, ताकि परिजनों को बीमारी के अगले स्तर के लिए तैयार किया जा सके।

दिशा-निर्देशों के अनुसार मरीज़ को हर तीन महीने में अपने न्यूरोलॉजिस्ट को दिखाना चाहिए। इसके अलावा उसको लगातार अपने फिजियोथेरेपिस्ट या ऑक्यूपेशनल थेरेपिस्ट के संपर्क में रहना चाहिए।

प्रत्येक विज़िट के दौरान अस्पताल में कुछ निश्चित परीक्षण / मूल्यांकन आयोजित किए जाते हैं, जिनके आधार पर मांसपेशिओं में आए बदलाव को अंदाज़ को मापा जाता है और बीमारी की स्थिति का पता लगाया जा सकता है।

रोग के बढ़ने की दर का मूल्यांकन करने के लिए अलग़ अलग़ अस्पताल (क्लिनिक) विभिन्न एवं विशिष्ट परीक्षण करते हैं। परन्तु नियमित जांच के लिए परीक्षणों के एक ही सेट के द्वारा मूल्यांकन किया जाना बहुत महत्वपूर्ण है, यह एक तुलनात्मक तस्वीर देता है। चिकित्सक के लिए यह जानना महत्वपूर्ण है कि आपके बेटे की मांसपेशियां कैसे काम कर रही हैं ताकि वे जल्द से जल्द सही इलाज़ शुरू किया जा सके। नियमित जाँच आगे की कार्रवाई की दिशा तय करने में सहायक होता है, यदि कोई असामान्य नतीज़े सामने आएं तो अतिरिक्त जाँच या अतिरिक्त परामर्श लिया जासके। कुछ प्रकार के व्यायाम और अत्याधिक थकावट, मांसपेशियों को क्षतिग्रस्त कर सकते हैं, इसलिए आवश्यक है कि कुछ ख़ास तरह की फिज़िओथेरपी की जाए। मांसपेशियों की ताकत और कामकाज करने की क्षमता को बनाए रखने और संकुचन को रोकने करने के लिए स्ट्रेच कसरत की आवश्यकता होती है।

ये सभी कसरतें या जो कसरतें नहीं करनी हैं इसका आंकलन एक फिजियोथेरेपिस्ट की देखरेख में किया जाना चाहिए। आपको अपने बेटे को हर ३ महीनों में अपने डॉक्टर या सेपेशियलिस्ट से मिलवाना (दिखाना) चाइये। अपने बच्चे को न्यूरोमस्कुलर / बाल स्पेशलिस्ट फ़िज़ियोथेरेपिस्ट हर 3 महीने में दिखाते रहना चाहिए।

इस नियमित मूल्यांकन में कुछ नियामत टेस्ट किये जाते हैं जो यह मांसपेशियों की स्थिति को दर्शाता है। ये टेस्ट मांसपेशियों की निम्नलिखित गुणों की जाँच करते हैं :

**बल**

विशिष्ट जोड़ों पर उत्पन्न होने वाली ताकत को विभिन्न तरीकों से मापा जाता है ताकि मांसपेशियों में बदलाव का पता लगाया जा सके। उदाहरण के लिए मायोमेट्री।

**जोड़ों के गति की सीमा**

यह जांचें जोड़ों की सिकुड़न या कसाव का टेस्ट करती हैं।क्या जोड़ों में सिकुड़न या कसाव पैदा होने लगा है ? क्या जोड़ों को चलने में दर्द होता है ? ये जांचें जोड़ों की गतिशीलता को बनाए रखने के लिए खिंचाव (स्ट्रेचिंग) कसरतों के चुनाव करने में सहायक सिद्ध होती हैं।

**कार्य करने के समय की जाँच (टाइम फंक्शन टेस्ट )**

ये टेस्ट भी मांसपेशियों की ताक़त की जाँच करने में सहायक होते हैं और ये टेस्ट वैश्विक स्तर पर प्रमाणित हैं। कई गतिविधियाँ आम तौर पर क्लीनिक में नियमित रूप से की जाती हैं जैसे फ़र्श से लेट कर उठने का समय, एक निश्चित दूरी तक चलने का समय आमतौर पर १० मीटर चलने के समय, कुछ निश्चित सीढियाँ चढ़ने का समय। यह मांसपेशियों में हो रहे बदलाव की महत्वपूर्ण जानकारी देते हैं कि स्थिति कैसे बदल रही है और उपचार कैसे प्रतिक्रिया दे रहे हैं। सामान्य तौर पर किये जानेवाले टाइम फंक्शन टेस्ट हैं :

गोवर्स समय, १० मीटर चलने के समय, 6 मिनट तक चलने की की पैदल दूरी, 4 सीढ़ियाँ चढ़ने और उतरने का समय और नार्थ स्टार अम्बुलेटरी असेसमेंट । यह बीमारी में अक्सर कुछ कार्यों को करने में औरों से अधिक समय लगता है और यह समय बीमारी बढ़ने के साथ बढ़ता जाता है। ये जांचे उसी समय का मूल्याङ्कन करते हैं।

**स्वयं की देखभाल गतिविधियाँ**

क्लिनिक को बच्चे के स्वयं की देखभाल करने से सम्बंधित जानकारी प्राप्त करना महत्वपूर्ण होता है। जैसे कितनी सहजता से बच्चा अपनी देख्भाल कर पता है ( उदाहरण के लिए दांतों को ब्रश करना, शौचालय का उपयोग करने के बाद

**ख़. मेडिकल चिकित्सा**

डी एम् डी के इलाज के लिए नई दवाओं पर लगातार बहुत सारे शोध हो रहे हैं। प्रेडनिसोलोन और डफलाजाकोर्ट के इस्तेमाल का मांसपेशियों की क्षमता को बचाए रखने में सकारात्मक प्रभाव देखा गया इस बीमारी के इलाज़ में नई नई दवाओं के आने से भविष्य में निम्नलिखित दिशा निर्देशों में बदलाव आ सकता है।

- स्टेरॉयड जन्हें ग्लुकोकोर्टिकोइड्स या कॉर्टिकोस्टेरॉइड्स भी कहा जाता है, डीएमडी में मांसपेशियों की ताकत और मोटर फ़ंक्शन में गिरावट की गति को धीमा करने के लिए एकमात्र दवाएं है।
- स्टेरॉयड के उपयोग का मुख्य लक्ष्य बच्चे को लम्बे समय तक स्वतंत्र रूप से चलने और उसकी स्वतंत्रता बनाए रखने में मदद करना है।
- स्टेरॉइड के इस्तेमाल से सांस, दिल और ड्रोन जैसी समस्याएं भी कम होती हैं।
- स्टेरॉइड थेरेपी स्कोलियोसिस के जोख़िम को काम करती है। (रीढ़ की वक्रता)
- स्टेरॉयड के शरीर पर होने वाले नुक़सान (साइड इफेक्ट्स) का पूर्वानुमान लगाकर और सक्रिय रूप से उनकी रोकथाम एवं इलाज़ करना आवश्यकता है।

***स्टेरॉयड कब शुरू करें?***

- स्टेरॉयड उपचार शुरू करने के लिए सबसे उत्तम समय है जब मोटर फंक्शन (जैसे दौड़ना, सीढ़ियां चढ़ना और उतरना, फ़र्श पर से उठना, चलना इत्यादी) में बहुत कम या कोई बदलाव नहीं दीखता है। इस अवस्था का प्लैटू फेज भी कहते हैं, लड़कों पतलून खींचना इत्यादि), घर और स्कूल में उसकी गतिविधियाँ। इससे उसे कुछ अतिरिक्त सहायता दी जा सकती है जिसकी उसे आवश्यकता होगी।

के मोटर फंक्शन में सुधार होना बंद हो जाता है परन्तु अभी स्थिति ख़राब होनी शुरू नहीं हुई है। यह आम तौर पर 4 से 6 साल की उम्र के बीच का समय होता है।

- स्टेरॉयड का उपचार शुरू होने से पहले राष्ट्रीय वैक्सीन टीकाकरण कार्यक्रम पूरा किया जाना चाहिए, इसके अलावा छोटी माता/छोटी चेचक (वेरीसेल्ला) का टीकाकरण भी जरुरी है।
- लड़कों और युवा वयस्कों (जो अब स्वतंत्र रूप से नहीं चल रहे हैं) में स्टेरॉयड का इस्तेमाल एक व्यक्तीगत निर्णय है और इसकी अपने चिकित्सक के साथ चर्चा करनी चाहिए।
- कुछ विशेषज्ञों का मानना है जिन लड़कों में स्टेरॉयड का उपयोग उस अवस्था में शुरू हो गया था जब वे स्वतन्त्र रूप से चल फ़िर रहे थे, उनको स्टेरॉयड का इस्तेमाल जारी रखना चहिये भलेही अब वे स्वतन्त्र रूप से चलने में असमर्थ हों।
- नॉन -एम्बुलेटरी (जो अब स्वतंत्र रूप से नहीं चल सकते हैं ) व्यक्तियों में इसका उद्देश्य होता है हाथों की ताकत को बनाए रखना। स्कोलियोसिस के बढ़ने की गति को धीमा करना और श्वसन और हृदय संबंधी कार्यक्षमता में गिरावट की दर को कम करना।

**विभिन्न स्टेरॉयड दवाइयाँ**

- प्रेडनिसोन (प्रेडनिसोलोन) और डिफ्लैजाकॉर्ट दो प्रकार की स्टेरॉयड हैं जो मुख्य रूप से डीएमडी में प्रयोग की जाती हैं।
- स्टेरॉयड का चुनाव उसकी उपलब्धता, लागत और दुष्प्रभावों पर निर्भर करता है।

- स्टेरॉयड की टैब्लेट मुख्य तौर पर दिन में एक बार लेनी पड़ती है।
- प्रेडनिसोलोन की प्रारंभिक खुराक 0.75 मिलीग्राम प्रति किलोग्राम प्रति दिन होती है तथा डिफ्लैजाकॉर्ट की 0.9 मिलीग्राम प्रति किलोग्राम प्रति दिन, प्रातः काल।
- कुछ बच्चों में थोड़ी समय (कुछ घंटों) के लिए व्यवहार में बदलाव देखा जासकता है, जैसे मनःस्थिति में बदलाव, अत्याधिक सक्रियता, एकाग्रता (ध्यान) की कमी , इन बच्चों के लिए, दवा को दोपहर या शाम के समय पर देनेसे इन परेशानियाँ कम हो सकती हैं।
- स्टेरॉयड की खुराक निर्धारित करने के लिए तीन बातों के बीच संतुलन बनाए रखना जरुरी है, १ बच्चे का उचित तौर से बढ़ना, २ स्टेरॉयड की प्रतिक्रिया कितनी अच्छी है तथा ३ दवा का दुष्प्रभाव जिसकी रोक थाम की जा सकती है। हर बार ३ महीने की नियमित क्लीनिकल जाँच के दौरान इन तीनो बातों पर ध्यान दिया जाता है।
- प्रेडनिसोलोन की अधिकतम ख़ुराक 30 मिलीग्राम प्रति किलोग्राम प्रति दिन तक रोक दी जाती है तथा डिफ्लैजाकॉर्ट ३६ मिलीग्राम प्रति किलोग्राम प्रति दिन पर।
- अगर स्टेरॉयड बच्चे के लिए असहनीय हो जाए या दुष्प्रभाव रोक थाम में कठिनाइयां बढ़ जाएँ तो इसकी खुराक कम करने की सलाह दी जाती है। परन्तु इस बात निर्णय स्पेश्यलिस्ट द्वारा दवा के साइड इफेक्ट के नियंत्रण का आकलन करने के बाद लिया जाता है।
- यदि स्टेरॉयड की ख़ुराक़ कम करने के बावजूद दुष्प्रभावों में ज़रा भी कमी न आए और अगर वे बढ़ते ही जातें है तो इस दवा को बंद कर के दूसरे इलाज़ की जरुरत होती है।

**स्टेरॉयड का इस्तेमाल और दुष्प्रभाव (साइड इफेक्ट्स)**

- लंबे समय तक स्टेरॉयड थेरेपी पर रहने के बाद स्टेरॉयड संबंधित दुष्प्रभावों का इलाज़ करना महत्वपूर्ण हो जाता है।
- स्टेरॉयड थेरेपी वर्तमान में डीएमडी के लिए दवा के द्वारा चिकित्सा का मुख्य आधार है इसलिए यह चिकित्सक या परिवार द्वारा हल्के में लिया जाना चाहिए। इसका इस्तेमाल स्पेशियलिस्ट की निगरानी में ही किया जाना चाहिए।
- अस्पताल में नियमित जाँच के समय मुख्य रूप से स्टेरॉयड थेरेपी संबंधित दुष्प्रभावों जैसे: बच्चे का उचित तौर से बढ़ना, वज़न, तस्र्ण अवस्था के बदलाव, हड्डियों का स्वास्थ्य, व्यवहार में परिवर्तन, मोतियाबिंद, इंट्राऑक्यूलर दबाव (ग्लोकोमा: आँख की बीमारी जिसमें पुतली फैल जाती है और दृष्टि धीरे- धीरे चली जाती है।
- अगर ख़ुराक़ में कमी के बावज़ूद साइड इफेक्ट्स का इलाज़ नहीं हो पता हैं, या दवाई अप्रभावी साबित होती है, फिर स्टेरॉयड थेरेपी को बंद करने के बारे में सोचने की आवश्यकता है। इन फैसलों को बच्चे, परिवार और स्पेशलिस्ट को मिल कर व्यक्तिगत रूप से करना चाहिए।
- स्टेरॉयड को कभी भी अचानक बंद नहीं करना चाहिए।

***सारिणी १: स्टेरॉयड के मुख्य दुष्प्रभावों का संक्षिप्त विवरण और उनको नियंत्रित करने के लिए क्या करना चाहिए।***

| **स्टेरॉयड संबंधित दुष्प्रभावों** | **विवरण** | **क्या किये जाने की आवश्यकता है?** |
|---|---|---|
| वजन का बढ़ना,<br>मोटापा | स्टेरॉयड थेरेपी शुरू करने से पहले आहार की सलाह ज़रूरी है। स्टेरॉयड लेने से भूख बढ़ाता है। | वजन बढ़ने से रोकना बहुत ज़रूरी है, इसके लिए आवश्यक है की पूरा परिवार मिलकर समझदारी से एक संतुलित आहार लेना चाहिए। |
| कुशिंगिंगोइड फीचर्स<br>"चंद्र-मुख" | चेहरे और गालों में भारीपन | खाने-पीने में पूरी सावधानी बरतनी चाहिए, चीनी और नमक का सेवन को कम करना चाहिए। |
| बच्चे का उचित तौर से बढ़ने में कमी "ग्रोथ रिटार्डेशन" | डीएमडी से प्रभावित लड़कों में दूसरों की तुलना में लम्बाई कम ही रहती है। | अस्पताल में नियमित जाँच के एक हिस्से के रूप में कम से कम हर 6 महीने में बच्चे की लम्बाई नापी जाती है। |
| उच्च रक्तचाप (हाइपर्टेंशन) | प्रत्येक क्लीनिकल जाँच के दौरान रक्तचाप (ब्लड प्रेशर) पर नज़र राखी जाती है। | यदि ब्लड प्रेशर बढ़ा हुआ मिलता है तो खाने में नमक की मात्रा और वजन को कम करना, ब्लड प्रेशर को नियंत्रित करने की दिशा में पहला कदम है। यदि इससे बात नहीं बनती है तो आपके चिकित्सक बीपी की दवाई शुरू की जा सकती है। |
| हिर्सुटिज़्म (अत्यधिक बालों का उगना) | स्टेरॉयड के इस्तेमाल से कभी कभी शरीर पर अत्यधिक बाल उग जाती हैं। | यह आमतौर पर गंभीर समस्या नहीं है दवा में बदलाव ही इसकेलिए पर्याप्त है। |
| मुँहासे और मस्से | किशोरावस्था में ये अधिक देखे जाते हैं। | ऊपर से लगाने की दवा का इस्तेमाल करें। |
| ग्लूकोज पर नियंत्रण न रहना | प्यास और मूत्र का अधिक लगना | क्लीनिकल जाँच के समय स्टिक टेस्ट के ज़रिये पेशाप में गुलूकोज़ की मात्रा की जाँच करनी चाहिए।<br>साल में एक बार रक्त में गुलूकोज़ की मात्रा की जाँच करनी चाहिए। |
| जठरशोथ (गैस्ट्राइटिस) / गैस्ट्रोएसोफिज़ियाल रिफ्लेक्स/ पेप्टिक अल्सर की बीमारी (पेट की परेशानी) | पेट में दर्द, पेट में जलन (जो रात में बढ़ जाती है ), भोजन या खट्टा तरल की मुँह में वापस आना, छाती में दर्द, गले में एक गांठ का एहसास होना। | इन लक्षण के होने पर ड्रग्स और एंटासिड का इस्तेमाल किया जा सकता है। |
| मोतियाबिंद | स्टेरॉयड मोतियाबिंद की शुरुवात का कारण बन सकता है। डिफ्लैजाकॉर्ट के साथ मोतियाबिंद का ख़तरा अधिक होता है। | मोतियाबिंद और नेत्र संबंधी दबाव की साल में एक बार आवश्यक है। नेत्र रोग विशेषज्ञ द्वारा इनका इलाज़ कराया जाना चाहिए। |
| हड्डियों में कमजोरी और फ्रैक्चर का ख़तरा बढ़ना। | प्रत्येक नियमित जाँच के दुराण किसी भी फ्रैक्चर और कमर में दर्द को रिपोर्ट करना चाहिए। | खून की जाँच, एक्स-रे और "डेक्सा स्कैन " के ज़रिये हड्डियों के स्वास्थ्य के बारे ममें पता लगाया जा सकता है।<br>रक्त में विटामिन डी के स्तर के आधार पर इसको दवा के तौर पर दिया जा सकती है, इस बात का ध्यान रखें कि आपकेआहार में कैल्शियम की मात्रा पर्याप्त हो। |
| रोग प्रतिरक्षा एवं एड्रिनल दमन | गंभीर संक्रमण के जोखिम से अवगत रहें और मामूली संक्रमण का तुरंत इलाज़ करवाएं। | छोटी माता/छोटी चेचक (वेरीसेल्ला) का टीका अवश्य लगवाएं। स्टेरॉयड लेने से पहले टी बी के लिए स्क्रीनिंग की जाती है, सभी लोग जो आपके पचे का इलाज़ कर रहे हैं उनको सूचित करें कि बच्चा स्टेरॉयड पर है। यह विशेष रूप से महत्वपूर्ण है कि लम्बे समय से स्टेरॉयड ले रहा व्यक्ति, किसी भी हल में 24 घंटे से अधिक समय तक अपनी दवाई न भूले। खासकर यदि वे बीमार या अस्वस्थ हैं। संक्रमण और सर्जरी की स्थिति में स्टेरॉयड की ख़ुराक़ की अधिक आवश्यकता हो सकती है। |
| मायोग्लोबिनुरिया (रबडोमायोलिसिस: मांसपेशियों में क्षति) | मूत्र लाल-भूरे रंग का दिखता है, क्योंकि इसमें मांसपेशियों के टूटे हुए प्रोटीन का अंश होता है। मूत्र के रंग में देखे गए इस बदलाव की रिपोर्ट विशेषज्ञ को देनी चाहिए। | ज़रुरत से ज़्यादा शारीरिक श्रम से बचें। तरल पदार्थ का सेवन खूब करें, इन लक्षणों के बने रहने पर गुर्दे की कार्य क्षमता की जाँच करवानी पड़ सकती है। |

**ग. हार्मोनल इलाज़**

डीएम् डी के उपचार के दौरान लिए गए स्टेरॉयड से कई हार्मोन पर नकारात्मक प्रभाव हो सकता हैं। जैसे, ग्रोथ हार्मोन (बच्चे का उचित तौर से बढ़ने में सहायक) और टेस्टोस्टेरोन (विलंबित यौवन/किशोरवस्था) बच्चे की आयु के अनुसार कद के न बढ़ने और किशोरावस्था के आने में देरी हो सकती है। इन परिस्थितियों में बाल चिकित्सक एंडोक्रिनोलॉजिस्ट (हॉर्मोन डॉक्टर) से परामर्श लेना ज़रूरी हो जाता है।

***छोटा कद और यौवन/ किशोरावस्था का देरी से आना***

स्टेरॉयड के इस्तेमाल से आमतौर पर ग्रोथ हार्मोन प्रभावित होते हैं। बच्चे का कद छोटा रह जाता है। छोटा कद और किशोरावस्था का देरी से आना, माता पिता के लिए चिंता का विषय हो सकता है। छोटा कद किसी अन्य चिकित्सीय समस्या का संकेत हो सकता है और टेस्टोस्टेरोन की कमी से हड्डियों की सेहत बिगड़ सकती है। इसलिए बच्चे की लम्बाई और किशोरावस्था पर लगातार नज़र रखना ज़रूरी है, एक बार बच्चा 9 साल की उम्र पार कर जाए।

किशोरावस्था पूर्ण रूप से आने तक, जब तक बच्चा पूरी लम्बाई प्राप्त न करले हर 6 महीने में लम्बाई/कद का आँकलन करके विकास की निगरानी की जानी चाहिए। यदि लम्बाई बढ़ने की गति 4 cm प्रति वर्ष से कम है तो आप अपने हॉर्मोन डॉक्टर या स्पेशलिस्ट से मिलें। यदि वृषण मात्रा 14 वर्ष की आयु में <4 सेमी$^3$ से कम है तो यह किशोरावस्था में आई देरी का संकेत है।

***ग्रोथ हार्मोनल थेरेपी***

अगर डॉक्टर को ग्रोथ हार्मोन में कमी के संकेत मिलते हैं तो बच्चे को उनकी निगरानी में हार्मोन दिए जा सकते हैं। ड्यूशेन से प्रभावित बच्चों में ग्रोथ हार्मोन के इस्तेमाल के बारे में कोई स्पष्ट जानकारी नहीं है।

***टेस्टोस्टेरोन थेरेपी***

टेस्टोस्टेरोन हार्मोन हड्डी के स्वास्थ्य,साथ ही साथ भावनात्मक विकास के लिए भी महत्वपूर्ण है। बाल चिकित्सक ही हार्मोन थेरेपी शुरू करने का निर्णय ले सकते हैं। हॉर्मोन डॉक्टर की निगरानी में कम खुराक पर उपचार शुरू किया जाता है। टेस्टोस्टेरोन हॉर्मोन कई रूपों में आता है, जैसे इंट्रामस्क्युलर इंजेक्शन, जैल और पैच। इसके सभी दुष्प्रभावों की चर्चा इस्तेमाल से पहले से आप से की जाती है।

***एड्रिनल ग्रंथि का संकट क्या है?***

एड्रिनल ग्रंथियां, गुर्दे के ऊपर स्थित होती हैं और कोर्टिसोल नामक एक हार्मोन का स्राव करती हैं। यह कोर्टिसोल शरीर को गंभीर बीमारी या चोट से निपटने में मदद करता है।

जब कोई व्यक्ति लंबे समय से स्टेरॉयड ले रहा होता है तो एड्रिनल ग्रंथियां कोर्टिसोल का स्राव करना बंद कर देती हैं, यह एड्रिनल ग्रंथि का दमन कहलाता है। एक बार स्टेरॉयड बंद करने के बाद, शरीर फिर से अपना कोर्टिसोल बनाने में हफ्ते या महीने लग जाते हैं। कोर्टिसोल के बिना शरीर किसी भी विषम परिस्थिति से निपटने में असमर्थ हो जाता है, जिसके परिणामस्वरूप एड्रिनल संकट उत्पन्न हो जाता है। इससे जान को खतरा होता है। इसलिए, स्टेरॉयड को अचानक बंद नहीं किया जाना चाहिए। मरीजों (लम्बे समय से स्टेरॉयड पर हैं ) के लिए एक योजना होनी चाहिए जो ये निर्धारित करे कि अगर बीमारी या अन्य कारणों से 24 घंटे से अधिक समय तक खुराक नहीं ले पाते हैं, जो इस छूटी हुई खुराक को कैसे लेना है ? और किस मात्रा में लेना है? किसी विकट बीमारी के समय या भयानक चोट लगने पर, सर्जरी होने पर की कितनी ख़ुराक स्टेरॉयड का इस्तेमाल करनी है? कब स्टेरॉयड की अतिरिक्त खुराक, या "स्ट्रेस डोज़," जरूरत हो सकती है।

एड्रिनल संकट के सामान्य लक्षण हैं : अत्यधिक थकान, सिरदर्द, मतली / उल्टी, रक्त में ग्लूकोस कि कमी एवं रक्तचाप में कमी और बेहोशी आ जाना।

**घ. फिजियोथेरेपी**

ड्यूशेन में फिजियोथेरेपी विभिन्न कसरतों के माध्यम से मरीज़ की लम्बे समय तक अधिकतम शारीरिक क्षमता को बनाए रखने में मदद करता है।

ड्यूशेन में फिजियोथेरेपिस्ट निम्नलिखित तरीकों से मदद कर सकते हैं :

- एक सुनियोजित ढंग से कसरतों के माध्यम से मांसपेशियों में खिंचाव करना ताकि जोड़ों में संकुचन और विकृति न आ जाए।
- शारीरिक जटिलताओं का जल्द से जल्द पहचान कर और कसरत के जरिये उनको कम से कम करना।
- सहायक उपकरणों को उनकी जरूत के हिसाब से उन्हें निर्धारित करना और इस्तेमाल करने की सलाह देना। (ऑर्थोसिस, कैलिपरस, व्हीलचेयर्स और स्टैंडिंग फ्रेम्स)
- अधिक से अधिक समय तक चलना फिरना बरक़रार रखने और उसमे आनेवाली दिक्कतों को सुलझाने की सलाह देते हैं।
- श्वसन कार्यक्षमता की लगातार निगरानी रखने के लिए, सांस लेने और स्राव को साफ करने के तरीकों पर तकनीकी सलाह देना।
- आपके फिजियोथेरेपिस्ट द्वारा सुझाई गई कसरतों को प्रभावी बनाए रखने के लिए उनका नियमित रूप से किया जाना आवश्यकता है।
- ये कसरतें आपको घर पर एक दिनचर्या से बांधतीं हैं। फिजियोथेरेपिस्ट के साथ मिलकर आप अपने बच्चे के लिए एक सही दिनचर्या निर्धारित करना महत्वपूर्ण है। जहां तक भी संभव इस दिनचर्या को इस प्रकार बनाएँ जो आपके परिवार की अन्य गतिविधियों में फिट बैठता हो। उदाहरण के लिए, यदि सुबह व्यस्त रहती हैं तो शाम के समय थेरेपी सेशन करना सबसे अच्छा हो सकता है, थेरेपी सेशन के लिए महत्वपूर्ण है कि वह नियमित दिनचर्या का हिस्सा हो।

  हालांकि कसरतें कभी भी पीड़ादायक नहीं होनी चाहिए, परन्तु खिचाव उत्पन्न करेने वाली कसरतें (स्ट्रेचिंग एक्सरसाइज) से मांसपेशी में एक अलग तरह का अनुभव हो सकता है। आपके बच्चे को इसके लिए आदी होना आवश्यक है। कुछ बच्चे स्वयं ही स्ट्रेचिंग का अभ्यास कर लेते हैं। कसरतें बहुत थकाने वाली नहीं होनी चाहिए और वजन अधिक होने पर और जरुरत से ज़्यादा व्यायाम करना उचित नहीं है। कोई भी गतिविधि जो एक बच्चा स्वेच्छा से और बिना थके हुए करता है, उसको प्रोत्साहित किया जाना चाहिए इससे एक सकारात्मक प्रभाव पड़ता है।

माता-पिता या बच्चे की देखभाल करने वाले व्यक्ति कसरतों का एक नियोजित कार्यक्रम शुरू कर सकते हैं जिसमें निम्नलिखित बातों का ध्यान दिया जाए:

- नियमित स्ट्रेचेस- स्वयं और / या मैनुअल स्ट्रेच के साथ-साथ दूसरे की सहायता से की गई स्ट्रेचिंग - ज्यादातर मांसपेशियों के समूह को के लिए कसने के लिए की जाती हैं। उदाहरण के लिए एड़ी का ऊपर-पीछे का हिस्सा (टेंडो एकलिस), जांघ की पिछले हिस्से की मासपेशियां (हंस्ट्रिंग्स)।
- तैराकी
- सायक्लिंग
- रात को सोते समय टखनों की संकुचन को धीमा करने के लिए ऑर्थोस (स्प्लिन्ट्स) पहनना।
- फ्लैट पैरों के लिए साधारण हल्के जूते में इनसोल का इस्तेमाल मददगार होते हैं।
- सामान्य खेल और फिटनेस, हृदय की कार्य क्षमता को लम्बे समय तक बनाए रखती हैं।

बीमारी के बाद के विकास के चरण में एक फिजियोथेरेपी कार्यक्रम शामिल कर सकता है :

- दूसरे की सहायता से  टेंडो एकलिस और जांघ की पिछले हिस्से की मांसपेशियों एवं कूल्हे की मांसपेशियों  की स्ट्रेचिंग कराइ जा सकती है।  साथ ही साथ कुछ स्वयं की जा सकने वाली स्ट्रेचिंग कसरतें।
- हाथो की मांसपेशियों के खिंचाव की कसरतें (स्ट्रेचिंग)।
- तैराकी
- रात को सोते समय टखनों की संकुचन को धीमा करने के लिए ऑर्थोस (स्प्लिन्ट्स) पहनना।
- पोजीशनिंग : मरीज के शरीर को ऐसी अवस्था में रखना जिसमें अधिकतम स्वास्थ्य लाभ हो सके।  यह पोजीशनिंग मांसपेशियों की मजबूती को बनाए रखती है साथ ही साथ संकुचन और मांसपेशियोँ में हो रहे दर्द को भी कम करती है।
- खेल खेल में बुलबुले उड़ाने और हवा से चलने वाले उपकरणों को बढ़ावा देना चाहिए।  ये कसरतें श्वास लेने क्षमता को बढ़ाते हैं।

**रात को पैरों में पहनने वाले बाहरी समर्थन (नाइट स्पलिंट):**

जैसा की नाम से प्रतीत होता है ये रात में पहनाए जाने के लिए डिज़ाइन किए गए हैं और आमतौर पर केवल एड़ियों के लिए होते हैं। स्प्लिंट बच्चे के जोड़ों को उचित स्थिति में रखकर संकुचन को धीमा करने में मदद करते है।  पॉलीप्रोपाइलीन सहित विभिन्न सामग्रियों से यह बनाया जाता है। यह पैर की उंगलियों से शुरू होकर, घुटनों के ठीक नीचे तक होता है।  वे आरामदायक और ठीक से फिट होना चाहिए, क्योंकि खराब फिटिंग वाले स्प्लिंट्स पहनने में बच्चे को परेशानी होती है और वह यह पहनना बंद कर देता है  स्प्लिन्ट्स का उपयोग करने से रोक सकते हैं।

रात को पैरों में पेहेन्ने वाले बाहरी समर्थन (नाइट स्पलिंट)

**मांसपेशियों की मजबूती बनाए रखने की जा सकने वाली कसरतों के कुछ उदहारण :**

**स्वयं की जा सकने वाली स्ट्रेचिंग कसरतें**

***अपने आप की जा सकने वाली कसरतें***

फ़र्श पर सीधे बैठते हुए, घुटने और टखने सीधे रखकर हाथ की उँगलियों से पैरों के पंजों को छूना

यह आपके जांघ की पिछली मांसपेशियों में खिंचाव पैदा करता है।

**पैरों के पंजे को चित्र में दिखाए गए स्टेप जैसी सीढ़ी पर रख कर एड़ी को फ़र्श की तरफ ले जाएं।**

**सीढ़ी की ऊंचाई लगभग ६ इंच हो और यह सुनिश्चित करें की सीढ़ी पूरी तरह से स्थिर हो।**

**यह आपकी पिंडलियों की मांसपेशियों में खिंचाव पैदा करता है।**

**पिंडलियों की मांसपेशियों में इस प्रकार से खिंचाव कपड़े या बैंड पैदा करता है।**

**फ़र्श या मेट पर कमर सीधी कर के घुटने और टखने सीधे रखकर बैठ जाइये।**

अब दोनों टांगों को बहार की तरफ़ जतना हो सके फैला लीजिये।

ऐसे ही थोड़ी देर ऐसे ही रुकिए (३० सेकंड), फिर से यह प्रक्रिया दोहराइए।

यह कसरत आपके जांघ की पिछली मांसपेशियों में खिंचाव पैदा करती है।

**दीवार की तरफ़ मुँह करके खड़े हो जाइये**

**दोनों बाहें कंधे के स्तर पर एकदम सीधे रखते हुए, दीवार को धक्का दें।**

**धक्का देते समय कन्धा, कोहनी और कलाई एकदम सीधी और बिना मुड़ी होनी चाहिए।**

**ऐसे ही थोड़ी देर ऐसे ही रुकिए (15 सेकंड), फिर से यह प्रक्रिया दोहराइए।**

**यह कसरत आपके कंधे की मांसपेशियों में खिंचाव और मज़बूती भी पैदा करती है।**

**दीवार की तरफ़ मुँह करके खड़े हो जाइये, दोनों बाँहों से दीवार पर रखते हुए तिरछे हो जाइये**

एक पैर को घुटने मोड़ें एड़ी को दिवार के एकदम पास रखते हुए पैर के पंजे को दिवार पर रखें।

दूसरा पैर एकदम सीधा रखें, अब दीवार को धक्का दें।

ऐसे ही थोड़ी देर ऐसे ही रुकिए (३० सेकंड), फिर से यह प्रक्रिया दोहराइए।

यह कसरत आपके पैरों की मांसपेशियों में खिंचाव और मज़बूती भी पैदा करती है।

**बाँह को कंधे के स्तर पर लेजाकर सीधा रखें। हथेली को कलाई पर नीचे की तरफ मोड़ के पीछे की तरफ धक्का दें।**

**इस प्रकार धक्का आप स्वयं के हाथ, बैंड अथवा कपड़े की सहायता दे सकते हैं।**

**यह कसरत आपकी कोहनी से कलाई के बीच की मांसपेशियों में खिंचाव पैदा करता है।**

## किसी की मदद लेकर की जासकने वाली कसरतें

**फ़र्श या मेट पर सीधे लेटे जाइये,**

**परिवार के किसी सदस्य की सहायता से एड़ी को सहारा देते हुए पैर के पंजे को टाँग की तरफ लेके के जाइये।**

**यह कसरत आपकी टख़ने मांसपेशियों में खिंचाव पैदा करता है।**

फ़र्श या मेट पर सीधे लेटे जाइये, परिवार के किसी सदस्य की सहायता से एड़ी को सहारा देते हुए टाँग को सीधा रखते हुए ऊपर की तरफ ले जाइये।

ऐसे ही थोड़ी देर ऐसे ही रुकिए (३० सेक्), फिर से यह प्रक्रिया दोहराइए।

यह कसरत आपके जांघ की पिछली मांसपेशियों में खिंचाव पैदा करती है।

## किसी की मदद लेकर कूल्हे की खिंचाव की जा सकने वाली कसरतें

फ़र्श या मेट पर पीठ के बल सीधे लेटे जाइये, परिवार के किसी सदस्य की सहायता से एक तरफ़ के कंधे को ज़मीन की तरफ़ दबाव दें।

परिजन के दूसरे हाथ से उसी तरफ़ की टाँग को हल्का सा कूल्हे और घुटनो को मोड़ते हुए उलटी तरफ़ को कूल्हे पर मोड़ते हुए धकेलें।

ऐसे ही थोड़ी देर ऐसे ही रुकिए (15 सेक्), फिर से यह प्रक्रिया दोहराइए।

यह कसरत पैर और कूल्हे की मांसपेशियों को में खिंचाव करती है।

इलूटीबीएल बैंड की स्ट्रेचिंग

फ़र्श या मेट पर पेट के बल उलटे लेट जाइये, परिवार के किसी सदस्य की सहायता से नीचे कमर पर एक हाथ से सहारा देते हुए,

परिजन के दूसरे हाथ से उसी एक टाँग को को घुटने पर मोड़ते हुए कूल्हे पर ऊपर की तरफ़ खींचें।

थोड़ी देर ऐसे ही रुकिए (15 सेक्), फिर से यह प्रक्रिया दोहराइए।

यह कसरत पैर की मांसपेशियों में खिंचाव पैदा करती है।

फ़र्श या मेट पर पेट के बल उलटे लेट जाइये, परिवार के किसी सदस्य की सहायता से नीचे कमर पर एक हाथ से सहारा देते हुए,

परिजन के दूसरे हाथ से उसी एक टाँग को को घुटने को सीधे रखते हुए कूल्हे की धुरी पर बाहर की तरफ़ खींचें।

थोड़ी देर ऐसे ही रुकिए (15 सेकंड), फिर से यह प्रक्रिया दोहराइए।

यह कसरत पैर की मांसपेशियों में खिंचाव पैदा करती है।

**मांसपेशियों मज़बूती प्रदान करने वाली कसरतें**

हमारे शरीर की मांसपेशियों के उचित विकास के लिए मांसपेशियों को मज़बूती प्रदान करने वाली कसरतें कराइ जाती हैं। इसके अंतर्गत मांसपेशियों को बाहर से लगाए गए बल के विपरीत चलाया जाता है।

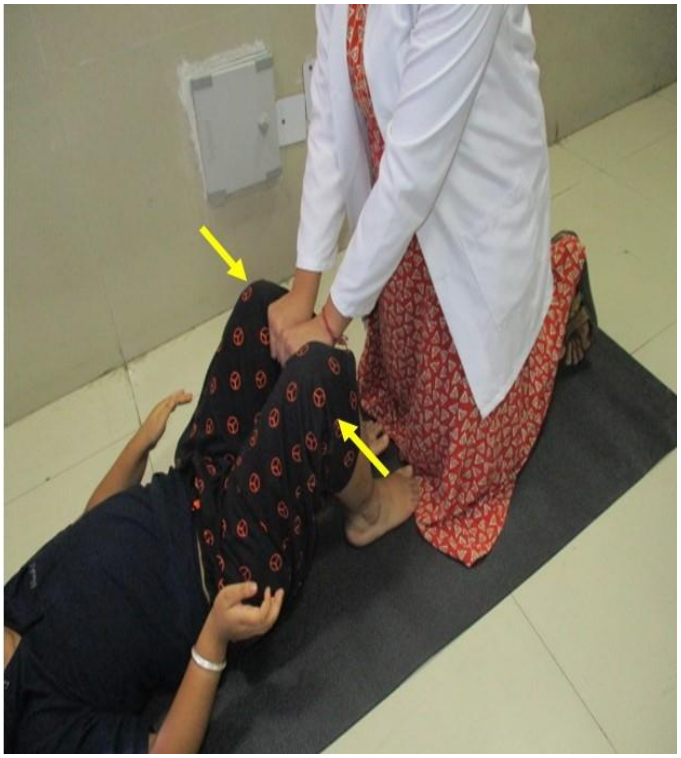
फ़र्श या मेट पर सीधे लेटे जाइये

पैरों को घुटने पर मोड़ लें, परिवार के किसी सदस्य को दोनों घुटनों के बीच में हल्क़े से दूरी बनाने के लिए कहें

अब अपने घुटनों को आपस में मिलाने की कोशिश करें।

ऐसे लगभग ३० सेकेंड ज़ोर लगाएं, फिर से यह प्रक्रिया दोहराइए।

यह कसरत आपके जांघ की मांसपेशियों को मज़बूती प्रदान करती है।



फ़र्श या मेट पर सीधे लेटे जाइये

पैरों को घुटने पर मोड़ लें और आपस में मिला के रखें,परिवार के किसी सदस्य को दोनों घुटनो के बाहरी हिस्से से अंदर की तरफ़ हल्क़े से ज़ोर लगाने के लिए कहें

अब अपने घुटनों को एक दूसरे से दूर करने की कोशिश करें।

ऐसे लगभग ३० सेकेंड ज़ोर लगाएं, फिर से यह प्रक्रिया दोहराइए।

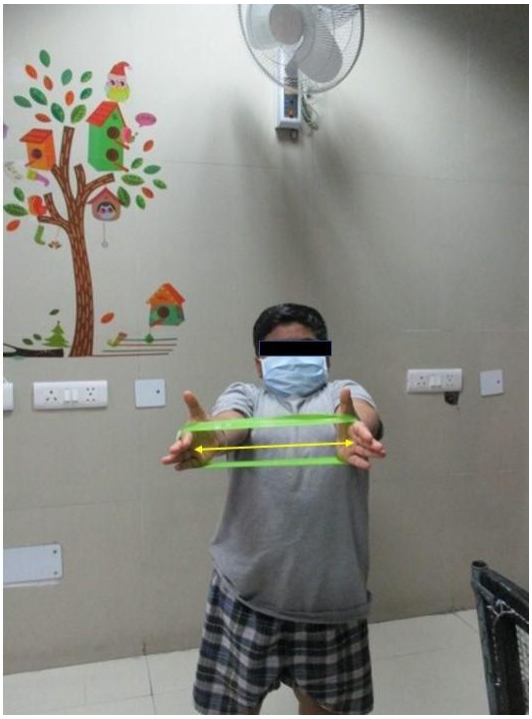
यह कसरत आपके जांघ की मांसपेशियों को मज़बूती प्रदान करती है।

फ़र्श पर सीधे खड़े हो जाइये

बाँह को कंधे के स्तर पर लेजाकर सीधा रखें। जैसा कि चित्र में दिखाया गया है, हथेलियों को विपरीत दिशा में ले जाते हुए थेरा बैंड को अधिक से अधिक खींचने की कोशिश करें।

ऐसे लगभग ३० सेकेंड ज़ोर लगाएं, फिर से यह प्रक्रिया दोहराइए।

यह कसरत आपके बाजुओं की मांसपेशियों को मज़बूती प्रदान करती है।



## सांस की मांसपेशियों को मज़बूती प्रदान करने वाली कसरत

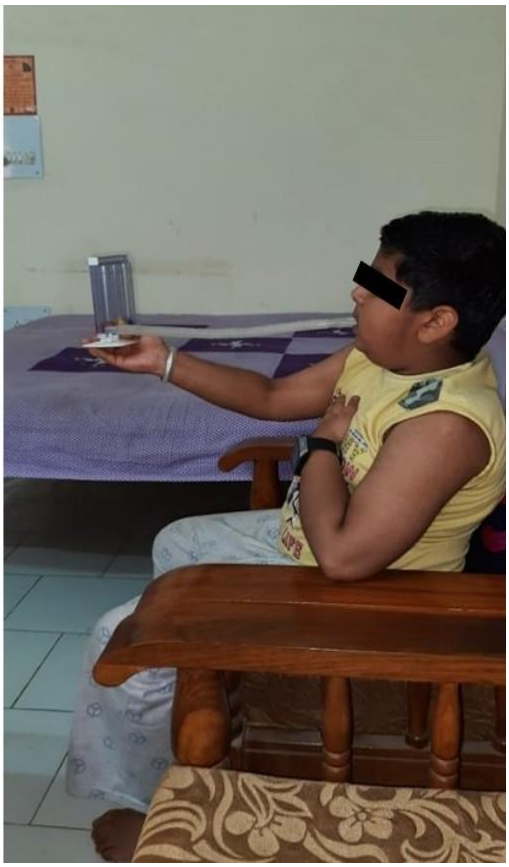
बच्चे को क़मर पर सहारा देते हुए  सीधा चेयर पर बैठाएँ,

फिर एक हाथ में स्पाइरोपमीटर को पकड़ें, हाथ सीधा होना चाहिए और स्पाइरोपमीटर आँखों के स्तर पर। दूसरे हाथ से पाइप मुँह में रखिये।

अब साँस अंदर की तरफ़ खीचें, साँस अंदर खेंचते ही स्पाइरोपमीटर की गेंदे ऊपर की तरफ़ जाएँगी

गेंदों को तीन सेक् तक ऊपर ही रोकने की कोशिश करें, इस कसरत को दस बार करें और दिन में ३-४ बार करें।

यह कसरत श्वास की मांसपेशियों को मज़बूती प्रदान करती हैं।



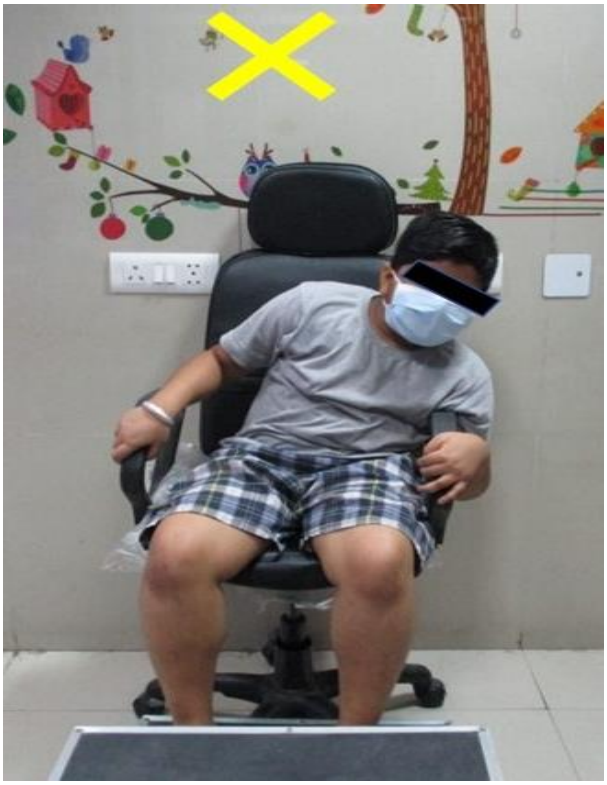
**पोस्चर**

एक विशेष स्थिति में हमारे शरीर के विभिन्न अंग परस्पर रूप से जुड़े होते हैं। इस को हम आसान या पोस्चर कहते हैं।

**बैठने का ढंग (पोस्चर)**

साँस लेने के दौरान पेफड़ों का बचाव और हड्डियों को विकृत टेढ़ा-मेढ़ा होने से रोकने के लिए बैठने के ढंग (पोस्चर) पर ध्यान देना बहुत महत्वपूर्ण है।

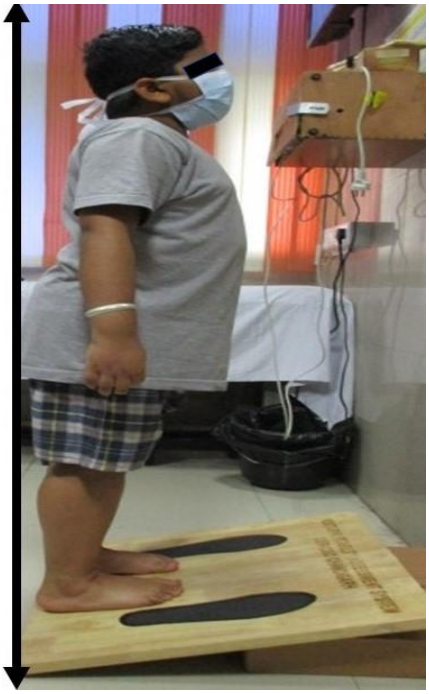
बैठते समय पांव और पैरों के बीच 90 डिग्री का कोण होना चाहिए। कुर्सी पर बैठने की जगह सख़त होनी चाहिए, आदर्श रूप से बहुत चौड़ा नहीं होना चाहिए। कुर्सी का टेक लेने वाला हिस्सा दृढ़ होना चाहिए। कुर्सी का यह हिस्सा या तो सीधा खड़ा या 10 डिग्री पर पीछे की तरफ झुका हुआ होना चाहिए। बच्चों को कुर्सी के पीछे का उपयोग करने के लिए प्रोत्साहित करना चाहिए, जिससे उनकी कमर को सहारा मिल सके। हाथों के लिए आर्मरेस्ट को उचित ऊंचाई और कमर से बहुत दूर नहीं होना चाहिए ताकि कोहनियों को आसानी से सहारा मिल सके। इस सहारे से कंधों में झुकाव या कुब्बड़ नहीं निकलेगा।



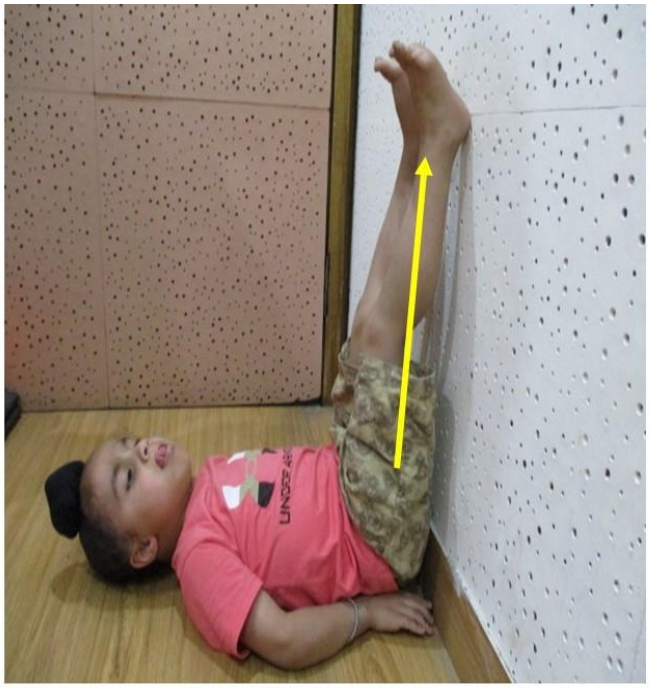
**पच्चर के आकार के लकड़ी पर बैठे और खड़े रहना।**

**लकड़ी के पच्चर पर बच्चों को सुबह और शाम आधे - आधे घण्टे इस तरीके से दीवार के सहारे खड़ा होना चाहिए।**

**ताकि पैरों की (पिंडलियों) मांसपेशियों में खिंचाव बना रहे।**



फ़र्श या बिस्तर पर लेट कर पैरों को दीवार के सहारे से खड़े कर के लेट जाएं, जैसा कि इनके दिए गए चित्र में दिखाया गया है। यह आराम करने का एक तरीका हो सकता है। इस प्रकार से लेटते हुए बच्चा किताब पढ़ सकता है या मोबाइल का इस्तेमाल कर सकता है। इस प्रकार से लेटना जांघों और टखनों की मांसपेशियों के संकुचन को धीमा करता है।



**फ़र्श या बिस्तर पर लेट कर पैरों को दीवार के सहारे  से खड़े कर लेना**

**उन्मुख लेटना (पेट के बल लेटना )**

पेट के बल लेटना जहाँ चेहरा नीचे की ओर होता है आराम करने के लिए अच्छा है। यह कूल्हों और घुटनों में विकसित होने वाले संकुचन को रोकने में मदद करता है।  पढ़ने या टेलीविजन देखते समय पेट के बल लेटा जा सकता है।

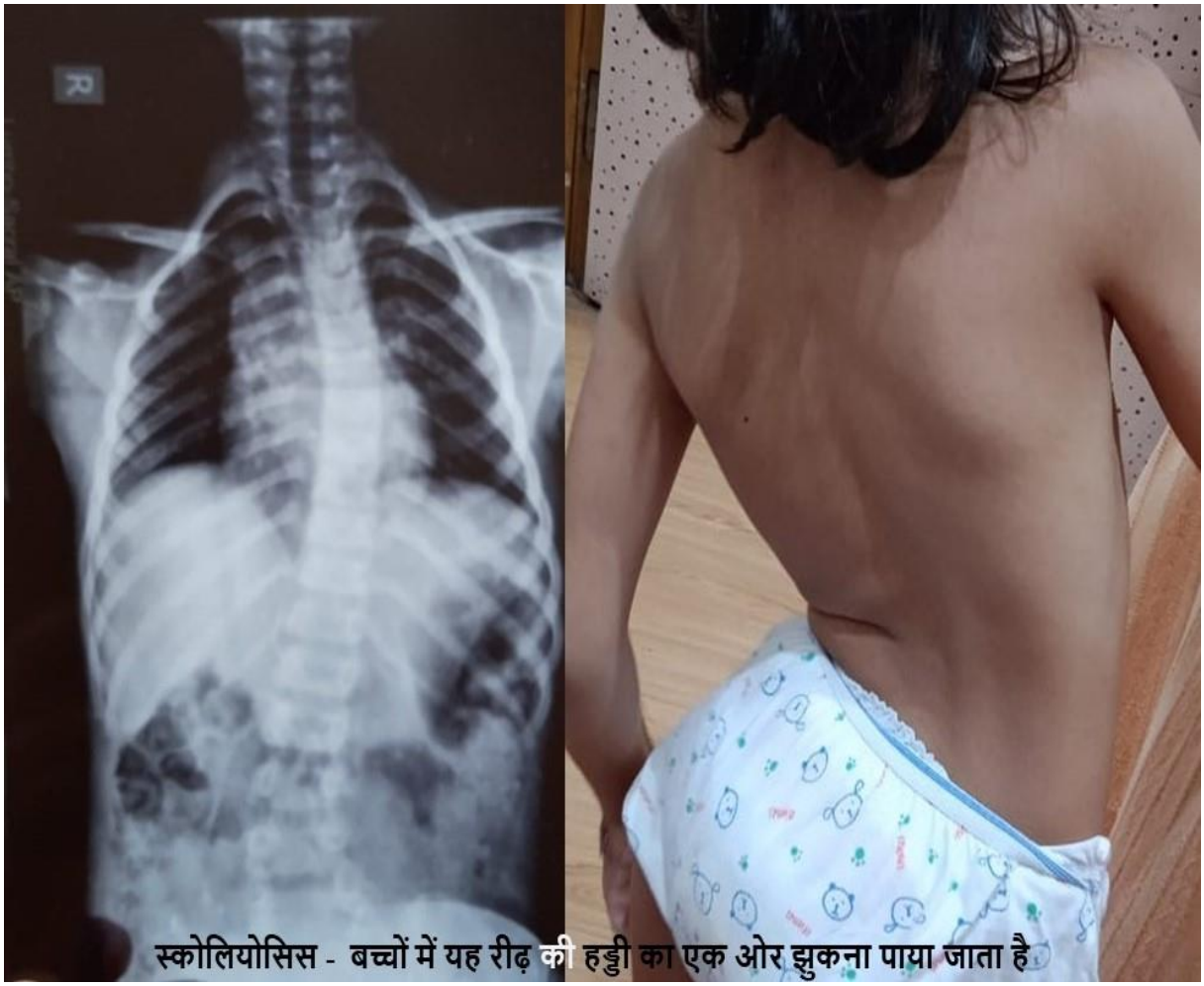
जब बच्चा फर्श या गद्दे पर उल्टा लेता हो तो एक छोटा तकिया कूल्हों के ठीक नीचे रख दें इससे कूल्हों के जोड़ों का प्रसार (एक्सटेंशन) प्रदान करता है।  इस प्रकार से लेटना कमर और जांघों की मांसपेशियों के संकुचन को धीमा करता है।

- बीमारी के शुरुआती दौर में ही स्ट्रेचिंग कसरतें शुरू कर देनी चाहिए।
- स्ट्रेचिंग कसरतें बीमारी पर अधिक प्रभावी ढंग से काम करतीं हैं अगर इनको मांसपेशियों में जकड़न या संकुचन स्थापित होने से पहले ही शुरू कर दिया जाए। बाद में स्ट्रेचिंग करने में बच्चे को दर्द होता है।
- सभी स्ट्रेचिंग कसरतों को को धीरे धीरे और बिना दर्द के किया जाना चाहिए।
- स्ट्रेच को 15से 30 सेकंड तक बनाए रखना चाहिए।
- दिन में 3 से 4 बार और एक बार में 10 से 15 या अधिक बार ये कसरतें करनी चाहिए।
- जब भी संभव हो, बच्चे को ही स्ट्रेचिंग कसरतें करने की ज़िम्मेदारी देनी चाहिए, यह उसकी दिनचर्या का एक हिस्सा होना चाहिए।

**ङ हड्डियों की देखभाल**

ड्यूशेन सीधे-सीधे हड्डियों के स्वास्थ्य को प्रभावित करता है। इस बीमारी मैं हड्डियाँ कमज़ोर हो जाती हैं, पीठ दर्द होता है, हड्डियों में खनिज के घनत्व में कमी आ जाती है जिससे बारंबार फ्रैक्चर होने की संभावनाएं बढ़ जाती है। मांपेशियों और हड्डियों में कमज़ोरी की वजह से रीढ़ की हड्डी एक ओर झुक जाती है जिसे स्कोलियोसिस भी कहते हैं। यह झुकाव समय के साथ बढ़ता जाताहै और देरतक सीधे बैठने में दिक्कत महसूस होती है। कमजोर मांसपेशियों, कम शारीरिक गतिविधियों, स्टेरॉयड थेरेपी और मांसपेशियों के असंतुलन के कारण हड्डियाँ कमजोर हो जाती हैं। हलके से गिरने पर भी हड्डी में फ्रैक्चर हो जाता है और रोगी का स्वतन्त्र रूप से चलना फिरना काफ़ी सीमित हो जाता है।

- डीएमडी से प्रभावित लड़के - कम आघात लगने पर भी भयानक अस्थि-भंग (फ्रैक्चर) के शिकार हो जाते हैं।
- डीएमडी से प्रभावित लड़के - कशेरुक फ्रैक्चर (बिना किसी लक्षण के).
- स्कोलियोसिस - 90% बच्चों में यह रीढ़ की हड्डी का एक ओर झुकना पाया जाता है।



स्कोलियोसिस - बच्चों में यह रीढ़ की हड्डी का एक ओर झुकना पाया जाता है

***हड्डी की स्वस्थ की निगरानी:***

- रीढ़, रीढ़ की हड्डी की वक्रता (झुकाव) और रीढ़ की हड्डी के आसपास छूने में दर्द के लिए, समय- समय पर नियमित जाँच आवश्यक है।
- एक स्पाइनल (रीढ़ की हड्डी) एक्स-रे बीमारी का पता लगने पर और एक स्पाइनल एक्स-रे जब बच्चा व्हील चेयर पर आश्रित हो जाता है, उस समय बहुत आवश्यक है। यह रीढ़ की हड्डी में विकृति को पकड़ने में सहायक सिद्ध होता है।
- विकृति के होने पर हर एक साल में एक्स-रे अवश्य करवाएं।
- शरीर में कैल्शियम, फास्फोरस, एएलपी, विटामिन डी की सही स्थिति जानने के लिए रक्त परीक्षण करवाना आवश्यक है।
- हड्डियों में खनिजों के घनत्व (बोन डैंसिटी) को डेक्सा स्कैन के माध्यम से साल में एक बार माप लेना चाहिए।
- दोहरी ऊर्जा एक्स-रे अवशोषण (डेक्सा स्कैन ) एक गैर इनवेसिव (किसी प्रकार का दर्द नहीं होता है) टेस्ट जो लंबी हड्डियों (आमतौर पर पैर या बांह) के अस्थि खनिज घनत्व को मापता है।

***स्कोलियोसिस की रोकथाम***

- हर समय आसन (ढंग से बैठने) का ध्यान रखें।
- जो लड़के अभी भी चल रहे हैं, उनमे दोनों पैर की असमान सिकुड़न को रोकने की ज़रुरत भी होती है।
- व्हीलचेयर में उचित आसन में बैठने की व्यवस्था होनी चाहिए जिस में रीढ़ और श्रोणि में समरूपता हो और रीढ़ की हड्डी के विस्तार का समर्थन करती हो।

- स्पाइनल ब्रेसिंग को रीढ़ की हड्डी को सही स्थिति में रखने के लिए उपयोग में लाया जाता, परन्तु इसे सर्जरी की जगह या फिर सर्जरी को टालने के लिए नहीं करना चाहिए।

स्पाइनल ब्रेसिंग

***स्कोलियोसिस का इलाज***

- जिन बच्चो में रीढ़ की हड्डी का टेढ़ापन (कोब कोण के रूप में जाना जाता है) २० से अधिक होता और वो अभी भी चल फिर रहे होते हैं, उनमें स्पाइनल सर्जरी की सलाह दी जाती है।

***हड्डी के फ्रैक्चर का इलाज़***

- हड्डी का फ्रैक्चर विशेष रूप से निचले अंग की हड्डी के फ्रैक्चर, काम करने की क्षमता को जारी रखने में प्रमुख बाधा है।
- इसलिए, सर्जरी के साथ उपचार पर विचार किया जाना चाहिए ताकि लड़के को जल्द से जल्द अपने पैरों पर वापस चल सकें।
- जो बच्चा अभी भी चलने फिरने में सक्षम है, उसके पैर टूटने (फ्रैक्चर) पर जितनी जल्दी हो सके इंटरनल फिक्सेशन कर पैर स्थिरता प्रदान करना ज़रूरी हो जाता है। ताकि बच्चा जल्दी से जल्दी चलना फिरना शुरू कर सके।
- जो बच्चा अभी चलने फिरने में सक्षम नहीं है, उसके पैर टूटने या किसी भी फ्रैक्चर को स्प्लिंटिंग या कास्टिंग के द्वारा ठीक किया जा सकता है। स्प्लिंटिंग या कास्टिंग के समय हाथ पैरों की गतिशीलता बने रहे इसका ध्यान देना चाहिए।

***कमजोर हड्डियों का इलाज़***

- नियमित शारीरिक गतिविधि बनाए रखना
- विटामिन डी की कमी हो जाती है, तो बच्चों को बहर से सुप्लिमेंट्स/ दवाईओं के तौर पर विटामिन डी की आवश्यकता पड़ती है।
- आहार में कैल्शियम की मात्रा अधिक होनी चाहिए। यदि आहार से जरुरी कैल्शियम पर्याप्त मात्रा में नहीं मिल पा रहा है तो आहार विशेषज्ञ से सलाह लेनी चाहिए। इसके अतिरिक्त बहर से कैल्शियम सुप्लिमेंट्स/ दवाइयाँ लेने चाहिए।
- यदि कोई फ्रैक्चर है, तो तुरंत अपने चिकित्सक को रिपोर्ट करें।

***सुरक्षा और गिरने से बचने के उपाय:***

- व्हीलचेयर के सुरक्षित इस्तेमाल पर मरीजों और परिवार के परिजनों को प्रशिक्षित करना महत्वपूर्ण है। व्हीलचेयर से गिरना चोट लगने का प्रमुख कारण है।
- व्हीलचेयर पर बैठते समय सीटबेल्ट का उपयोग हर समय किया जाना चाहिए।

- घर पर व्हीलचेयर के सहज उपयोग के लिए कुछ सुरक्षा के उपाय किए जाने आवश्यक होते हैं, जैसे : फ़र्श को साफ़ रखें, फ़र्श पर दरियाँ, खिलौने एवं रस्सियाँ आदि बिखरे न पड़े रहें।
- घर से बाहर जाते समय असमान सतहों पर विशेष सावधानी बरतें।
- फिसलन वाली सतहों पर चलने पर गिरने से बचाव के लिए बच्चे को बिना फिसलन वाला जूता पहनना चाहिए।
- व्हीलचेयर से बिस्तर, कार की सीट या किसी अन्य स्थानांतरण को बहुत सावधानी से करें।

***घर में सुरक्षा की दृष्टिकोण से किये जाने योग्य संभावित बदलाव:***

- शॉवर या बाथटब में फिसलन रहित मैट का उपयोग।
- शॉवर या नहाने के स्थान पर पकड़ने के लिए बार्स अथवा डण्डों को लगवाएं।
- स्नान के लिए बाथ सीट या अन्य अनुकूली उपकरण का उपयोग करें।
- सीढ़ियों के किनारे पर फिसलन रहित ट्रेड्स का इस्तेमाल करें।
- सीढ़ी के दोनों किनारों पर सहारे के लिए पकड़ने के लिए रेलिंग का इस्तेमाल करें।

**सीढ़ियों के किनारे पर फिसलन रहित ट्रेड्स**

**सीढ़ी के किनारों पर रैंप और हैंड्रिल**

**स्नान के लिए बाथ सीट या अन्य अनुकूली उपकरण**

### च. दिल और सांस लेने में सहायक मांसपेशियों की देखभाल

#### हृदय संबंधी- प्रबंधन - हृदय की देखभाल

दिल की मांसपेशियों में भी डिस्ट्रोफिन मौजूद होता है, इसलिए डिस्ट्रोफिन की कमी से हृदय की मांसपेशियां भी प्रभावित होती हैं। बाद में बीमारी के बढ़ने के साथ हृदय की मांसपेशियां भी शामिल हो जाती हैं।

डीएमडी में हृदय, बीमारी के बाद के चरणों में प्रभावित होता नज़र आता है:

- कार्डियोमायोपैथी - दिल की मांसपेशियों का प्रभावित होना
- दिल के धड़कने की लय में गड़बड़ी- घबराहट
- हृदय की गति का रुक जाना

हृदय में असर अक्सर बहुत धीमें धीमें और चुपचाप होता है, यह खतरनाक लक्षणों के विकास के बिना है। परन्तु इन बदलावों को जल्द से जल्द देखा जाना चाहिए और शीघ्र उपचार किया जाना चाहिए।

कार्डियक मैनेजमेंट का उद्देश्य है, दिल की मांसपेशियों के बिगड़ती कार्यक्षमता का जल्द से जल्द पता लगाना और उपचार करना। हृदय की कार्यक्षमता का आधारभूत मूल्यांकन बीमारी की पुष्टि के समय या फिर कम से कम 6 वर्ष की आयु तक किया जाना चाहिए।

मूल्यांकन में काम से काम एक इलेक्ट्रोकार्डियोग्राम (ईसीजी) और इकोकार्डियोग्राम किया जाना चाहिए। बाद में दस साल की उम्र तक हृदय की कार्यक्षमता का नियमित मूल्यांकन हर दो साल में कम से कम एक बार होना चाहिए। 10 वर्ष की आयु के बाद कार्डियक मूल्यांकन वार्षिक या उससे पहले होना चाहिए, जैसे भी बीमारी के लक्षण संकेत देते हैं। यदि इस नॉन इनवेसिव (बिना दर्द के ) होने वाले कार्डियक टेस्ट में कुछ असामान्यता का पता चलता है, तो कम से कम हर 6 महीने में या अधिक निगरानी की आवश्यकता हो सकती है। बिना दर्द के हृदय की कार्यक्षमता की जाँच ईसीजी, इकोकार्डियोग्राम और कार्डियक मैग्नेटिक रेजोनेंस इमेजिंग (एम् आर आई) के द्वारा की जा सकती है। कार्डिएक एमआरआई दिल की संरचना और कार्यों की सटीक छवियां देता है। यह हृदय की मांसपेशियों में छोटे से छोटे बदलाव कोभी पकड़ लेता है।

ड्यूशेन की वाहक (कैरीयर) महिलाओं को अपने हृदय की जाँच वयस्क होते ही करा लेनी चाहिए और उसके बाद हर 3-5 साल में। कैरीयर, महिलाओं में यह जाँच ज़रूरी है क्यूंकि इनके हृदय में भी रोगियों के दिल की समस्याओं के समान ही अंतर पाया जा सकता है।

- बच्चे में हृदय की कार्य क्षमता में क्लिनिकली कमी के साथ-साथ जाँच में यदि कार्डियोमायोपैथी पायी जाती है तो यह संकेत है दवाई शुरू करने का।
- 10 साल की उम्र तक कार्डियक फंक्शन सामान्य होने के बावज़ूद, दवाइयाँ देनी चाहिए क्योंकि इससे दिल को सुरक्षित रखने में मदद मिलती है ।
- जो बच्चे स्टेरॉयड की थेरेपी पर होते हैं, उनको हृदय के दृष्टिकोण से विशेष रूप से उच्च रक्तचाप (उच्च रक्तचाप) की निगरानी रखनी चाहिए।

#### श्वास की मांसपेशियों की देखभाल

जैसे जैसे लड़के बड़े होते जाते हैं, सांस लेने और खांसने की मांसपेशियां कमजोर होती जाती हैं। जिससे उनको छाती के संक्रमण होने का खतरा बढ़ जाता है। यह अक्सर खाँसी और छाती की मांसपेशियों के कमज़ोर होने के चलते होता है। बाद में सांस की मांसपेशियों की लगातार कमजोरी की वजह से सोते समय भी सांस लेने में कठिनाई महसूस होती है। इससे नींद में गड़बड़ी होती है और दिन के वक़्त बार-बार सिरदर्द बना रहता है। जैसे-जैसे वे बड़े होते हैं, उन्हें दिन के दौरान भी सांस लेने में दिक्कत महसूस हो सकती है। जैसा कि यह प्रत्याशित समस्याओं के साथ एक लगातार बढ़ने वाली बीमारी है, इसलिए श्वसन की देखभाल एक योजनाबद्ध और सक्रिय तरीके से करनी ज़रूरी है।

डीएमडी से प्रभावित लड़कों में सांस लेने में तकलीफ होने वाले संकेतों को देखना या पहचानना बहुत जरूरी है। यदि

माता-पिता अपने बेटे में निम्नलिखित समस्याओं को देख रहे हैं, तो उन्हें चिकित्सक को रिपोर्ट करने की आवश्यकता है।

- स्पष्ट रूप से मामूली ऊपरी श्वसन संक्रमण के बाद लम्बे समय तक बीमार रहना। उदाहरण के लिए, सामान्य सर्दी- जुकाम का बढ़ जाना और छाती में कफ़ जम जाना और बाद में एंटीबायोटिक चिकित्सा की आवश्यकता पड़ना।
- बच्चे का सामान्य से अधिक थका हुआ रहना तथा ज्यादातर समय ध्यान की कमी रहना।
- सांस लेने में तकलीफ होती है, जैसे कि उसकी सांस उखड़ी उखड़ी रहती है या उसे एक साँस में वाक्य को पूरा करने में कठिनाई होती है।
- हर समय या सुबह सिर दर्द रहता है।
- सीधे लेटकर सोने में परेशानी होती है इसलिए ज़्यादातर जागता रहता है
- सोने पर बुरे बुरे सपने आते हैं और अचानक उठ जाता है, उसकी साँस तेज चल रही होती हैं और दिल बहुत तेज धड़क रहा होता है।

**श्वास की मांसपेशियों की निगरानी**

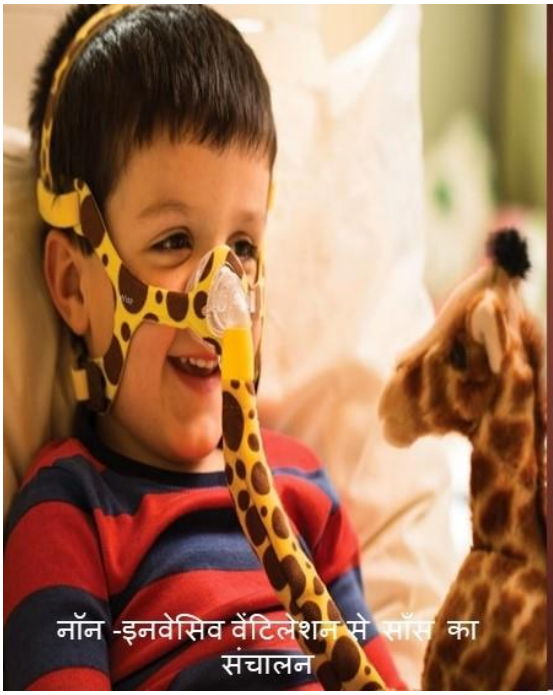
यह आवश्यक है कि अभी जब डी एम् डी से प्रभावित बच्चा चल रहा है, एक फेफड़ों (पल्मोनरी) की कार्यक्षमता का परिक्षण कर लिया जाए।

- वार्षिक फेफड़े के कार्य परीक्षण (फोर्स्ड वाइटल कैपेसिटी )
- नींद में कैपोग्राफी और ऑक्सीजन संतृप्ति निगरानी।

फेफड़ों (पल्मोनरी) की कार्यक्षमता की जाँच मुख्य रूप से उन बच्चों में महत्वपूर्ण हो जाती है जो चलना फिरने में असमर्थ हो जाते हैं। इसमें फेफड़े के कार्यों का मूल्यांकन, खांसी की क्षमता, नींद के दौरान ऑक्सीजन के स्तर का अध्ययन किया जाता है। ये मूल्यांकन हर ६ महीने में करने की सलाह डी जाती है।

***समस्याओं की रोकथाम***

- फेफड़ों में प्रवेश करने वाली हवा की मात्रा को बढ़ाना है ही मुख्य उद्देश्य होता है। सांस लेने की मांसपेशियों की कसरत (गहरी साँस लेना) फेफड़ों में प्रवेश करने वाली हवा की मात्रा को बढ़ाने में मदद करतीं हैं।
- बाद में जैसे-जैसे बीमारी बढ़ती है साँस लेने के लिए मदद की ज़रुरत पड़ती ह। शुरू में सिर्फ रात में बाहर से वायु-संचालन (नॉन -इनवेसिव नोक्टर्नल असिस्टेड वेंटिलेशन वाया बाई-लेवल ऐरवे प्रेशर या इंटरमिटेंट पॉजिटिव प्रेशर वेंटिलेशन (IPPV), या Bi-PAP) की आवश्यकता पड़ती है परन्तु बाद लक्षणों के बढ़ने के साथ दिन में भी बाहर से वायु-संचालन (गैर-इनवेसिव डेटाइम असिस्टेड वेंटिलेशन) की ज़रुरत पड़ती है।
- रोगी और परिवार की आवश्यकता या पसंद के अनुसार इनवेसिव वेंटिलेशन का विकल्प भी है। इनवेसिव वेंटिलेशन में सर्जिकल रूप से एक ट्यूब ("ट्रेकियोस्टोमी ट्यूब") को साँस की नली में रखा जाता है।


नॉन -इनवेसिव वेंटिलेशन मे साँस का संचालन


इनवेसिव वेंटिलेशन: सर्जिकल रूप से एक ट्यूब को साँस की नली ("ट्रेकियोस्टोमी ट्यूब")

***प्रतिरक्षण (टीकाकरण)***

- न्यूमोकोकल वैक्सीन: निमोनिया से बचाव में मदद करता है। 2 साल से अधिक उम्र के बच्चे में दिया जाता है और दो खुराक में 8 सप्ताह का अंतर रखा जाता है।
  वार्षिक इन्फ्लुएंजा टीका: इन्फ्लूएंजा का टीका वर्ष में एक बार लगाया जाना चाहिए।

  दोनों टीकों को स्टेरॉयड के साथ इलाज लेरहे व्यक्ति को देना चाहिए, हालांकि टीकाकरण की प्रतिरक्षा प्रतिक्रिया उन व्यक्तियों में कम हो सकती है।

***छाती में संक्रमण का इलाज़***

- यदि छाती में संक्रमण होता है, तो एंटीबायोटिक दवाओं, छाती की फिजियोथेरेपी और खांसी करने में सहायता कर के शीघ्र उपचार करने की सलाह दी जाती है।
- गैर-इनवेसिव वेंटिलेशन की मदद से सांस लेने की प्रक्रिया को बनाए रखने के लिए एक बहुत ही महत्वपूर्ण तरीका है।
- नियोजित सर्जरी के समय श्वास पर विशेष ध्यान देना आवश्यक है।

**छः आहार का सही चुनाव**

बीमारी की पुष्टि होने से लेकर जीवन भर अच्छा और पुष्टिक आहार अत्यधिक आवश्यक है। संतुलित आहार, वज़न बढ़ने और कुपोषण से सदा बचाव करता है।

पंजीकृत आहार विशेषज्ञ से प्रति दिन के लिए आवश्यक कैलोरी का मूल्यांकन कर परामर्श लेना अत्यन्त महत्वपूर्ण है। यह मूल्याँकन बच्चे की लम्बाई, उम्र और काम करने की गतिविधि के स्तर के अनुसार से किया जाता है। उम्र के हिसाब से वजन या बॉडी मास इंडेक्स (बीएमआई) को राष्ट्रीय पर्सेंटाइल चार्ट पर 10वें और 85वें पर्सेंटाइल के बीच होना चाहिए।

स्वस्थ शरीर बनाए रखने के लिए स्वास्थ्यवर्धक और संतुलित आहार खाना, जिसमें विभिन्न प्रकार के स्थानीय खाने सम्मिलित कियेजाने चाहिए। परिवार को संतुलित खाने के बारे में जानकारी देते समय यह ज़रूरी है कि स्थानीय और मौसमी खाद्य पदार्थों को प्राथमिकता दें खाने में परामर्श के हिसाब से कैलोरी, प्रोटीन, तरल पदार्थ, कैल्शियम, विटामिन डी, और अन्य पोषक तत्व होने चाहिए, ये मूल्याँकन कम से कम साल में एक बार आवश्यक है। अधिक से अधिक तरल पदार्थ का सेवन कब्ज और गुर्दों को दोष प्रभाव से बचाने के लिए जरुरी है।

- बीमारी  की पुष्टि के समय, स्टेरॉयड शुरू करते समय, बच्चे का चलना फिरना बंद होने पर और जब खाना निगलने में समस्या होने लगे; इन सभी बदलावों के समय खान -पान पर विशेष ध्यान दैनिकी ज़रूरत होती है। अच्छा हो इस दौरान अगर आहार विशेषज्ञ का परामर्श ले लिया जाए।
- यह परामर्श गैस्ट्रोपारेसिस ( बीमारी बढ़ने पर पेट देर से ख़ाली होता है) के दौरान भी ज़रूरी होता है ।  इस अवस्था में खाने के बाद पेट में दर्द, जी मिचलाना, उल्टी, भूख न लगना और जल्दी से पेट भरा हुआ महसूस होता है।
- यदि बच्चे का वजन अचानक से बहुत कम हो जाए, तो हो सकता है  हृदय या श्वसन की समस्याएं बढ़ गई हैं या फिर खाना निगलने में दिक्कत आ रही हो।  इस सूरत में भी परामर्श लेना आवश्यक हो जाता है।

**हर बार अस्पाल में दिखते समय**

पंजीकृत आहार विशेषज्ञ द्वारा मूल्यांकन। वजन और ऊंचाई की निगरानी; जो बच्चे अब खड़े नहीं हो सकते हैं उनके लिए वैकल्पिक ऊंचाई का अनुमान उपयोग लाया जाना चाहिए।

**हर 6 महीने में चलने फिरने में असमर्थ होने के बाद**

खाना निगलने में दिक्कत, कब्ज, गैस्ट्रोइसोफ़ेगल रिफ़्लक्स (खट्टी डकारों) और गैस्ट्रोपारेसिस से आधारित प्रश्न किये जाने चाहिए।

**हर साल:** खून में 25-hydroxyvitamin डी और आहार में कैल्शियम की मात्रा की जाँच होनी चहिये।

आहार की योजना हमेशा पंजीकृत आहार विशेषज्ञ के साथ चर्चा के बाद ही तैयार की जानी चाहिए।  हालांकि कुछ सामान्य बातें हैं जिन्हें स्टेरॉयड ले रहे ड्यूशेन के बच्चों को सदा ध्यान में रखना चाहिए।

- संतुलित आहार में अनाज, दालें, दूध, सब्जियां और फल शामिल होने चाहिए।
- थोड़े-थोड़े अंतराल पर बच्चे को बार बार खिलाएं।
- सुबह नाश्ते में केवल दूध नहीं करना चाहिए, दूध के साथ रोटी, सैंडविच और सब्जी का दलिया भी देना चाहिए।
- फलों को कभी भी भोजन का विकल्प उसका न समझें। फलों को अलग से दें।
- बच्चों को फलों का रस पीने के बजाय पूरे फल खाने के लिए प्रोत्साहित करें। इसमें रेशा होता है।
- बादाम, अखरोट, किशमिश, मुन्नके आदि जैसे मेवे को अपने दैनिक आहार में शामिल करें।
- सब्ज़ियों का पोहा, अंकुरित दालें, बेसन चीला, सलाद आदि जैसे पौष्टिक स्नैक्स को बढ़ावा दें।
- खाने को निर्धारित समय पर ही दें, बीच में कभी भी खाने को कुछ न दें।

**आहार योजना का एक नमूना**

| **भोजन का समय** | **खाद्य पदार्थ** |
|---|---|
| बहुत सवेरे | बादाम (4-5 रात भर भिगो कर); अख़रोट (1-2), किशमिश या मुन्नके (4-5 रात भर भिगो कर) |
| सुबह 7:30 am | दूध, अनाज के साथ (दलिया, सूज़ी) (1 कटोरी)<br>या<br>काम तेल वाले भरवां पराठे/रोटी (1) + दही (1 कटोरी) |
| मिड- डे -11:00 | काम तेल वाले भरवां पराठे/रोटी/बेसन चीला  (1) +सब्ज़ी या दाल (1 कटोरी)+ फ़ल<br>या सब्जी उपमा/सब्जी इडली/सब्जी पोहा (1 कटोरी) |
| लंच -2:30 | रोटी / चावल पुलाव, (½ or1 कटोरी) + दाल/पनीर/दही (रायता या लस्सी) (1 कटोरी) +<br>सब्ज़ी (1 कटोरी) |
| शाम 5:00 | दूध/ चावल या सूज़ी खीर / पनीर / हलवा/नमकीन दलिया (1 कटोरी) |
| रात का खाना -8:00 to 9:00 | रोटी / खिचड़ी (½ or1 कटोरी) + दाल/पनीर (½कटोरी) + सब्ज़ी (1/2 कटोरी) +  सलाद (1/2 प्लेट) |
| खाने के बाद  9:30 | दूध 1 गिलास |

## 3. ड्यूशेन के मरीज़ों में आपात का की स्थिति आने पर कैसे कार्य करें?

जब कोई भी चिकित्सीय आपात स्थिति उत्पन्न होती है तो एक डीएमडी बच्चे की देखभाल में कुछ बातों पर ध्यान देने की आवश्यकता होती है।

- डीएमडी का इलाज़, दवा जो वर्तमान में चल रही है, किसी भी हृदय या साँस लेने की असामान्यता की उपस्थिति को बच्चे को भर्ती कर रही इकाई से साझा करें।
- बच्चे की ड्यूशेन की  देखभाल करने वाली टीम की जानकारी आपातकालीन देखभाल इकाई को दी जानी चाहिए। इससे उन्हें उचित समय पर आवश्यक देखभाल करने की अनुमति मिलेगी।
- बीमारी के पिछले सालों में बार बार होने वाली बिमारियों अथवा समस्याओं के बारे में आपात स्थिति में बच्चे की देखभाल कर रही टीम को बताएं।  जैसे : निमोनिया, हार्ट फ़ेल, गुर्दे की पथरी, पेट का आंशिक पक्षाघात (गैस्ट्रोपरेसिस) इस में पेट खाली होने में देरी लगता है।
- आपके पास जोभी  नवीनतम हृदय या साँस की कार्यक्षमता जाँच के रिकॉर्ड्स जहाँ तक संभव हो अपनी आपातकालीन टीम से साझा करें।जैसे स्पिरोमेट्री, ईसीजी, और कार्डियक ईको।
- अगर कोई भी आपातकालीन सर्जरी करने का निर्णय लिया गया है तो सबसे पहले अपने एनेस्थेटिस्ट को डीएमडी के बारे में जानकारी दें।  इन मरीजों में एनेस्थेसिया से संबंधित जोखिम अधिक होते हैं।
- स्टेरॉयड की अवधि और खुराक, स्टेरॉयड का दीर्घकालिक से उपयोग स्पष्ट रूप से अपनी आपातकालीन (ईआर) टीम को सूचित किया जाना चाहिए।
- टीम को यह स्पष्ट करना महत्वपूर्ण है कि क्या बच्चा स्टेरॉयड की खुराक लेने से चूक गया है और कब तक।
- स्टेरॉयड शरीर की किसी भी विषम परिस्थिति से निपटने को कम कर सकता है, इसलिए विषम परिस्थिति में स्टेरॉयड की अतिरिक्त खुराक की आवश्यकता हो सकती है।
- स्टेरॉयड से संबंधित विषम दुष्प्रभाव किंचित ही आपातकालीन स्थिति की तरह सामने आते हैं।

***आपातकालीन स्थितियाँ***

***टटी हुई हड्डी / फ्रैक्चर***

डीएमडी से प्रभावित लड़कों में हड्डियों के टूटने का खतरा अधिक होता है।  पैर की हड्डी टूटने से बच्चे की भविष्य में ना चल- फिर पाने की संभावनाए बढ़ जाती हैं।  फिजियोथेरेपिस्ट और देखभाल करने वाली टीम को बताएं अगर कोई फ्रैक्चर हो तो, वे आवश्यकता पड़ने पर सर्जन से बात कर सकते हैं।

- यदि कोई अभी भी चल रहा है तो अक्सर सर्जरी, कास्ट (सांचा) की तुलना में एक बेहतर विकल्प होता है।
- इस परिस्तिति में फिजियोथेरेपिस्ट के इनपुट यह सुनिश्चित करने के लिए महत्वपूर्ण होते हैं कि लड़का जल्द से जल्द अपने पैरों से वापस चलने लग जाए।

***साँस लेने में परेशनियाँ***

- नवीनतम सांस लेने की क्षमता का परिक्षण (पल्मोनरी फंक्शन टेस्ट) के रिकॉर्ड की कॉपी रखें। ये रिकॉर्ड आपातकालीन स्तिति में बच्चे

का ज़ल्द से ज़ल्द इलाज़ शुरू करने में सहायक होंगे।

- माता -पिता अथवा फ़िज़ियोथेरेपिस्ट द्वारा सीने की फिजियोथेरेपी इस नाजुक समय की प्रमुख आवश्यकता हो सकती है।
- कभी-कभी तुरंत एंटीबायोटिक की आवश्यकता हो सकती है।
- जिनकी साँस लेनेकी कार्यक्षमता कम होती है, उनकी सांस लेने वाली मांसपेशियों को संक्रमण के दौरान अतिरिक्त सहायता की आवश्यकता पढ़ सकती है।
- यदि आपके फेफड़ों की कार्यक्षमता कम है तो ऑक्सीजन देने से सांस लेने के लिए शरीर की ड्राइव कम हो सकती है। ऐसा होने पर रक्त में कार्बन डाइऑक्साइड का स्टार खतरनाक स्तर तक बढ़ सकता है। यह घातक हो सकता है, और जीवन के लिए खतरा भी हो सकता है इसलिए ऑक्सीजन का इतेमाल अत्यधिक सावधानी के साथ किया जाना चाहिए और कार्बन डाइऑक्साइड की निगरानी रखनी चाहिए।
- देखभाल के दौरान ओपियेट्स अथवा अन्य बेहोश करने वाली दवाओं के उपयोग की आवश्यकता पड़ सकती है।
- साँस लेने के लिए बहार से समर्थन की आवश्यकता पड़ सकती है। यह समर्थन लम्बे समय तक भी पड़ सकता है।
- अगर एनेस्थीसिया की आवश्यकता हो तो जहाँ तक हो सके इंजेक्शन द्वारा ही एनेस्थीसिया दें। सांस के द्वारा एनेस्थीसिया देने से बचना चाहिए। सक्सीनिलकोलिन ड्यूशेन में वर्जित है और नहीं देना चाहिए।

**4. कौन से अस्पतालों में ड्यूशेन के मरीज़ों के लिए उचित देखभाल उपलब्ध है?**

सभी डी एम् डी रोगियों को सभी चरणों में अच्छी से अच्छी देखभाल मिलनी चाहिए। ड्यूशेन की देखभाल बहुत व्यापक होती है इसमें शरीर के कई हिस्सों की देखभाल शामिल है। बीमारी, उससे सम्बंदित जटिलताओं की जाँच और चिकित्सीय आयामों की प्रगति के साथ ड्यूचेन से प्रभावित लोगों की जीवन काल बढ़ गया है। इसलिए उनके जीवन की गुणवत्ता को सर्वोत्तम बनाए रखने की आवश्यकता महसूस हुई है।

कोई भी चिकित्सक जिसको बच्चे को प्रारंभिक जांचो के दौरान मांसपेशियों के अपविकास का संदेह होता है तो उसे अन्य जांचे भी करनी चाहिए ताकि बीमारी का सही सही पता लगाया जा सके। मांसपेशियों के अपविकास का संदेह पर मरीजों को उन केंद्र में भेज देना चाहिए जहाँ इस प्रकार की बिमारिओं के इलाज़ की सुविधा उपलब्ध हो। एक बार बीमारी की पुष्टि हो जाने के बाद पुरे इलाज़ की के रूप रेखा बनाई जाती है जिसकी खंड ग में चर्चा की गई है। डी एम् डी की देखभाल एक बहु-आयामी देखभाल है, देखभाल करने वाली टीम में विभिन्न लोग (विभाग) शामिल हैं:

- बाल रोग विशेषज्ञ (न्यूरोलॉजी)
- फेफड़ों के रोग विशेषज्ञ
- हृदय रोग विशेषज्ञ
- बाल एंडोक्रिनोलॉजिस्ट विशेषज्ञ
- फिजियोथेरेपिस्ट
- हड्डी रोग और पुनर्वास विशेषज्ञ
- जठरांत्र चिकित्सक
- डी एम् डी देखभाल समन्वयक

कभी-कभी देखभाल के सभी डोमेन एक ही जगह पर प्राप्त करना मुश्किल होता है, इस स्थिति में विशिष्ट देखभाल प्राप्त करने के लिए विभिन्न अन्यत्र अस्पतालों में इलाज़ प्राप्त कर सकते हैं। आमतौर पर डीएमडी रोगी को एक साथ अलग-अलग विशेष देखभाल की आवश्यकता होती है इसलिए ये अच्छा होगा अगर एक ऐसे स्वास्थ्य केन्द्र की पहचान की जाए जहाँ आवश्यकता पड़ने पर सभी प्रकार की वांछित इलाज़ संभव हों।

कुछ देशों में इस प्रकार के कार्यकारी समूह होते हैं जो मरीज़ो को इस प्रकार के बहु आयामी अस्पतालों के पहचान करने में परिवारों की मदद करते है।

भारत में हमारे पास ऐसे सहायता समूहों की कमी है लेकिन चीजें लगातार बदल रही हैं। भारत के सभी स्नातकोत्तर अनुसंधान संस्थानों में ड्यूशेन की देखभाल से सम्बंधित सभी सुविधाएँ उपलब्ध हैं। इसलिए, इन संस्थानों के साथ निरंतर और नियमित रूप से संपर्क में रहना चाहिए। डीएमडी उपचार के लिए नई दवाएं और इलाज़ पर लगातार शोध कार्य किया जा रहा है। ये सरे केंद्र इन सभी शोध कार्यों अथवा संबंधित एजेंसियों द्वारा कार्यक्रम या नीतियों को कार्यान्वित इकाइयाँ हैं इसलिए इन केंद्रों से जुड़ा रह कर यथोचित देखभाल महत्वपूर्ण हैं।

## खंड घ : अस्पताल में देखभाल करने वाली टीम की भूमिका

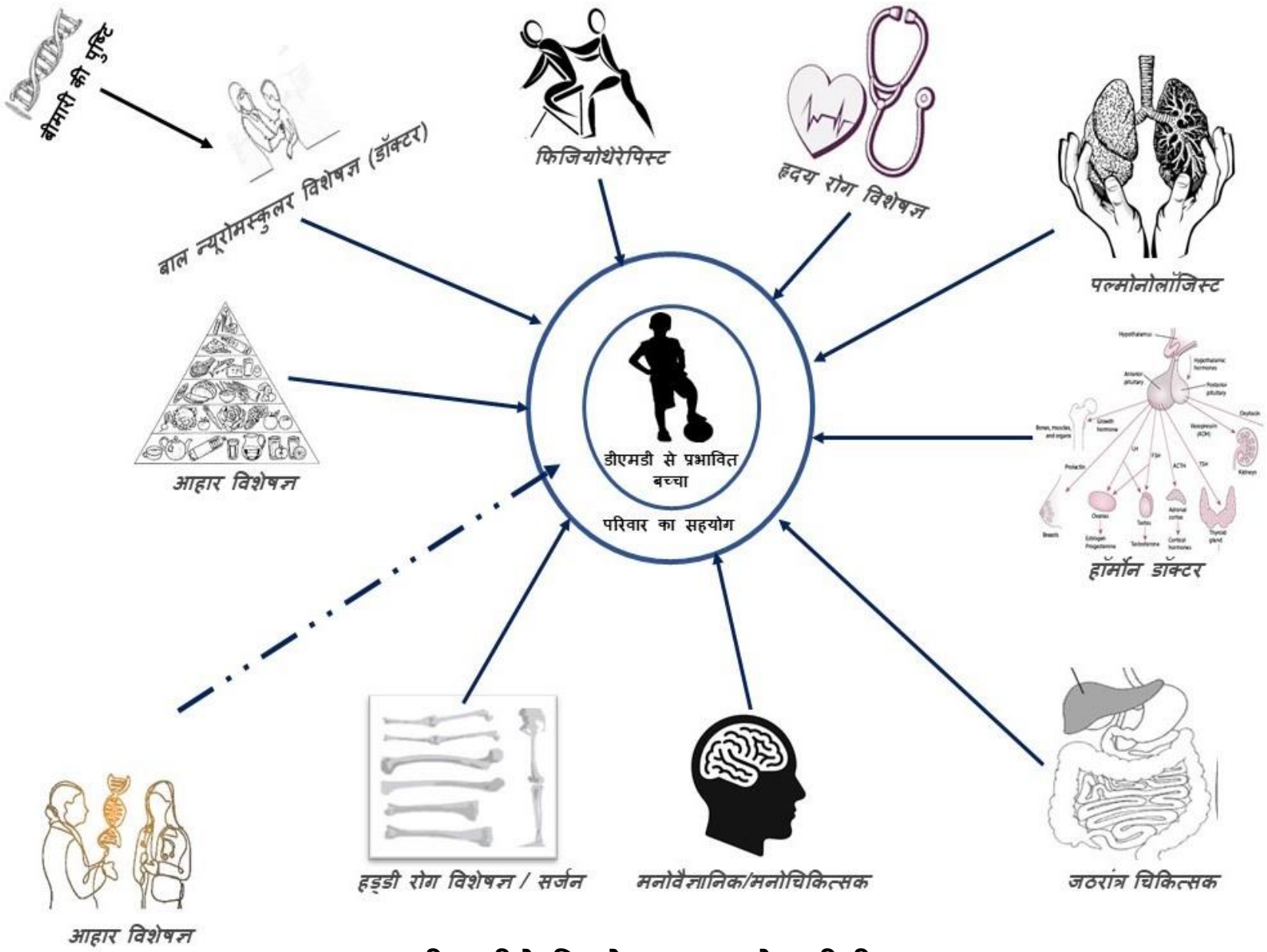
### 1. देखभाल के लिए एक टीम की आवश्यकता क्यों है?

डीएमडी की बीमारी बढ़ने के साथ विभिन्न अंग शामिल हो जाते हैं इसलिए रोग के बढ़ने के साथ रोग के इलाज़ में बहु-अनुशासनात्मक देखभाल महत्वपूर्ण हो जाती है। इस टीम में न्यूरोलॉजी, कार्डियोलॉजी, पल्मोनोलॉजी, एंडोक्रिनोलॉजी, मनोविज्ञान के विशेषज्ञ और उप -विशेषज्ञों और कई अन्य सहायक टीम के सदस्यों की आवश्यकता होती है।

टीम के सदस्यों द्वारा उचित समय पर मिलकर, एक समन्वित तरीके से किये गए प्रयास हर लड़के और परिवार के परिजनो के जीवन की गुणवत्ताको बनाए रखने में सहायक होते हैं। सभी देखभाल करने वाले सदस्यों के साथ में मिल कर काम करने से उपलब्ध संसाधनों का ज़्यादा से ज़्यादा उपयोग हो पता है। यदि स्वास्थ्य केंद्र में देखभाल के लिए एक समन्वयक है तो वह मरीजों की बहुआयामी देखभाल को व्यवस्थित करने में मदद कर सकता है। देखभाल कर रही टीम के सदस्यों और परिवार के बीच में एक सेतु बन समन्वयक इलाज़ को सहज और तेज़ बना सकता है। हर विशेषज्ञ या उप विशेषज्ञ की आवश्यकता बीमारी के हर चरण में नहीं होती है, इलाज़ कर रही टीम की सूची में शामिल हैं:

- ***बाल न्यूरोमस्कुलर विशेषज्ञ (डॉक्टर) :*** न्यूरोमस्कुलर विशेषज्ञ /पुनर्वास विशेषज्ञ केयर टीम के लीडर होते हैं। ये विशेषज्ञ इलाज़ की सारे महत्वपूर्ण निर्णय लेते हैं। यही व्यक्ति रोग की प्रगति पर नज़र रखता है। जिन अस्पतालों में जहां देखभाल करने वाली टीम में समन्वयक उपलब्ध नहीं होता है, वहां बाल न्यूरोमस्कुलर विशेषज्ञ इलाज़ की आवश्यक सभी प्रक्रियाओं में समन्वय रखते हैं।
- ***देखभाल-समन्वयक:*** आवश्यक सभी प्रक्रियाओं में समन्वय रखते हैं। रोगी, और देखभाल करने वाली टीम के सभी सदस्यों के साथ बातचीत करते हैं। समन्वयक क्षित करने के साथ साथ इलाज़ के लिए ज़रूरी संसाधनों से संबंधित जानकारी भी प्रदान कर सकते हैं। जैसे जीवन के स्तर को बेहतर बनाना, वित्तीय सहायता, अन्य पुनर्वास संसाधन।
- ***फिजियोथेरेपिस्ट:*** फिजियोथेरेपी बीमारी के पता चलने के समय से मददगार सिद्ध होती है। फिजियोथेरेपी विभिन्न कसरतों के माध्यम से मरीज़ की मांसपेशियों की लम्बे समय तक क्षमता को बनाए रखने में मदद करता है। हाथ पैरों के जोड़ों की गतिशीलता को क्षमता के अनुसार अधिकतम बनाए रखता है।
- ***व्यावसायिक थेरेपिस्ट***: थेरेपिस्ट, लोगों को दैनिक जीवन की गतिविधियों को करने के लिए सक्षम करता है। यह बच्चे और माता-पिता को स्वाव्लम्बी बनाने में सहायक होते हैं। थेरेपिस्ट, फिजियोथेरेपिस्ट के साथ मिलकर विभिन्न उपकरणों को मरीज़ के हिसाब से अनुकूल बनाने में मदद कर सकते हैं।
- ***हृदय रोग विशेषज्ञ (दिल की देखभाल) :*** एक हृदय रोग विशेषज्ञ बीमारी के बढ़ने पर बच्चे के दिल की देखभल का ख्याल रखते हैं। एक बार बीमारी का पता चलने के बाद 6-12 महीने पर हृदय की जाँच करने की जरूरत होती है।
- ***पल्मोनोलॉजिस्ट (फेफड़े की देखभाल) :*** पल्मोनोलॉजिस्टआपकी श्वास और खाँसी की मांसपेशियों का ख्याल रखते हैं। एक फेफड़ों (पल्मोनरी) की कार्यक्षमता का जाँच 6-12 महीने के अंतर पर करने की जरूरत होती है।
- ***आनुवांशिक परामर्शदाता :*** आनुवांशिक परामर्शदाता माता-पिता को भविष्य में अपने परिवार की योजना बनाने में मदद करते हैं। यह बीमारी को पीढ़ी दर पीढ़ी आगे बढ़ने से रोकते हैं।

- ***बाल चिकित्सक एंडोक्रिनोलॉजिस्ट (हॉर्मोन डॉक्टर)*** डीएम् डी के उपचार के दौरान बच्चों में हार्मोन्स के पर नकारात्मक प्रभाव हो सकता है। जो बच्चे की आयु के अनुसार कद के न बढ़ने और किशोरावस्था के आने में देरी, जैसी दिक्कतों पैदा हो सकती हैं। इन दिक्कतों की देखरेख ये विशेषज्ञ करते हैं।
- ***जठरांत्र चिकित्सक (गैस्ट्रोएन्टेरोलॉजिस्ट)***: गैस्ट्रोएन्टेरोलॉजिस्ट, बीमारी के बढ़ने के साथ पेट और आँतों की बिमारियों (जटिलताओं) की देखभाल/इलाज़ करते हैं।
- ***मनोवैज्ञानिक/मनोचिकित्सक :*** मनोवैज्ञानिक/मनोचिकित्सक मरीज़ की भावनात्मक, व्यवहारिक, शैक्षिक स्तर को इष्टतम बनाने का ध्यान रखते हैं।
- ***हड्डी रोग विशेषज्ञ / सर्जन:*** हड्डियों और जोड़ों की देखरेख में योगदान देते हैं।
- ***आहार विशेषज्ञ:*** आहार विशेषज्ञ आवश्यकता के अनुसार बच्चे के आहार और पोषण की योजना बनाने में मदद करते हैं। यदि रोगी लंबे समय से स्टेरॉयड पर है तो विटामिन डी और कैल्शियम के सेवन पर विशेष ध्यान देने की आवश्यक है।
- ***पुनर्वास विशेषज्ञ/डॉक्टर,*** फिजियोथेरेपिस्ट के साथ मिलकर मांसपेशियों की क्षमता, खिंचाव और कार्यक्षमता को बनाएरखने के लिए काम करते हैं। बीमारी के अंतिम चरण में वे सभी उपाय करते हैं जो जीवन की संभव गुणवत्ता को सर्वोत्तम बनाए रखने में मदद करते हैं।




**डीएमडी के लिए देखभाल करने वाली टीम**

## 2. ड्यूशेन के इलाज़ में परिवार के सदस्यों की क्या भूमिका है?

एक परिवार वह इकाई है, जहाँ एक बच्चा अपने माता-पिता एवं भाई- बहनो के साथ रहता है, यह जीवन का सबसे केंद्रीय और स्थिर हिस्सा होता है। परिवार सबसे पहला वह परिवेश है जहां से ड्यूशेन बच्चे को प्रथम देखभाल मिलती है। भारतीय संस्कृति में किसी भी व्यक्ति की बीमारी उसके परिवार की बीमारी होती है। परिवार के सदस्य मरीज़ की देखभाल करने में बहुत महत्वपूर्ण भूमिका निभाते हैं। डीएमडी से प्रभावित बच्चों की देखभाल एक योजना बद्ध तरीके से करने में परिवार के सदस्य की महत्वपूर्ण भागीदारी होती हैं। देखभाल करने वाले / परिवार के सदस्य बच्चे की दैनिक गतिविधियों जैसे स्वयं की देखभाल, स्कूली शिक्षा, इधर-उधर जाने में सहायता और भावनात्मक सहायता प्रदान करते हैं। बीमारी के बढ़ने के साथ देखभाल देने के प्रयास में भी वृद्धि हो जाती है, अब देखभाल में अधिक समय लगता है। परिवार के सदस्यों पर देखभाल का बोझ बढ़ जाता है। परिवार की भागीदारी के बिना रोगी को किसी भी प्रकार की चिकित्सीय या अन्य देखभाल नहीं दी जा सकती है। परिवार को शामिल कर के रोगियों को बढ़ती बीमारी के अनुरूप ढ़ाला जा सकता है, इलाज़ को उनके अनुसार सुविधाजनक बनाया जा सकता है।

मुख्य या प्राथमिक देखभालकर्ता वह है, जो रोगी को सबसे अधिक देखभाल प्रदान करता है। उसके पास देखभाल से जुड़ी अधिकांश जिम्मेदारियां होती हैं। माँ ही अक्सर मुख़्य या प्राथमिक देखभालकर्ता होती है और उसका देखभाल करने वाली टीम में प्रमुख़ योगदान होता है। बीमारी के विभिन्न चरणों में प्राथमिक देखभाल करने वाले का सभी प्रकार के इलाजों की योजना बनाने में भागीदारी आवश्यक है।

देखभाल की सभी गतिविधियों को दो प्रमुख श्रेणियों में बांटा जासकता है : एक एम्बुलेटरी चरण जिसमें बच्चा चल- फिर सकता है और दूसरा गैर-एम्बुलेटरी चरण : जब बच्चा चलने फिरने में सक्षम नहीं होता है और इधर-उधर जाने के लिए व्हीलचेयर पर निर्भर हो जाता है।

एम्बुलेटरी चरण में की जाने वाली देखभाल :

- दिनभर स्वयं की देखभाल करने में सहायता प्रदान करना। बीमारी के पहले दो चरणों में इन गतिविधियों में काम समय और न्यूनतम प्रयास लगता है। परन्तु बाद के चरणों में ये स्पष्ट रूप से बढ़ जाते हैं।
- स्कूल छोड़ना और वापस लेकर आना। स्कूल के दौरान, स्कूल की दैनिक गतिविधियों में सहायता प्रदान करना।
- डीएमडी में मांसपेशियों की कसरत के लिए सक्रिय और निष्क्रिय दोनों तरह के अभ्यास में बच्चे की मदद करना।
- माताएँ बच्चों के साथ मांसपेशियों के खेल में पूरी तरह से शामिल हो सकती हैं।
- पोषण विशेषज्ञ द्वारा आहार योजनाओं पर अमल करना।
- अस्पताल जाने के दौरान लगातार साथ रहना और अपना सहयोग देना।

गैर-एम्बुलेटरी चरण में की जाने वाली देखभाल :

- घर के अंदर या घर के बाहर सभी प्रकार के स्थानान्तरण/सरकाना, जैसे बिस्तर से व्हीलचेयर पर, व्हीलचेयर से टॉयलेट सीट, टॉयलेट सीट से व्हीलचेयर, व्हीलचेयर से वाहन इत्यादि।
- व्हीलचेयर की सहायता से आवत-जावत बनाए रखने में सहयोग देना। (व्हील चेयर गतिशीलता)
- पढ़ने और लिखने में मदद करना।
- स्वयं की देखभाल, स्कूल की शैक्षणिक या गैर-शैक्षणिक गतिविधियों में उनकी सहायता करना।
- बाद के गैर-एम्बुलेटरी चरण में जब बच्चा बिस्तर पर पड़ जाता है तो उसे अतिरिक्त देखभाल की ज़रूरत होती है। जैसे : पोस्ट्युरल ड्रेनेज, साँस लेने में

सहायता, बिस्तर गतिशीलता बनाए रखना एवं इसकी ट्रेनिंग देना।

**बीमारी के दौरान भी लंबे समय तक आराम और गतिहीनता से बचना चाहिए क्योंकि इससे बच्चे  की दैनिक गतिविधि करने की क्षमता एकदम से घाट जाती है।**

एक महिला द्वारा  देखभाल करना सबसे बड़ी बाधा सिद्ध होती है क्योंकि बीमारी के अंतिम चरणों में बहुत समय और श्रम की आवश्यकता होती है और माँ के लिए अकेले संभालना मुश्किल हो जाता है। इसलिए ये ज़रूरी है की देखभाल की जिम्मेदारियों को परिवार के सदस्यों के बीच में साझा किया जाए ताकि एक व्यक्ति विशेष पर पूरा बोझ न आए।

बीमारी बढ़ने के साथ, देखभाल में शामिल होने वाले परिवारों को समय-समय पर देखभाल के लिए निरंतर परामर्श और प्रशिक्षण की आवश्यकता होती है। यह प्रशिक्षण मुख्य रूप से बाल न्यूरोमस्कुलर विशेषज्ञ अथवा समन्वयक द्वारा दिया जा सकता है। देखभाल करने में आनेवाली बाधाओं को दूर करने के उपाय हैं:

- सहायक उपकरण प्रदान करना जैसे: व्हील चेयर
- सहायक उपकरणों पर प्रशिक्षण एवं परामर्श।
- घर और अपने आस पास के परिवेश को बीमारी के अनुरूप ढालने के लिए समाज को इनके बारे में जानकारी प्रदान करना।
- मनोवैज्ञानिक और भावनात्मक मुद्दों पर बच्चे एवं परिवार के सदस्यों को परामर्श।
- स्कूल की गतिविधियों में सहायता के लिए "दूसरा शिक्षक" प्रदान किया जा सकता है।
- विकलांग व्यक्तियों (ADIP) योजना के तहत सहायता: सभी डीएमडी रोगियों को मोटराइज्ड ट्राइसाइकिल और व्हीलचेयर प्रदान की जाती हैं। माली मदद (सब्सिडी) 25,000 / - रुतक है और 16 वर्ष पूरे होने पर मिल सकती है, कोई भी योजना में ऑनलाइन पंजीकरण कर सकता है।

## 3. ड्यूशेन की देखभाल में क्या बाधाएं आतीं हैं?

लम्बी बीमारी की देखभाल से संबंधित दो घटक होते हैं: एक है स्वास्थ्य कर्मियों द्वारा दी जाने वाली विशिष्ट देखभाल जो अस्पतालों तक ही सीमित होती है; दूसरी है घर अथवा अनौपचारिक देखभाल जो परिवार के सदस्यों या दोस्तों द्वारा प्रदान की जाती है।

क्षेत्रीय स्तर पर डाक्टरों और परिवार के सदस्यों में जागरुकता की कमी एवं बीमारी के बारे में सीमित जानकारी की वजह से बीमारी की पुष्टि होने में बेहद कठिनाइयों का सामना करना पड़ता है। इन सभी कारणों से इलाज़ शुरू करने में खासी देरी हो जाती है। यह ड्यूशेन के इलाज़ में प्रमुख बाधाओं में से एक। लोगों को उन अस्पतालों के बारे में पता नहीं है जहां डीएमडी के इलाज़/देखभाल से संबंधित स्वास्थ्य सुविधाएं उपलब्ध हैं और ये संख्या में भी बहुत कम हैं। राष्ट्रीय या राज्य स्तर पर किसी भी सार्वजनिक / सरकारी साइट पर ऐसे संस्थानों की कोई पहचान या सूची उपलब्ध नहीं है। कुछ देशों में ऐसी वेबसाइट्स या सूचना पोर्टल उपलब्ध हैं जहाँ ऐसे संस्थानों के बारे में जानकारी मिल जाती है, जो मान्यता प्राप्त केंद्रों के रूप में डीएमडी देखभाल प्रदान करने में सक्षम हैं।

अगर कुछ लोगों को में इन सुविधाओं के बारे में पता है तो वे बहुत दूर स्थित हैं। अस्पतालों से दूरी अधिक होने की वज़ह से मरीजों को बार बार चिकित्सा संस्थानों में आने में दिक्कत होती है। कभी कभी नियमित जाँचों में भी देरी हो जाती है और कभी कभी मरीज़ अस्पताल लौट के आता ही नहीं है (लॉस टू फॉलो अप)। डीएमडी से जुड़ी शारीरिक दुर्बलताओं के कारण स्वास्थ्य केंद्र में आकर डॉक्टर को दिखाना एक भारी बाधा बन सकता है। समय और पैसा दोनों ही महत्वपूर्ण कारण हैं जो स्वास्थ्य देखभाल की मांग में एक बाधा के रूप में कार्य करते हैं।

डीएमडी की देखभाल में लगी टीम के सदस्यों के बीच समन्वय का आभाव, स्वास्थ्य परामर्श दाताओं और परिवार के सदस्यों के बीच अपर्याप्त संचार/बातचीत ज़रूरी देखभाल और इलाज़ के घटकों के अनुपालन में गिरावट लाती है। दुर्लभ बीमारियां चिकित्सा समुदाय के लिए कई चुनौतियां पेश करती हैं, लेकिन पिछले कुछ वर्षों में, अनुसंधान के प्रयासों से इलाज़ में कई सुधार आए हैं। सरकार भी इन बिमारियों की तरफ ख़ासा ध्यान दे रही है।

इलाज़ का दूसरा प्रमुख घटक "होम केयर" है, घर पर की जानेवाली देखभाल। घर पर देखभाल आमतौर पर परिवार के सदस्यों द्वारा जाती है और यह अनौपचारिक देखभाल कहलाती है। देखभालकर्ता विभिन्न प्रकार की सहायता प्रदान करते हैं जैसे: रोगी को जाँच के लिए अस्पताल लेकर आना, घर पर आहार और दवाओं का प्रबंधन करना, कपड़े पहनाना और सामाजिक गतिविधियों में शामिल करने में मदद करना। रोगियों में जीवन की गुणवत्ता बनाए रखने के लिए होम केयर का बहुत महत्व है। आमतौर पर माँ ही मुख्य रूप से घर पर देखभाल करती है, हालांकि देखभाल करने में ममता और और संतुष्टि अनुभव करतीं हैं फिर भी कभी-कभी वे भी कई बार तनाव और हताश महसूस करती हैं । समय कम मिल पाने के कारण, खुद की और अपने अन्य बच्चों की देखभाल के लिए या नौकरी/आजीविका करने में सक्षम नहीं होने की वजसे से वे कभी कभी हताश भी हो जाती हैं।

घर पर देखभाल के दौरान माता पिता को मुख्य रूप से निम्नलिखित अड़चने आती हैं :

- मनोवैज्ञानिक समस्याएं : यह मूल रूप से भविष्य में आनेवाली समस्याओं और बीमारी से सम्बंधित अनिश्चितताओं की आशंका से उत्पन्न होती हैं ।

- कई बार माता-पिता बीमारी की पुष्टि होने के बावज़ूद उस पर विश्वास नहीं करते हैं और अन्य-अन्य जगहों से राय मश्वरा करते रहते हैं, इससे इलाज़ के लिए ज़रूरी समय और पैसे दोनों की क्षति होती है।
- बच्चे को गोद में उठाने, उठा के इधर उधर ले जाना या करवट देना आदि शारीरिक कठिनाइयाँ भी आती हैं।
- देखभाल करने वाला व्यक्ति परिवार में अतिरिक्त भार का अनुभव कर सकते हैं क्योंकि देखभाल करने वाले पर मानसिक एवं शारीरिक तनाव पड़ता है। इसकी वज़ह से उसके स्वास्थ्य पर नकारात्मक प्रभाव पड़ता है जो देखभाल करने में बाधक सिद्ध होता है।
- विकलांगता को ध्यान में रखते हुए घर पर किये गए संशोधनों और रोगी को सहायता प्रदान करने के लिए संबंधित सहायक उपकरणों पर आने वाला ख़र्चा।
- स्ट्रेचिंग की कसरतें, आहार में संशोधन, सहायक उपकरणों का उचित तरीके से उपयोग और रोगी का स्वयं अपनी देखभाल करने से संबंधित आदि उचित प्रशिक्षण एवं जानकारी का अभाव।
- अवास्तविक या 'अंधा आशावाद' के बाद ना उम्मीदी और निराशा।

यह रोग पूरे परिवार को प्रभावित करता है। ड्यूचेन से प्रभावित व्यक्ति और उनके परिवारों के लिए मनोवैज्ञानिक और भावनात्मक मदद महत्वपूर्ण है। बच्चे की देखभाल कर रही टीम से यह साझा करना महत्वपूर्ण है कि आपका परिवार इस बीमारी से कैसे सामना कर रहा है, विशेषकर न्यूरोमस्कुलर टीम, ताकि आवश्यकता पड़ने पर परामर्श की व्यवस्था की जा सके। इस परामर्श में माता-पिता तनावपूर्ण परिस्थितियों से उबरने के लिए प्रशिक्षित किया जाता है। कभी कभी व्यवहार संबंधी बदलाव केलिए भी थेरेपी भी दी जाती है।

डीएमडी केयर टीम द्वारा देखभाल करने वालों को नियमित प्रशिक्षण और परामर्श सत्र देना, दूर के रिश्तेदारों और स्कूल को भी इस देखभाल में भागीदार बनाकर, ड्यूशेन से प्रभावित बच्चों की देखभाल को आसान बनाया जा सकता है।

## 4. तरुणावस्था आने पर ड्यूशेन की देखभाल में इलाज़ से सम्बंधित किन बदलावों की आवश्यकता पड़ती है?

अब देखभाल बच्चे  की नहीं वयस्क की करनी है !! बच्चे के तरुणावस्था में आते ही उसकी मानसिक एवं मानसिक जरूरतें बदल जाती हैं।  उम्र के इस पड़ाव में मरीज़ के अपने इलाज़ के सभी निर्णयों में भागीदारी बढ़ने के साथ स्वयं के देखभाल की ज़िम्मेदारी भी बढ़ जाती है।

इस  बदलाव के समय डीएमडी की देखभाल में कुछ बदलावों को शामिल करना ज़रूरी हो जाता है, जैसे : शिक्षा, व्यावसायिक प्रशिक्षण, व्यक्तिगत देखभाल, सामाजिक स्तरपर सम्बन्ध स्थापित करना आदि।

अक़्सर प्रश्न उठता है "हमें इस देखभाल में बदलाव की आवश्यकता क्यों है? जब बाल न्यूरोलॉजिस्ट अनुभवी हैं और रोगी को इतने लंबे समय से देख रहे हैं !! तो क्या वयस्क होने पर देखभाल में बदलाव की आवश्यक है?

बचपन से माता पिता द्वारा लम्बी देखभाल की वज़ह से अन्य चुनौतियां उत्पन्न हो जाती हैं।  वे बच्चे के प्रति अतिसंवेदनशील हो जाते हैं इस कारण बच्चे में स्वतंत्र व्यवहार का विकास नहीं हो पता है। बच्चा छोटी सी छोटी चीज़ के लिए माता पिता पर निर्भर रहता है परन्तु जा उम्र का यह बदलाव आता है तो उसे अपनी ज़रूरतों के बारे में आवाज़ उठाना मुश्क़िल लगता है।

इलाज़/देखभाल में उन्नत्ति के कारण डीएमडी से प्रभावित लोगों का जीवन लम्बा हो गया है इसलिए अधिकांश चिकित्सा विशेषज्ञों के लिए डीएमडी के साथ रहने वाले पुरुषों  की समूचित देखभाल एक नई चुनौती है।  वयस्कों में बीमारी के बारे में ज्ञान नियमित रूप से उपलब्ध नहीं है। अन्तः डीएमडी के वयस्कों की उचित देखभाल बहुत कठिन है क्योंकि इन रोगियों को अभी-अभी महत्त्व मिलना शुरू हुआ है और इसके लिए कुछ ही सेंटर्स/ विशेषज्ञ उपलब्ध हैं। वर्त्तमान स्वास्थ्य व्यवस्था में, विभिन्न विभागों के बीच में रोगी की देखभाल के लिए समन्वय अपनेआप में एक बड़ी चुनौती है। डीएमडी के वयस्कों में विभिन्न अंग प्रणालियों के प्रभावित होने के कारण इलाज़ और भी जटिल हो जाता है।

कुछ बदलाव बहुत महत्वपूर्ण होते हैं, माता-पिता को बच्चों को इलाज़ दौरान उनको निर्णय लेने में मदद करनी चाहिए। उनसे सम्बंधित सभी प्रकार के फैसले लेने में उनको प्रोत्साहित करना चाहिए, जैसे शिक्षा एवं व्यवसाय के विकल्प का चयन; दैनिक जीवन में गतिविधियाँ, गृह संशोधन और परिवहन के साधन। बदलाव का यह चरण वयस्क के रूप में डीएमडी बच्चे की स्वायत्तता और स्वतंत्रता को विकसित करने के बारे में है।

## 5. देखभाल के दौरान आनेवाली चुनौतियों पर किस तरह काबू पाया जा सकता है?

माता-पिता को 13 से 14 वर्ष की आयु आते आते इस बदलाव के बारे में सोचना शुरू कर देना चाहिए और मन में एक योजना बनानी शुरू कर देनी चाहिए। प्रभावित बच्चा इस भविष्य की इस योजना का हिस्सा है :

- कौन सी सेवाएं प्रदान करने की आवश्यकता है, उन्हें कौन प्रदान करेगा, और उनके लिए पैसा कहाँ से आएगा।
- रोजगार/शिक्षा के लिए क्या योजनाएं हैं (आप क्या करना चाहते हैं)
- आत्मनिर्भर जीवन (अधिक से अधिक आत्मनिर्भर रहने पर महत्त्व देना)
- वित्तीय योजनाएं दीर्घकालिक होनी चाहिएँ।

देखभाल समन्वयक या सामाजिक कार्यकर्ता यदि डीएमडी देखभाल के सेंटर पर उपलब्ध है तो माता-पिता को पूरी देखभाल करनेवाली टीम के सदस्यों के बीच समन्वय स्थापित करने में मदद मिलती है। टीम के सदस्य रोगियों को विभिन्न उपकरणों और संसाधनो के बारे में बताते हैं जो उनकी रोज़मर्रा की गतिविधियों को काफ़ी आसान कर सकते हैं। साथ ही साथ वे मरीज़ों को इनको प्राप्त करने में सहायता भी प्रदान करते हैं। ये उपकरण बढ़ती हुई बीमारी के साथ मुकाबला करने में हमारी मदद करते हैं।

**वयस्क मरीज़ में देखभाल के घटक हैं:**

**इलाज़**

- माता-पिता को चाहिए कि वे अपने बच्चे को उसके स्वास्थ्य से संबंधित मामलों में यथाशीघ्र भाग लेने के लिए प्रोत्साहित करें। ताकि जब रोगी 14-15 वर्ष की आयु तक पहुंचे तो वह अपने इलाज़ के आवश्यकताओं में रुचि लेना शुरू कर और उनकी स्वयं वकालत भी करने लगे । इससे मरीज अपने स्वास्थ्य के प्रति जिम्मेदार मेहसूस करेगा।
- संवेदनशील समस्याएं जैसे असहजता, चिंता और बीमारी के बढ़ने से उत्पन्न कठिनाइयाँ किशोरावस्था और वयस्कों में आम हैं। इन मुद्दों को डीएमडी केयर टीम के साथ साझा करना चाहिए ताकि जब भी जरूरत हो, समय पर हस्तक्षेप किया जा सके।

**शिक्षा और रोजगार**

- बच्चे को उनकी रुचियों, प्रतिभाओं और भविष्य में नौकरी के बारे में जानने के लिए प्रोत्साहित किया जाना चाहिए।
- जिम्मेदारी का भाव बचपन से ही डाला जा सकता है, यह भावना बच्चे को घर के कामों और स्वयं की देखभाल करने में मदद करती है। साथ ही बच्चा आत्मविश्वास का अनुभव करता है।
- प्रभावित लोगों को सहायक प्रौद्योगिकी (एटी) का उपयोग एवं गतिशीलता बनाए रखने में मदद करने वाले उपकरणों (व्हीलचेयर) का प्रशिक्षण देने केलिए शैक्षिक सेटिंग आदर्श वातावरण है। ये उन्हें भौतिक बाधाओं को दूर करने और दुनिया तक अपनी पहुंच बनाए रखने में मदद कर सकता है।

माता-पिता को व्यावसायिक प्रशिक्षण की सुविधा प्रदान करनी चाहिए।

**आवास और दैनिक जीवन की गतिविधियों के लिए सहायता**

परंपरागत रूप से भारत में, डीएमडी से प्रभावित बच्चों की अधिकांश व्यक्तिगत देखभाल परिवार के सदस्यों द्वारा ही होती है। हालाँकि, जैसे-जैसे ये बच्चे बड़े होते हैं, उनके देखभाल करने वाले परिजन भी उम्रदर हो जाते हैं। अक्सर वे शारीरिक देखभाल प्रदान करने में काफ़ी कठिनाई का अनुभव करते हैं।. इस सन्दर्भ में बाहर से देखभाल करने वाले व्यक्ति को नियुक्त करने की आवश्यकता पड़ सकती है।

एडीएल के साथ सहायता ली जा सकती है, ये वे उपकरण हैं जो दैनिक जीवन की गतिविधियों को करने में मदद करते हैं तथा जीवन की गुणवत्ता में उल्लेखनीय सुधार करते हैं। परन्तु रोगी और देखभाल करने वाले दोनों को ही इनके इस्तेमाल के लिए प्रशिक्षित करना आवश्यक है।

**परिवहन**

एक स्थान से दूसरे स्थान तक जासकने की क्षमता सीधे सीधे किसी भी व्यक्ति की स्वायत्तता और स्वतंत्रता को प्रभावित करता है। यही उसके रोजगारएवं शिक्षा के लिए अवसर और उसकी सामाजिक भागीदारीको भी तय करते हैं। व्यक्ति अपनी न्यूरोमस्कुलर केयर टीम के साथ सुरक्षित परिवहन विकल्प/विकल्पों पर चर्चा कर सकता है, इनमें शामिल हैं:

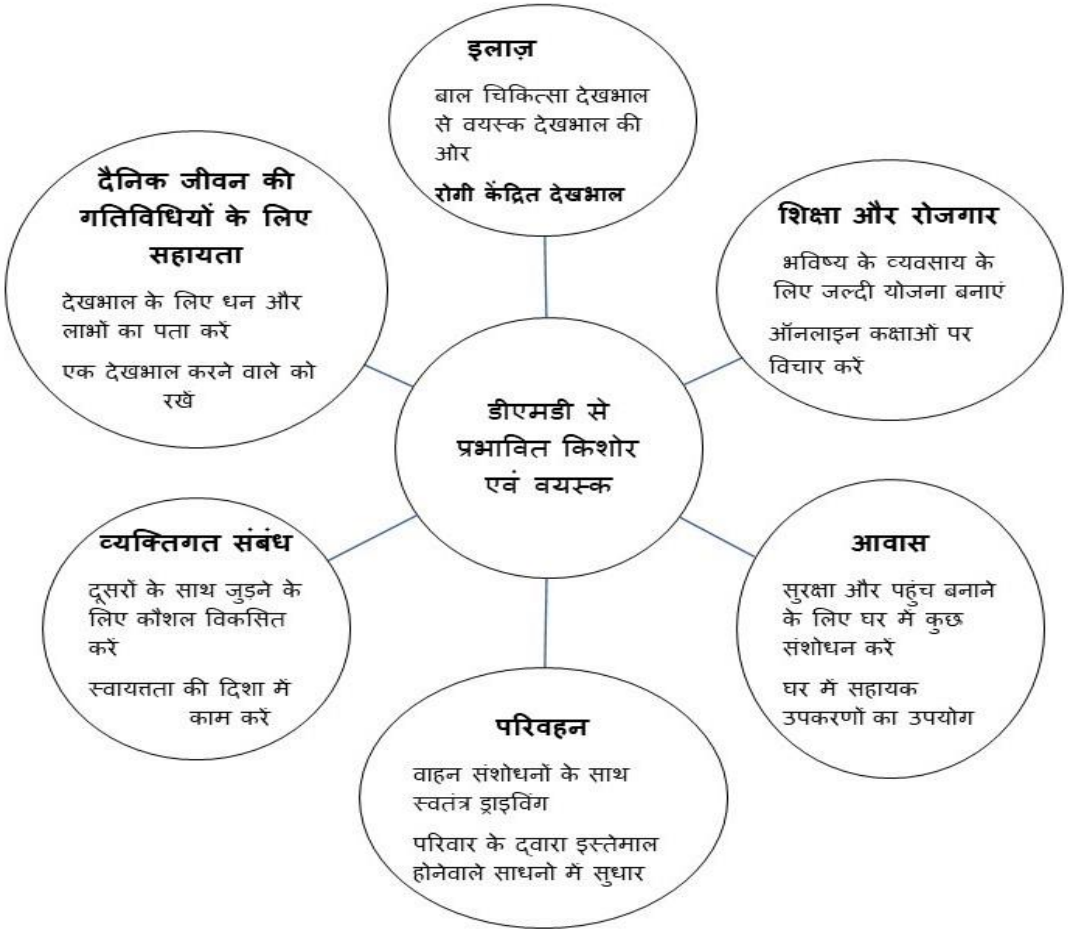
• वाहन संशोधनों के साथ स्वतंत्र ड्राइविंग

• परिवार के द्वारा इस्तेमाल होनेवाले साधनो में सुधार

• सार्वजनिक परिवहन में प्रदान की जाने वाली सुविधाएँ।

**व्यक्तिगत संबंध**

- स्वास्थ्य, मानसिक कल्याण और जीवन की गुणवत्ता सुनिश्चित करने के लिए सामाजिक संपर्क अत्यंत महत्वपूर्ण हैं।
- स्कूल, साथियों के साथ दैनिक बातचीत का अवसर प्रदान करती है। यदि किशोर या वयस्क प्रतिदिन इस तरह की बातचीत नहीं कर रहे हैं तो उन्हें साथियों, स्वयंसेवी समूहों और सामाजिक समूहों से जुड़ने और जुड़ने के लिए प्रोत्साहित किया जाना चाहिए।




किशोर एवं युवा अवस्था में डीएमडी से प्रभावित होने पर योजना बनाने के प्रमुख क्षेत्र

## खंड ङ: ड्यूशेन से प्रभावित लोगों की सहायता के लिए सहायता समूह

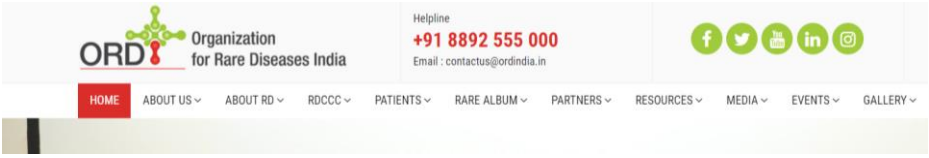
**आर्गेनाइजेशन फॉर रेयर डिज़ीज़ इंडिया ; ORDI (ओ. आर. डी. आई) (Organization for rare diseases India)**

यह एक रोगी-केंद्रित, गैर-लाभकारी संगठन है। इस का उद्देश्य भारत भर में दुर्लभ बीमारियों वाले रोगियों के स्वास्थ्य में सुधार करना है। इस की स्थापना 2014 में हुई थी।

इस संगठन का मुख्य उद्देश्य समाज की असमानताओं को कम करना और यह सुनिश्चित करना है कि दुर्लभ बीमारियों से पीड़ित लोगों को भी सभी संसाधनों का सामान्य लोगों की तरह ही उपभोग करने का अवसर मिले।

संगठन इन बिमारियों के बारे में लोगों में जागरकता पैदा करता है इनसे प्रभावित लोगों के लिए अलग से नीतियां बनाने की पैरवी करता है। नीतियाँ बन जाने पर उनको लागू करने में योगदान देता है। क्लिनिकल ट्रायल को भी बढ़ावा देता है।

ओ. आर. डी. आई. ऐसे अस्पतालों की पहचान करता है जो, रेयर डिज़ीज़ केयर कोआर्डिनेशन सेंटर (आर. डी. सी. सी.) कहलाते हैं। इन केंद्रों का उद्देश्य है, इन बीमारियों से प्रभावित मरीज़ों को चिकित्सा विशेषज्ञों, बीमारी की जाँच सुनिश्चित करने वाले केंद्रों तथा बीमारी से सम्बंधित अन्य सभी स्वास्थ्य केंद्रों से जोड़ना। मरीज़ों को इलाज़ के सभी विकल्पों, किराय पे देखभाल करने वालों की जानकारी और क्लिनिकल ट्रायल के लिए राष्ट्रीय अथवा अंतर्राष्ट्रीय पूँजी का इंतज़ाम करना भी इन्ही केंद्रों काम है। भारत का सर्वप्रथम आर. डी. सी. सी. केंद्र बंगलौर में स्थित है।




**आर. डी. सी. सी. केंद्र विभिन्न गतिविधियों का संचालन करता है जैसे:**

1. रोगियों, देखभाल करने वालों, डॉक्टरों आदि के बीच में समन्वय।
2. बीमारी से सम्बंधित जाँचों, इलाज़, देखभाल, किसी विशेष इलाजों में मरीज़ों को मदद करना।
3. विशेषज्ञों के साथ मिलकर रोगी/देखभाल करने वाले को बीमारी के बारे में जानकारी प्रदान करना।
4. डॉक्टर, छात्र और नर्सिंग स्टाफ को बीमारी से सम्बन्धित प्रशिक्षण और शिक्षा कार्यक्रम में सम्मिलित करना।
5. वित्तीय और गैर-वित्तीय सहायता

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**भारत में मांसपेशियों से सबंधित बीमारियोँ में मरीज़ों को सहायता प्रदान करने वाले संगठन :**

1. इंडियन एसोसिएशन ऑफ़ मस्कुलर डिस्ट्रॉफी
2. डिस्ट्रॉफी अन्निहिलेशन रिसर्च ट्रस्ट
3. आशा एक होप
4. मस्कुलर डिस्ट्रॉफी एसोसिएशन इंडिया

**इंडियन एसोसिएशन ऑफ़ मस्कुलर डिस्ट्रॉफी (आई. एम. डी. आर. सी.)** की स्थापना 1992 में हिमाचल प्रदेश में एक समाजसेवा सेवा संगठन ने की थी। यह एसोसिएशन भारत में मस्कुलर डिस्ट्रॉफी से पीड़ित लोगों को राहत और पुनर्वास प्रदान करता है और जन जागरूकता भी पैदा करता है। वर्तमान में यह फिजियोथेरेपी, हाइड्रोथेरेपी, आनुवंशिक और मनोवैज्ञानिक परामर्श, आहार योजना और मनोरंजक गतिविधियों के माध्यम से 25 रोगियों के लिए साइट पर आवासीय देखभाल प्रदान करता है। आर्थिक रूप से कमज़ोर रोगियों को स्वास्थ्य सम्बन्धी देखभाल और व्हील चेयर नाममात्र कीमत पर या मुफ़्त में प्रदान की जाती है।

संपर्क करने के लिए:

पता: आई एम् डी आर सी ग्राम कोठों, गवर्नमेंट सीनियर सेकेंडरी स्कूल के निकट, सोलन, हिमाचल प्रदेश 173212, फोन: 094180 54877; http://iamd.in/

### डिस्ट्रॉफी अन्निहिलेशन रिसर्च ट्रस्ट, मस्कुलर डिस्ट्रॉफी के पूर्ण रूप से समाप्त करने की आकांक्षा

डिस्ट्रॉफी अन्निहिलेशन रिसर्च ट्रस्ट [डी. ए. आर. टी. /DART] ड्यूशेन मस्कुलर डिस्ट्रॉफी (DMD) पर केंद्रित भारत की पहली अनुसंधान प्रयोगशाला है। इस की स्थापना २०१२ में श्री. आर. एस. आनंद में की थी। इसकी स्थापना उन्होंने अपने बेटे को डी एम् डी की पुष्टि (2003) होने के बाद की।

डी. ए. आर. टी. का उद्देश्य मस्कुलर डिस्ट्रॉफी से प्रभावित बच्चों और उनके परिवारों को बीमारी से लड़ने में मदद करना है।

- मस्कुलर डिस्ट्रॉफी के बारे में जानकारी प्रदान करना और बीमारी के बारे में लोगों में जागरूकता बढ़ाना।
- बीमारी से ग्रसित बच्चों को शिक्षा, रोजगार और सार्वजनिक स्थानों को मुख्य धारा में सम्मिलित करने के लिए कार्य करना।
- बच्चों और परिवारों के लिए परामर्श सहायता प्रदान करना।
- चिकित्सा सलाह और पुनर्वास सहायता
- विकलांग बच्चों की जरूरतों के बारे में जागरूकता बढ़ाने के लिए गैर सरकारी संगठनों के साथ मिलकर काम करना।

संपर्क करने के लिए:

पता: 295, 14 क्रॉस रोड, ए जी एस लेआउट, डॉलर्स कॉलोनी, आर.ऍम.वी. २ण्ड स्टेज, बेंगलुरु, कर्नाटक 560094, फोन: 080 2341 2725; वेबसाइट: http://www.dartindia.in/

### आशा एक होप *फाउंडेशन एमएनडी/एएलएस इंडिया*

आशा एक होप फाउंडेशन एक गैर-लाभकारी संगठन है जो मोटर न्यूरॉन रोग (एमएनडी) से पीड़ित लोगों और उनके परिवारों को सहयोग करता है। यह फाउंडेशन वैश्विक स्तर पर भारत का प्रतिनिधित्व करता है। कोई भी इस संस्थान से मुंबई, बैंगलोर और कोलकाता के ऑफिसेस के माध्यम से जुड़ सकता है। इस का उद्देश्य है एमएनडी को पूरी तरह से ठीक करना और इलाज योग्य बनाना।

संस्था काम करती है :

- प्रभावित व्यक्ति को दैनिक गतिविधियों को जारी रखने में उन्नत यांत्रिक सहायता और आधुनिक प्रौद्योगिकी सहायता सेवाएं प्रदान करके।
- इन बिमारियों से जुड़ी सभी प्रकार के अनुसंधानों को बढ़ावा देना।
- विभिन्न आर्थिक और सामाजिक धारणाओं वाले रोगियों और देखभाल करने वालों की जरूरतों को पहचान और उनको पूरा करना।

संपर्क करने के लिए:

पता: 402, गुरुप्रभा अपार्टमेंट, एस. बी. रोड, दादर वेस्ट, मुंबई - 400028, महाराष्ट्र, भारत; , फोन: 098197 41333, 81045464793; वेबसाइट: http://ashaekhope.com/;

Email: ashaekhope@gmail.com, contact@ashaekhope.com

### मस्कुलर डिस्ट्रॉफी एसोसिएशन इंडिया

इस संगठन की स्थापना सन २००० हुई थी। तब से यह मस्कुलर डिस्ट्रॉफी से प्रभावित लोगों की मदद कर रही है। इस संगठन में रोगी, माता-पिता, डॉक्टर, वैज्ञानिक, स्वयंसेवक और वे सभी शामिल हैं जो प्रभावितों लोगों की मदद करने में सकारात्मक योगदान दे सकते हैं।

एसोसिएशन प्रदान करता है:

- मरीजों और माता-पिता को मनोवैज्ञानिक सहायता
- बीमारी के बारे लोगों में जागरूकता फैलाना,
- इन बिमारियों से जुड़ी सभी प्रकार के अनुसंधानों को बढ़ावा देना।
- सरकारी निकायों के साथ बातचीत कर के रोगियों के स्वास्थ्य सुधार के लिए हिमायत करना।
- पुनर्वास के उपाय: जरूरतमंदों को व्हील चेयर उपलब्ध कराना
- माता-पिता और करीबी रिश्तेदारों के लिए परामर्श सत्र आयोजित करना

संपर्क करने के लिए:

पता: न्यू नंबर: 6, ओल्ड 21, फोर्थ क्रॉस स्ट्रीट, शास्त्री नगर, अड्यार, चेन्नई - 600 020 तमिलनाडु , इंडिया.; फोन: 73586 82881; वेबसाइट: http://mdindia.net/;

Email: mdindia.net@gmail.com

## खंड च: निष्कर्ष

"बेंगलुरु के करणवीर आनंद सिंह ड्यूशेन मस्कुलर डिस्ट्रॉफी (डीएमडी) से पीड़ित हैं। उन्हें तीन साल की उम्र में ही पता चल गया था की वे डीएमडी से प्रभावित हैं। वह अठारह साल की उम्र में 2019 में सीबीएसई की कक्षा 12 की परीक्षा में शामिल हुए, इतना ही नहीं उन्होंने परीक्षा में 94.8% अंक भी हासिल किए। उनके लिए यह आसान नहीं था। डीएमडी की बढ़ती हुई चुनौतियों के बावज़ूद उन्होंने परीक्षा में झंडे गाड़ दिए। अब वह अपने कॉलेज जीवन और अपने उज्जवल भविष्य की तरफ़ अग्रसर हैं।"

इस पुस्तिका को पढ़ने के बाद आपको यह स्पष्ट हो जाएगा कि यह बीमारी कैसे होती है? कैसे बढ़ती है? बीमारी के विभिन्न चरण क्या हैं ? उम्र बढ़ने के साथ साथ शरीर के अन्य कौन-कौन से अंग प्रभावित होते हैं, और इससे सम्बंधित उपचार एवं सलाहें क्या हैं? ताकि आप अपनी आवश्यकताओं को आगे बढ़ कर बच्चे की इलाज़ कर रही के टीम सामने रख सकें। ऐसा करने से आपके लिए इलाज़ की एक बेहतर योजना तैयार करने में मदद मिलेगी। आपकी यह भागीदारी डॉक्टर द्वारा दी गई सलाहों का उचित तरीक़े से पालन करने में सहायक होगी। आपकी भागीदारी आपके बच्चे और पूरे परिवार के जीवन को ख़ुशनुमा और बेहतर बनाने में योगदान देगा। अपने बच्चे के इलाज़ में आपकी अहम् भूमिका है। आपकी भागीदारी बच्चे के स्वास्थ्य सम्बन्धी रखरखाव तक ही सीमित नहीं बल्कि आपकी इच्छा अनुसार आप अपनी इलाज़ की पद्धति का चुनाव कर सकते हैं।

आप अपने बच्चे का सिर्फ इलाज़ ही अपितु चहुमुखी विकास कर सकते हैं, ताकि भविष्य में वह अधिक से अधिक समय तक आत्मनिर्भर बना रहे।

आशा है कि यह पुस्तक बच्चे और परिवार को ड्यूशेन की इस साथ यात्रा में मदद करेगी। किसी भी स्वयं सेवी समूह, न्यूरोमस्कुलर सेंटर, और अन्य ड्यूशेन सम्बन्धी समूहों, परिवारों और दोस्तों के साथ जुड़ना हमेशा अच्छा होता है, जो हर कदम पर आपकी मदद करते हैं। इस सफर में कोई अकेला नहीं है।

डॉ अमरजीत सिंह

डॉ तितिक्षा सिराड़ी

डॉ नवीन संख्यान

डॉ रेनू सुथार

डॉ शंकर प्रिंजा

डॉ पूजा कांसरा

डॉ मनीषा मालवीय (पी टी)