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Merosin Deficient Congenital Muscular Dystrophy- A Case Study

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***Abstract*-** Merosin Deficient Congenital Muscular Dystrophy (CMD) is a rare and highly severe type of muscular dystrophy. Mutations in the LAMA2 gene have been identified as the cause of congenital Merosin-deficient CMD. Purpose of the study is to highlight the importance of functional diagnosis and patient centric interventions for improving quality of life in patient with Merosin deficient congenital muscular dystrophy. This case study examines 14-year-old female presented with progressive muscle weakness and delayed motor milestones noticed in early infancy with preserved language and social milestones. With physiotherapy rehabilitation patient has shown Functional improvement on paediatric QFL inventory and FIM scores. This highlights the importance of functional diagnosis and patient centric interventions for improving quality of life in patient with Merosin deficient congenital muscular dystrophy.

***Index terms*: Merosin Deficient Congenital Muscular Dystrophy, functional diagnosis, quality of life**

I. Introduction-

Muscular dystrophy (MD) is a collective term referring to a group of inherited neuromuscular diseases. Congenital Muscular Dystrophies are with recessive autosomal inheritance, characterized by altered musculature noticed at birth[1]. From which Merosin-deficient congenital muscular dystrophy is an autosomal recessive form of muscular dystrophy showing presence or deficiency of the protein Merosin, affecting 1 in 30,000–100,000 individuals and accounts for 24–37% of all congenital muscular dystrophies. It is caused by mutations in the *LAMA2* gene, which encodes laminin-alpha 2, a component of heterotrimeric laminin-211, also known as Merosin. Laminin-211 is expressed in basement membranes of striated muscle, placental trophoblasts, Schwann cells, neuromuscular synapses and brain cells. Complete loss of laminin-211 results in disruption of structural stability and signal transduction of the extracellular matrix. This manifests in failed regeneration, fibrosis, apoptosis, chronic inflammation and muscle wasting[2].

The clinical presentation includes hypotonia, atrophy, and muscle weakness, or with minimal changes, and the presence of joint retractions with proximal regions most affected, diminished reflexes, and good cognition. Most individuals do not acquire a gait, but are able to remain seated and this is the main form of travel. The respiratory impairment is intense and this is closely related to the prognosis of the disease, as well as the spinal deformities.1,2 delayed motor development; most never achieve independent ambulation. Most patients also have periventricular white matter abnormalities on brain imaging, but mental retardation and/or seizures occur only rarely (summary by Xiong et al., 2015)[3] In addition to pharmacological treatment, these patients require a multidisciplinary team for rehabilitation. Physiotherapy is one of the resources present in this process and should be started early, as soon as it is diagnosed, even if this is in the first months of life. Helping with functional diagnosis, preventing the deformities caused by the disease, preserving the autonomy

of the patient, avoiding pain and muscle fatigue, preventing pulmonary restriction and its complication is key for this and should be more central in the treatment of patients.

To date, there is no effective pharmacological treatment available for Merosin Deficient Congenital Muscular Dystrophy patients and current treatment options focus on symptom relief.[2] The involvement of patients is also fundamental in this respect to define these measures correctly, as any treatment should eventually improve their health and quality of life. Thus, patient centric interventions and goals can be planned by physiotherapist for improving quality of life in patient with Merosin deficient congenital muscular dystrophy.

Although the effectiveness of interventions with physiotherapy on patients with neuromuscular diseases is evident today, but studies on patients with CMD are scarce and limited. Most clinical studies for muscular dystrophy utilize a standard protocol for measurement exploring pathophysiology, muscle strength and timed tasks. However, we propose that examining broader components of health as emphasized by the International Classification of Functioning, Disability and exchange of knowledge leading to a broader understanding of the pathophysiology, clinical course and novel treatment options for Merosin Deficient Congenital Muscular Dystrophy is therefore of great importance for both patients and their parents, caretakers, clinicians and researchers.

II. Objective

To highlight the purpose and effectiveness of functional diagnosis and to plan of physiotherapy intervention accordingly. To verify the effect of physiotherapy intervention on quality of life and functional independence. in addition to providing information and assisting therapists in drawing up a plan of care for patients with Merosin-deficient CMD, striving for greater independence for these individuals.

III. Methods

This work of a case study with an interventional and assessment clinical prospective. The informed consent was taken from the parent.

Case presentation -14-year-old female studying in 9th standard, born of a non consanguious marriage born at full term weighing 3 kg, it was a natural childbirth with cephalic presentation. There were no pre-natal and/or perinatal complications gives complains of progressive muscle weakness and delayed motor milestones since early infancy. With regard to her motor development, she showed developmental delay and hypotonia since his earliest days of life, she sat with support at year of 1 and without support with 3 years, when she also began to speak. The family sought a paediatrician who requested a muscle biopsy examination and investigations which showed Creatine kinase is elevated and white matter changes are detected in the brain MRI. Muscle biopsy -dystrophic changes with complete laminin α2 deficiency and thus diagnosed with Merosin deficient congenital muscular dystrophy at 7 years of age.

Physical examination shows muscle atrophy and generalized Hypotonia, Hyporeflexia, thoracolumbar scoliosis, arthrogrypotic changes in knee (FFD -15 degree) and elbow (FFD- 20 degree). The range of motion of the upper limbs and lower limbs is reduced. She presents with thoracolumbar S shaped scoliosis and hyper lordosis of the lumbar spine shortening of the flexor muscles of both hips, knee flexors and planta flexors. The sensory and cognitive functions are preserved. Speech, vision and hearing are normal. Chest expansion is reduced at axillary level. She presents no cardiac, or associated digestive impairments.

Patients strengths –She is a happy social child, Enjoys music, good receptive language skills. She wears a AFO in a house for walking and uses wheelchair outside, requires assistance to get up into standing position. At school provides the physical assistance for motor activities and after school she attends outpatient therapy. Good cognitive abilities

Contextual factors –Supportive and realistic parents and have ffinancial resources to provide services and equipment's.

Areas of concern-Requires assistance for postural change to get up from supine to sitting and sit to stand, No community ambulation independently ,Unable to do over head activities,Inability to assume standing from chair sitting, Inability to generate and sustain the muscle forces ,Inadequate postural control,Imbalance of patterns of motor activation, Secondary impairments (deformities, muscle tightness and decreased range of motion)

IV. Outcome measures

All evaluations were implemented at the beginning and at the end of the program of physiotherapy. Outcome measures used Pediatric quality of life inventory (PedsQL) and Functional independence measure (FIM)

Pediatric quality of life inventory (PedsQL)[4] -The Pediatric Quality of Life Inventory (PedsQL) is an instrument to assess HRQoL in children and adolescents aged 2 to 18. It consists of generic core and disease-specific modules . The PedsQL™ 3.0 Neuromuscular Module was developed by Dr. James W. Varni and a disease-specific module for measuring children's quality of life with Neuromuscular diseases by assessing their neuromuscular disease, communication difficulties, and family resources. This instrument is acknowledged as a validated health outcome measure in patients with NMDs (Cronbach's alpha = 0.89/parent proxy-report, 0.88/child self-report; intraclass correlation coefficients/ICC = 0.89/parent proxy-report, 0.81/child self-report)

The Functional Independence Measure (FIM)[5] – It is an instrument that was developed as a measure of disability for a variety of populations and is not specific to any diagnosis. The FIM instrument includes measures of independence for self-care, including sphincter controltransfers, locomotion, communication, and social cognition[1] Is an 18-item, seven-level, ordinal scale intended to be sensitive to changes over the course of a comprehensive inpatient medical rehabilitation program. Uses the level of assistance an individual needs to grade functional status from total independence to total assistance. The tool is used to assess a patient's level of disability as well as a change in patient status in response to rehabilitation.

V. Interventions

The child did physiotherapy thrice a week for 60 minutes per session for 8 weeks. The functional goals were established considering parents and patients goals which focused particularly on to increase use of upper extremity for self-care, school skills and to become more independent in mobility (getting up to stand and walking). Each session consisted of a period of exercises for the trunk control and for muscle activation of the lower abdominal, improving truncal control, Postural correction, breathing exercises, improving mobility of joints, decrease reliance on upper body for support while sitting and standing, Postural control in sitting and standing, Functional activity training of transfers like sit to stand with play activities.

Intervention summary: -

Goals	Interventions
Activities to improve mobility of joints and muscle flexibility	Mobilization of hip and trunk and shoulder; Passive stretching for hip flexors, knee flexors, planta flexors.
Improving truncal control	Scoliosis correction exercises, Movements to activate abdominals and lateral flexors. Core muscle activation. Postural correction exercises in front of mirror and with vestibular ball.
Increasing postural stability in sitting	Postural balance control static and dynamic exercises.
Exercises of functional training	Sit too stand with mod support, standing control with static exercises
Respiratory care	Breathing exercises aimed at greater thoracic mobility Chest expansion exercises Incentive spirometer Coughing techniques

VI. Results

Physiotherapy management patient has shown Functional improvement on Paediatric QFL inventory with scores moving from score 4 (always almost difficult) to 3(often difficult) and FIM scores from maximal assistance to moderate assistance. (Table-1)

Demonstrated increased symmetry in weight bearing and also demonstrated effective co activation of head, neck and trunk with improved overhead reach.

Table-1

Outcome measures	Pre intervention	Post intervention
Pediatric QFL inventory	Score 4 (Always Almost Difficult)	3 (Often Difficult)
FIM scores	Maximal Assistance	Moderate Assistance

VII. Discussion -

This case study describes a physiotherapy program and importance of functional diagnosis which is used to improve muscle activation, especially the functionality during daily activities for a child with Merosin-deficient congenital muscular dystrophy. The results included improvement in motor function, improvement in increased symmetry in weight bearing and also demonstrated effective co activation of head, neck and trunk with improved overhead reach.

Ansved T. eta also suggested positive effects of mild to moderate exercise programs on muscular strength without causing significant muscle damage[6]. Aksu Yıldırım S et al also suggested combined use of upper/lower extremity exercises with neck and body exercises is preferred in clinical practice due to the ability to spread force from strong muscles to weak muscles and relieve fatigue[7].

Craig Mc Donald suggested that improvement seen in muscular dystrophy is because, exercises resulted in different physiological benefits such as increased skeletal muscle mass, mitochondrial amount, and enhanced lactate degradation. Physical activity increases the production of the myokine (cytokine generated by skeletal muscle). These effects lead to increased neuromuscular transmission efficiency, endurance, strength, and functional activities[8]

Theoretically, the clinical outcomes and prognosis is poor and might reach to terminal stage in adolescence

But Recent advances says that supportive pulmonary, orthopaedic care for patients with neuromuscular disorders have significantly affected prognosis, long-term management, and rates of survival; however, these interventions can make it even more difficult to predict outcome and life expectancy in patients with congenital muscular dystrophy. Providing well-coordinated multidisciplinary care and creating strong provider–patient relationships and individualized care plans are essential throughout the changing course of the disease. Muscle impairments, Psychological well being and clinical considerations should be given attention, as it is important to have a diagnosis which not only highlights the underlying pathological condition but also the physical and functional deficit that the patient encounters

The ICF model provides areas of intervention that can enhance the participation of children whose functional well-being is at risk. Interventions that focus on multiple aspects of the child and address participation afford children the opportunity to continue to engage in age and developmentally appropriate activities, and ultimately may serve to promote overall health [9].

Physical therapist needs to identify the impairments related to functional limitation, its underlying structural source and its influence on the contextual factors (environmental and personal) and accordingly plan an intervention to maximize performance in all the domains.

VIII. Clinical Application

Importance of functional diagnosis, patient and parent centric goals for improving quality of life

X. References

1- Richard M Lovering,et al, The Muscular Dystrophies: From Genes to Therapies Phys Ther. 2005 Dec; 85(12): 1372–1388.
2- Hubert J.M. Smeets,' Merosin deficient congenital muscular dystrophy type 1A: An international workshop on the road to therapy 15–17 November 2019, Maastricht, the Netherlands Neuromuscul Disord. 2021 Jul; 31(7): 673–680. doi: 10.1016/j.nmd.2021.04.003
3- Xiong, H., Tan, D., Wang, S., Song, S., Yang, H., Gao, K., Liu, A., Jiao, H., Mao, B., Ding, J., Chang, X., Wang, J., Wu, Y., Yuan, Y., Jiang, Y., Zhang, F., Wu, H., Wu, X. Genotype/phenotype analysis in Chinese laminin-alpha-2 deficient congenital muscular dystrophy patients**.** Clin. Genet. 87: 233-243
4- https://www.pedsql.org/PedsQL-Scoring.pdf
5- Cameron MH, Monroe L. Physical Rehabilitation-E-Book: Evidence-based examination, evaluation, and intervention. Elsevier Health Sciences; 2007
6- Ansved T. Muscle training in muscular dystrophies. Acta Physiologica Scandinavica. 2001; 171: 359–66
7- Aksu Yıldırım S, Erden Z, Kılınç M. Nöromusküler hastalıklarda proprioseptif nöromusküler fasilitasyon ve ağırlık eğitiminin etkilerinin karşılaştırılması. Fizyoterapi Rehabilitasyon. 2007; 18(2): 65–71.
8- Craig M McDonald, Physical Activity, Health Impairments, and Disability in Neuromuscular DiseaseAmerican journal of physical medicine & rehabilitation / Association of Academic Physiatrists 81(11 Suppl):S108-20 DOI:10.1097/01.PHM.0000029767.43578.3C
9- Roxanna M Bendixen, Participation and quality of life in children with Duchenne muscular dystrophy using the International Classification of Functioning, Disability, and Health Health and Quality of Life Outcomes volume 10, Article number: 43 (2012)
10- Ching chang et al Consensus Statement on Standard of Care for Congenital Muscular Dystrophies, November 15 ,2010 PubMedhttps://doi.org/10.1177/0883073810381924
11- Cinthya Patrícia de Albuquerque Santos et al Aquatic physical therapy in the treatment of a child with merosin-deficient congenital muscular dystrophy: case report, DOI: 10.5935/0104-7795.20160020
12- MK Thong, Congenital muscular dystrophy due to laminin α2 (merosin) deficiency (MDC1A) in an ethnic Malay girl Neurology Asia 2017; 22(2): 155 – 159