Merosin Deficient Congenital Muscular Dystrophy: A Case Study

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ABSTRACT

Introduction: Congenital muscular dystrophies are a group of genetic neuromuscular disorders with muscle weakness presenting at birth or early infancy. 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.

Methodology: A case study of 14-year-old female presented with progressive muscle weakness and delayed motor milestones noticed in early infancy with preserved language and social milestones. Examination revealed generalised hypotonia and hyporeflexia, thoracolumbar scoliosis, arthrogrypotic changes are seen in knee and elbow but good cognitive function. Creatine kinase is elevated and white matter changes are detected in the brain MRI. Muscle biopsy showed dystrophic changes with complete laminin α2 deficiency by immunohistochemistry. According to studies the clinical outcomes and prognosis is poor and might reach to terminal stage by 19-20 years of age. With physiotherapy rehabilitation patient has shown Functional improvement on paediatric QFL inventory and FIM scores Muscle impairments, Psychological wellbeing 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. Highlighting the importance of functional diagnosis and patient centric interventions for improving quality of life in patient with Merosin deficient congenital muscular dystrophy.

Conclusion: 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.

Keywords: Merosin Deficient Congenital Muscular Dystrophy, Functional diagnosis, Quality of life

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