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Case Report

Myopathic Disorder in an 8-Year-Old Male with Familial Predisposition: A Case Report

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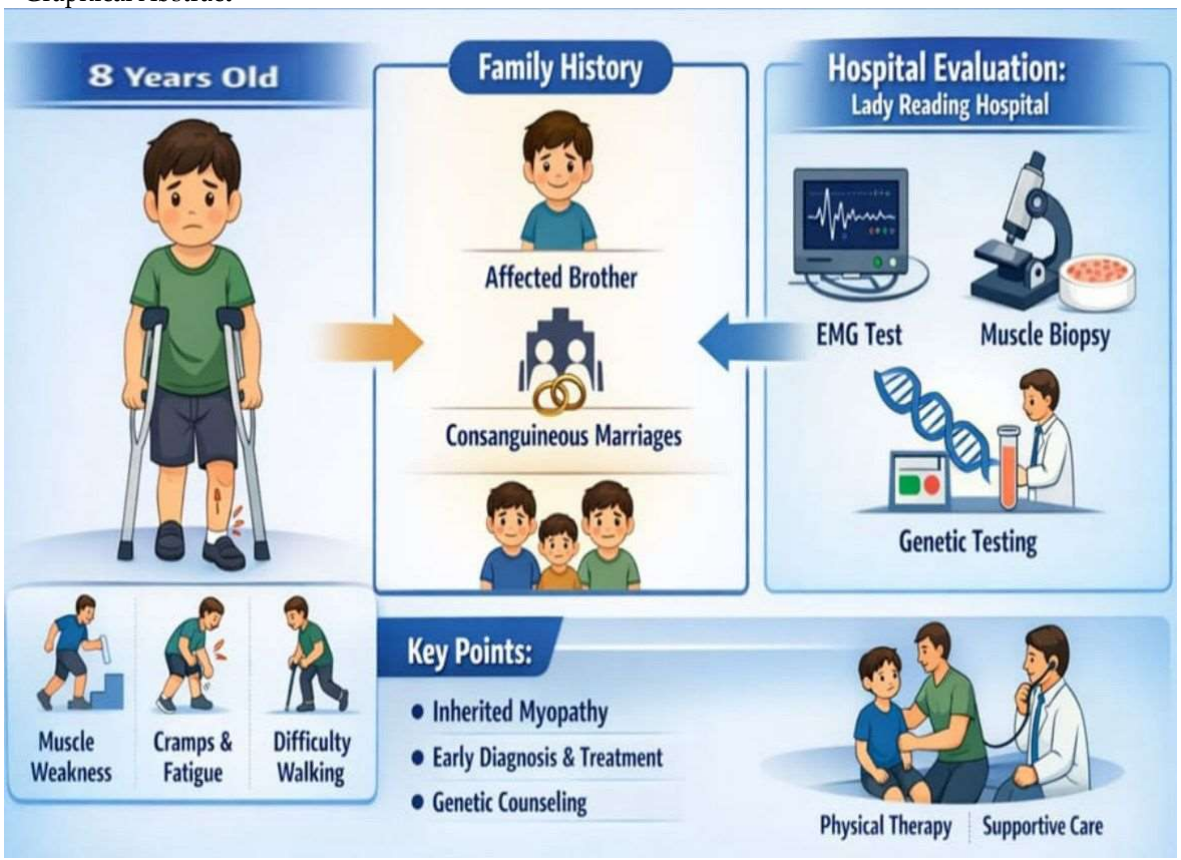
Graphical Abstract



Abstract: Myopathic disorders in children are a heterogeneous group of neuromuscular conditions characterized by progressive muscle weakness, fatigue, and impaired motor function. Early recognition is crucial for diagnosis and management. We report the case of an 8-year-old boy from Charsadda presenting with progressive weakness in the muscles of the hands and legs, accompanied by fatigue, cramps, spasms, and movement difficulties. The patient has a positive family history, including an affected brother and consanguineous marriages within the family. The patient was evaluated at Lady Reading Hospital (LRH), highlighting the importance of clinical recognition, family screening, and early supportive care in pediatric myopathies.

Keywords: Myopathy, childhood myopathy, muscle weakness, cramps, fatigue, familial myopathy

Case Presentation

An 8-year-old male residing on Charsadda Road, presented to LRH with progressive muscle weakness over the past eight months. The patient's parents reported that he initially had trouble in performing routine activities, such as running or climbing stairs, which gradually progressed to involve both the upper and lower limbs. He also experienced fatigue, muscle cramps, spasms, and difficulties with movement, affecting his daily activities and school performance. Family history was significant, with one brother also affected by a similar condition and a background of consanguineous marriages, raising suspicion for a hereditary myopathic disorder. The patient has three sisters and four brothers. There was no reported history of acute illness, trauma, or developmental regression prior to symptom onset.

Physical examination revealed weakness predominantly in the proximal muscles of both upper and lower limbs. There were signs of muscle cramps and mild difficulty with coordinated movements. Laboratory and neurophysiological investigations were recommended to confirm the diagnosis and characterize the myopathic disorder, considering the familial pattern and clinical features.

Discussion

Pediatric myopathic disorders are characterized by progressive muscle weakness and impaired motor function (Cassandrini et al., 2017). The presence of fatigue, cramps, and movement difficulties in this patient are typical features of inherited or congenital myopathies (Petty et al., 2003). Familial clustering, especially in the context of consanguineous marriages, suggests a possible autosomal recessive inheritance pattern, which is common in certain regions. Early diagnosis is critical for guiding management, which may include physical therapy, supportive care, and monitoring for complications such as contractures or respiratory involvement (King et al., 2013). Genetic counseling is particularly important in families with multiple affected members to inform future reproductive decisions. Evaluations at tertiary centers, such as LRH, allow for specialized investigations, including electromyography, muscle biopsy, and genetic testing, which can confirm the diagnosis and guide prognosis. This case underscores the need for heightened clinical awareness of pediatric myopathies, particularly in regions with high rates of consanguinity, and highlights the importance of family history in early identification and intervention.

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