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FAMILIAL BETHLEM MYOPATHY IN A PEDIATRIC PATIENT: A CASE REPORT

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Introduction. Bethlem myopathy is a rare subtype of congenital muscular dystrophy characterized by progressive muscle weakness and joint contractures. It is a milder phenotype of the spectrum of collagen VI-related myopathies, which are caused by mutations in genes encoding collagen VI proteins, namely *COL6A1*, *COL6A2*, or *COL6A3*. Clinical manifestation may begin at any time from the prenatal period to late adulthood, presenting a range of symptoms from mild muscle weakness to severe disability impacting the individual's mobility and quality of life.

Case description. A 13-year-old patient presented with progressive weakness affecting both arms and legs, accompanied by an inability to stand flat-footed, walk on heels, squat and rise, and experiencing calf pain after walking longer distances. Additionally, the patient struggled with lifting and carrying various objects. Since the age of 2, an abnormal gait characterized by toe walking had been observed. At 11 years old, the patient underwent Achilles tendon surgery. Physical examination revealed scapular winging, elbow contractures, muscle atrophy in the arms and legs, and follicular hyperkeratosis in the humeral and thigh regions. Elevated levels of creatine kinase were noted, along with myopathic changes evident on electroneuromyography. At 15 years old, muscle biopsy demonstrated myopathic changes without dystrophinopathy-related immunohistochemical alterations.

At the age of 20, next generation sequencing analysis revealed a heterozygous variant c.1053+1G>A; p.(?) of *COL6A2* gene (NM_001849.4) which is reported as pathogenic.

The patient's mother, her mother, brother, and niece reported similar symptoms. Segregation analysis in affected family members revealed the same variant of *COL6A2* gene.

Conclusions. We present a rare case of familial Bethlem myopathy with symptoms evident from early childhood. Understanding the clinical presentation and progression of Bethlem myopathy in children is crucial for early detection, effective management, and genetic counseling. We highlight the importance of a multidisciplinary approach involving neurologists, orthopedist, geneticists, and rehabilitation specialists in providing care for patients with Bethlem myopathy, particularly in pediatric cases where early intervention can significantly impact quality of life.

Keywords. Bethlem myopathy, Collagen type VI-related myopathy, *COL6A2*, Joint contractures, Proximal muscle weakness.