COLLAGEN VI MUTATION RELATED DISORDER BETHLEM MYOPATHY: A RETROSPECTIVE COHORT STUDY

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Background: Collagen VI related myopathies with a mutation in the COL6 genes manifest as a phenotypic continuum of rare disorders ranging from mild Bethlem myopathy to severe Ullrich congenital muscular dystrophy, characterized by early onset muscle weakness, proximal joint contractures, distal joint laxity, delayed motor milestones and a variable ability for ambulation.

Materials/Methods: A retrospective cohort study was conducted with 23 patients from two pediatric institutions with a confirmed diagnosis of Bethlem myopathy. Charts were reviewed for demographic data, age of disease presentation and diagnosis, COL6 genotype, genetic diagnosis method, ambulation status, physical limitations, musculoskeletal abnormalities, other systemic co-morbidities, advanced imaging and screening diagnostics, previous surgical interventions, and progression of disease.

Results: The average age of our population was 11.65 years-old (3-19 years-old). Initial presentation of symptoms was 4.18 years-old, diagnosis was at an average age of 8.22 years. 73.9% of patients required assistive mobility devices (orthoses, braces, walkers, or wheelchairs). 30.4% had scoliosis and 39.1% developed acetabular dysplasia. 96.6% of patients developed foot and ankle deformities such as hindfoot varus, equinovarus, cavus, and pes planus. 86.9% of patients had muscle contractures, the most common locations being the ankle (55%) and elbow (35%). Only 21.7% of patients had a chief presenting complaint of pain. 65.2% of patients underwent at least one surgical procedure, the most common being a muscle biopsy (73.3%). 33.3% of patients underwent a posteromedial-lateral equinovarus releases and 20% required an Achilles tendon lengthening.

Conclusions: Our study population demonstrated and characterized progression of motor impairments requiring assisted devices even when presenting with only mild motor function issues initially. The most common physical manifestations requiring surgical treatment was foot and ankle deformities, whereas surgical intervention to correct scoliosis or acetabular dysplasia was relatively rare. Patients and families need to be counseled on possible clinical course of this disorder and requirement for ambulation aids.

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