CASE STUDY

96. Genetically Confirmed Schwartz–Jampel Syndrome: An Ultra-Rare Case of Congenital Myotonia with Osteochondrodysplasia from India

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Background: Schwartz–Jampel syndrome is a rare autosomal recessive disorder caused by mutations in the HSPG2 gene, leading to perlecan deficiency. Perlecan is a heparan sulfate proteoglycan critical for cartilage integrity, basement membrane stability, and neuromuscular transmission. The disorder manifests with skeletal dysplasia and persistent myotonia, while cognition and lifespan are usually preserved. Only about 150 cases have been reported worldwide. We report a four year old child with severe osteochondrodysplasia and generalized myotonia, highlighting the therapeutic benefit of botulinum toxin A in managing both blepharospasm and perioral myotonia.

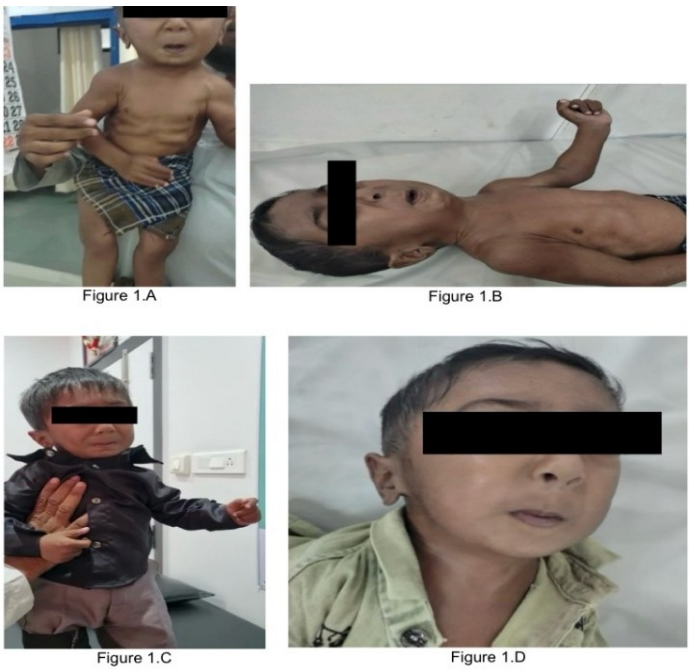
The Case: A male neonate, born full term to consanguineous parents, was referred on day one of life with suspected skeletal dysplasia. Birth weight was 3.0 kg (−0 to +1 SD), with macrocephaly (>+2 SD) and short length (<−3 SD). Dysmorphic features included a narrow thorax, bent femora and tibiae, retrognathia, long philtrum, and ichthyosis. Infantogram revealed rhizomesomelia with irregular metaphyses of long bones.

At four years, he presented with gross motor delay due to sustained skeletal muscle contractions, failure to thrive, and gait difficulty. Anthropometry remained <−3 SD for height, weight, and head circumference. Examination showed generalized myotonia of facial and skeletal muscles, pursed lips, micrognathia, thoracic kyphosis, hip abduction deformities, and hypertrophied muscles. Neurological assessment revealed hypertonia with areflexia, while speech and cognition were preserved. The differential diagnosis includes muscular dystrophies, skeletal dysplasias, myotonic disorders, congenital myopathies, and cramp–stiffness syndromes such as stiff person syndrome and Isaac's syndrome. However, the coexistence of osteochondrodysplasia with persistent myotonia, supported by whole exome sequencing which revealed a homozygous likely pathogenic variant in the HSPG2 gene (c.5014+2T>C), confirmed the diagnosis. Parental carrier testing showed that both parents were heterozygous carriers of the mutation.

Management included carbamazepine and regular physiotherapy, which provided partial benefit. To address disabling blepharospasm and perioral myotonia that impaired feeding, botulinum toxin A injections were administered to the orbicularis oculi and orbicularis oris. This improved both vision and feeding ability, demonstrating an effective extension of botulinum toxin use in this condition. Parents were counseled regarding malignant hyperthermia risk with anesthesia and advised genetic counseling for future pregnancies.

Conclusion: Schwartz–Jampel syndrome should be suspected when skeletal dysplasia coexists with generalized myotonia, particularly in children of consanguineous unions. Supportive management includes physiotherapy and anticonvulsants, and botulinum toxin A, though primarily used for blepharospasm, can also enhance feeding through perioral muscle relaxation. Prognosis is generally favorable, as symptoms stabilize after adolescence and cognition remains intact. However, morbidity arises from progressive contractures and skeletal dysplasia, speech and feeding difficulties from facial and oropharyngeal myotonia, restrictive lung disease due to a narrow thorax, and the psychosocial burden of visible deformities with reduced mobility. Early recognition, multidisciplinary care, and family counseling, including genetic counseling for future pregnancies, are essential to optimize outcomes in this rare disorder.

Figure 1. Clinical Photographs.



Legend: Clinical Photographs of the patient. (A, B) At presentation, showing myotonic facies with pursed lips and marked muscle wasting. (C, D) Four months following treatment, showing improved facies and nutritional status.

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