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Case Report OMICS International

Shwartz Jampel Syndrome: Rarest of the Rarest Case/A Rare Cause of Blepharospasm

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Abstract

We report a case of 4 year old girl presenting with the inability to open her left eye, which was identified as blepharospasm. She was also found to have pursed lips, micrognathia and puckered facial appearance, all of which are characteristic of Schwartz jample syndrome. Schwartz jample syndrome is a rare disorder that harbors a mutation in the HSPG gene on chromosome 1.

Keywords: Blepharospasm; Myotonia; Facial dysmorphism; Shwartz jampel syndrome

Introduction

Schwartz Jampel Syndrome (SJS), also known as chondrodystrophic myotonia is a very rare autosomal recessive congenital disorder associated with generalised myotonic myopathy, joint contractures, bone dysplasia and blepharophimosis. The prevalence rate has been reported to be less than 1 per million, with total reported cases found to be about 129 [1]. The primary cause was attributed to a mutation in the HSPG2 gene in the 1p34-p36 region on chromosome 1. The encoded protein, Perlecan, is a multifunctional proteoglycan present in basement membranes of cartilage and muscles [2,3]. SJS has been classified into two types – Type 1 and Type 2. Type 1 has two recognized subtypes - 1A and 1B, both manifest in childhood but differ in the intensity of dysplasia which is moderate in 1A and severe in 1B. On the contrary, Type 2 displays the most severe form of dysplasia occurring in neonatal period with extremely high mortality [4]. The majority of signs and symptoms are observed in the second or third year of life [5]. Several authors have reported that SJS is a progressive disorder, with musculoskeletal impairments peaking at adolescence but the life expectancy of such individuals remains normal. We describe a 4 year old girl with SJS, for the purpose of helping clinicians in better understanding of this rare musculoskeletal disorder and early recognition in children.

Case Presentation

A 4 year old girl of a third degree consanguineous marriage presented with complaints of inability to open her left eye since age 3. She had an unremarkable birth and a normal antenatal developmental history. Her elder siblings share similar phenotypic features with less severity (Figure 1).

On examination, she was found to have a unilateral narrowed left sided palpebral fissure secondary to blepharospasm. She also had bilateral tarsal epicanthal folds and a pseudo-exotropia (Figure 2). Her Best Corrected Visual Acuity was 6/6 in both eyes with an otherwise normal anterior and posterior segments in both eyes.

She had a 'fixed' dysmorphic facies with narrowed palpebral fissure, blepharospasm, tarsal epicanthal folds, pursed lips, low set ears and prominent mental creases (Figure 2). Systemic examination revealed a short neck, short stature and myotonia with hypertrophy of the limb muscles (Figure 3).

No joint contractures, kyphosis or mental retardation were identified in the child.

On laboratory investigations, raised serum aldolase and creatinine levels were identified. Electrophysiological tests were conducted with normal nerve conduction tests but significant repetitive discharges on electromyography from the biceps, mentalis and orbicularis oculi muscles were observed (Figure 4).

Considering all these findings, a clinical diagnosis of Shwartz Jampel Syndrome was made. The child was referred to a pediatric orthopedician for planned muscle relaxation exercises. In view of the mild and unilateral blepharospasm, no medical intervention at present was decided for the patient. However, future need for Botulinum Toxin A injections to relieve severe blepharospasm was discussed with the patient's family with advice to review routinely at 6 monthly intervals.

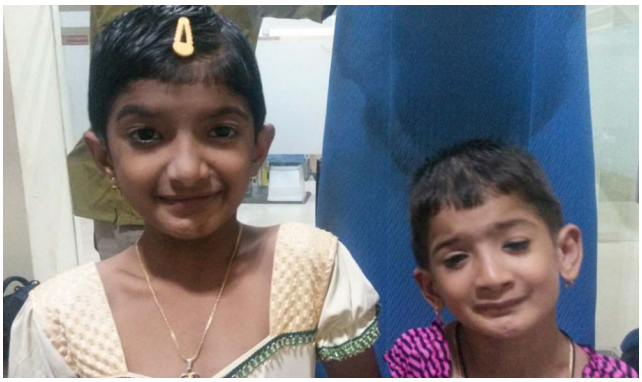

Figure 1: Characteristic 'fixed' facies in both siblings.

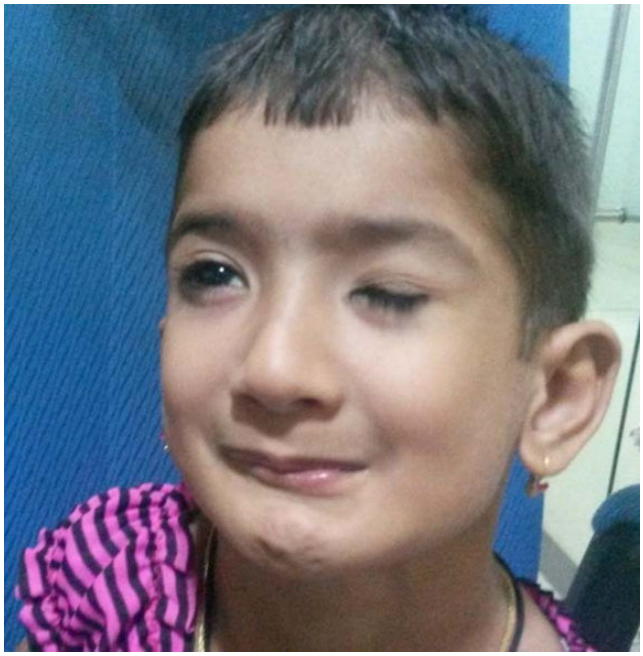

Figure 2: Ocular and facial features.

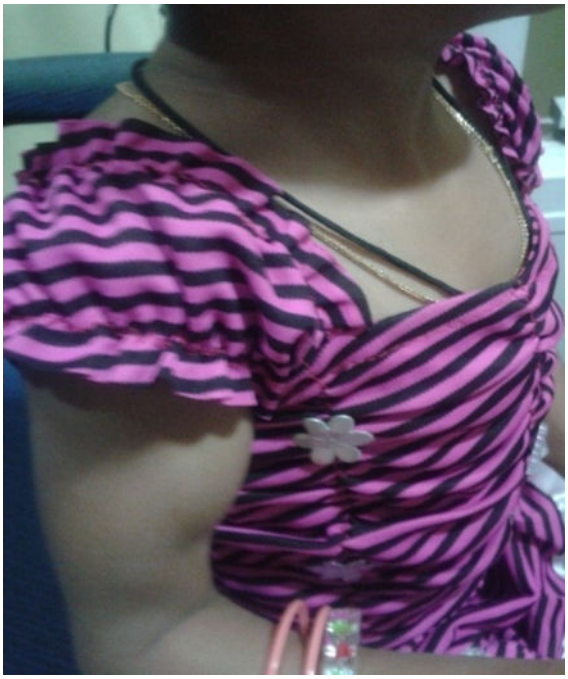

Figure 3: Limb and trunk abnormalities.

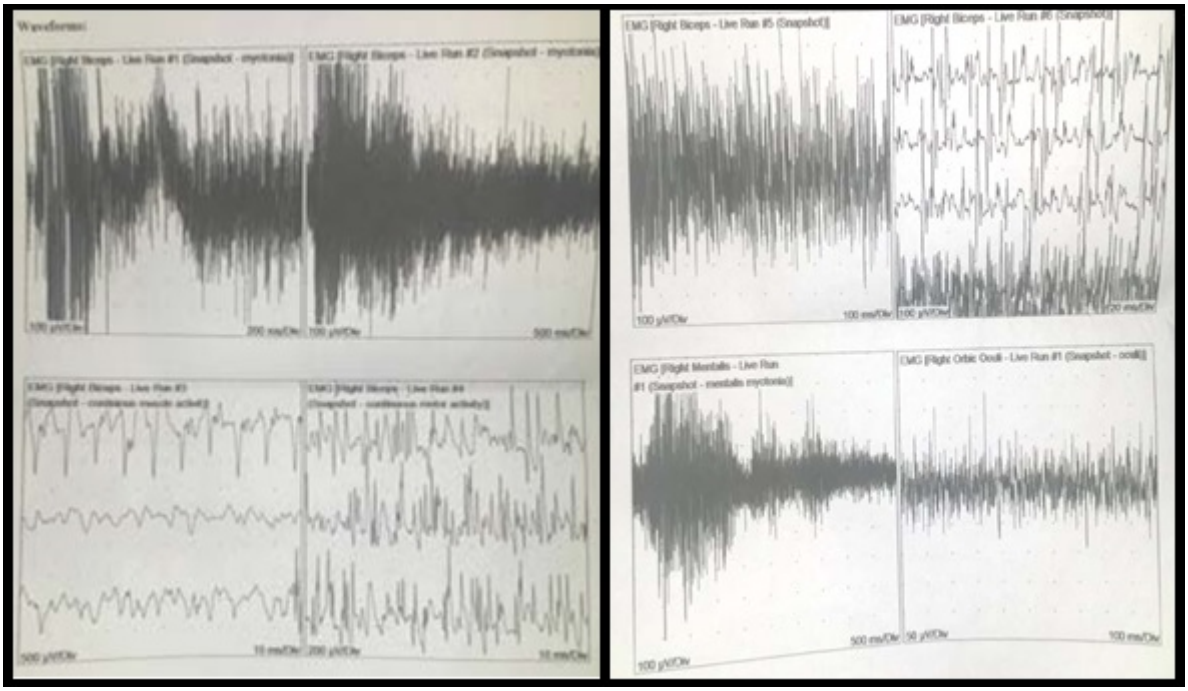

Figure 4: Electrophysiological tests showing repetitive discharges from biceps, mentalis and orbicularis muscles.

Discussion

A diagnosis of SJS can be arrived on the presence of distinct electrophysiological and clinical features. The characteristic phenotypic features include short stature, mask like facies, pinched face, long philtrum, low set ears, high pitched voice, kyphoscoliosis, bowing of long bones, joint contractures, muscular hypertrophy, increased muscle tone and blepharophimosis [6-8]. Eye abnormalities like small

palpebral fissures, strabismus, nystagmus, microphthalmus, inverse epicanthus, ptosis and hypermetropia have been reported in literature [9]. Skeletal abnormalities occur as a direct result of altered perlecan concentration in the basement membranes of muscles and cartilage. As perlecan is known to strengthen the extracellular matrix structure, its absence or deficient levels lead to impairment of cartilage matrix resulting in skeletal dimorphisms [3]. Explanations for the mechanism of abnormal neurological findings and electrical hyper-excitability in the current literature are conflicting. Studies have suggested that perlecan acts as an acceptor site for Acetylcholine esterase (AChE) molecules (responsible for elimination of Acetylcholine) at the Neuromuscular Junctions (NMJ) [3,10]. Truncated levels of perlecan will decrease the clustering of AChE which leads to inadequate elimination of Acetylcholine at the NMJ, resulting in hyperexcitability of muscles and myotonia. Conversely, it have been also suggested that the origin of electrical imbalance could be at the neural level [11]. Myotonia could be attributed to defective Na+ channel mechanism, where the synchronised Na+ channel opening is delayed, continuing well after surface membrane repolarisation has occurred. The differential diagnosis of SJS include disorders like Freeman-Sheldon syndrome, Marden-Walker Syndrome, Seckel syndrome, familial myotonia, congenital myotonic dystrophy, myotonia congenita, Duchenne dystrophy, Morquio's syndrome and various other contractual disorders [12,13].

Management of SJS is mostly symptomatic and may rarely involve surgical intervention. The primary goal of treatment is to decrease the abnormal muscle activity which can be achieved through anticonvulsants (phenytoin, carbamazepine) and antiarrhythmic drugs (procainamide). Physiotherapy, gradual stretching, warming exercise and massage may help to relieve muscular contractures. In cases of crippling and immobility, orthopaedic surgical intervention may be required. Ophthalmological abnormalities and blepharospasm can be dealt surgically by myectomy, levator recession, lateral canthopexy, levator tucking, partial corrugator muscle excision and Botulinum toxin-A injection in the orbicularis muscle.

Patients of SJS often need additional psychological and psychosocial support due to their unique physical appearance. There is a risk of iatrogenic addiction to muscle relaxants which require counselling on drug related side effects. The therapies of SJS are targeted towards specific symptoms and ophthalmologists are required to diagnose and treat ocular issues. As different specialities are involved, ophthalmologists need to work as a team with other associated professionals of different specialities to provide comprehensive treatment.

Conclusion

We presented this rare case to help clinicians easily identify Schwartz Jampel syndrome in neonates and children. The characteristic phenotypic skeletal deformities, facial dysmorphisim with blepharospasm and myotonia are highly specific for the diagnosis of SJS. Electromyography and genetic analysis will help in the confirmation of diagnosis. Prenatal ultrasound evaluation can also aid in early identification of SJS.

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