

Case Report

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Schwartz Jampel Syndrome- A Case Report



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Abstract

We describe a case of Schwartz-Jampel syndrome with the objective of highlighting this rare presentation of blepharophimosis. The syndrome is diagnosed by the typical dysmorphic features

Keywords: Blepharophimosis; Schwartz-jampel; Syndrome; Myotonia

Introduction

Blepharophimosis is a general diminution of palpebral fissure in all its dimensions. The lids usually show ptosis, dystopia canthorum, lateral displacement of the lateral puncti, or abnormalities of the lashes such as ditichiasis or misdirected and stiff lashes. The other ocular defects associated with congenital blepharophimosis include strabismus, nystagmus, amblyopia, microphthalmus, anophthalmus, epicanthus inversus, microcornea and hypermetropia [1,2]. Schwartz-Jampel syndrome, an autosomal recessively transmitted disease, is a rare presentation of blepharophimosis.

Case History

A 2 year old male child, having dysmorphic features was referred from the department of Pediatrics for Ophthalmic assessment. The child was the first born of healthy non-consanguineous parents after an uneventful pregnancy. His mental and motor development was normal and he acquired independent walking at 16 months. Fine pincer grasp developed by 9 months of age. At the age of 2 years the child could talk only two words with meaning. Social development of the child was poor because of his abnormal appearance and poor language development. The parents noted the abnormal facial expression at the age of 18 months.

On examination the child had a short stature. The head posture was normal. Forehead did not show excessive wrinkling. The child had blepharophimosis (Figure 1). Lid crease was present. The child also had hypertrichosis. The globe examination was normal. The extraocular movements were normal. There was no refractive error. Fundus was normal. The child demonstrated pursing of lips giving him a 'whistling face' appearance and restricting his mouth opening (Figure 2). The shape of the chest

was abnormal with sternal protrusion and sub-costal retraction (Figure 3). There was stiffness of his abdominal wall. The upper and lower limbs demonstrated hypertonia. The deep tendon reflexes were exaggerated. He had a waddling gait. The child had a high pitched voice (Figure 4).



Figure 1: Rigid abdominal wall.



Figure 2: Whistling face look .



Figure 3: Blepharophimosis.



Figure 4: Generalised stiffness over trunk and abdominal wall. Hypertrophy of Deltoid and thigh muscles.

Discussion

Schwartz-Jampel syndrome is a rare autosomal recessively transmitted disease, characterized by generalized myotonic myopathy, typical facial features, skeletal dysplasia, contracture of joints, growth retardation and bone maturation delay [3]. However a few cases showing dominant inheritance have also been reported. It is classified into 3 types based on age and severity

1. Type 1A
2. Type 1B
3. Type 2

Type 1A

The type 1A disease is diagnosed in mid-childhood with recognition of myotonic facies with convex profile, short palpebral fissure, telecanthus, dimpling or quivering of the chin, prominent eyebrows, low hairline, low-set ears, flat base of the nose, micrognathia, microstomia, sometimes high-arched palate. The child exhibits progressive myotonia, muscle wasting and orthopaedic problems with decreased linear growth myotonia plateaus by mid childhood. Additional findings reported in a few cases are myopia, hypertrichosis, and strabismus. The continuous myotonia is probably responsible for both muscular hypertrophy and peculiar facial appearance.

Type 1B

Type 1B is more severe than 1A, Bone dysplasia is present at birth. Long bones are shortened, femurs are dumbbell shaped. Bone epiphyses are large and vertebral bodies are flat.

Type 2

Type 2 disease is more severe. Onset is neonatal, there is short limb dysplasia and long bones are bowed. Early death is frequent [4].

The diagnosis is predominantly on the basis of the typical dysmorphic facies [5]. EMG showing continuous discharges further supports the diagnosis. The gene defect in SJS type 1 is located in the 1p34-p36 of chromosome 1, whereas it is different in type 2 [6,7]. Perlecan the major proteoglycan of basement membranes is altered in patients with Schwartz-jampel syndrome [8]. However, a significant amount of molecular heterogeneity exists, genomically and proteomically, within SJS type 1. Currently no known correlation exists between the specific mutations found and the specific features of a given case However, the new mutations found by Stum et al. In 2006 have been discovered so recently that not enough time has elapsed to explore such possibilities. The new findings should be important tools to help find correlations among genetic variants, perlecan forms and levels, and clinical subtypes. Other facts yet unknown also may influence the severity and the specific characteristics of the disease [5]. The genetic tests for perlecan gene are not easily available in the commercial laboratories.

The child was diagnosed as having type 1A type of Schwartz-Jampel syndrome since the typical facial features became manifest at the age of 18 months. The old pictures of the child taken on his first birthday showed normal facial features. Medications that have been found useful in myotonic disorders such as phenytoin and carbamazepine may help to reduce the abnormal muscle activity. Warm baths are helpful in reducing stiffness. Botox injections are reportedly found useful to relieve blepharospasm.

Patients are generally treated with Carbamazepine 20-30mg/kg body weight and most of them show improvement. Carbamazepine probably works by inhibiting neuronal sodium channels and may have direct effects on neurotransmitter systems. Orbicularis oculi myectomy, levator aponeurosis resection and lateral canthopexy are some surgical procedures which may be tried if the response to carbamazepine or botox is not adequate. The parents of the child were educated regarding the genetic nature of the disease and were referred to the geneticist. This particular child has not reported for follow-up as he belongs to a remote village far from our hospital and is probably reporting for follow-up at a nearby city.

Conclusion

Schwartz-Jampel syndrome is a rare cause of blepharophimosis. The condition can be managed with medications in most of the

cases. Surgery may be required if the condition does not improve with drugs.

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