CASE REPORT

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Epibulbar Dermoid and Type 1 Duane Syndrome Coexistence in a Patient with Goldenhar Syndrome

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ABSTRACT Duane syndrome results from a dysplastic abducens motor neurons with aberrant innervations of the lateral rectus muscle by the oculomotor nerve. It presents with a variety of clinical features including diplopia and amblyopia. Duane syndrome is a congenital strabismus syndrome occurring in isolated or syndromic forms. While isolated Duane syndrome cases constitute 70% of all Duane syndrome cases, congenital anomalies such as Goldenhar syndrome, Okihiro syndrome, Wildervanck syndrome and Moebius syndrome accompanied with Duane syndrome in 30% of all cases. Goldenhar syndrome is a rare congenital anomaly involving defective development of the structures derived from the first and second branchial arch. It is also known as the oculoauriculovertebral dysplasia due to the association of eye anomalies with vertebral or ear anomalies. Here we reported a case of GS in a 12 year old male patient with the features of accessory tragus, preauricular apendages and epibulbar dermoid with coexistence of Type I DS.

Keywords: Duane syndrome; goldenhar syndrome; epibulbar dermoid; oculovertebral dysplasia

Goldenhar Syndrome (GS) is a rarely seen congenital anomaly comprising with the clinical features of epibulbar or limbal dermoids, accessory tragus or auricular appendages and vertebral anomalies.[1] Specific forms of ocular motor disturbances, such as Duane Syndrome (DS) or double elevator palsy might be rarely seen in this syndrome.[1,2]

Duane Syndrome is a congenital ocular motility disorder characterized by limited abduction and/or limited adduction. Abnormal development of the 6th cranial nerve may be responsible for this disorder but the exact cause of DS is still unknown. A mutation in *CHN1* gene causes the disorder in a small group of patients. The *CHN1* gene plays an important role in the early development of the nerves by instructing a protein which is critical for the management of the nerves responsible for the extraocular muscles innervation. Mutations of the *CHN1* gene leads to destructive nerve development and restricted eye movement.[3]

Herein reported is a case of GS with the features of preauricular apendages and epibulbar dermoid with coexistence of Type I DS. By presenting this case, we also gained an opportunity to review the ocular aspects of this syndrome.

CASE REPORT

A 12 year-old male patient admitted with complaint of left sided swelling of bulbar conjunctiva since birth. The child was born of a full-term normal

delivery from a non consanguineous marriage and there was no history of any maternal illness or drug use during pregnancy. Written informed consent was obtained from the patient and his family for the presentation.

Right sided preauricular appendages along the line joining the tragus and the angle of the mouth and lateral cleft-like extension of right corner of the mouth was noted during examination (Figures 1, 2). The patient had undergone surgery for right sided accessory tragus 4 years ago and the remnant scar tissue was seen on cutaneous examination (Figure 1).

Ocular examination demonstrated the presence of yellowish, hairy, soft dermoid with 6 mm x10 mm size on the lower temporal aspect of the left bulbar conjunctiva (Figure 3). Extra ocular movements of the left eye showed limitation of abduction and normal adduction. The left eye demonstrated fissure narrowing at adduction and fissure widening at abduction (Figure 4). The right eye did not constitute any ocular motility disorder. Hertel exophthalmometry reading was 16.8 mm at primary position, 17 mm at abduction and 17 mm at adduction on the right eye. Hertel exophthalmometry reading was 16.9 mm at primary position, 17.1 mm at abduction and 14.8 mm at adduction on the left eye. Refractive status, fundus examination and intraocular pressure of the both eyes were normal.

The patient was underwent surgery for excision of the mass. It was observed that the posterior extension of the fatty mass has no adherence with the lateral rectus muscle. Histological examination of the mass confirmed the diagnosis as an epibulbar dermoid.

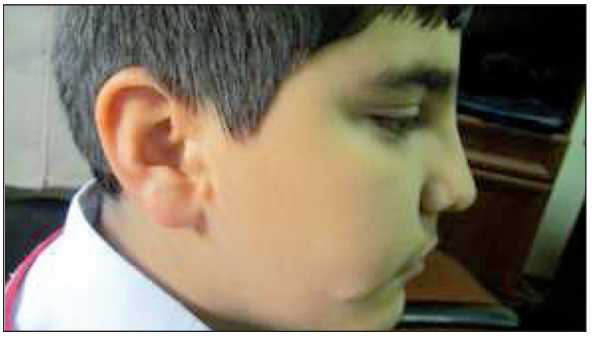

FIGURE 1: Right sided preauricular appendages along the line joining the tragus and the angle of the mouth and the scar tissue of excised accessory tragus.

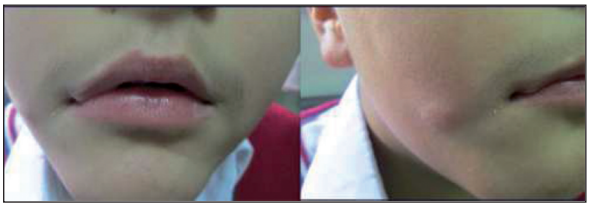

FIGURE 2: Cleft-like extension of right corner of the mouth in patient with Goldenhar syndrome.

The patient's mental and physical development was normal. He had no deafness and any acoustic problems. There were no associate anomalies of the vertebra. Blood count and urine analysis were normal. Examination of the cardiovascular, respiratory, gastrointestinal and genitourinary systems revealed no abnormality. We pre-diagnosed the patient as a case of GS on the basis of accessory tragus, preauricular appendages, epibulbar dermoid and type I DS. Genetic consultation of the patient was confirmed the diagnosis.

DISCUSSION

Oculoauriculovertebral dysplasia was first decribed by *Arlt* in 1845. Subsequently, in 1952 *Goldenhar*

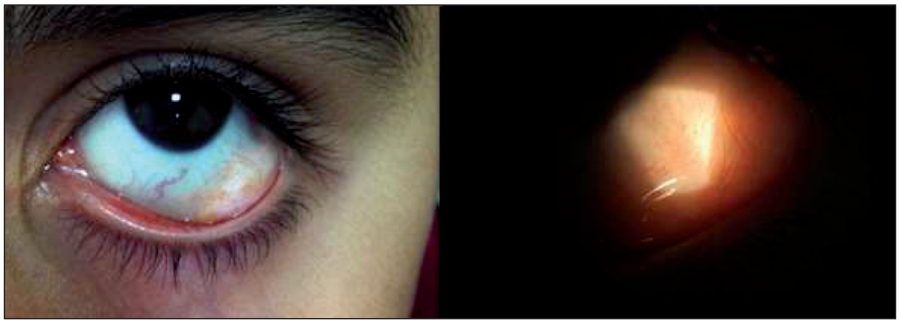

FIGURE 3: The macroscopic and the microscopic view of yellowish, hairy, soft dermoid with 6 mm x10 mm size on the lower temporal aspect of the left bulbar conjunctiva.

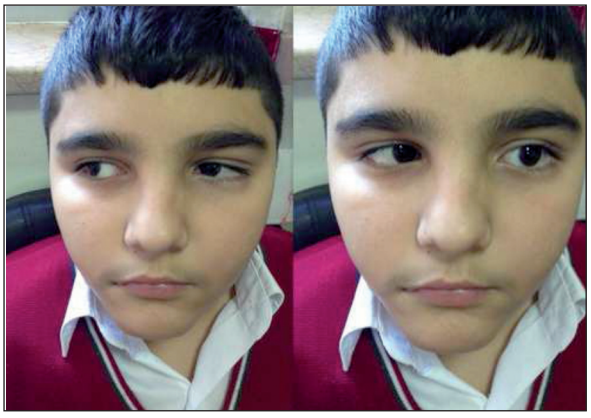

FIGURE 4: Extraocular motility of the left eye showed limitation of abduction and normal adduction. Extraocular motility of the right eye is normal.

reported a patient with mandibular hypoplasia, accessory tragus, epibulbar dermoids and named this group of features Goldenhar Syndrome.[1] GS is also known by various terminologies such as hemifacial microsomia or first and second branchial arch syndrome is seen predominantly in male with a ratio of 3:2.[1,2] The incidence is reported 1:3500-1:5600 and this is higher (1:1000) in children with congenital deafness.[2]

Defective development of the first and second branchial arches is thought to be responsible for the physical features.[1,2] The majority of the cases are sporadic. Autosomal dominant, autosomal recessive and other inheritance patterns have been suggested. However chromosomal studies have revealed normal in all cases.[2,3] Maternal use of drugs such as tamoxifen, thalidomide, retinoic acid and cocaine during pregnancy and maternal diabetes, rubella and influenza have been suggested as etiologic factors.[4,5] In our case, there was no history of any maternal illness or drug use during the pregnancy.

The first branchial arch is formed of two divisions. The first division forms maxilla, lateral portion of lip and cheek and anterior of the ear and the second one develops lower part of face, mandible, lower lip, part of middle ear. The external ear and auricle develops from the second branchial arch.[4] Ear anomalies such as accessory tragus, preauricular appendages, microtia and hearing loss are seen 40% of the cases.[6,7]

Other facial and physical features such as cleft lip and palate, mandibular hypoplasia, macrostomia, micrognathia, tracheoesophageal fistula, umbilical hernia, urological and anal anomalies are found in various ratios.[5-7] Calcification of falx cerebri and pseudotumor cerebri are the rarer reported anomalies.[4] In addition to the macrostomia our patient showed an appendage near the angle of the mouth which has not been reported in the literature yet. The presence of heart disease accounts for 50% and vertebral anomalies such as scoliosis and hemivertebra acoounts for 40%.[1,5-7] Our case did not associate with any cardiovascular or vertebral anomaly.

The classical ocular features of GS are microphthalmia, epibulbar dermoids, limbal dermoids, iris and choroid coloboma.[4,6,7] Dermoids are seen in 50% of cases concomitantly. Lipodermoids are seen rare than dermoids.[1,5] Other unusual ocular features consist anophthalmos, glaucoma and ocular motility problems like DS.[1,7] *Caca et al* reported a case of GS with preauricular appendages and Type I DS.[7] *Verma and Faridi* described a GS case associated with Type I DS and double elevator palsy concomitantly. [8]

The exact etiology of ocular motility disturbances with GS still remains unknown. *Barry et al* reported that the coexistence of epibulbar dermoids and DS is arised from the mutation of SALL-4 gene and this coexistence is not only seen in SALL-4 mutation and GS.[9]

The prognosis of GS is generally good in exception of any systemic coexistences as heart defects.[10] Minimal systemic findings with more than one ocular sign is apparently seen in our case. Goldenhar syndrome should bring to mind in cases with ocular features such as lipodermoid and Duane syndrome coexistence and without manifest systemic findings. A multidisciplinary approach is required to manage the associated anomalies. Reconstruction surgeries can be done for the anomalies of eyes, ears, cleft palate and mandibular hypoplasia. The dermoid cysts can cover the visual axis or more commonly can cause astigmatism, but our patient had no such problem. Children with DS should have eye examination to treat the possible refractive error.

Informed Consent

Written informed consent was obtained from the patient and his family.

Source of Finance

During this study, no financial or spiritual support was received neither from any pharmaceutical company that has a direct connection with the research subject, nor from a company that provides or produces medical instruments and materials which may negatively affect the evaluation process of this study.

Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

This study is entirely author's own work and no other author contribution.

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