RARE ASSOCIATION OF DUANE WITH GOLDENHAR SYNDROME: A CASE REPORT

Deepak Choudhury, Jayashree Dora, Pramod Kumar Sharma, Nisha Jha
Department of Ophthalmology, Veer Surendra Sai Institute of Medical Science and Research, Burla, Sambalpur, Odisha, India

ABSTRACT

Goldenhar syndrome is a rare congenital defect. Common clinical features include: Limbal dermoid, pre-auricular skin tag & strabismus. It affects between 1 in 3500 to 1 in 7000 live births. Duane syndrome is congenital rare type of strabismus. Limitation of adduction & abduction & narrowing of palpebral fissure is seen. A 20 year old Hindu male presented to our outpatient department with chief complains of fleshy mass in both inter-palpebral regions with F.B. sensation. Face was asymmetrical & head was turned to left side. Limbal dermoid was present in both eyes. On examination of right eye (RE): - Limitation of both abduction and adduction. On attempted adduction, decreased interpalpebral fissure height (IPF) & retraction of globe occurred. Left eye (LE) examination revealed limitation of abduction. On adduction, decreased IPF occurred. Excision of dermoids of both eyes were done. Conclusion: - Goldenhar Syndrome with type 3 Duane’s in RE & type 1 Duane’s in LE. This is a very rare association.

KEYWORD

Goldenhar syndrome, Duane’s retraction syndrome, limbal dermoid, strabismus, Electromyography

INTRODUCTION

Goldenhar was first noticed by Canton in 1861, and later by Von Arlt in 1881 however it went unnoticed. It was in 1952 that Maurice Goldenhar described it and it was named after him. It consisted of preauricular appendages, fistulas and epibulbar dermoids. Gorlin et al. in 196 included vertebral anomalies also as one of the manifestations of this syndrome and thus suggested the name Oculoauriculovertebral dysplasia. It results probably by development defects of first and second brachial arches. Goldenhar syndrome is a condition with prevalence ranging from 1:3500 to 1:7000 live births. About 2/3rd of patients show vertebral anomalies too. Other unusual associated features comprise ocular motility problems like Duane retraction syndrome besides rare accomplishments as lacrimal duct stenosis and iris and choroid colobomas.

Duane retraction syndrome is a unique restrictive type of strabismus characterized by co-contraction of medial and lateral rectus muscles due to anomalous innervations of one of the extraocular muscle antagonists during embryogenesis. Alexandrakis and Saunders found that in most cases the abducens nucleus and nerve are hypoplastic, and the lateral nerve is innervated by a branch of oculomotor nerve. It is characterized by variable limitation or absence of abduction, variable restriction of adduction, and retraction of the globe with narrowing of the palpebral fissure on adduction. Huber compartmentalized it into three types based on clinical findings and electromyographic recordings:

(i) Duane type I consists of limited or absent abduction with relatively normal adduction.
(ii) Duane type II consists of limited or absent adduction with relatively normal abduction.
(iii) Duane type III consists of both limited adduction and abduction.

The first type is more common.

Here we present a case of Goldenhar syndrome with type III Duane in right eye and type I Duane in left eye. This association is very rare.

CASE REPORT

A 20 year old Hindu young male presented to our outpatient department (OPD) on 22nd July 2014 with the complaint of presence of fleshy mass in both the eyes since birth and foreign body sensation of both eyes for 10 days.
There was no increase in the size of the mass since birth. No other family member had similar symptoms. There is no past history of any ocular surgeries or trauma.

On general examination, patient was conscious and oriented. His vitals were stable. Examination of cardiovascular, central nervous system, chest and abdomen revealed no abnormalities. His face was asymmetrical due to mandibular hypoplasia (Figure-1). He also had preauricular skin tags in both side (Figure-1). He had a head tilt to left in primary position of gaze. Best corrected visual acuity was 20/20 in both eyes. Epibulbar masses were present in both the eyes (Figure-1). In RE the mass was 3mm x 2mm present near the lateral canthus and in LE it was 4mm x 3mm present at lateral limbus.

![Figure 1: showing epibulbar mass with preauricular tags bilaterally with mandibular hypoplasia](image)

On examination of ocular motility, in primary gaze a head tilt to the left side was there. In right eye (RE) there was a restriction of both abduction and adduction. On attempted adduction, decreased interpalpebral fissure height (IPF) with retraction of eyeball into the socket was observed (figure-3) and on attempted abduction IPF increased in RE (Figure-4). On examination of left eye (LE) limitation of abduction was seen (Figure-3). Patient was able to adduct his LE. However, on adduction decreased IPF occurred and on attempted abduction increased IPF was observed. No abnormality of upward and downward gaze was seen in both the eyes.

![Figure 3: On attempted adduction of RE- decresed IPF](image)  ![Figure-4: On attempted abduction of RE-increased IPD](image)

Detail anterior segment (AS) examination with slitlamp revealed no significant findings. Dilated fundus examination was normal. Patient was advised to do complete haemogram, X-ray of pelvis and spine, Computerized Tomography (CT) scan of brain and orbit and Electromyograph (EMG) of extraocular muscles. X-ray of pelvis and spine showed scoliosis. CT scan showed atrophy of extraocular muscles. EMG showed activation of medial and lateral rectus muscle simultaneously.
The patient was treated by excision of epibulbar masses bilaterally under peribulbar anaesthesia. On histopathological examination of the mass it was confirmed as dermoid. On follow up after 1 month no recurrence of the mass was found. As the visual acuity was good the patient denied to undergo strabismus surgery for the treatment of head tilt.

DISCUSSION

This case is very interesting because of the unusual association of Duane’s with typical Goldenhar syndrome which is usually unilateral. Bilateral involvement is seen only in 5-8% cases of Goldenhar syndrome. The incidence of Goldenhar syndrome has been reported to be between 1:3500 and 1:5600, with a male: female ratio of 3:2. Goldenhar syndrome usually presents in the childhood than adulthood and has no sex predilection. The exact pathogenesis of Goldenhar syndrome is not known. Chromosomal studies are found normal in all cases. Baun and Feingold reported this syndrome to be sporadic in 90% of cases while positive family history was reported in the maternal grandmothers and mother of two cases in their series. In this case other family members of the patient were not affected. Ingestion of drugs such as thalidomide, retinoic acid, tamoxifen and cocaine by the pregnant mother may be related to the development of this syndrome. Maternal diabetes, rubella, and influenza have also been suggested as etiologic factors. In our case, there was no history of maternal drug intake, any febrile illness or diabetes during pregnancy.

In Goldenhar syndrome, ocular anomalies especially bilateral dermoids are seen in 60% of the cases, vertebral anomalies in 40% of the cases, and ear anomalies also in 40% of the cases. Other systemic features are found in about 50% of the patients. Tetralogy of Fallot and ventricular septal defects are the most common cardiovascular anomalies associated with it. Our patient presented late because of the absence of cardiovascular and central nervous system anomalies. Other syndromes associated with multiple preauricular tragi include Treacher-Collins syndrome, Wolf-Hirschhorn syndrome, Nager's acrofacial dysostosis, Wildervanck syndrome (cervicooculoacoustic syndrome), Townes-Brocks syndrome and Delleman syndrome.

Among strabismus patients the incidence of DRS is probably not more than 5%. Most cases are sporadic, but familial cases have been estimated at 10%. Agenesis of the abducens nucleus or paradoxical innervation of the lateral rectus muscle, which subsequently causes a cocontraction of the horizontal rectus muscles may be causative. The frequent association of DRS with other congenital anomalies suggests a teratogenic event occurring between the fourth to eighth week of gestation as an etiological factor. Electromyography reveals that a paradoxical anomalous innervation of lateral rectus muscle is the underlying cause. This paradoxical innervation may be due to anomalous contact of lateral rectus with branches of third nerve if abducent nerve is absent or defective. The EMG finding corresponds well with the anatomical observations of abnormal innervations. EMG of this patient shows contraction of lateral and medial recti.

The dermoid cysts usually impinge on the visual axis but more commonly interfere with vision by causing astigmatism, but this patient had no such problem. Dermoid excision is associated with surgical complications like persistent epithelial defects (40%) and peripheral corneal vascularization and opacity (70%). But these...
complications do not outweigh the cosmetic and visual benefits of dermoid excision in selected patients\textsuperscript{14}. In this case excision of bilateral dermoid was done. Scott et al in their study of 19 patients with Duane’s syndrome showed that treatment with appropriate horizontal muscle recession in order to relieve abnormal head position or a significant tropia in primary position can eliminate the abnormal head position in 79\% cases and improvement in cases\textsuperscript{15}. This patient denied to undergo strabismus surgery.

\begin{center}
\textbf{CONCLUSION}
\end{center}

This patient is a case of Goldenhar Syndrome with type 3 Duane’s syndrome in RE & type 1 Duane’s syndrome in LE. This is a very rare association. Excision of dermoid in Goldenhar syndrome can be done for cosmetic purposes.

\begin{center}
\textbf{REFERENCES}
\end{center}