Goldenhar Syndrome: 3 Case Reports

Nashrin N¹, Gogoi B², Deka B³

¹Post Graduate Student, Regional Institute of Ophthalmology, Guwahati Medical College and Hospital, Guwahati
²Professor, Regional Institute of Ophthalmology, Guwahati Medical College and Hospital, Guwahati
³Assistant Professor, Regional Institute of Ophthalmology, Guwahati Medical College and Hospital, Guwahati

Abstract: Goldenhar syndrome is a term synonymously used with “oculoauriculo vertebral syndrome” is a rare birth defect that is apparent at birth. Described as early as 1950’s by Goldenhar it initially comprised of malformation of ears and ocular abnormalities, it was only in 1963 that vertebral anomalies were included as signs of this syndrome.¹ This work reports 3 cases of Goldenhar Syndrome who presented to our institute. All the cases are female in our case report which shows an increased preponderance in females and also absence of systemic abnormalities in these cases.

Keywords: Goldenhar Syndrome, limbal dermoid, polydactyly, lid coloboma

1. Introduction

Goldenhar Syndrome (Oculoauriculo vertebral dysplasia with hemifacial microsomia) is a rare congenital anomaly involving the first and the second brachial arches.² It is a congenital defect characterized by a heterogenous constellation of malformations classically involving the face, eyes and ears. This condition was first described by Dr Maurice Goldenhar in 1952. In 1963, Gorlin et al. named this syndrome as oculo - auriculo - vertebral syndrome (OAVS) due to the presence of additional vertebral anomalies.³ Unilateral involvement is seen in 70% - 80% of cases. Most cases of OAV are sporadic, autosomal dominant transmission is reported for 1% - 2% of the cases. A few persons reported with autosomal recessive inheritance has been reported. Aetiology of the syndrome remains unclear. Currently a deficiency in mesodermal formation or defective interaction between neural crest or mesoderm is suggested as possible eitiology. Different factors also contribute to the disease such as: ingestion of some drugs (Cocaine, Thalidomide, Retinoic acid and Tamoxifen), environmental factors (Insecticides, Herbicides) and maternal diabetes.⁴

Case 1:
A 1 year old girl born of non-consanguineous marriage came to our department for assessment of dysmorphic facial features. The patient was a normal full time vaginal delivery with no perinatal complications except for low birth weight (2Kg).

2. On Examination

Patient had facial asymmetry-hypoplastic maxilla and mandible, bilateral limbal dermoids-left limbal dermoid being much larger than the right ,right upper lid coloboma, preauricular tags on left, malformed pinna left ear and microtia right ear and polydactyly left hand. BERA revealed conductive hearing loss. Larger dermoid interferes with the visual axis causing astigmatism and predisposing to secondary strabismus from anisometropic amblyopia. Hence, the dermoid was excised and superficial keratectomy done. As the right upper lid coloboma was small, management was deferred till the ripe age of 2 to 4 years when the eyelids have grown to a more manageable size and more tissue is available for reconstruction, particularly if they have a good Bell’s phenomenon.
which lies in the blastogenesis period which are responsible for external ear abnormalities, as development of anterior ear primordium takes place from first branchial arch, and the second arch modifies to posterior ear primordium. Also, the outer ear canal is derived from the dorsal portion of first branchial cleft. The other mechanism may be related to an anomaly in migration defect of neural crest cells or could be due to predisposed genetic determinant.5

The oculoauriculovertebral “spectrum” (OAVS) includes a variable combination of microtia, hemifacial microsomia, lateral facial cleft, epibulbar dermoid and upper eyelid coloboma [Gorlin et al., 1990]. This condition is thought to affect structures developed from the first and second branchial arches. Because there is no agreement on minimal diagnostic criteria, the phenotype overlaps many genetic and teratologic syndromes.

However, according to Kumar et al. [1993], the minimum criteria for diagnosis of OAVS are the presence of at least two of the following: otic hypoplasia, hemifacial microsomia, lateral facial cleft, epibulbar dermoid and / or upper eyelid lid coloboma, and vertebral anomalies (fusion of vertebral body or spine, and segmentation abnormality or “butterfly” vertebra). All of our cases satisfies the criteria as stated by Kumar et al.(1993). Also, our first case presented with two uncommon presentations like polydactyly and upper lid coloboma. Our second case presented with two common presentations like limbal dermoid and pre-auricular tags. The third case was also a simple case of Goldenhar syndrome without any systemic abnormalities.

The frequency of Cardio Vascular Manifestations (CVM) was found to be 5 - 58 % showing great variability.8 Children with Goldenhar’s Syndrome have a high incidence of congenital malformations of the cervical spine including odontoid hypoplasia that puts them at particular risk during general anaesthesia.9 It has been estimated that there is a 12% incidence of platybasia and occipitalization of Cl in children with Goldenhar Syndrome.10 All our cases were free from any systemic abnormalities as discussed above.

Dermoids tend to increase in size with time, so to prevent the risk of astigmatism; amblyopia and strabismus excision of limbal dermoids along with superficial keratectomy was done.

Surgical treatment of lid coloboma requires surgical repair and spectacle correction. For small defects that do require suturing (up to 25%), direct closure may suffice, although for moderately sized defects (25%–50%), where a severing of the upper crus of the lateral canthal tendon is required for satisfactory closure. For larger defects (50% or more of the eyelid), available options include the Cutler-Beard procedure, an eyelid rotational (switch) flap, or a tarsomarginal graft.11

Severe anomalies of the mandible require reconstruction with bone graft. In case of microtia or other ear defects needs extensive ear reconstruction to be done within 6 - 8 years of age.
4. Conclusion

Life expectancy is usually normal in patients suffering from Goldenhar Syndrome in absence of serious systemic abnormalities. Hence in all the above three cases, it can be said that the patients will be able to lead a normal life.

5. Declaration of Patient Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity.

6. Financial Support and Sponsorship

Nil

7. Conflicts of Interest

There are no conflicts of interest.

References