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Goldenhar Syndrome: 3 Case Reports

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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 (Oculoauriculovertebral 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 in70% - 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 weigh (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.

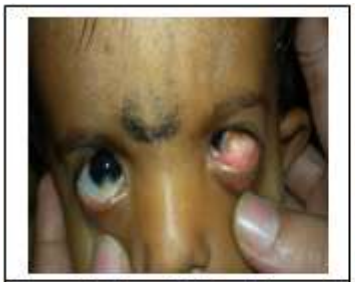

Figure 1: Bilateral limbal dermoids (Left larger than right)

Figure 2: Pre auricular tags & Mandibular Hypoplasia

Figure 3: Right upper lid coloboma

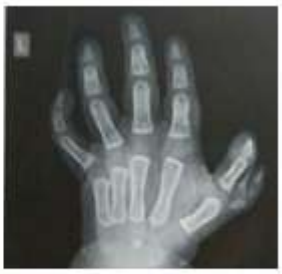

Figure 4: Polydactyly (X-ray left hand)

Case 2:

The patient presented in our department, with a growth in the left eye and pre-auricular tags. The growth was a limbal dermoid located in the lower temporal quadrant. Fundas examination was normal. The parents & siblings are normal. The systemic examination was found to be normal. BERA was done which came out to be normal. The limbal dermoids was excised and superficial keratectomy done. Histopathological examination of the limbal dermoid shows dermolipoma.

Figure 5: Limbal dermoid (Left eye)

Figure 6: Pre auricular tags

Figure 7: H& E (4X) of dermoid showing skin with adnexal structures.

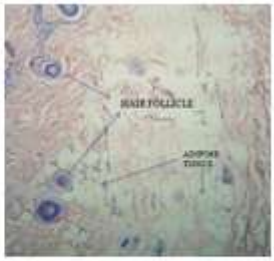

Figure 8: H& E (40 X) of dermoid showing adipose tissue and hair follicle

Figure 7

Case 3

Our third case is a female chid of fourteen months age who presented with limbal dermoid on right eye and preauricular tags on left side. She was operated for cleft lip on 2/6/2016. She was born at the gestational age of 8 months by normal vaginal delivery. She was of 2.05 kg at birth. BERA and other reports regarding systemic examination came to be normal. The dermoids was excised and superficial keratectomy done. Histopathological examination revealed dermolipoma.

Figure 7: Pre auricular tags

Figure 8: Limbal dermoid

3. Discussion

The incidence of Goldenhar Syndrome has been reported to be 1:3500 to 1:5600 with a male: female ratio of 3:2 in various literatures.[5] We report three cases of Goldenhar Syndrome and all of them are female. Hence, it seems to be a common occurrence in females also. One of our cases also presents with bilateral limbal dermoids which is an uncommon presentation. Also the first case presents with polydactyly which is seen to be an uncommon association with Goldenhar Syndrome.

Patients of Goldenhar Syndrome may present with various clinical features ranging from mild to severe forms. Two pathophysiologic mechanisms have been proposed for the syndrome (OAVS) which includes, reduction in the blood flow resulting in focal hemorrhage in the first and second branchial arches around 30-45 days of gestational age, 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.[6]

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 eye lid coloboma, and vertebral anomalies (fusion of vertebral body or spine, and segmentation abnormality or ‘‘butterfly’’ vertebra).[7]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 platyblasia and occipitalization of C1 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

[1] Sharma JK, Pippal S K Raghuvanshi S K, Shitij A.Goldenhar – Gorlin's Syndrome: A case report. Indian J Otolaryngol Head Neck Surg .2006 Jan; 58(1):97-
[2] Riyaz A, Riyaz N.Goldehar Syndrome with unusual features.Indian J Dermatol Venerol Leprol 1999; 65:143-44.
[3] Vinay C, Reddy R S, Uloopi K S, Madhuri V, Sekhar R C. C raniofacial features in Goldenhar Syndrome. J Indian Pedod Prev Dent, 2009; 27: 121-4.
[4] Siddique MAR, Hossain J, Abedin MJ, Parvez M.J Bangladesh Coll Phys Surg 2010; 28: 193-195.
[5] Mehta B, Nayak C, Savant S,Amladi S. Goldenhar Syndrome with unusual features.Indian J DermatolVenereol 2008; 74:254-56.
[6] Patil NA, Patil AB. Goldenhar syndrome: Case report. IJSS Journal of Surgery. 2015; 1:18-20.
[7] Stoll, C. Viville, B. Treisser A. Gasser B. (1998), A family with dominant oculoauriculovertebral spectrum. Am. J. Med. Genet., 78: 345–9.
[8] Nakajima H.,Goto G., Tanaka N., Ashiya H., Ibukiyama C., Goldenhar Syndrome Associated with Various Cardiovascular Malformations. JpnCirc J 1998; 62 : 617-620.
[9] Healey D, Letts M, Jarvis JG. Cervical spine instability in children with Goldenhar's syndrome. Can JSurg. 2002 Oct; 45(5):341-4
[10] Gibson JN, Sillence DO, Taylor TK. Abnormalities of spine in goldenharssyndrome. J Pediatr Orthorp 1996; 16: 344-9.
[11] Tawfik HA, Abdulhafez MH, Fouad YA. Congenital upper eyelid coloboma: embryologic, nomenclatorial, nosologic, etiologic, pathogenetic, epidemiologic, clinical, and management perspectives. Ophthalmic Plast Reconstr Surg. 2015; 31:1–12.

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