ISSN No:-2456-2165

Goldenhar Syndrome an Ophthalmological Perspective: A Case Series

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Abstract:- Goldenhar syndrome is a rare congenital condition affecting 1:3500 - 1:5600 with a male to female ratio of 3:2 births. Goldenhar syndrome is represented by abnormal development of the eyes, ears, and spine. Children with Goldenhar syndrome may have partially formed or absent ears, benign eye growths like Epibulbar and limbal dermoids and spinal deformities like scoliosis. The condition may also affect facial structure and other body organs, with most deformities affecting only one side of the body.Spine and rib cage deformities are common, with about half of people with Goldenhar syndrome having congenital scoliosis. Spine anomalies can lead to incomplete growth and pulmonary disorders.Here, we are reporting three cases of Goldenhar's syndrome, where almost all the classical signs of this rare condition were present.

Keywords:- *Deformity, Epibulbar Dermoid, Goldenhar's syndrome, Epicanthusinversus, Duane syndrome.*

I. INTRODUCTION

A. CASE -1

A four-year-old male child studying in preschool, brought by his guardian in ophthalmology department at Tertiary Care Hospital, with the complaint of lid defect, yellowish mass in left eye since birth. Patients complaining of yellowish mass in left eye which insidious progressive not associated with any pain. No history of any chronic illness. The patient has personal history of sleep appetite and bowel bladder normal. With birth history of Pre-Term Normal Delivery, NICU admission for 3 days for jaundice and given phototherapy.General examinationrevealed that the patient was underweightwith delayed speech with other normal milestones.The patient was the first child of a consanguineous marriage with second degree relative and with no pregnancy related complications.**Ocular examination**reviled that visual acuity was (RE 6/36, LE 6/36), Right eye anterior segment was normal.Left eye medial upper lid coloboma (approximately size 3mm*3mm) with trichomegaly.Yellowish soft mass on temporal side of left eyes (Approx 4mm*4mm) with hair follicles whose posterior limit could not be seen.Exposure keratitis in left eye.On palpation, mass was soft, non-tender.The rest of Anterior Segment was normal. Extra ocular movements present in all directions (Full and Free).Retinoscopy and Fundoscopy were normal.**ENT examination** revealed **preauricular tag** on left side. BERA test reports were normal, both ears normal hearing.Otoacoustic Emission was normal and the child was not advised any intervention for auricular tag at present.On recent 2D echo report were normal.Rest of the systemic Examination was normal.The laboratory investigations were within normal limits.Radiological Investigations were normal.The clinical diagnosis of case was **Goldenhar Syndrome**.Cause was sporadic in this case.

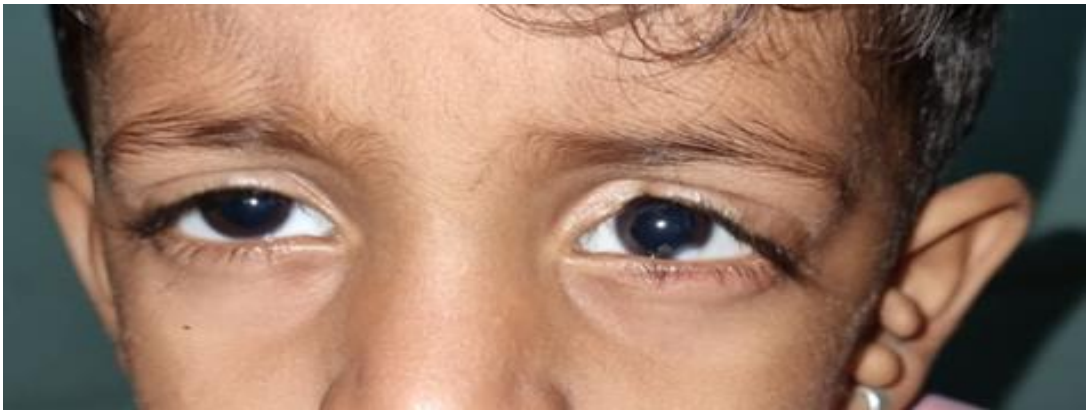

Fig. 1: Photograph- showing LE lid coloboma with redness

ISSN No:-2456-2165

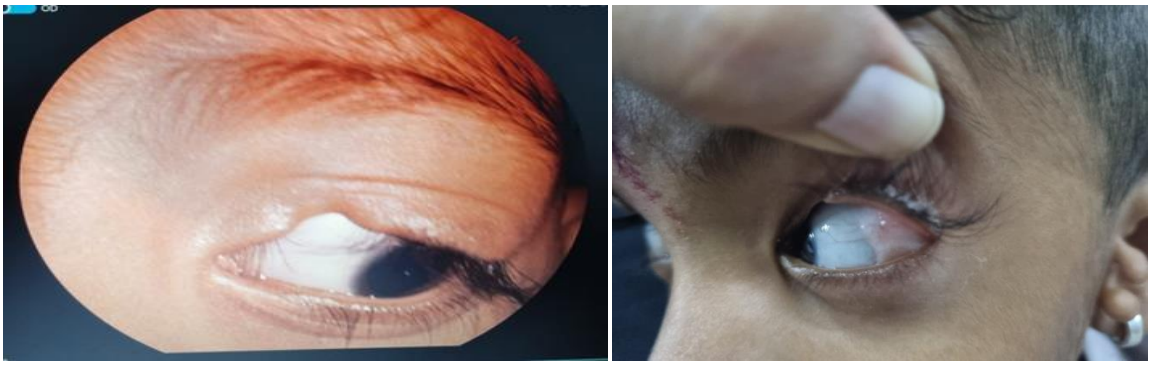

Fig. 2: Photograph- ocular features showing trichomegaly and lipodermoid

Fig. 3: Showing Left ear preauricular tag

Fig. 4: Image of epidermal dermoid with hair follicle

B. CASE – 2

An 18 months old female child broughtby her mother at Tertiary Care Hospital,with the complaint of yellowish mass inthe eye since birth.The patient was the only child of a non-consanguineous marriage with motherbeing elderly primigravida and havingpolyhydramnios during Antenatal Period. With birth history evidence of Full-Term Normal Delivery,No NICU admission.General examination revealed that the patientwas well nourished with normal developmentalstatus and milestones.On ocular examinationVisual acuity was fixating and following light.A yellowish soft mass on temporal side of both eyes (Right sided mass was approx. 3mm * 3mm, left sided mass 4mm*3mm) whose posterior limit could not be seen. Left sided mass was encroaching over cornea slightly (5-6 o'clock). Epicanthus inversus in both eyes.On palpation, it was soft, non-tender. The rest of Anterior Segment wasnormal.Extra ocular movements present in all directions.Retinoscopy and Fundoscopy were normal.ENT examination revealed preauricular tag on left side and left side hemimandibular hypoplasia.Otoacoustic Emission was normal and the child was not advised any intervention for auricular tag at present. On dental examination crowding of teeth with mandibular hypoplasia on left side was noted.The child was diagnosed with Atrial Septal Defect on paediatric transthoracic echocardiography at 5 months of age. A 6 monthly follow up for the same was advised.No other abnormalities were detected.Rest of the systemic Examination was normal.The laboratory investigationswere within normal limits.Radiological Investigationswere normal.The clinical diagnosis of this case being Bilateral Ocular Dermolipoma (atypicalpresentation of Goldenhar Syndrome), considering the bilateral presentation and novertebral abnormalities.The cause being sporadic or fertilization of overripe or aged ovum in the case.

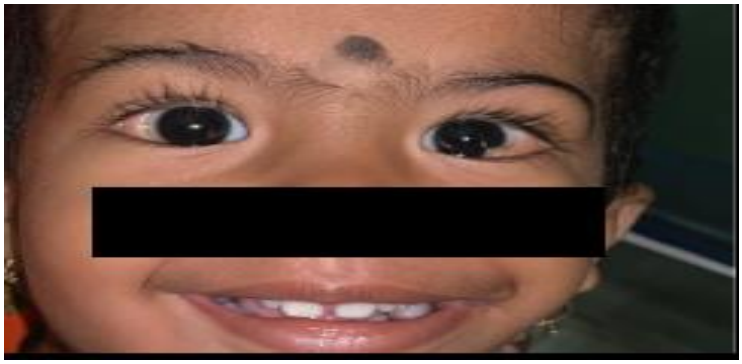

Fig. 5: Both eye lipodermoid

Fig. 6: hemimandibularhypoplasia

Fig. 7: Preauricular tag

C. CASE – 3

A6-year-old female child referred to ophthalmology department by peadiatric department at Tertiary Care Hospital, with the complaint of lid defect, Discharge, redness and yellowish mass in right eye and Discharge in left eye since birth.The patient was the second child of a non-consanguineous marriage with no pregnancy related complications.Birth History include Full Term Normal Delivery and no NICU admission.**General examination** revealed that the patient was underweight with isolated language defect with other normal milestones. On o**cular examination v**isual acuity was RE (6/12) and LE (6/9). Right eye medial upper lid coloboma (approximately size 7mm*3mm) with tricomegaly. Yellowish soft mass on temporal side of right eyes (approx 4mm * 4mm) with hair follicleswhose posterior limit could not be seen.Exposure keratitis in right eye. Discharge with Nasolacrimal duct obstruction in both eyes.On palpation, mass was soft, nontender.The rest of Anterior Segment was normal.Extra ocular movements present in all directions.Retinoscopy and Fundoscopy were normal.**ENT examination** revealed **preauricular tag** on both side with atresia of left external auditory canal and right sided **maxillary and hemimandibular hypoplasia**.Child was not advised any intervention for auricular tag at present. On dental examination c**rowding of teeth** with mandibular hypoplasia on right side was observed.The child was diagnosed with Ventricular Septal Defect, coarctation of aorta with B/L SVC on pediatric transthoracic echocardiography at 1 year of age and 6 monthly follow up for the same was advised. On recent 2D echo report previous VSD is closed Rest of the systemic Examination was normal.The laboratory investigations were within normal limits.Radiological Investigations were normal.The clinical diagnosis of case is **Goldenhar Syndrome**, with congenital heart disease.Cause was sporadic in this case.

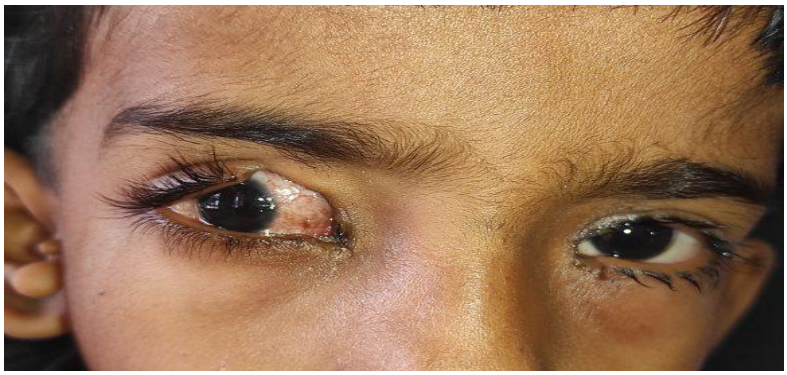

Fig. 8: Photograph- showing RE lid coloboma with redness and BE Discharge

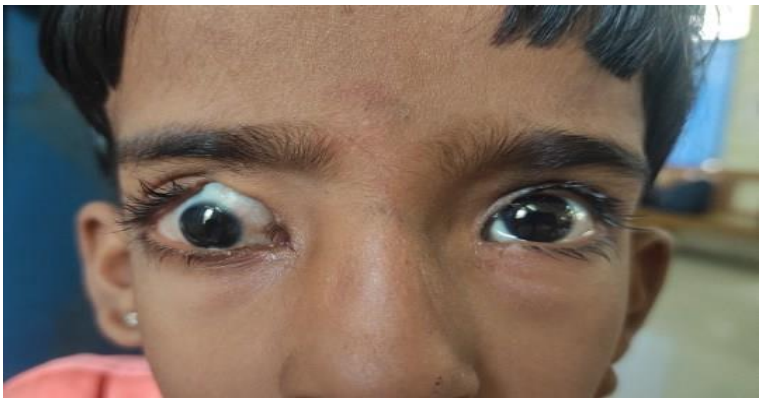

Fig. 9: Photograph- ocular features Lid coloboma

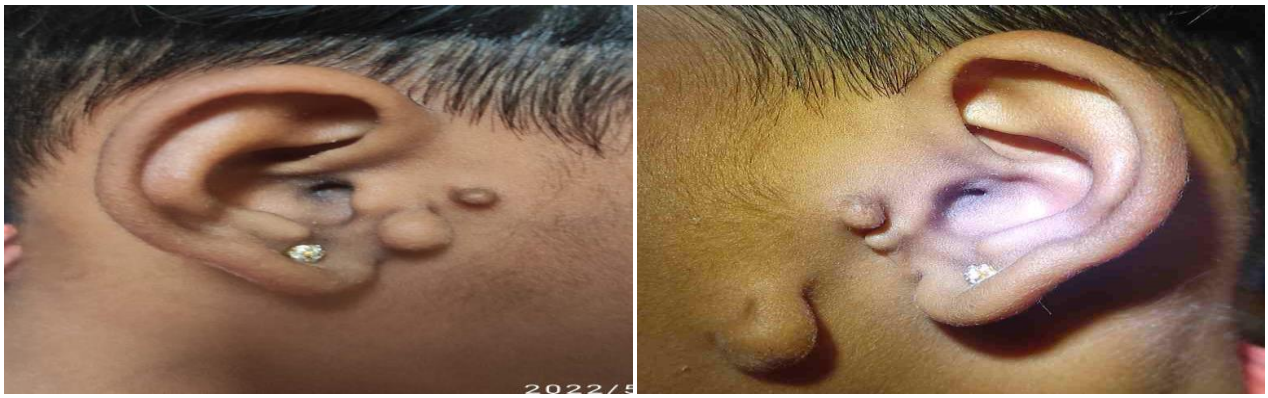

Fig. 10: Right ear and Left ear with EAC atresia &Preauricular tag

Fig. 11: Right sided maxillary & hemimandibular hypoplasia, crowding of teeth

ISSN No:-2456-2165

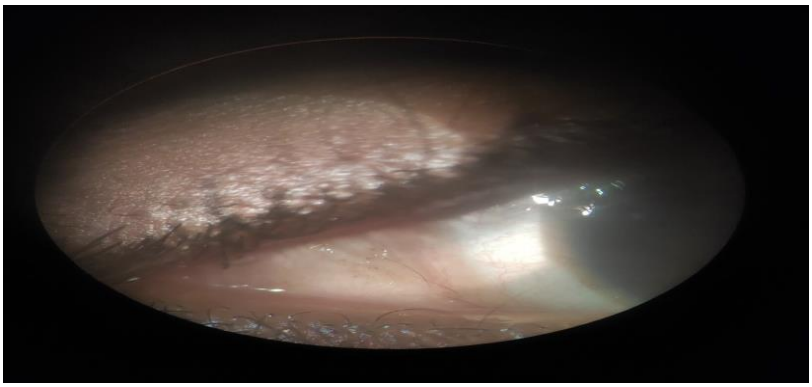

Fig. 12: Slitlamp image of epidermal dermoid with hair follicle

Table 1: Comparison of the clinical features in the all cases

Features	Case 1	Case 2	Case 3
Hemifacial hypoplasia	-	+	+
Epicanthus inversus	-	+	-
Mid face hypoplasia	-	+	+
Microphthalmia	-	+	+
Ocular dermoid	+	+	+
Coloboma	+	+	+
Crowding of teeth	-	+	+
Accessory tragi	+	+	+
Microotia	-	+	+
Cleft lip and palate	-	-	-
Vertebral abnormalities	-	-	+
EAC atresia	-	-	+
Systemic abnormalities	-	+	+
AtrialSeptal Defect	-	+	-
Ventricular Septal Defect	-	-	+
Nasolacrimal duct obstruction	-	-	+
Exposure keratitis	+	+	+

II. DISCUSSION

Goldenhar's syndrome is a rare, inherited condition characterized by accessory tragie, mandibular hypoplasia, and ocular dermoids. It is also known as oculoauriculo-vertebral dysplasia due to additional vertebral anomalies.The syndrome has a multifactorial aetiopathology, including Chromosomal aberrations and Fertilization of overripe or aged ovumthat can disrupt blastogenesis.[(1)]The incidence is higher in among children with congenital deafness.The exact cause is unknown, but it could be due toabnormal mesodermal migrations, disturbed mesodermal migration, or other factors. [(2)] Most cases are sporadic. Autosomal dominant, autosomal recessive, and multifactorial modes of inheritance have been suggested, but chromosomal studies have not revealed any abnormalities. A previous study conducted by Kallen et al., also found that majority of the patients were sporadic. Autosomal dominant, autosomal recessive and multifactorial modes of inheritance have also been suggested. Chromosomal studies did not reveal any abnormality.[(3)]

III. ETIOLOGY

Ingestion of drugs during pregnancy such as retinoic acid, thalidomide, tamoxifen, and cocaine. Gestational diabetes, rubella and influenza have also been known risk factors. [(4,5)] Heavy alcohol consumption during pregnancy is also one of the risks of this syndrome. [(6)] In our cases, there was no history of any maternal drug intake, any febrile illness or gestational diabetes which had occurred during pregnancy. In our cases causewas sporadic.

IV. DIAGNOSIS

Classic triad of Goldenhar syndrome Mandibular hypoplasia, Ocular and auricular malformations, and Vertebral anomalies.The classic features of this syndrome include such as Facial asymmetry, ENT anomalies, Vertebral, Cardiac and CNS anomalies. Epibulbar and limbal dermoidsare the ophthalmic hallmarks of Goldenhar syndrome. Most common site inferotemporal limbus. Eyelid colobomas may occur. Other abnormalities include microphthalmia, cataract, and iris abnormalities.It is mostly unilateral in nature, it can also present bilateral with the right side being more common affected with respect to the left.[(2)]

ISSN No:-2456-2165

Tetralogy of Fallot and ventricular septal defects are the most common cardiovascular anomalies. [7]similar findings were reported in our cases. Anopthalmos, facial palsy, calcification of falx cerebri, undescented testes, and association of Goldenhar syndrome with Turner's syndrome and glaucoma are the rare reported associations.

Goldenhar syndrome may be also associated with various conditions such as cleft lip and palate, crowding of teeth, macrostomia, micrognathia, neck webbing, tracheoesophagial fistulas, urologic anomalies, hypoplastic vagina, anopthalmos, facial palsy, and Turner's syndrome.

V. DIFFRENTIAL DIAGNOSIS

Treacher-Collin syndrome, Wolf- Hirschhorn syndrome, Townes-Brocks syndrome, and Delleman syndrome are other syndromes linked to multiple preauricular tragic, with Treacher Collin syndrome causing maxillary and mandibular hypoplasia but not ocular and aural anomalies.

VI. MANAGEMENT

Goldenhar syndrome, or hemifacial microsomia, is a cosmetic condition affecting the eyes and ears. Treatment varies based on age and systemic associations, with cosmetic procedures such as rib bone grafts for mandibular hypoplasia, bone distraction and osteogenesis for underdeveloped maxilla, and surgical corrections for cleft lip and palate. Reconstruction surgeries for the external ear can be performed at 6-8 years, while milder cases can be done in early teens. Social workers can provide support and guidance to children and their families, including accessing community resources, making decisions about surgeries, and adjusting for facial asymmetry. The prognosis is good in uncomplicated cases without systemic associations. However, severe cases may require immediate interventions from birth, such as obstructive sleep apnea, jaw problems, and vision problems.[2]

In our cases treatment for exposure keratitis was given, advices Lid reconstruction surgery for epibulbar dermoidOphthalmic intervention is not required at present since it is not causing any vision defect or amblyopia.In future the patientsmay require excision of dermolipoma and removal of preauricular tag.Awareness to prevent ocular complications such as exposure keratitis and to monitor the progression of dermoid. Multidisciplinary treatment approachand long term follow up is required to assess psychosocial impact on child and her mental health and for counselling of parents.The caseshave been presented to increase the awareness of this entity in general public.

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