



“GOLDENHAR SYNDROME- A RARE CASE REPORT”

Ophthalmology

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KEYWORDS

Epibulbar dermoid, disc hypoplasia, facial abnormality.

INTRODUCTION

Goldenhar Syndrome also Called as Oculo-auriculo-vertebral Dysplasia. The Incidence of this syndrome is about 1 in 5600 live births. Male to Female preponderance is 3:2. Most of these cases are sporadic and multifactorial etiology that includes nutritional & environmental factors that result in disturbance of blastogenesis. An Autosomal Dominant pattern is also observed. 10-30% have bilateral manifestations.

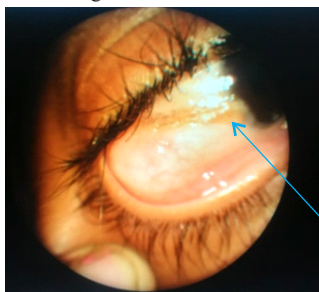
Case report

A 10 years old boy, presented with cosmetic complaint of right eye flashy and hairy mass with keratinization in temporal area in lower fornix since birth, which is not increased in size with age and having 6/6 vision. Patient was born through full term normal vaginal delivery. Mother not give any history of drug intake, diabetes or hypertension during pregnancy. He has an Elder male sibling with no obvious clinical illness.

On clinical examination subsequent finding are present :-

Ocular examination

a) Epibulbar dermoid in temporal area in lower fornix with Hypoplasia of lateral wall of orbit on Right Side.

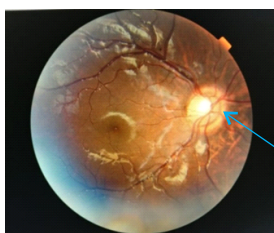


Epibulbar dermoid



Hypoplasia of lateral wall

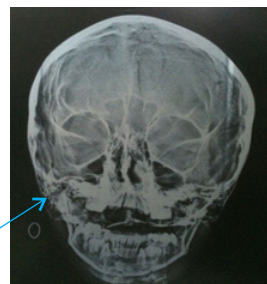
b) On right eye fundus examination, showing disc hypoplasia, peripapillary tessellated fundus with normal macula.



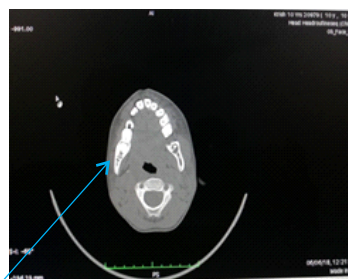
Disc hypoplasia

Cranio-facial examination

a) Patient had Facial Asymmetry due to Mandibular and Maxillary Hypoplasia showing in CT Scan.



Maxillary hypoplasia



Mandibular hypoplasia

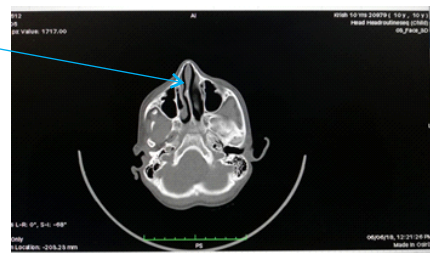
b) On auricular examination Preauricular tags on Right Side



Preauricular tags

c) On nasal cavity examination, Deviated Nasal Septum (DNS) is seen.

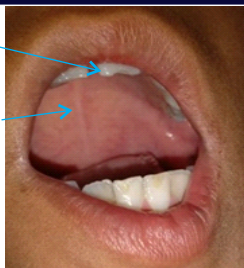
Disc hypoplasia



d) Oral cavity examination, High arched palate with tooth enamel abnormality are present

High arched palate

Tooth enamel abnormality



Multicystic dysplastic kidney, Ectopic kidney and Malformed or Absent ureters

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Skeleton examination

Winging of Scapula on Left side is present.



Winging of Scapula

No Neurological, Genitourinary and Cardiac Anomalies are seen.

Investigation profile of patient under normal range are :- Liver function test, Renal function test, Chest x-ray, ECG, Urine examination, Abdominal USG.

DISCUSSION

Goldenhar Syndrome is a rare disorder, has a wide range of manifestations including ocular, auricular, craniofacial, vertebral, cardiac, renal and central nervous system anomalies.

Classical triad of syndrome includes -

- 1) Hemifacial microsomia
- 2) Epibulbar dermoid
- 3) Deformity of ears with pre auricular appendage

Types of syndrome includes -

- 1) Mild- Oculo-auriculo-vertebral dysplasia
- 2) Intermediate - Hemifacial microstomia
- 3) Severe - Goldenhar syndrome (Systemic association)

Various Ocular manifestations are :- Epibulbar dermoid (75% of cases), hypoplasia of orbital wall, Coloboma, Microphthalmos, Cataract, Iris anomalies, Anophthalmos, Optic nerve Hypoplasia, dacryocystitis, nystagmus and Squint.

Various Auricular manifestations are :- Preauricular skin tags, Accessory auricle and Anotia (rare).

Various Facio-dental anomalies are :- Hypoplasia of malar bones, Zygomatic arch and mandible, Macrostomia, Micrognathia, bifid tongue, Cleft lip, Cleft palate, Temporo-mandibular joint malformation.

Various Vertebral anomalies are :- Fusion of posterior elements of cervical vertebrae, Hemivertebral loss and Spina bifida (rare) .

Various Central nervous system manifestations are :- Frontal hypodensities (most frequent), Asymptomatic Hydrocephalus, Corpus callosum dysgenesis, Arnold chiari malformation and facial nerve palsy.

Various Cardiac defects are :- Ventricular septal defects ,Patent ductus arteriosus, Fallot's tetralogy, Coarctation of Aorta and pulmonary stenosis.

Various Uro-genital anomalies are :- Renal agenesis, Hydronephrosis,