Original Research Paper

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Ophthalmology

A RARE CLINICAL CASE OF GOLDENHAR SYNDROME

Dr. Supriya Sureshappa Tadkase* Resident doctor at KEM Hospital, Mumbai. *Corresponding Author

Dr. Sayali Mahajan Assistant Professor at KEM Hospital, Mumbai.

KEYWORDS :

Introduction:

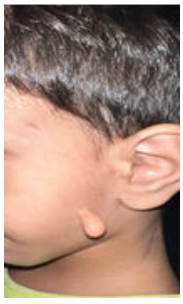
- Goldenhar syndrome (GS) is a congenital disease first described in 1952 by ophthalmologist Maurice Goldenhar. In the literature, we can find many other synonyms of this defect including oculo-auriculo-vertebral syndrome (OAVS), facio-auriculo-vertebral syndrome or GoldenharGorlin syndrome.[1] It is characterized by impaired development of structures such as eyes, ears (with or without hearing loss), lip, tongue, palate, mandible, maxilla and deformations of the teeth structures. Because these parts of the face derive from branchial arches, and it is also classifi ed as 1st and 2nd branchial arch syndrome. In this syndrome, abnormalities localize in the internal organs such as heart, kidneys, in the central nervous system or in the skeleton and different vertebral defects are observed.[2-4] According to some authors for this reason other name like hemifacial microsomia shouldn't be used interchangeably while referring to this syndrome.[5,6] Various studies have shown that this defect occur from 1:3500 or 1:5600 to 1:45 000 live births.[7,8]

Case report:

- Informant : mother
- 4 year old female
- Mass in left eye since childhood
- Slowly progressive and gradually increasing in nature.
- History of hearing loss since birth
- Left sided pre auricular tag and skin tag over left cheek since birth
- Child is a known case of seizure disorder
- Developmental delay (at present achieved all developmental milestones except speech)

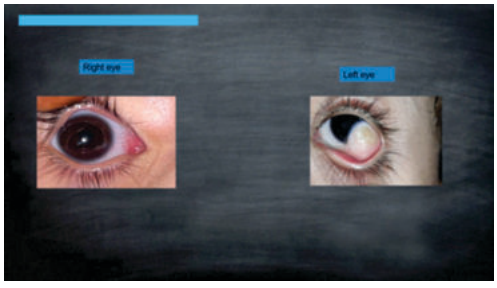
History of present illness:

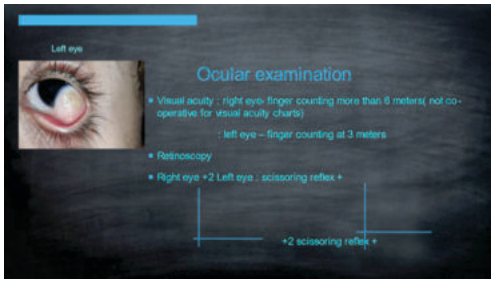
- No other ophthalmological complaints, ocular injuries, other systemic illness and other topical or systemic medication use
- No significant anti natal history
- Birth history: full term normal vaginal delivery and no history of NICU admission
- Fully Immunized till date
- No history of consanguineous marriage
- No significant family history



Examination:

- General examination: child is conscious , well-oriented Facial asymmetry with mild mandibular hypoplasia Lipoma over left cheek Low set ears Left sided pre auricular tag
- Cardiovascular system: within normal limit
- Respiratory system: within normal limit
- Nervous system : within normal limit
- Dental assessment : mandibular hypoplasia
- ENT Assessment: bilateral profound hearing loss





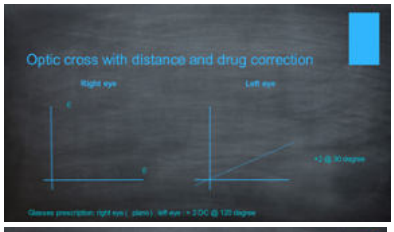
Right eye	Ocular examination	Left eye
Lid – normal	Ocular adnexa	Lid – normal
Clear	Conjunctiva and cornea	A nodular, soft, localised, opaque, yellow-white , elevated, infero-temporal corneo-limbo-conjunctival lesion, deeply infiltrating into cornea without any pigmentations, measuring approximately 5mm x 3 mm x 1mm in size with hair follicle on its surface
Well formed, normal depth	Anterior chamber	Well formed, normal depth
Normal colour and pattern	Iris pattern	Normal colour and pattern
single, circular, reacting to both direct and consensual light reflex	pupil	single, circular, reacting to both direct and consensual light reflex
Clear	lens	Clear

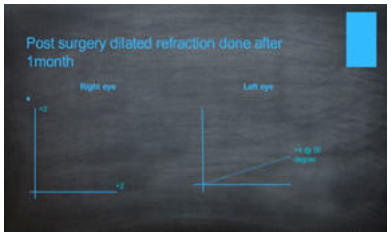
Active movements full range	Extra ocular movements	Active movements full range
13mmhg with rebound tonometer (i-care)	Intraocular pressure	13mmhg with rebound tonometer (i-care)
0.2 cup: disc ratio pink disc Macula : within normal limit Foveal reflex : present	Posterior segment	0.2 cup: disc ratio pink disc Macula : within normal limit Foveal reflex : present

- Provisional diagnosis of left eye choriostoma and right eye within normal in a case of goldenhar syndrome
- Excisional biopsy was advised.
- Patient underwent the procedure of left eye limbal dermoid mass excision with 0.02% mitomycin C application with a amniotic membrane graft implantation under general anaesthesia.
- The excised tissue sent for histo pathological examination.

Histopathological report:
- Section shows tiny fragmented tissue bit focally lined by stratified squamous epithelium with underlying fibrofatty stroma showing adnexal structure, with mild chronic inflammation.
- Finding suggestive of limbal dermoid cyst.

Post operative management:
Systemic antibiotics and anti-inflammatory medications given for 1 week Locally left eye drop tobramycin 0.3% w/v + dexamethasone0.01% six times a day given in tapering dosage with lubricating eye drops.





- Advised constant use of glasses and right eye patching for 3 hours a day.
- Follow up after 2 months in ophthalmology opd and close follow up in pediatric, ENT and dental opd.

CONCLUSION:
- Patients with GS due to a large variety of abnormalities and different severity of symptoms pose a challenge for clinicians. All of this necessitate an individual approach to each single patient and involvement a team of specialists in treatment planning. It is a complex, long-lasting, multidisciplinary process and should be divided into stages, according to patient's age, as well as the extent and severity of observed abnormalities. Neonatologists and pediatricians are involved in care of these patients from the onset.

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