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Goldenhar Syndrome: An Unusual Case Report

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Abstract:- Goldenhar syndrome is a rare and sporadic condition of unknown etiology characterized by a combination of anomalies: oculo-auriculo-vertebral (OAV) dysplasia, auricular appendices, dermal epibulbar cysts and malformation of the ears.Here, we report an unusual case of Goldenhar syndrome with skin lesion suggestive of xeroderma pigmentosa.Patient with Goldenhar syndrome has inconsistent presentation and can affect the various parts of the body.Multidisciplinary approach is mandatory for early diagnosis and managementto avoid from severe complications in later stage of life.

Keywords:- *Goldenhar syndrome, Oculoauriculovertebral, Preauricular tags, Vertebrae.*

I. INTRODUCTION

Goldenhar syndrome is a rare congenital condition of unknown etiology which was first reported by a Swiss OphthalmologistMaurice Goldenhar in 1952.[1]It is also known as oculo-auriculo-vertebral (OAV) dysplasia which mainly arises from defects in the first and second brachial arches. [1,2]Few literatures reported that the frequency of Goldenhar syndrome would be 1:35,000-1:56,000 with a male to female ratio of 3:2.[3]Various etiological factors; abnormalities of chromosomes, neural crest cells, ingestion of drugs during pregnancy such as thalidomide, cocaine, retinoic acid, tamoxifen, exposure to tobacco or herbicides, malnutrition, maternal diabetes and intake of alcohol would be plausible caused for the evolution of the disease..[4]
Xeroderma Pigmentosum (XP) is a rare autosomal recessive genetic disorder due to defective in DNA repair which leads to extremeclinical and cellular hypersensitivity to ultraviolet radiation.Few studies reported that approximately 80% and 20% of XP have ocular and neurological complications respectively.[5]

II. CASE REPORT

Aneleven-year-oldfemale patient presented with chief complaints of diminution of vision along with painless swelling in the righteye since birth. On examination her best corrected visual acuity was 6/36 and 6/6 in the right eye and left eyerespectively.Ocular examinationof both eyes revealed full extraocular movements in all cardinal gazes. Slit lamp bio-microscopic examination divulgedmultiple brown black pigmented conjunctival lesions in both eyes whereasright eye had skin colored round lesion suggestive of limbal dermoid (5 x 5 mm) in infero-lateral regioncovering half of the cornea. (Fig 1) Anterior segment of the left eye as well as posterior segment of both eyes were normal.On further examination of the right ear two pre auricular skin tagswere seen whereas the left ear was normal. (Fig2). However, she didn't give any history suggestive of hearing impairment. Audiometry along with X-ray para nasal sinus (PNS) waswithin normal limit. On oral examination microdontia was seen (Fig 3)Cardiovascular, central nervous and Musculo skeleton system examination were within normal limit. Patient was referred to higher center for further examination to rule other systemic anomalies.Examination of skin revealed black brownish hyperpigmented lesion in sun exposed area suggestive of xeroderma pigmentosa. (Fig 4) Patient was sent for dermatological consultation for further management but she didn't come for follow up.

Without above findings patient was diagnosed with Goldenhar Syndrome with multiple skin lesions suggestive of xeroderma pigmentosa.

III. DISCUSSION

Goldenhar syndrome is a rare and sporadic condition of unknown etiology characterized by a combination of anomalies: oculo-auriculo-vertebral (OAV) dysplasia, auricular appendices, dermal epibulbar cysts and malformation of the ears.[1,2]

The incidence of this syndrome is 1:3000 to 1:26,500 witha male predominance.GHS is a raredisorder of multiple congenital anomalies and are consistent with autosomal dominant, recessive and multifactorial patterns of inheritance. [3]GHS might be a result of fertilization of an over ripe ovum and hereditary pattern being the causative agent for it. [6]Previous report hypothesised thatembryologicallymesoblasts are being affected by some abnormal mechanism which in turnaffects the branchial and vertebral development systems late in the first trimester resulting the syndrome with multiple anomalies.[7]Baum and Feingold et al. stated that Goldenhar syndrome occurs in sporadic fashion during embryogenesis state which could be elucidated by epigenetic change, reduced penetrance or somatic mosaicism. [8,9,10]Gomez et al. postulated that implementation of cholecystographybetween the fourth and sixth weeks of pregnancy would be the etiological hallmark of the syndrome. [11]

In classic Goldenhar syndrome patient shows characteristic ocular,auricular,facial and vertebral features as given in following table. [12]

Ocular manifestations	Epibulbar dermoid or lipo dermoid (mostly bilateral), colobomas of the upper eyelid, iris, choroid and retina. Other eye anomalies (microphthalmia, anophthalmia, cataract, astigmatism, antimongoloid obliquity of palpebral fissures and blepharophimosis.
Auricular manifestations	Preauricular skin tags, microtia, middle and internal ear anomalies
Facial and oral manifestations	Unilateral facial hypoplasia, prominent forehead, hypoplasia of the zygomatic area, maxillary and mandibular hypoplasia, unilateral macro stomia
Vertebral anomalies	Vertebral column anomalies, rib anomalies and anomalies of extremities
Miscellaneous anomalies	Congenital heart disease, ventricular septal defect, anomalies of the urogenital (ectopic kidney), gastrointestinal system, anomalies of central nervous system, mental retardation

Management consist supportive multidisciplinary approach with the help of pediatrician, dental surgeon, dermatologist, ophthalmic and ear surgeons. Early diagnosis and prompt management would be recommended in each case of Goldenhar syndrome.

IV. CONCLUSION

GHS is associated with various congenital anomalies. Multiple approachesare required for prompt diagnosis and management to avoid from severe complications as well as to lessen the emotional, physical and financial burden of children.

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FIGURES:

Fig 1: Right eye showing inferotempotal dermoid covering nearly half of cornea.

Fig 2: Two pre auricular skin tags present in the right side.

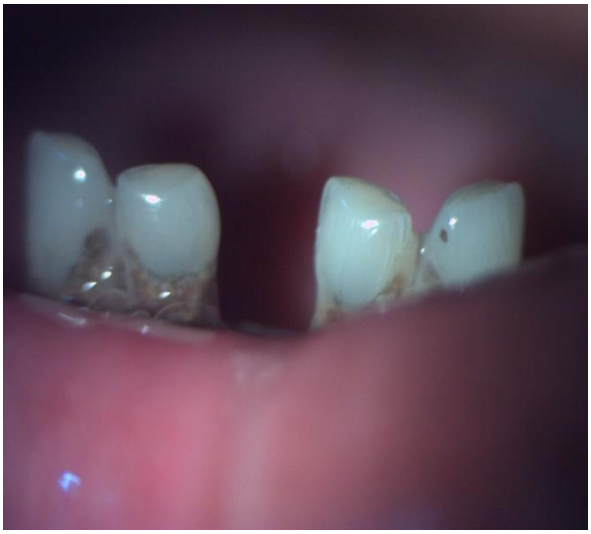

Fig 3: Showing microdontia

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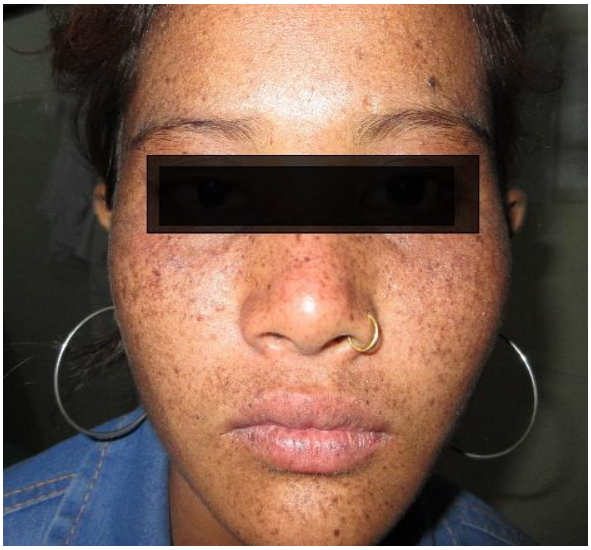
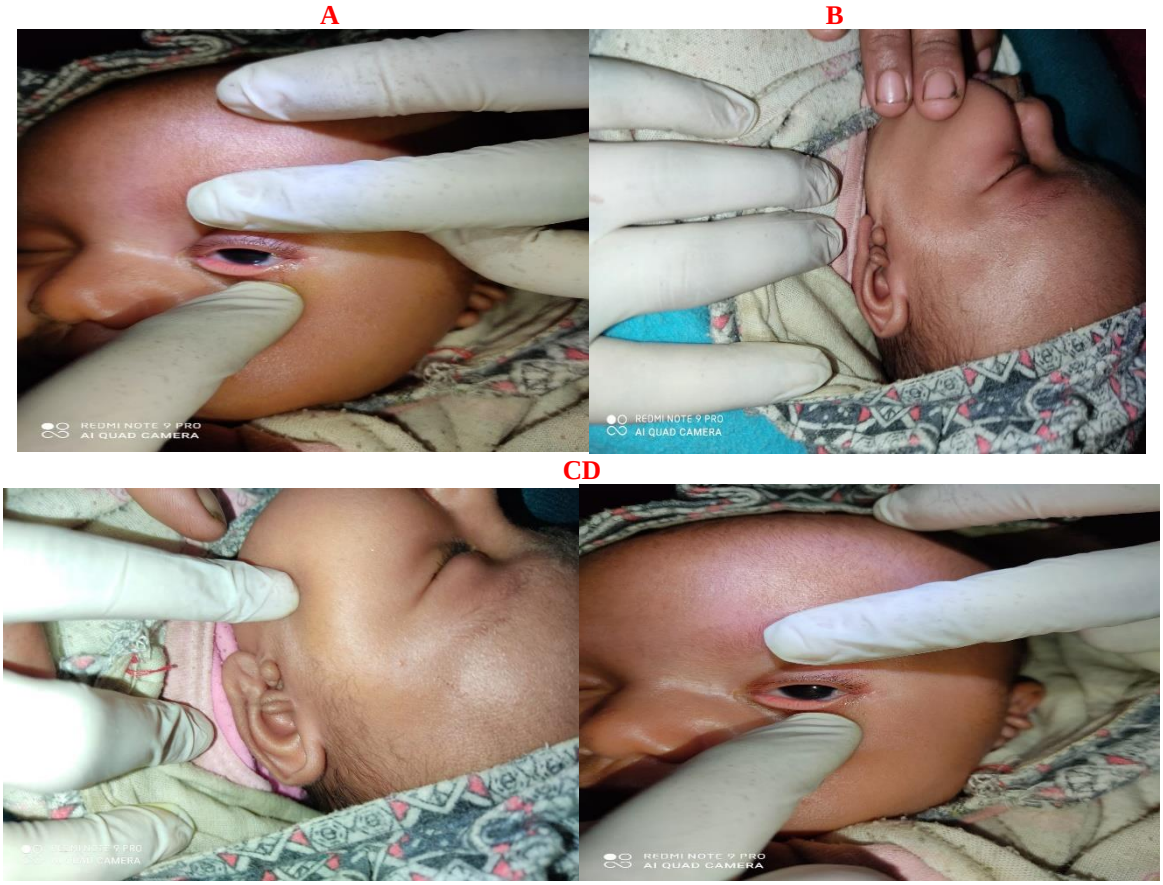


Fig 4: Examination of skin revealed black brownish hyperpigmented lesion in sun exposed area suggestive of xeroderma pigmentosa.

Picture A, B, C, D shows second case of Goldenhar syndrome in a new born child.



Picture A and D shows a one-month-old child with left eye temporal limbal dermoid. Picture B and C shows a same child with left side preauricular skin tag suggestive of Goldenhar syndrome.