GOLDENHAR SYNDROME: A VERY RARE CASE REPORT

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ABSTRACT

A 20 year old female presented with a swelling arising from lateral canthus of Left Eye which was gradually increasing in size for last one year. On examination, an accessory auricle was found in left ear. The case was diagnosed as Goldenhar syndrome and was planned for excision of the mass and histopathological examination.

INTRODUCTION

Goldenhar syndrome is also known as Oculoauriculovertebral dysplasia. It was named in 1952, when Dr. Goldenhar wrote about a number of facial problems that tend to occur together[5]. It is a rare clinical syndrome comprising of a triad of ocular dermoid, preauricular skin tag or accessory auricle and vertebral dysplasia[1].The frequency is 1/3,000 to 1/5,000 live births[2,3].The male to female ratio is 3:2.Pericentric inversion of chromosome 9 is one of the most common structural chromosomal aberration with its incidence of 15-25%[4].

CASE REPORT

A 20 year old female presented to our department with c/o a swelling at outer canthus of left eye which was gradually increasing in size and growing towards limbus.

On examination, the mass was soft, subconjunctival and movable over sclera.An accessory auricle was found on left side. Ocular movement was normal in all directions. Visual acuity was 20 / 20 and fundus was within normal limits. There was mild hypoplasia of muscles of face and cheek bones of the left side.

The mass was diagnosed as Dermolipoma and the case was diagnosed as Goldenhar syndrome. Right eye was normal in all aspects.The mass was planned for excisional biopsy. It was excised and suturing of conjunctiva was done.The excised tissue was sent for histopathological examination and diagnosis of Dermolipoma was confirmed from the histopathological report.

DISCUSSION

The first case of Goldenhar epibulbar dermoid cyst and congenital malformation was reported in 1952[5].There after, Gorlin and Pindborg [6] included vertebral anomalies in this syndrome.It has multifactorial etiology including nutritional and environmental issues resulting in disturbance of blastogenesis involving first and second brachial arches[7].Some studies have suggested a disturbance in neural crest cells[8].Vascular abnormality particularly haemorrhage and expanding haematoma formation in region of stapedial artery have been implicated as possible

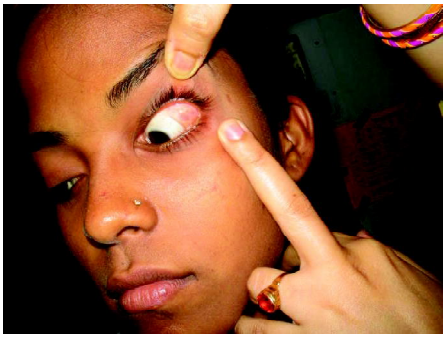

Figure-1 Dermo lipoma involving upper lateral fornix & canthus

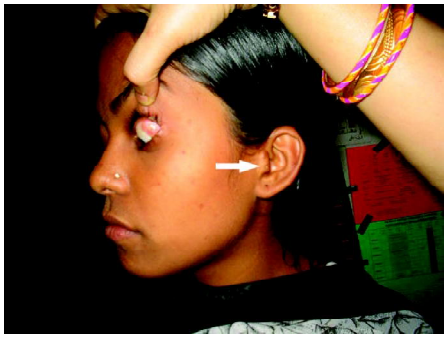

Figure-2 Pre auricular tag shown in white arrow

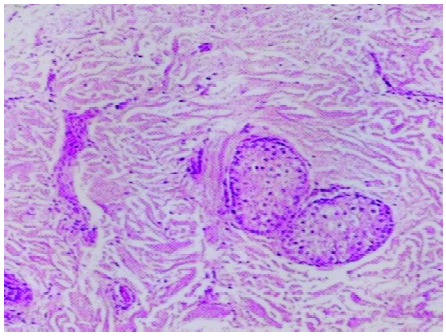

Figure-3 HE Stain showing collagen matrix with embeded dermoid tissue sorrounded by stratified squamous epithelium

environmental factors[9].Use of some drugs like Cocaine, Thalidomide, Tamoxifen, Retinoic acid during pregnancy can lead to this condition [8,9].

The diagnosis is based mainly on clinical aspects associated with systemic conditions and radiological findings. Our patient had anomalies of ear like accessory auricle like what is reported by Barbosa et al[8].Other ocular anomalies of this syndrome include coloboma of upper eyelid, uvea, choroids, microcornea and subconjunctival cyst[1].Auricular anomalies are usually unilateral and include posterior and inferior positions of ear, microtia, hypoplasia of external ear canal with or without hearing loss[1].

Hemivertebra, cuneiform vertebra and occipitation of atlas are most commonly seen vertebral anomalies[1]. Micrognathia, coronary heart disease, unilateral facial anomalies are sometimes seen.

CONCLUSION

Goldenhar syndrome; also known as Oculo-Auricular-Vertebral Syndrome is a congenital malformation of the jaw, cheek and ear associated with vertebral defects. There is deformity of the external ear and abnormal smallness of that half of the face. Coloboma of the upper eyelid with epibular dermoids in the fornix are frequent. The ear deformities range from tags in front of the ear, to atresia of the external auditory canal, abnormalities in the size and shape of the ear, and even anotia.. It is associated with anomalous development of the first branchial arch and second branchial arch. It is prevalent in males (70%) eyes. There is very little evidence to explain why Goldenhar Syndrome occurs. In most cases, Goldenhar Syndrome appears to occur randomly. However, in some cases, positive family histories have been present that have suggested autosomal dominant or rarely recessive inheritance. In addition, some researchers suggest that the disorder may be caused by the interaction of many genes, possibly in combination with environmental factors - multifactorial inheritance. Children with Goldenhar Syndrome usually look forward to a long life and normal intelligence .Cosmetic surgery is needed for correcting the deformities along with treatment for deafness as and where required.

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