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RESEARCH ARTICLE

GOLDENHAR SYNDROME A CASE REPORT

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ABSTRACT

A 12 year old patient presented with Limbal dermoid, Pre- auricular tags, Fused C3-C4 and C5-C6 vertebrae diagnosed as Goldenher Syndrome.

INTRODUCTION

Goldenhar syndrome (Kansky Jack, 2000) is rare inherited condition also known as Oculo-auriculo-vertebral syndrome with incidence of 1 in 3600 to 1 in 26000 live birth. This has a multifactorial etiopathology that includes nutritional, environmental and genetic factors and it is associated with incomplete development of the ear, nose, soft palate, lip and mandible. It is named after American Ophthalmologist Maurice Goldenhar (1924-2001). It is associated with anomalous development of the first and second branchial arches (Touliatow *et al*., 2006) Common clinical features (Martini- Junior *et al*., 2010) are Limbal Dermoids, Pre-auricular skin tags and vertebral anomalies (Engizo *et al*., 2007)

Purpose

To report a rare case of GOLDENHAR SYNDROME

MATERIALS AND METHODS

This is an observational study of an individual interesting case. The place of study is Sarojini Devi eye hospital which is a tertiary eye care hospital attached to Gandhi Medical College. A 12 yr old female patient born out of non-consanguinous marriage presented with mass in the right eye since birth.

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No history of similar complaints in the family. On examination visual acuity of right eye is 6/60 improving with pin hole to 6/6 and in left eye it is 6/6. There is a Limbal dermoid in right eye extending from 6 to 9'o clock position. Right Hemifacial atrophy with atrophy of cheek muscles is observed. There are pre-auricular tags in front of right ear.

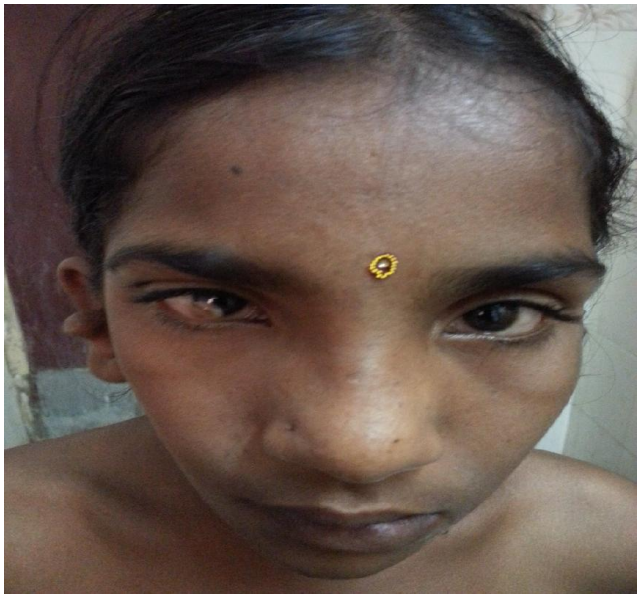

Fig.1. Showing Limbal dermoid in RE, Pre auricular tags & hypoplasia of right side of face

Fig.2. Shows Pre auricular tags & Limbal dermoid in RE

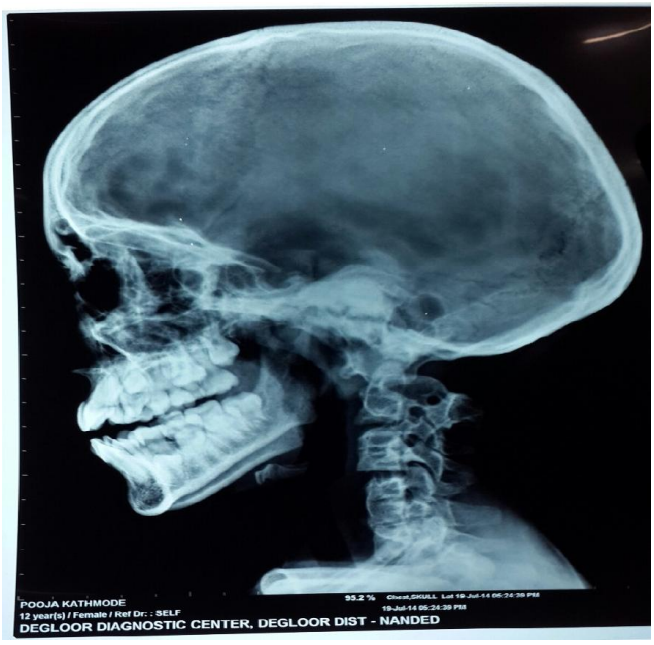

Fig.3. X-ray shows fused C3-C4 & C5-C6

Investigation

X-Ray skull AP and Lateral showed fused C3-C4 and C5-C6 cervical vertebrae

RESULTS

From above clinical findings and investigations she was diagnosed to have Goldenhar Syndrome and we have planned for excision of limbal dermoid and referred to plastic surgeon for further management.

Conclusion

Goldenhar syndrome is rare inherited condition which has multifactorial etiology and multisystem involvement along with ocular manifestations, so ophthalmologists has a role in diagnosing this condition.

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