CASE REPORTS

GOLDENHAR SYNDROME WITH UNUSUAL FEATURES

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We report two cases of Golderhar syndrome in whom bilateral ocular and auricular changes were observed. One patient also had hypertelorism, macrophthalmia and bulbous nose.

Key Words : Goldenhar syndrome, Hypertelorism, Macrophthalmia

Introduction

Goldenhar syndrome (oculo-auriculo-vertebral dysplasia with hemifacial microsomia) is a rare congenital anomaly involving first and second branchial arches. Though a male predominance is documented, we report two female patients with Goldenhar syndrome. Bilateral ocular and auricular changes were observed in both patients. One patient had hypertelorism and bulbous nose and the other had cleft lip and palate.

Case Reports

Case 1

A 2 - month-old girl, the only child of non-consanguinous parents, was referred to the Paediatric department for evaluation of cleft lip and cleft palate (fig.1). She was born of a full-term normal vaginal delivery and her antenatal period was uneventful except for feeding problems.

She was an SGA (Small for Gestational Age) baby with head circumference 35cms, length 40cms and weight 2.2Kg. In addition to cleft lip and palate she had bilateral epibulbar dermoids and coloboma of upper eyelids and bilateral preauricular tags. She also had micrognathia . No vertebral anomalies were observed.

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Case 2

An 8-year-old girl, the second child of non-consanguinous parents was evaluated for facial dysmor phism (Fig.2). Her scholastic performance was normal.

She had bilateral epibulbar dermoids, upper eyelid coloboma and hypertelorism. Other features were bulbous nose, bilateral preauricular tags and macrostomia with cleft-

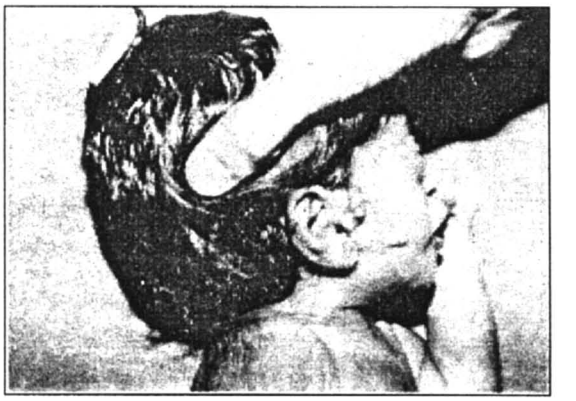

Fig.1. A 2-month-old girl with Goldenhar syndrome

like extension of the corners of mouth. Her cardiovascular and other systems were normal.

Discussion

Goldenhar syndrome, an autosomal dominant condition was described by Goldenhar in 1952.[1] The prevalence is 1:4000 live births.

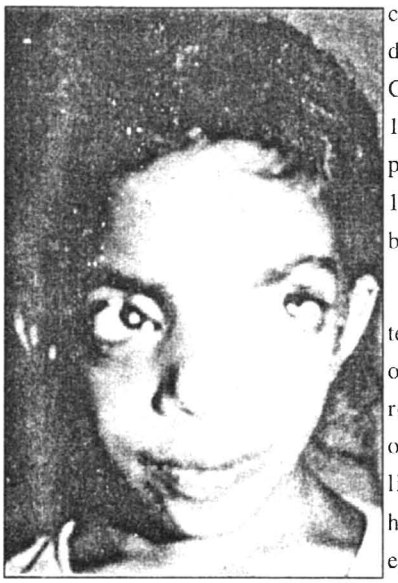

Fig.2. Goldenhar syndrome : Note hypertelorism, bilateral epibulbar dermoids and bulbous nose

The characteristic features of this syndrome include ocular changes [2] like microphthalmia, upper eyelid coloboma, epibulbar dermoid, micrognathia, macrostomia and microtia with preauricular tags in a line from the tragus to the angle of mouth. In 70% of cases the anomalies are asymmetric and unilateral.[3] But our patients had bilateral anomalies, which are rather unusual. Other unusual features were macrophthalmia and hypertelorism in the second patient. Hypertelorism is often associated with trisomy 22 and such patients are likely to have aneuploidy as well.[4] Chromosome analysis will help to find out such an association. Unfortunately this could not be done in our patients. In Goldenhar syndrome the nostrils are poorly developed giving a 'parrot-like' appearance.[5] But our 8 year old girl had a bulbous nose which has not been reported so far.

References

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