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GOLDENHAR'S SYNDROME (OCULO-AURICULO-VERTEBRAL DYSPLASIA)

N. K. Mehdiratta, D. R. Singh, P. C. Misra and S. K. Singh

SUMMARY

Oculo-auriculo-vertebral syndrome (Goldenhar's syndrome) was found in two cases, who had epibulbar dermoids, pre-auricular skin tags and hypomandibulosis. One case also had left sided complete congenital inguinal hernia.

(*Key Words* : Epibulbar dermoids, Pre-auricular appendages, Hypomandibulosis, Colobomata)

In 1932 Goldenhar reviewed 19 cases of oculo-auriculo-vertebral syndrome (Goldenhar's syndrome). Till 1966 only 40 cases were noted in the literature and in the ensuing decades only a few more cases have been added (Reddy and Rao, 1987; Bhattacharya et al., 1987). It is a complex developmental defect in which branchial arch structures are conspicuously involved, but it also affects the parachordal mesoblast (McKenzie, 1958 and Schultz, 1967). Cases have been described without vertebral anomalies (Sugar, 1966; Reddy, 1971 and Papp, 1974) and auricular defects. Epibulbar dermoids or lipodermoids are constant features and their presence differentiates the syndrome from mandibulo-facial dysostosis (Treacher Collin's syndrome).

Case Reports

Case No. 1 : A $3\frac{1}{2}$ years old boy, first issue, was born normally at full term. Mother aged 20 years was healthy and there was no history of taking any drugs during pregnancy. Examination revealed presence of pale yellow epibulbar dermoids at corneo-scleral junction. It was of 3 mm in diameter and situated at 9 o'clock position on the right side while it was of 5 mm in diameter and situated at 3 o'clock position on the left side. These exhibited smooth granular surfaces. The child had no visual or fundus problems. Accessory auricular tubercles were present on the right side in front of the tragus and on left side, the helix was deformed in addition to two tubercles one at base of helix and other being in front of the lobule (Fig. 1).

Case No. 2 : A 2 years old baby, second issue was born normally at full term. Mother aged 22 years was healthy and there was no history of taking any drugs during pregnancy. Examination revealed presence of a pale yellow epibulbar dermoid at 7 o'clock position. It exhibited smooth granular surface. Pre-auricular skin tag was present on the left side (Fig. 3).

Both the cases had mandibular hypoplasia which was more marked in Case No. 1. In addition, the Case No. 1 had left sided complete congenital inguinal hernia (Fig. 2). No spinal deformity was detected on x-ray spine of both the cases.

Discussion

Goldenhar's syndrome is complex and involves multiple tissues and shows considerable variability in its expression. Of the ocular, auricular and vertebral anomalies the first two are more constant while the vertebral may not be present in some cases. Epibulbar dermoids or lipodermoids are most important diagnostic features, which are bilateral in about 75% cases. They are yellow or white, with smooth granular or fine hairy surface. Dermoids usually lie in the lower outer quadrant while lipodermoids are normally found in upper quadrant. Coloboma is present in 60% of the cases. In our cases, except coloboma all above

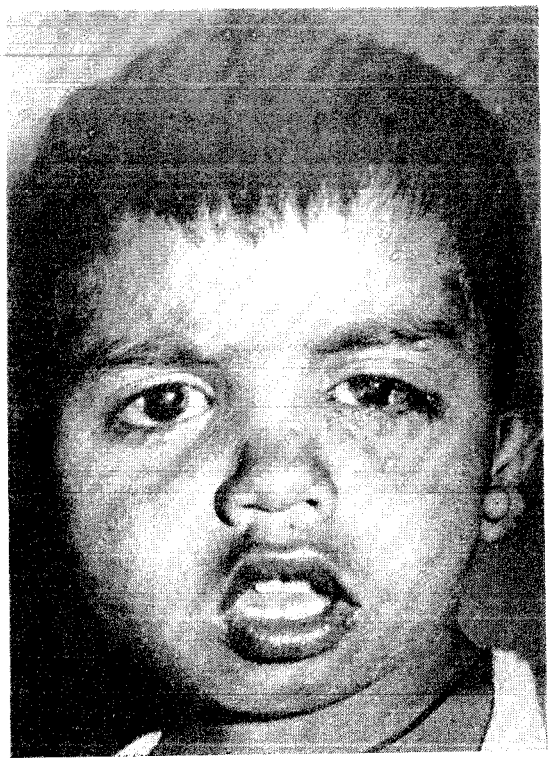

Fig. 1. Case No. 1 : Goldenhar's Syndrome.

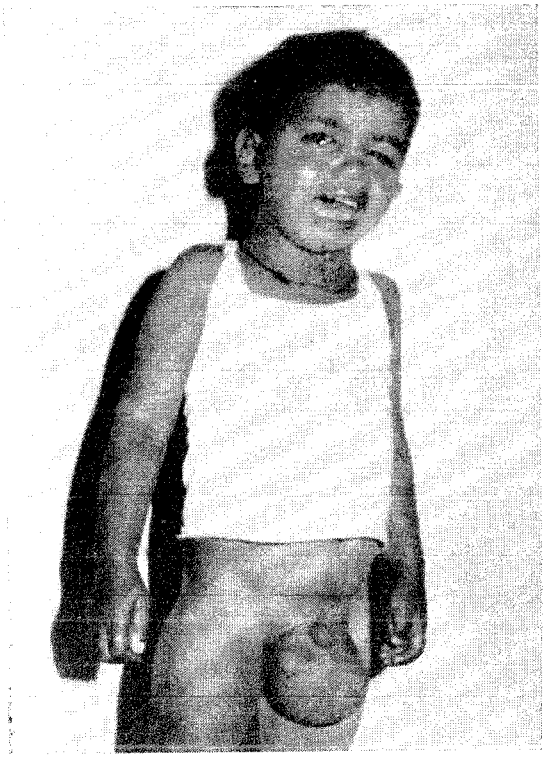

Fig. 2. Case No. 1 : Goldenhar's Syndrome with complete inguinal hernia left side.

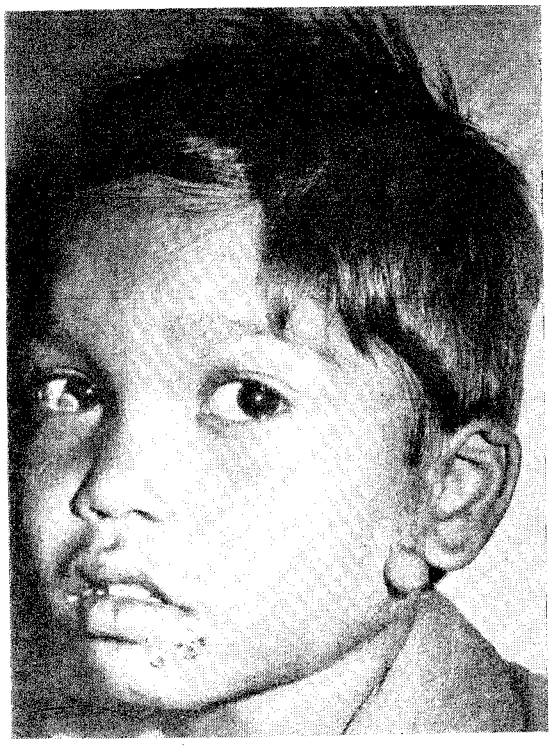

Fig. 3. Case No. 2 : Goldenhar's Syndrome.

abnormalities were present. Other ocular anomalies described by Gorlin (1963), such as microphthalmia, microcornea, anophthalmos and colobomata of the choroid were absent.

Multiple preauricular appendages along with helical deformities were present in our cases. Blind fistulae unilateral or bilateral, often found in these were not seen. Microtia and deafness with or without disturbances of external auditory meatus may also be present. In our cases deformity of pinnae were present. There may be macrostomia due to fibrous band between the angle of mouth and tragus. Both these features were absent in our cases.

Other features, such as, high arched palate, bifid tongue and double lingual frenulum were not seen in our cases.

Skeletal manifestations are not infrequent and tend to involve specially the vertebral column such as occipitalization of the atlas, synostosis of two or more vertebrae. No vertebral malformations were present in our cases. Though mental retardation has been described but in our case intelligence was commensurate with age. No chromosomal abnormality or hereditary patterns have been detected. Paufique et al. (1968) thought that this could be due to genes undergoing mutation. Papp et al. (1974) believed the syndrome could be due to environmental effects.

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