Goldenhar syndrome (oculo-auriculo-vertebral dysplasia) in a twin new born baby

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A case of oculo-auriculo-vertebral dysplasia (Goldenhar syndrome) in one of the twin babies is reported for the first time from India.

Key words : Goldenhar syndrome; dizygotic twins.

The Oculo-auriculo-vertebral dysplasia, described for the first time by Goldenhar1 is a disorder affecting the structures which develop from I and II branchial arches. It is characterised by epibulbar dermoid cyst and preauricular skin appendages with or without vertebral defects.2

This syndrome has always been reported in single born babies from India2-4 but in western literature 3 cases have been described in one of the twins.5 We have also studied a case of Goldenhar syndrome in one of the male twins delivered at JN Medical College Hospital, AMU, Aligarh. The other male twin baby was apparently normal which is quite interesting and is the first case of its kind from India.

Case report

A thirty years old 3rd gravida healthy mother delivered male twins per vaginum on 22-12-83 following full term uneventful gestation. The first male twin baby was born by breach presentation, cried soon after birth, weighed 2.7 kg and had an uneventful neonatal period.

The other male baby weighing 1.6 kg was born by vertex presentation and cried soon after birth; Apgar score at 1 minute was 8/10. On clinical examination he was designated as small for date full term baby. He had left sided facial prominence, depressed nasal bridge and a small cystic swelling on the right side of the forehead just above the lateral end of the right eyebrow. The nose was deformed and deviated towards right side but both nostrils were patent.

The preauricular skin appendages were present on both sides in front of the tragus measuring from 5 mm to 8 mm in length and 3-4 in number, however the oto-spic examination was normal. There was no hypoplasia of the jaw or macrostomia. Both lower eyelids had coloboma and near the lateral canthus of the left eye, a creamy white, pea sized epibulbar dermoid was found arising from the con-
Fig. 1. Male twin babies; baby suffering from Goldenhar syndrome was S.F.D.

junctiva of the lower eye lid. Fundus examination revealed no abnormality. All neonatal reflexes were normally elicited. Radiographs of the skull were normal but those of vertebral column showed hemivertebrae from C2 to T6. No evidence of occipitalisation of atlas could be seen. Other systems were apparently normal on clinical examination. There was no family history of such congenital anomaly and the two elder sibs were normal. There was no history of drug intake, infection and exposure to x-rays in this pregnancy or previous foetal and neonatal loss. Baby behaved normally in the neonatal period and regained birth weight by first week.

Discussion

Goldenhar syndrome is a complex developmental defect which usually involves the first and second arch structures alongwith the parachordal mesoblast. It consists of external ear malformations, ocular dermoids or lipodermoids, colobomata of the eyes, asymmetric facial under development, macrostomia and vertebral defects, although cases have been described without structural vertebral anomalies.

Gorlin et al emphasized the presence of epibulbar dermoid, which may be bilateral in 75 per cent cases and its presence eliminates the possibility of mandibulofacial dysostosis (Treacher Collin's syndrome) and hemifacial microstomia. Thus our case has all the principal features of this syndrome. The external and internal ears may be absent and associated with sensorineural deafness, but in our case audiopalpebral response was normal. Other features not present in our case but reported by others are maxillary and mandibular hypoplasia, high