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GOLDENHAR'S SYNDROME (A CASE REPORT)

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SUMMARY

Goldenhar's syndrome or occulo-auriculo-vertebral dysplasia is characterised by the presence of abnormalities of the first and second branchial arches accompanied by epibulbar dermoids or lipodermoids and vertebral anomalies. Other associated congenital anomalies may be frequently present. A case of Goldenhar's syndrome with macrostomia, transverse facial cleft and telepes calcaneovalgus deformity is being presented.

Goldenhar's syndrome (occulo-auriculo-vertebral dysplasia) is a variant of the first and second branchial arch syndrome and is specifically characterised by epibulbar dermoids and/or lipodermoids, unilateral or bilateral pretragal blind ended fistulae, auricular appendages and vertebral anomalies. Etiologically it is probably due to a vascular catastrophy between 3rd to 5th week of foetal life affecting the development and differentiation of the first and second branchial arches. Other congenital anomalies in the face accompanying the typical Goldenhar's syndrome are not uncommon.

Case Report

A 3 month old male child presented with disfigurement of ear, angle of mouth, neck, eye and foot all of the right side. The baby was one of the twin issues of the parents, the other baby, a female child, was completely normal. The parents had no similar congenital anomaly but the maternal uncle had a receding chin and a blindness in one eye, the cause of which could not be ascertained.

An examination of the baby revealed a normal shape of the calvaria with open fontanels. There was micrognathia (Fig. 3) with a deviation of the chin to the right and a smaller horizontal and vertical ramus of the mandible on the right side (Fig. 1). The right molar prominence was less marked (Fig. 5). The baby also had a cervical and upper thoracic scoliosis.

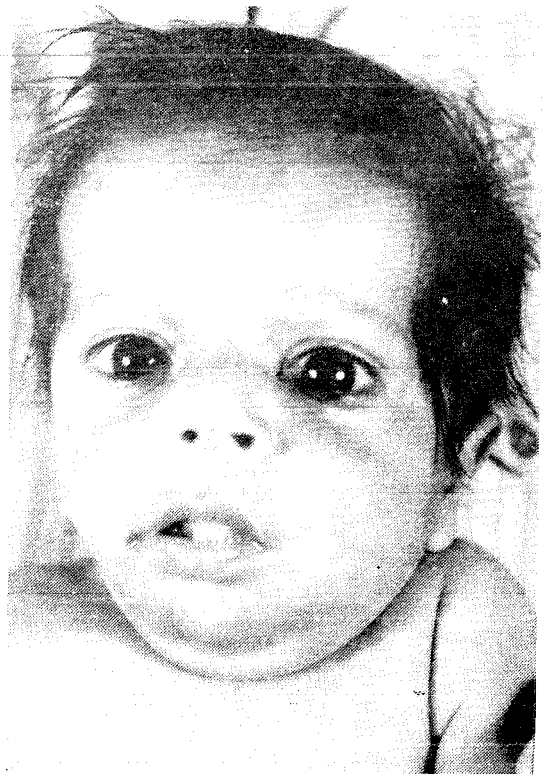
The entire orbital rim could be felt without any bony defect and the palpebral apertures were equal. The right eye however had an epibulbar dermoid (Fig. 2).

The external ear on the right side had a grade II deformity (Meurmann, 1957) (Fig. 3 and 4). The tragus, antitragus, helix, antihelix were not clearly distinguishable and the crumpled cartilagenous mass had a more horizontal lie on the profile than the opposite ear. From what appeared to be the lobule, a ridge extended to the right angle of mouth (Fig. 4) and there was a pretragal blindly ending fistula. There was macrostomia on the right side (Fig. 1 and 4) and the right ala of nose was thinner and moved less with respiration.

The right feet showed a telepes calcaneovalgus deformity (Fig. 6). An X-ray of the cervical spine showed hemivertebrae C_{6-7} and fused 1st and 2nd ribs on the right side.

Discussion

Occulo-auricular dystrophy was for the first time described by Von Arlt in 1895 quoted by Reddy and Rao, 1987; the eponymn Goldenhar's syndrome was coined in 1952. Goldenhar drew the attention of association of epibulbar



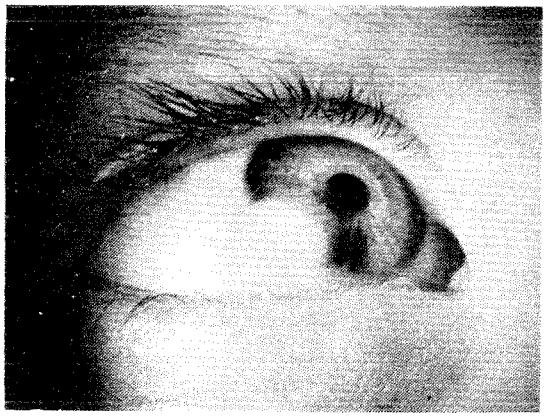


Fig. 2. An epibulbar dermoid in the lower temporal quadrant of the globe.

←Fig. 1. Front view of the face showing a grossly asymmetrical face, macrostomia, a heavy lower lid and a thinner alar rim with a smaller nostril on the right side.

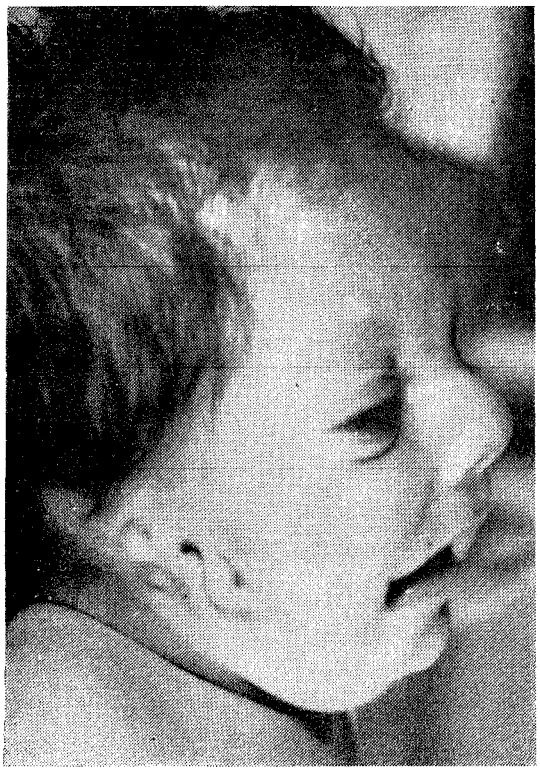


Fig. 3. The profile of face showing a grossly disfigured external ear with a more horizontal lie than normal.

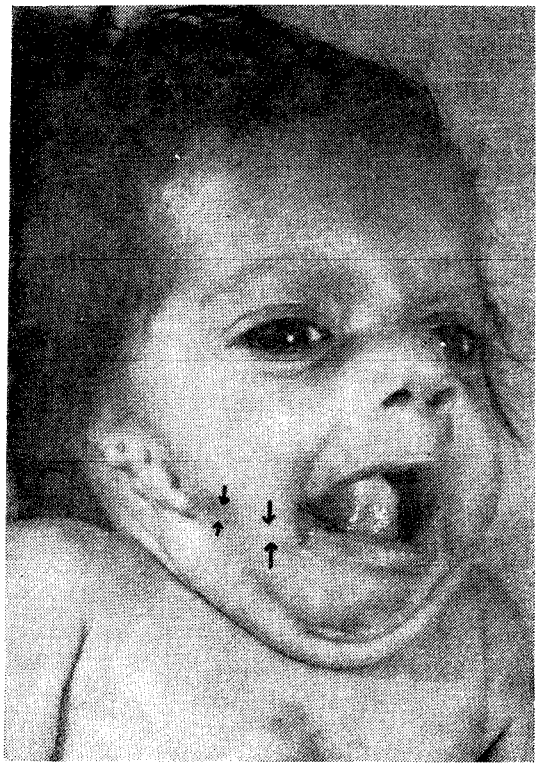


Fig. 4. A deformed external ear and micrognathia on the right side. In between the lobule of ear and micrognathia is a ridge—the abortive transverse facial cleft.

Fig. 5. A bird's eye view showing a depressed right malar prominence.

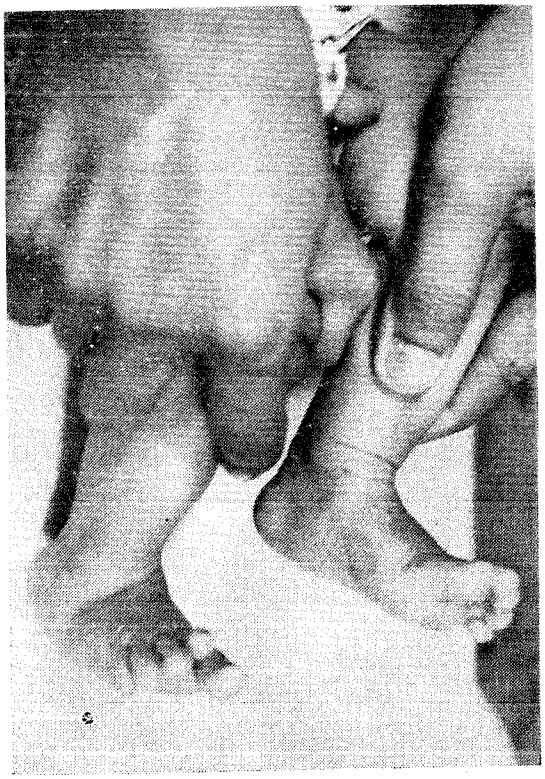
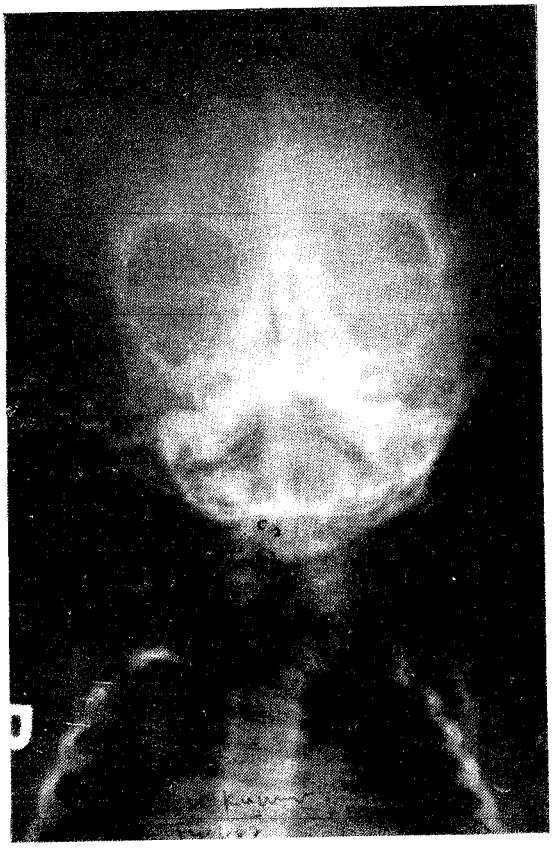
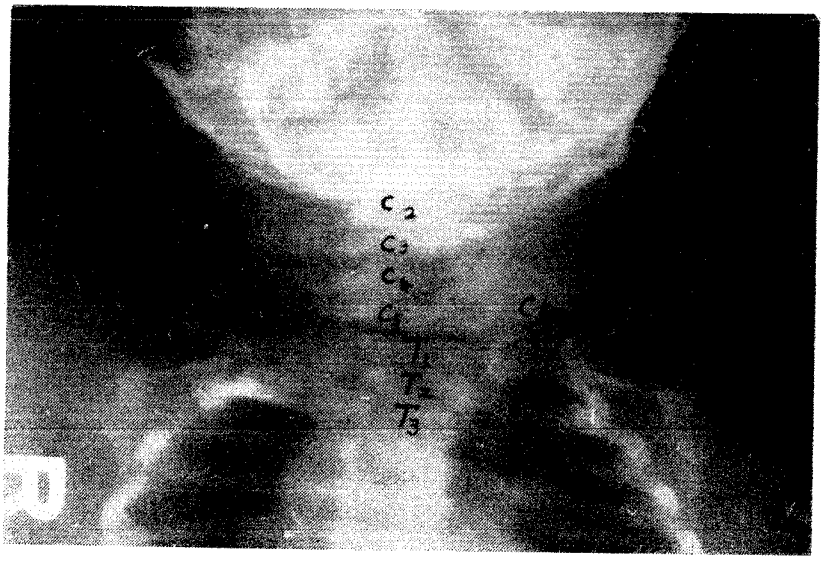


Fig. 6. Telepes calcaneo-valgus deformity in right foot.



(a)



(b)

Fig. 7a & b. X-ray spine showing hemivertebrae C_6 and C_7 and fused 1st and 2nd rib on the right side.

dermoids, lipodermoids, auricular appendages and/or pre-auricular fistula. Subsequently Gorlin and Pindborg (1969) added vertebral anomalies to this symptom complex and called the syndrome 'occulo-auriculo-vertebral dysplasia'. There is no evidence of any hereditary pattern and chromosomal studies do not reveal any abnormality (Reddy and Rao, 1987).

Epibulbar dermoid in the lower temporal quadrant of the globe is a constant feature. Their presence is because of abnormal differentiation of pleuripotent embryonic cells located between the edge of the optic cup and ectoderm. When lipodermoids are present, they are usually in the upper temporal quadrant. Colobomas in upper lid are fairly common.

The auricular deformities are variable and are a flag sign for middle and internal ear disturbances. The degree of severity of auricular disfigurement has no relation to the hearing capability or the anatomical structure of middle ear. Hearing loss, which is conductive in origin, can be checked by audiometry and middle ear morphology by a tomogram.

Vertebral and costal anomalies in association with first and second branchial arch syndrome are rather rare and Grab (1965) reported 11 out of 102 patients with congenital anomalies in vertebrae and/or ribs. Hemivertebrae, fused rib and scoliosis which were seen in the present case were all reported by Grab in his 11 cases. Reddy and Rao (1987) presented 6 cases but vertebral/costal anomalies were seen in none of them

The skull is often asymmetric with frontal bossing. The hairs are often situated low on the brow, the mild malar hypoplasia and receding chin, as seen in our patient, gives the individual a parrot like face with gradual development. Micrognathia is fairly common being present in 60% of the cases (Converse, 1977).

Other oral anomalies include cleft lip and palate (Gosserez, 1968) bifid uvula, bifid tongue, and double lingual frenulum (Goldenhar, 1952; Gorlin and Pindborg, 1964; Nyhan 1973; Jaworska and Dudkiewicz, 1974).

Macrostoma is another common accompaniment which has been reported by Reddy and Rao (1987) in 4 out of 6 cases and by Jaworska and Dudkiewicz (1979). What was

peculiar in our case was a remanent of a transverse facial cleft (Tessier's 7 cleft) extending from the angle of mouth to the deformed auricular cartilage and lobule.

Vascular defects of the stapedial artery may account for the maldevelopment of first and second branchial arches (McKenzie and Craig, 1955). The stapedial artery, a temporary vascular supply for the primodia of first and second branchial arches, appears as a collateral of the hyiod artery and anastomoses with pharyngeal artery; it is ultimately replaced by the finite external carotid system. Posivillo (1973) produced phenotypes of craniofacial microsomia in mouse and monkey by administering triazene and thalidomide respectively and a spreading haematoma prior to the formation of stapedial artery was demonstrated. The extent of the haematoma was directly related to the extent of deformity in the Ist and IInd branchial arch derivatives. So a vascular insult at a critical time i.e. 3rd to 6th week of intra uterine life involving the stapedial artery is probably the cause of first and second branchial arch syndrome, of which Goldenhar's syndrome is a variant. Our patient was born to a hypertensive mother who took Alpha methyl dopa during this critical period of development. How the drug is related to the aetiology is yet to be seen.

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