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Goldenhar's Syndrome

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THE oculo-auriculo-vertebral dysplasia syndrome of Goldenhar, (Goldenhar 1952) is characterized by three main features.

1. Epibulbar dermoid or dermolipoma of the eye
2. Auricular anomalies
3. Vertebral anomalies

Besides the above constant features, other associated anomalies may be present. Two cases are presented.

CASE REPORTS

Case 1.

H.K.L., 19 year old Chinese female was referred to the Eye Clinic, University Hospital, Kuala Lumpur, in July, 1975, with a mass below the right zygomatic arch and a fleshy dermolipoma in the right eye. There was no relevant family history.

She was found to have the three characteristic features of Goldenhar's syndrome.

Ocular findings in the right eye were an epibulbar dermolipoma covering the temporal half of the cornea (Fig. 1) and a concomitant divergent squint with amblyopia. The visual acuity was reduced to counting fingers.

General examination showed a right hemifacial microsomia, five preauricular skin appendages placed along a line extending from in front of the

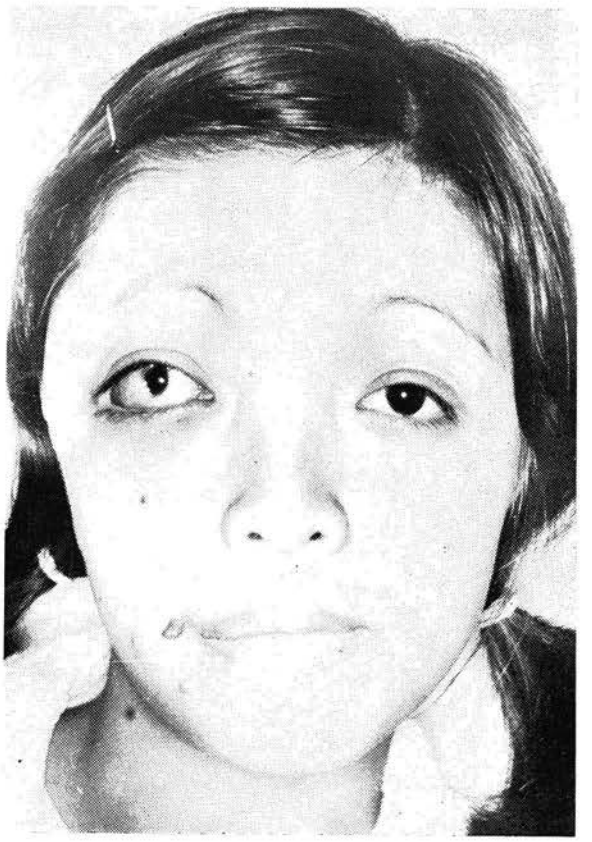

Fig. 1.
Right hemifacial microsomia, with skin appendages, dermolipoma in right eye with divergent squint.

ear to the angle of the mouth and a mass with overlying hypopigmented skin below the right zygomatic arch (Figs. 1 & 2).

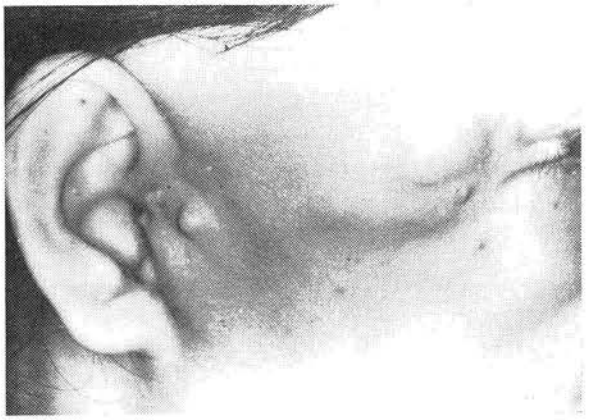

Fig. 2
Mass below right zygomatic arch and preauricular appendages.

Neurological examination revealed exaggerated knee and ankle jerks and bilateral positive Babinski. The reflexes of the upper limb and abdomen were normal. There was no sensory and motor involvement nor any sphincteric disturbances.

Radiographs of her facial bones and jaw demonstrated a right hypoplastic maxillary antrum and nasal cavity, a deviated nasal septum (Fig. 3) and three unerupted right upper molar teeth (Fig. 4). Radiographs of the cervico-dorsal spine revealed scoliosis of the dorsal spine with thinning and flattening of the pedicles of C_7 to T_5 vertebrae (Fig. 5) and the lateral view showed an increased

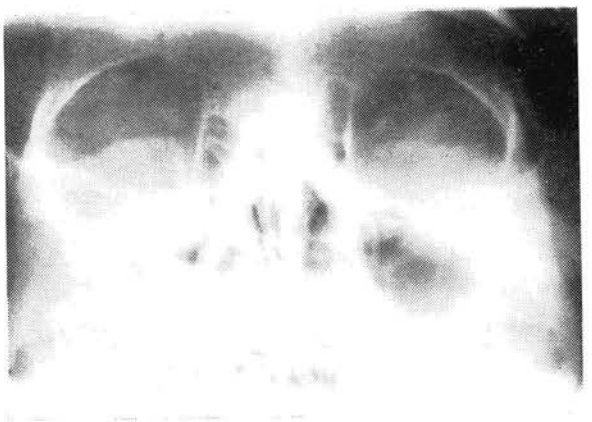

Fig. 3 Occipito-mental view of facial bones. Note the deviated nasal septum, hypoplastic right maxillary antrum with three unerupted molar teeth and hypoplastic right nasal cavity.

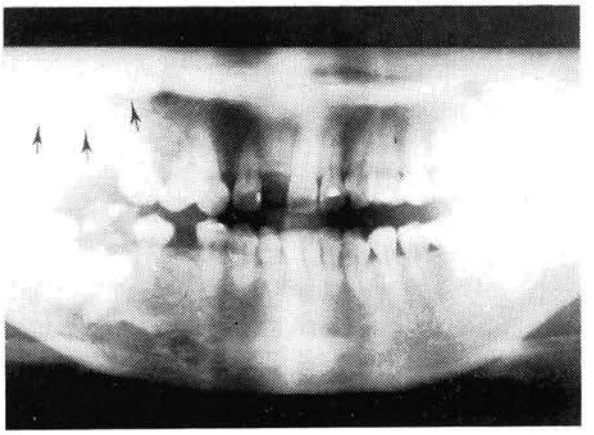

Fig. 4 Panoramic tomogram of the mandible. Note the three unerupted right upper molar teeth (indicated by arrows).

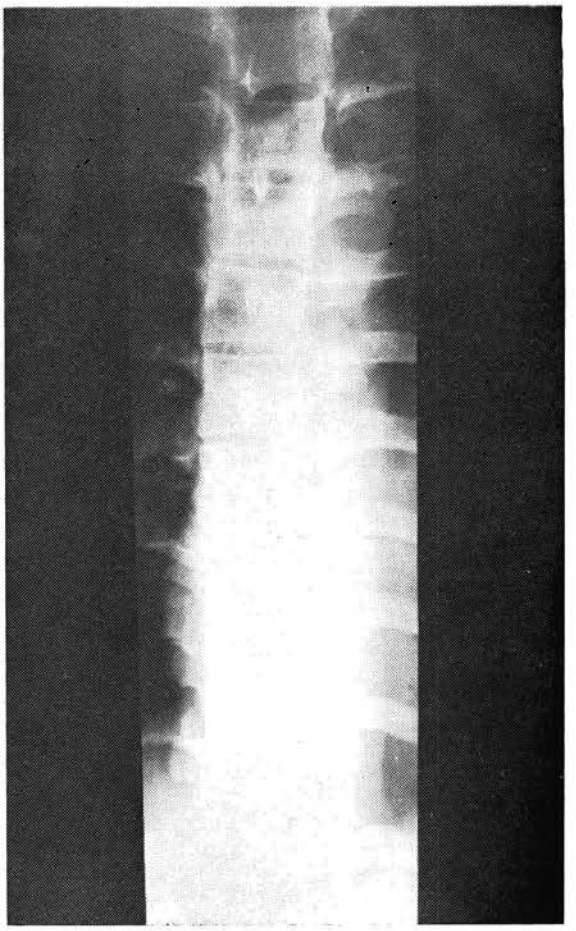

Fig. 5 Anteroposterior projection of dorsal spine. Note the flattened and thinned pedicles of C_7 to T_5 vertebrae and the increased interpedicular distance at the vertebral levels.

sagittal diameter of the spinal canal (Fig. 6). These radiographic findings suggested the presence of an intramedullary tumour of the spinal cord. A myelogram (Fig. 7) confirmed the intramedullary tumour extending from C_7 to T_5 vertebrae.

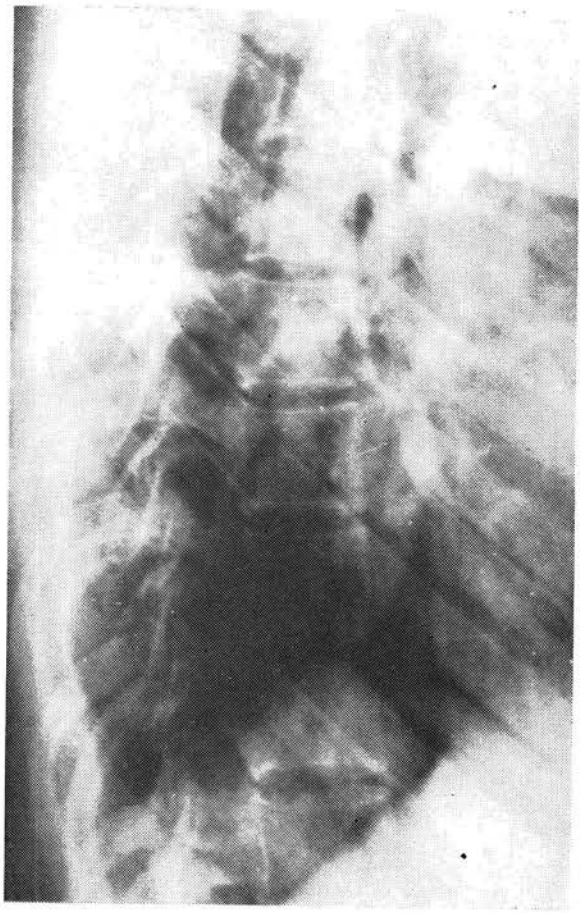

Fig. 6 Lateral projection of dorsal spine. Note the increased sagittal diameter of spinal canal from C_7 to T_5 vertebral levels.

Case 2

M.S., a six months old female Indian child was seen at the Eye Clinic, in August, 1975. There was no history of maternal illness during pregnancy. She was a normal full-term delivery. Her developmental milestones were slightly delayed. There was no relevant family history.

The eye examination showed a left inferior limbal dermoid (Fig. 8).

General examination revealed four left preauricular skin appendages and a left hare-lip with an anterior cleft of the hard palate (Fig. 9).

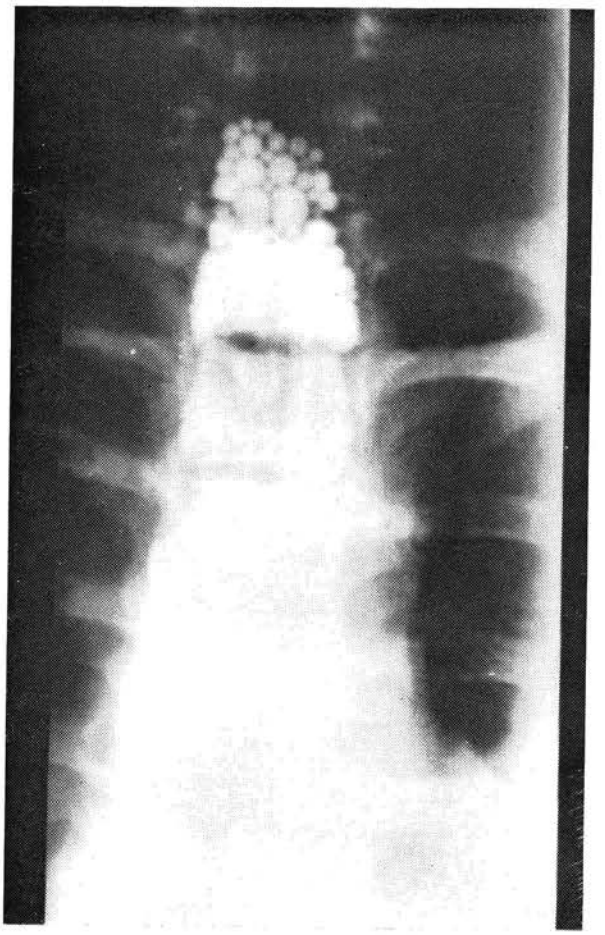

Fig. 7
Antero-posterior spot view of myelogram done in prone position shows a widened half shadow of spinal cord from C_7 to T_5 vertebral levels.

Radiographs of the facial bones and jaw were normal. Her chest radiograph revealed no abnormality.

Radiographs of her spine (Fig. 10) revealed spina bifida of T_1, T_2 and T_3 vertebrae with a hemivertebra at T_4 level.

Discussion

Goldenhar's Syndrome presents with three essential features which are ocular, auricular and vertebral. Other associated anomalies that have been described are varied in nature.

Ocular Anomalies. In the series by Baum et al (1973) a dermoid cyst or dermolipoma was bilateral in 23% and unilateral in 53% of the cases. Upper

lid colobomas were found to be unilateral in 21% and bilateral in 3% of the cases. Other ocular abnormalities described are Duane's retraction syndrome, anophthalmos, corneal anaesthesia and decreased tear formation.

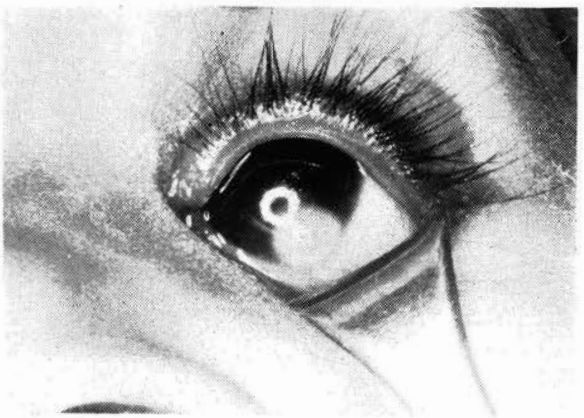

Fig. 8 Case 2
Inferior limbal dermoid of left eye.

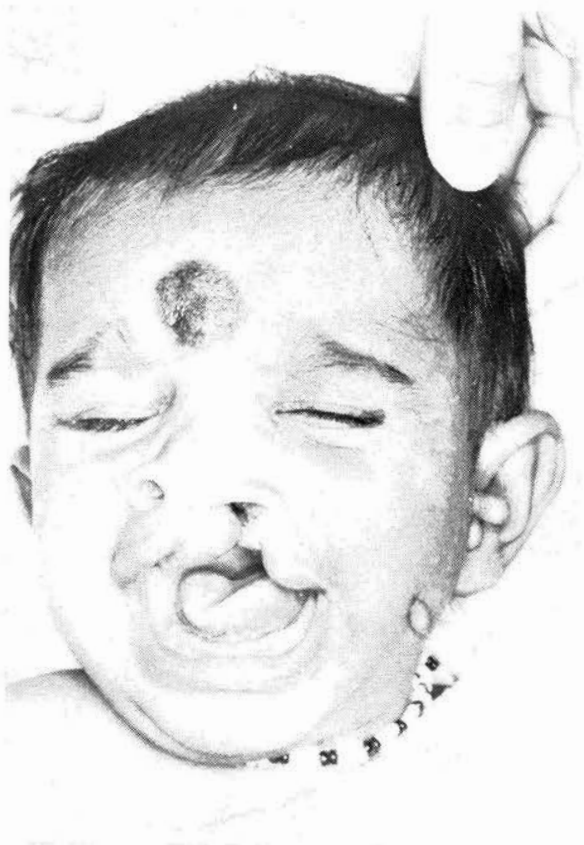

Fig. 9 Case 2
Left harelip with cleft palate and preauricular skin appendages.

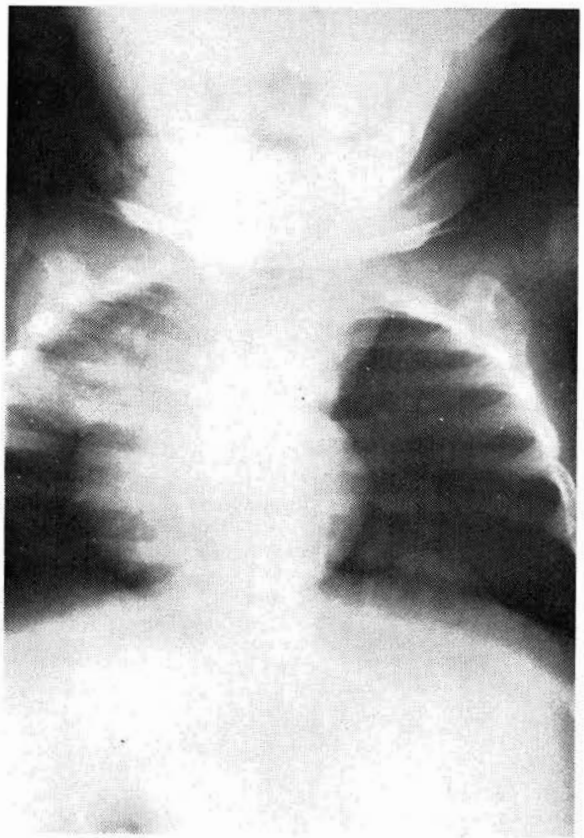

Fig. 10 Case 2.
Antero-posterior projection of dorsal spine shows spina bifida of T_1, T_2 and T_3 vertebrae and hemi-vertebrae at T_4, level.

Auricular Anomalies. The most constant feature is preauricular appendages of which 70% are unilateral. Other anomalies are posteriorly placed ears, microtia, aplasia or stenosis of the external auditory meatus. Hearing loss mainly unilateral, is usually a conduction defect (Baum, 1973).

Vertebral Anomalies. Vertebral anomalies vary; commonly seen are hemi-vertebrae, cuneiform vertebrae, narrowed disc spaces, spina bifida, scoliosis, and supernumerary thoracic or lumber vertebrae (Bowen, 1971).

Associated systemic manifestations. The cardio-vascular system is most commonly involved. Ventricular defect, Fallot's tetralogy, patent ductus

arteriosus, coarctation of the aorta and right bundle branch block have been described (Baum, 1973). Inguinal hernia, umbilical hernia, rectovaginal fistula, imperforate anus, high arched palate, bifid tongue and uvula and cleft palate have all been noted. Other associations include hydrocephalus, mental retardation and epilepsy (Bowen, 1971).

The first patient had an intramedullary spinal lesion. This association has not been previously repo rted. Syringomyelia and glioma of the spinal cord were considered but the absence of a definite sensory level, the site and extent of the lesion together with the lack of symptoms make the diagnosis of syringomyelia unlikely. The aetiology of this lesion is not confirmed as the patient was reluctant to undergo further investigation.

Summary

Two cases of Goldenhar's Syndrome are reported. Both cases presented with the three classical features of the syndrome together with associated anomalies. An intramedullary spinal tumour as an associated anomaly, as seen in Case 1, has not been reported previously.

Acknowledgements

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