

OCULO-AURICULAR CRANIAL DYSPLASIA*†

BY

J. S. GUPTA, S. D. GUPTA, AND S. K. PRASHAR

From the Department of Ophthalmology, Institute of Post-Graduate Medical Education and Research, Chandigarh, India

GOLDENHAR (1952) reviewed the literature relating to a syndrome consisting of epibulbar dermoids or lipodermoids, auricular appendices, and certain skeletal anomalies. Forty such cases or variants thereof have so far been recorded and Sugar (1966) added three more. We have recently seen an infant with ocular epibulbar dermoids, auricular appendices, congenital hydrocephalus, meningo-encephalocele, and a nasal deformity due to malunion of the medial and lateral nasal folds on the right side. It would appear that hydrocephalus has not previously been described in association with the oculo-auricular syndrome of Goldenhar.

Case Report

A male infant aged 4 months was brought to the Ophthalmological Department in January, 1967. There were several epibulbar dermoids on the limbus in both eyes (Fig. 1). The right conjunctiva was congested and some muco-purulent discharge was visible; this appeared to be due to stenosis of the naso-lacrimal duct. The eyelids were normal. Movements were full so far as these could be assessed. The pupils reacted, and the eyes could follow bright objects. The fundi showed no papilloedema or optic atrophy.

The left tragus (Fig. 2) was malformed and a few separate tubercles (auricular appendices) were situated anteriorly.

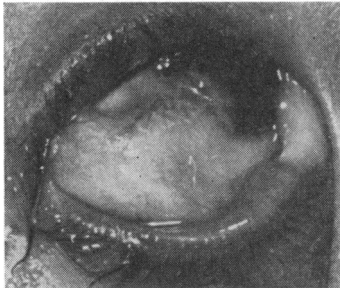


FIG. 1.—Epibulbar dermoids in lower temporal region of right eye.

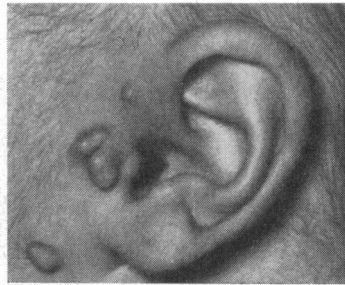


FIG. 2.—Malformed left tragus with several auricular tubercles.

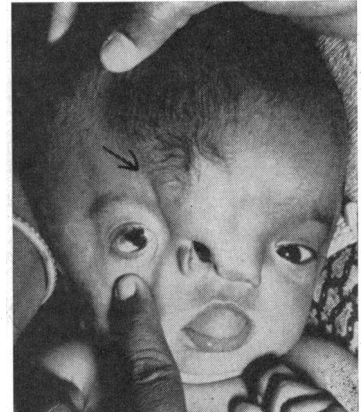


FIG. 3.—Hydrocephalus and malformed right nostril (ala of the nose). Arrow indicates meningo-encephalocele.

The right nostril (nasal ala) was malformed. The head was obviously enlarged (Fig. 3), the circumference being 52.5 cm. at the level of the external occiput. Both the anterior and posterior

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† Address for reprints: Dr. J. S. Gupta, 6J/7, Sector 24A, Chandigarh 23, India.

fontanelle were open and bulging outwards. The contour of the skull was irregular and on the right side a large bony defect was easily palpable through which one could feel a soft swelling (meningo-encephalocele) which became large and tense every time the child cried (Fig. 3, arrow).

The palate was highly arched. The central nervous system revealed no abnormality, and all four limbs moved freely with normal power. X rays showed no abnormality in the spine, chest, and pelvis, but the appearance of the skull confirmed the diagnosis of an internal hydrocephalus.

This was a second child of the family; the elder child was normal and there was no history of consanguinity.

Comment

Of the ocular anomalies in Goldenhar's syndrome, epibulbar dermoids are the most important, being bilateral and multiple in most cases. A palpebral coloboma is also frequently present. We have recently seen a girl about 12 years old with a coloboma of the lid and auricular appendices but without epibulbar dermoids. The skeletal anomalies are usually confined to the vertebral column, but in the patient described above the skull showed various deformities in association with an internal hydrocephalus. The malformed nasal ala and high arched palate were additional features.

Summary

A case of Goldenhar's syndrome is reported with epibulbar dermoids, auricular appendices, internal hydrocephalus, meningo-encephalocele, and a nasal deformity.

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