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Oculo-auriculo-vertebral Dysplasia (Goldenhar's syndrome)

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Summary

Osahon AI and Ibhanesebhor SE. Oculo-auriculo-vertebral dysplasia (Goldenher's syndrome). *Nigerian Journal of Paediatrics* 1995; **22:64.** Two cases of oculo-auriculo-vertebral dysplasia (Goldenhar's syndrome) are reported because of the extreme rarity of the condition. The first case was a five-month old male twin infant who had the ocular, auricular and vertebral manifestations of the syndrome. The second case was a nine-day old male infant with several features of the auricular component of the syndrome. The baby also had some degree of lumbar spinal curvature, presumed to be caused by vertebral anomalies.

Introduction

OCULO-AURICULO-VERTEBRAL dysplasia (Goldenhar's syndrome) was first reported by Von Arlt.[1] The syndrome comprises the classical triad of epibulbar dermoids (or lipodermoids), auricular appendages and vertebral anomalies.[2] Other terms that have been applied to this disorder include the first arch syndrome[3] and the first and second arch syndrome.[4] Twenty years ago, Odiase[5] reported one case of Goldenhar's syndrome from the same hospital as the present cases. At that time, the author noted that the case had not been reported previously in Nigeria and that only a few cases had been reported world-wide. It is evident from this small number of reported cases that the syndrome is relatively rare and for this reason of its extreme rarity, two other cases seen in the University of Benin Teaching Hospital (UBTH) are reported.

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Case Reports

Case 1: (UBTH No 272226) KU, a five-month old male second twin infant, was referred from a local private hospital to the UBTH with the complaint of multiple congenital anomalies. The child was the product of a full term, normal pregnancy and delivery. The first twin sib-

ling, who had no obvious congenital malformation, died from severe birth asphyxia few hours after birth. There were five other siblings who were reported to be alive and well. Both parents of no consanguinity, were alive and well. There was no family history of previous congenital abnormalies.

Physical examination revealed a well-nourished and active male infant, weighing 5.1 kg. There was no pallor of the mucosae. Heart sounds were normal and no murmurs were present. Breath sounds were normal and there were no adventitiae. There was facial asymmetry due to hypoplasia of the left side of the face (Fig 1). There was epibulbar dermoid in the inferior temporal region of the conjunctiva that encroached upon the cornea and a corneal opacity in the superior third of the cornea, just above the dermoid. The left malformed ear had a pre-auricular tag as well as being microtic (Fig 2). The abdomen was essentially normal except for an uncomplicated bilateral inguinal hernia. Radiograph of the lumbo-sacral spine revealed multiple hemi-vertebrae (Fig 3) and also dorso-lumbar scoliosis.

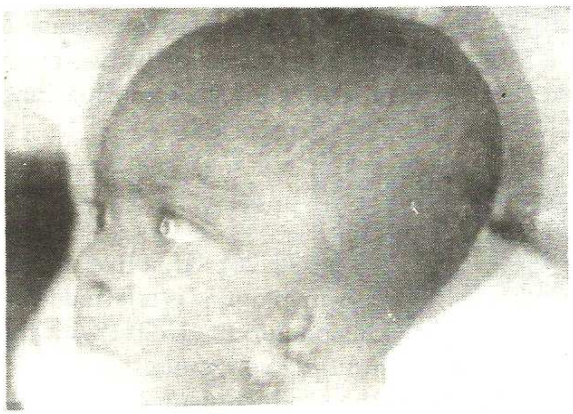

Fig 1: Case 1 with facial asymmetry. Note the left malformed and microtic pina.

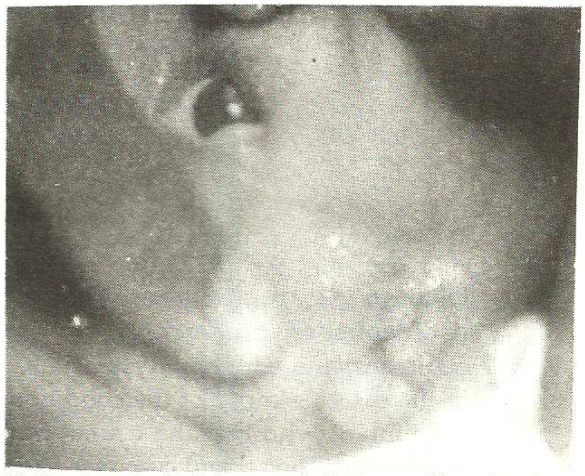

Fig 2: Case 1 with left pre-auricular tag, malformed and microtic ear. Note also the left epibulbar dermoid in the inferior temporal region of the conjunctiva and corneal opacity.

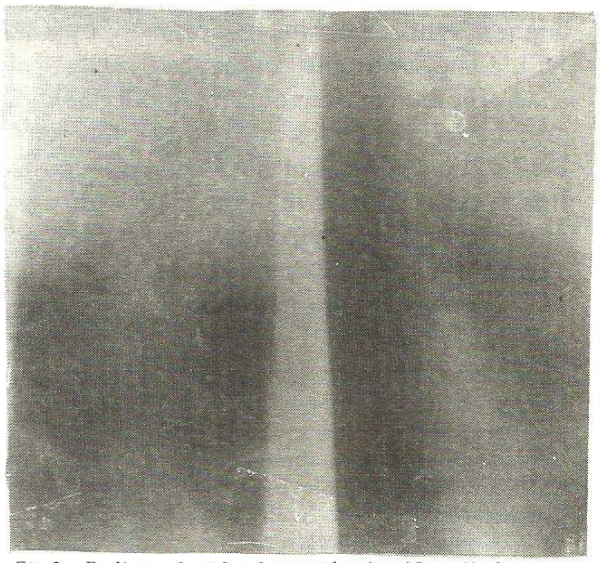

Fig 3: Radiograph of lumbo-sacral spine (Case 1) showing multiple hemi-vertebrae.

Case 2: (UBTH No 3318) BO, a nine-day old male newborn infant, was referred from a local maternity hospital to the UBTH, with a history of respiratory distress from birth. According to the referring physician, delivery was by spontaneous vertex approach to a 22-year old mother. Pregnancy was said to be normal. Physical examination revealed a term baby (birthweight = 2700gm), with a length of 46cm and occipito-frontal diameter of 35cm. There were hypoplasia of the right maxilla, anteriorly-displaced and a microtic right pinna, absence of the right external auditory meatus, left pre-auricular tags and microstomia. There was a mild degree of curvature in the lumbar spine. After a full sepsis work-up, the child was treated with parenteral cloxacillin and gentamycin, but he died suddenly seven days after admission at the age of 16 days. No skeletal radiographs were taken before death. Necropsy revealed no congenital anomaly of the internal organs.

Discussion

In Goldenhar's syndrome, the most constant aural manifestation is the pre-auricular appendage;[2] other less constant features include microtia, aural sinuses, deafness and absent or defective external auditory meatus. Of our two cases, the first one had the pre-auricular appendage or tags, microtia and malformed pinna, while the second case had pre-auricular tags in the left, right microtia and absence of right auditory meatus. With regard to the ocular manifestations, conjunctival dermoid is said to be the most constant.[3,6,7] As described above, our Case 1 had this ocular manifestation, while Case 2 did not. The vertebral component of the syndrome was present in our Case 1 and not in Case 2, from whom no vertebral radiograph was obtained before death. It is however, pertinent to note that this child had some degree of lumbar scoliosis. There can be no doubt, that this second case also had the syndrome, although there were no ocular manifestations, nor vertebral radiograph to confirm anomalies in the lumbar vertebrae. Absence of one or two of the three classical features of the syndromes, has been reported by other workers.[1,3,6]

Aknowledgements

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