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Oculo-Auriculo-Vertebral Dysplasia (Goldenhar's Syndrome)

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Summary

Odiase, V. O. N. (**1976**). *Nigerian Journal of Paediatrics*, **3** (1), 23. **Oculo-Auriculo-Vertebral Dysplasia** (**Goldenhar's Syndrome**). A case of oculo-auriculo-vertebral dysplasia (Goldenhar's syndrome) in a two-year old female child is described. The main complaint in this patient was macrostomia which superficially resembled cancrum oris, a common clinical condition in Nigerian malnourished children of this age group.

'OCULO-AURICULO-VERTEBRAL dysplasia' (eponym: Goldenhar's syndrome) is a combination of multiple congenital malformations involving mainly the eyes, ears, mouth and vertebral column. According to Goorlin *et al.*, (1963) and Smithells (1964), the syndrome was first recognized by Arlt (1845). In 1952, Goldenhar, after whom the condition is often called, reported three cases of his own, and reviewed the literature on 16 previously reported cases. The abnormalities constituting the syndrome are so typical that cases may be easily diagnosed at birth. This paper describes a case of this condition which, to our knowledge, has not been reported previously in a Nigerian.

Case Report

E. A. (UBTH 6790), a two-year old female, was referred to the University of Benin Teaching Hospital (UBTH) because of a defect of the right angle of her mouth which had been present since birth. The child had otherwise been quite well and there was no history of any infection of the affected cheek. She was the product of a full-term pregnancy which was supervised throughout in the antenatal clinic of the local hospital. The mother denied any illness during the pregnancy and only took the drugs prescribed for her at the antenatal clinic. Delivery was normal, and the neonatal period uneventful.

The baby was breast-fed from birth to the age of eighteen months. The development of the child was said to be normal; she started to sit, crawl and walk at five months, seven months and twelve months respectively. Dentition started at the age of seven months.

The patient was the seventh of eight children, all of whom were reported to be normal. Both parents were well. There was no family history of congenital abnormalities.

Physical examination revealed a well-nourished active female child of normal size. The facial appearance was distorted by a right-sided macrostomia which extended three centimetres into the cheek (Fig. 1). As a consequence of

this defect, there was constant dribbling of saliva which produced a patchy hypopigmentation of the right side of the lower lip. The jaws appeared normal and there was no dental malocclusion. There was frontal bossing and extension of hair well forward into the glabella, producing the so-called "hair tongue". There was bulbar epidermoid in the lower outer quadrant of the right eye (Fig. I). There was no coloboma of the eye lids. In the right ear there were several preauricular tags or appendages, a sinus above the tags, and a thick fleshy ridge in the anterior wall of the external auditory meatus (Fig. 2). Bone and air conduction of sound in both ears were normal.

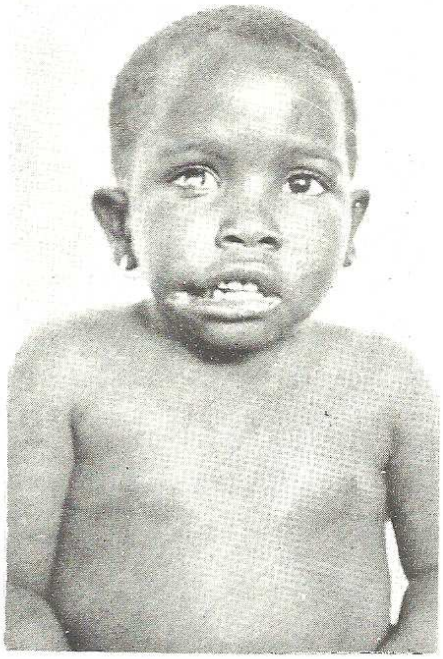

Fig. 1. Facial appearance of a two-year old girl with right macrostomia and bulbar epidermoid in the lower quadrant of the right eye.

The child walked with a lop-sided gait, and an examination of the back revealed a right-sided scoliosis. No abnormality could be detected in the cardiovascular, respiratory, nervous or gastrointestinal systems.

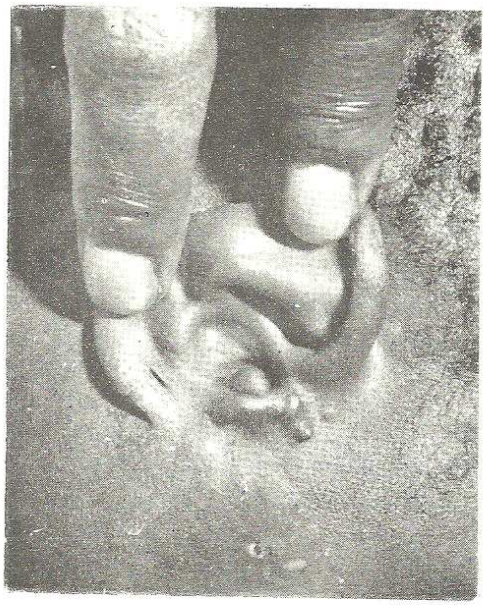

Fig. 2. Right ear of the patient showing several pre-auricular appendages and a thick fleshy ridge in the anterior wall of the external auditory meatus. Note also the sinus above appendages.

Investigations revealed a haemoglobin of 11.4 gms per 100 ml; packed cell volume of 34 per cent; Hb genotype AA; serum urea 21 mg per cent, and normal electrocardiogram. Skeletal radiographs showed posterior fusion of the right 8th and 9th ribs, and hemivertebrae involving D10–12 and L_1. (Fig. 3). The cervical spines were normal.

Discussion

The term, oculo-auriculo-vertebral dysplasia, first suggested by Gorlin *et al.*, (1963) is very descriptive of a typical case of the syndrome. The most obvious features of the syndrome are the preauricular appendages, the conjunctival dermoid, and the vertebral anomalies (McKenzie 1958; Smithells, 1964; Dumas and Charles, 1968). All these features occurred in the present case.

Macrostomia, which was the presenting complaint in the present case, has also been described by other workers (Mahnekce, 1956; Tranos, 1954), and according to Gorlin *et al.*,

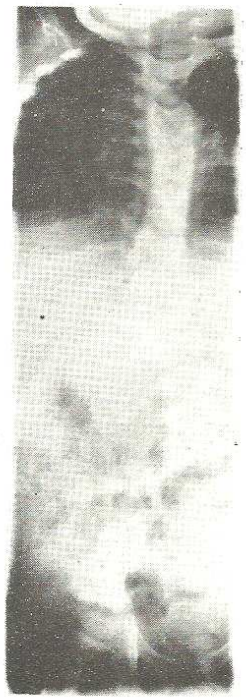

Fig. 3. Radiograph of the spine showing hemi-vertebrae involving D10–12 and L1.

(1963) this feature occurs in about 40 per cent of the reported cases. It is noteworthy that in the present case the macrostomia was not a sharp and well-defined lesion of the mouth as described by others, but it superficially resembled healed cancrum oris which is a common clinical condition among malnourished Nigerian children.

Aural manifestations, including microtia, aural appendages and sinuses, deafness and absent or defective external auditory meatus, are said to be fairly common features of this syndrome, the most constant feature being the preauricular appendages (Gorlin *et al.*, 1963). Ear tags by themselves are fairly common congenital anomalies and may not be particularly disfiguring so that patients as in the present case, may tend to ignore them. However, their presence should lead to further investigations (Dumas and Charles, 1968), since they may be accompanied by serious abnormalities.

McKenzie (1958) has included oculo-auriculo-vertebral syndrome in a group of congenital malformations which he calls the 'First arch syndrome'. However, in the differential diagnosis of the former syndrome two common malformations namely: mandibulofacial dystosis and hemi-facial microstomia, must be excluded. In this respect Gorlin *et al.*, (1963) have summarized the differential features, pointing out that, epibulbar dermoids, lipodermoids, vertebral and aural anomalies are constantly present in the oculo-auriculo-vertebral syndrome but absent in the other two conditions.

The aetiology of oculo-auriculo-vertebral dysplasia is unknown. However, it is generally agreed that there is no evidence of a hereditary factor, nor are chromosome abnormalities found in the syndrome (Gorlin *et al.*, 1963; Dumas and Charles, 1968).

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