Oxycephaly in an African Child Associated with other Abnormalities

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Oxycephaly is one of the craniostenoses, a group of conditions characterised by a premature closure of the sutures of the vault and base of the skull. As the brain grows, the capacity of the cranium has to be made good partly by an overgrowth of the great wings of the sphenoid, which bulge the skull laterally above the ears, but chiefly by an increased height of the vault. This characteristic has led to the synonyms tower skull, steeple head, turret skull, etc.

"The theories as to the aetiology and pathology are voluminous, complex and inconclusive" (Buckley and Yakovlev, 1948). There is a strong hereditary and familial factor which, together with the fact that the condition is much commoner in males and the common association with other congenital abnormalities, strongly suggest that it has a genetic origin.

In 1906 Apert described a closely analogous deformity under the name "acrocephalo-syndactaly." In addition to the characteristic appearance of oxycephaly, webbing of the fingers and toes is present. Crouzon in 1929 described a condition in which oxycephaly is associated with hypoplasia of the maxilla and absent or rudimentary maxillary sinuses.

As will be seen later, all three conditions were present in the following case.

Case Report

An African female child aged five years was admitted to hospital on 16th March, 1957, and discharged on 30th March, 1957, having been treated for acute bronchitis.

Family History.—There was no family history of congenital abnormality on either side of the family so far as could be ascertained by questioning the parents, both relatively unintelligent Africans. Examination of the three siblings showed them to be normal in all respects.

Personal History.—This was difficult to elicit, because although the child showed blatant abnormality, the mother apparently regarded it as quite unexceptional. However, the following milestones were obtained:—

At birth: Mother noticed webbing of fingers and toes (Fig. 1), and during the first year realised that there was some abnormality of the skull. Crawling was delayed until the second year, as was the breast feeding. Walking was delayed until the third or fourth year and the child is still unsteady on its feet. The child had a vocabulary of four or five words which had been gradually acquired since the second year. The child was incontinent of faeces and urine.

Examination showed a well-nourished African child with height 33 inches and circumference of the skull 19 inches. The skull showed typical high forehead, the highest part of the skull being at the anterior fontanelle, from which part there was a backward slope (Fig. 2). The antero-posterior diameter was much reduced. Exopthalmos was present with divergent strabismus. The lower jaw appeared relatively large and protruding. The upper and lower incisors did not articulate and the lower lip protruded (Figs. 3 and 4).

The expression was vacant and there was a trickle of saliva from the angle of the mouth.

The patient was found to be suffering from an acute bronchitis. An upper respiratory tract infection was also present. The bronchitis responded readily to treatment, but the patient was discharged with a mucopurulent nasal discharge little altered by treatment.

Mentally the patient was retarded, and it was our impression that the physical and mental development was roughly equivalent to that of an African child of eighteen months.

X-ray showed typical "digital markings," with thinning of skull bones. The W.R. of the infant and mother was negative.

Summary and Conclusions

As has been seen in this case, oxycephaly was present, together with hypoplasia of the maxilla and webbing of the fingers and toes. As far as the writer knows, this may be the first occasion on which the condition has been recorded in an African. The oxycephaly and associated abnormalities appear to be identical with such cases as described in the European.

REFERENCE

Illingworth. Vol. 9, British Encyclopaedia of Medical Practice.

Acknowledgment

I wish to thank Dr. D. M. Blair, O.B.E., Director of Medical Services, Southern Rhodesia, for his permission to publish this paper.