CASE REPORTS AND TECHNICAL NOTES

FAMILIAL SCAPHOCEPHALY*

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There have been many articles in the medical literature relating to craniosynostosis, in all its varied forms, since Virchow first described this entity and its symptoms in 1851. Numerous authors have discussed the clinical syndrome and the value of surgical intervention. There have been many modifications and improvements upon Lannelongue's first linear craniectomy for this condition, reported in 1890. Most authors, while admitting that most cases are sporadic in type, readily agree to the familial character of this disorder. Nonetheless, there are very few documented cases of familial or hereditary craniosynostosis. It is the purpose of this paper to present two families with hereditary scaphocephaly, and to request other authors to add to the literature by bringing to light other instances and other varieties of the familial form of this disorder.

In 1956, Greig suggested that hereditary examples are restricted, since those slightly afflicted do not seek medical advice and those grossly affected are not acceptable in marriage. Since then, Duguid, 2 in 1929, reported familial scaphocephaly in 4 siblings; Hilson 4 reported scaphocephaly in a brother and sister; and Murphy 5 reported scaphocephaly in a father and son. Greig, 6 in his review of the literature, mentioned a total of 10 families, with 18 instances of familial oxycephaly.

CASE REPORTS

Family "N." A 3½-year-old Caucasian male was brought to the neurosurgical clinic because of breath-holding spells, followed by loss of consciousness, then a generalized seizure.

Neurologic and general physical findings appeared normal except for the shape of the head, which was typical of scaphocephaly. Roentgenograms revealed premature closure of the sagittal suture (Fig. 1). A sibling, aged 2 years, also had breath-holding episodes with loss of consciousness, but no motor-seizure phenomenon. This child showed the same shaped head and closure of sutures (Fig. 2) as his brother. The third, and only other sibling, is a 4-week-old female. She had an elongated head (Fig. 3) and essentially nonexistent anterior fontanel, though the sutures could be palpated as open through the scalp. Roentgenograms of this child's head showed all the sutures to be open (Fig. 4), despite the abnormally shaped head and the extremely small anterior fontanel. Circumferences of the heads of the children were 20½", 20½", and 15", in order of age.

Review of the family history revealed that the father, paternal cousin, two paternal uncles, and paternal

* The opinions expressed in this article are those of the authors and not necessarily those of the United States Navy.
Family "S." Two children, aged 7 months and 9 years, the product of one mother and separate fathers, both presented clinical evidence of scaphocephaly confirmed radiologically (Figs. 5 and 6).

Surgery was recommended for the older child, when an infant, by another neurosurgeon, but was declined by the mother. This child is doing well at the age of 11 years, mentally and neurologically, except for the cosmetic appearance. There was, however, some pallor of the optic discs on examination without evidence of visual disturbances.

Surgery was recommended, and accepted, for the younger child. Bilateral linear parasagittal craniectomies were performed at the age of 8 months. This child, at the age of 8½ years, appears normal neurologically and cosmetically and does not have the dolichocephalic appearance of his half brother.

There is no history of craniosynostosis in any other members of this family. There are no other siblings.

SUMMARY

The above cases demonstrate hereditary passage through the male parent in the first family and through the female parent in the second family. As is true in most cases of scaphocephaly, the majority of the patients were male, but indications exist that at least 2 female members of family "N" were involved. At least 10 of the 28 patients with oxycephaly mentioned in Greig's review were female. It is also of interest to note that the youngest member of family "N" already had assumed a scaphocephalic shape of head, even though the sagittal suture remained open both to palpation and on roentgenograms.

CONCLUSION

The occurrence of familial scaphocephaly and oxycephaly cannot be doubted, as it is well documented in the literature. It is generally accepted that the other forms of craniosynostosis do appear...
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on a familial, as well as sporadic, basis. However, as yet there are no case reports in the literature to support this supposition. We hope this report will stimulate others to list, in print, their cases of familial brachycephaly, plagiocephaly, and trigonocephaly.

REFERENCES