An unusual form of familial acrocephalosyndactyly

ID YOUNG AND PS HARPER

From the Department of Medical Genetics, University Hospital of Wales, Heath Park, Cardiff

SUMMARY A family is described in which at least six members have an unusual form of acrocephalosyndactyly showing autosomal dominant inheritance. The most characteristic feature in the more severely affected individuals is duplication of the distal phalanx of the hallux. Review of family photographs suggests that the cosmetic outcome in apparently affected infants may be much better than anticipated.

Craniosynostosis may occur in isolation or as a component of a large number of pleiotropic malformation syndromes. The acrocephalosyndactylies comprise a group of hereditary disorders characterised by premature fusion of cranial sutures in association with distal limb abnormalities. In addition to neurological complications secondary to the premature synostosis, considerable cosmetic problems may occur. This report describes an unusual form of acrocephalosyndactyly showing autosomal dominant inheritance in a family, in which the ultimate facial appearance of affected members may be much more pleasing than might otherwise be expected.

Case reports

The family pedigree is shown in fig 1.

Case 1 (III.16), the proband aged 10 years, is asymptomatic and of normal intelligence. Review of early photographs shows marked frontal bossing and maxillary hypoplasia with gradual normalisation of facial appearance, as shown in fig 2. Examination revealed brachycephaly with hypertelorism (canthal index = 44.2), a normal palate, mild soft tissue syndactyly in both hands, and broad halluces, as shown in fig 3. Both height and weight lie on the 25th centile, whereas head circumference (50.6 cm) is below the 10th centile. Skull x-ray showed coronal suture synostosis with prominent copper beating. In both halluces there was an accessory epiphysis lying lateral to the interphalangeal joint, with partial duplication of the distal phalanx, as shown in fig 4.

Case 2 (II.6), the proband's father, is also asymptomatic. Examination revealed mild maxillary hypoplasia with marginal hypertelorism (canthal index = 41), brachydactyly of the hands, and bilateral cutaneous syndactyly of the fourth and fifth toes. Skull x-ray was normal.

Case 3 (III.1), aged 18 years, is a healthy and attractive female. At birth marked frontal bossing was present, as shown in fig 5. At the age of 5 years bilateral decompressive craniectomies were performed because of severe headaches with visual disturbance and radiological signs of raised intracranial pressure (copper beating and bulging of the middle fossae). Examination revealed mild hypertelorism (canthal index = 42.1), a high arched palate, and a double left hallux, as shown in fig 3.

Cases 4 (III.2), 5 (III.4), and 6 (II.1) all show mild features of the disorder. Case 4 had telecanthus with brachycephaly, a high arched palate, and broad laterally deviated halluces. Case 5 also showed these features in addition to bilateral ptosis. Case 6 showed maxillary hypoplasia with antimongoloid slanting of the eyes and a prominent glabella. In addition, he had broad laterally deviated halluces. All of these individuals are of normal intelligence.

FIG 1 Family pedigree.

Received for publication 13 November 1981
An unusual form of familial acrocephalosyndactyly
concerned by the alarming facies of their newly born baby, can be advised that the ultimate cosmetic outcome may be much better than expected. Cases 1 and 3 both presented an unusual appearance during early infancy but are now able to conceal a mild degree of frontal bossing behind a strategically placed fringe so that their facial appearance is not unattractive.

More serious is the history of probable raised intracranial pressure in case 3, for which bilateral decompressive craniectomies were performed at the age of 5 years. Similar surgical intervention has been reported in at least four patients with Pfeiffer syndrome5-7 and in the proband of the large family described by Robinow and Sorauf,3 although in this case the indication may have been cosmetic rather than neurological. There would thus seem to be a strong case for reviewing these children regularly during their early years with the option of surgical intervention if there is evidence of persistently raised intracranial pressure. In the absence of such evidence the indications for surgical action should be carefully considered since the majority of affected individuals in this family have reached adult life with normal intelligence and acceptable physiognomy without surgery.

The authors are most grateful to Professor C O Carter for helpful discussion and for bringing to their attention the distinction between the Pfeiffer and Robinow-Sorauf syndromes.

References


Requests for reprints to Professor P S Harper, Department of Medical Genetics, University Hospital of Wales, Heath Park, Cardiff CF4 4XW.
An unusual form of familial acrocephalosyndactyly.

I D Young and P S Harper

doi: 10.1136/jmg.19.4.286

Updated information and services can be found at: [http://jmg.bmj.com/content/19/4/286](http://jmg.bmj.com/content/19/4/286)

**Email alerting service**

*These include:*

Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to: [http://group.bmj.com/group/rights-licensing/permissions](http://group.bmj.com/group/rights-licensing/permissions)

To order reprints go to: [http://journals.bmj.com/cgi/reprintform](http://journals.bmj.com/cgi/reprintform)

To subscribe to BMJ go to: [http://group.bmj.com/subscribe/](http://group.bmj.com/subscribe/)