Case report

Journal of Medical Genetics 1988, **25**, 491–493

Auralcephalosyndactyly: a new hereditary craniosynostosis syndrome

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SUMMARY A family is described in which craniosynostosis is associated with characteristic pinnae, a short columella, and symmetrical syndactyly of the fourth and fifth toes, inherited as an autosomal dominant condition. Various dominantly inherited syndromes involving craniosynostosis have been identified, but the constellation of findings in this family suggests a new syndrome different from those previously described.

Case report

The proband was three weeks old when she was seen for evaluation of craniosynostosis. She was the product of an uncomplicated term pregnancy, with prolonged labour requiring caesarean section, to a 20 year old gravida 2, para 1 (one spontaneous abortion) mother. The birth weight was 3400 g and her head was noted to be brachycephalic with a high vertex. Skull radiographs showed bilateral coronal craniosynostosis. There were no other perinatal problems and she was otherwise well.

The family history was significant for craniosynostosis, as shown in the pedigree (fig 1). The mother was born with right coronal synostosis, a left club foot, and mild syndactyly of the fourth and fifth toes. She had no surgery for her craniosynostosis. Three brothers were born with bilateral coronal craniosynostosis. Two of them subsequently had craniectomies and developed mild mental retardation and hearing loss. The third affected brother died of congenital heart disease in infancy. The mother's father and some of his sibs were considered to have similar head shapes by the family but never sought medical evaluation.

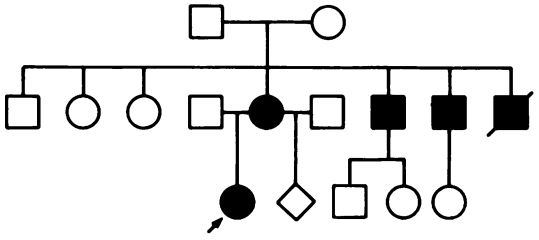

FIG 1 *Family pedigree.*

Received for publication 2 September 1987.
Revised version accepted for publication 23 December 1988.

On physical examination at three weeks of age, the proband was an alert, active infant girl (fig 2). The head circumference was 37 cm. There was frontal bossing, with the maximum diameter displaced anteriorly, and a flattened occiput. The sagittal suture was wide, soft, and continuous from the anterior to the posterior fontanelle. Supraorbital and glabellar ridging were present. The palate was high arched and narrow with prominent alveolar ridges. The pinnae were unusually shaped being relatively narrow inferiorly. The columella was short and the insertion of the alae nasae extended below the insertion of the columella. The neck, chest, abdomen, back, and perineal area showed no abnormalities. The extremities, however, had bilateral simian creases and cutaneous syndactyly of the fourth and fifth toes. Neurological examination was normal except for head lag on pulling from a supine position.

Examination of the mother showed brachycephaly and facial asymmetry with a greater prominence of the right side of the face. There was also cutaneous syndactyly of the fourth and fifth toes, the columella was short, and the pinnae were shaped similarly to those of the proband (fig 3).

The karyotype of the proband was normal female. Skull radiographs showed complete synostosis of the coronal sutures but widened lambdoidal and sagittal sutures and a foreshortened anterior fossa. Radiographs of the hands, feet, and long bones were normal. A CT scan of the head showed petrous pyramids concave posteriorly and a ventricular system generous but within normal limits. Audiological assessment indicated normal hearing. The patient subsequently had cranial synostectomy and dural plication without complications. Follow up at six months of age showed persistent brachycephaly

but improvement in the facial appearance. Neurodevelopmental evaluation was normal.

Discussion

The facial appearance of the proband was typical of bilateral coronal craniosynostosis such as occurs in various dominantly inherited craniosynostosis syndromes.[1] A number of these syndromes (such as Crouzon syndrome) are not associated with the presence of syndactyly and have distinguishing features which make it possible to exclude them as possible diagnoses for this patient.

Four characteristic craniosynostosis syndromes involve syndactyly of various digits, but none of them involves both the symmetrical syndactyly of

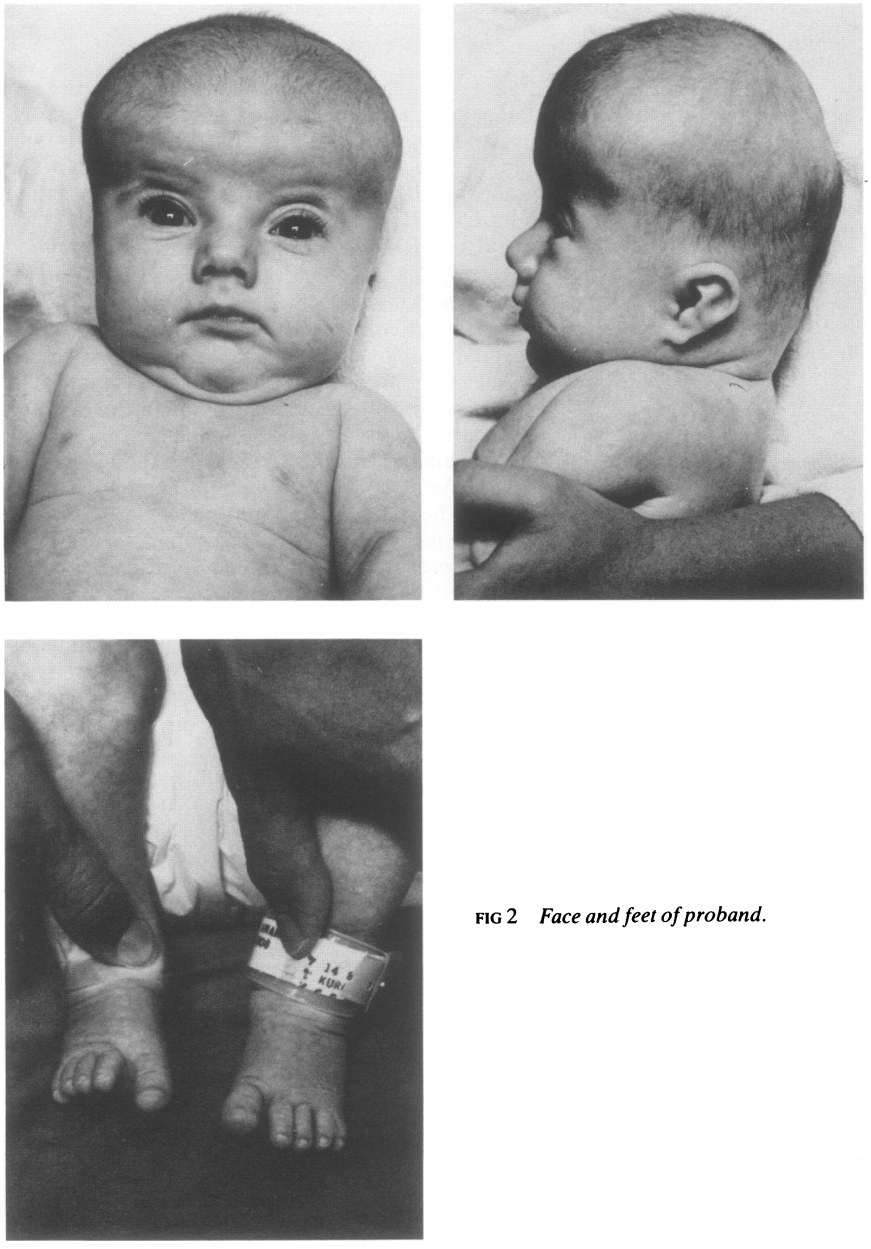

FIG 2 *Face and feet of proband.*

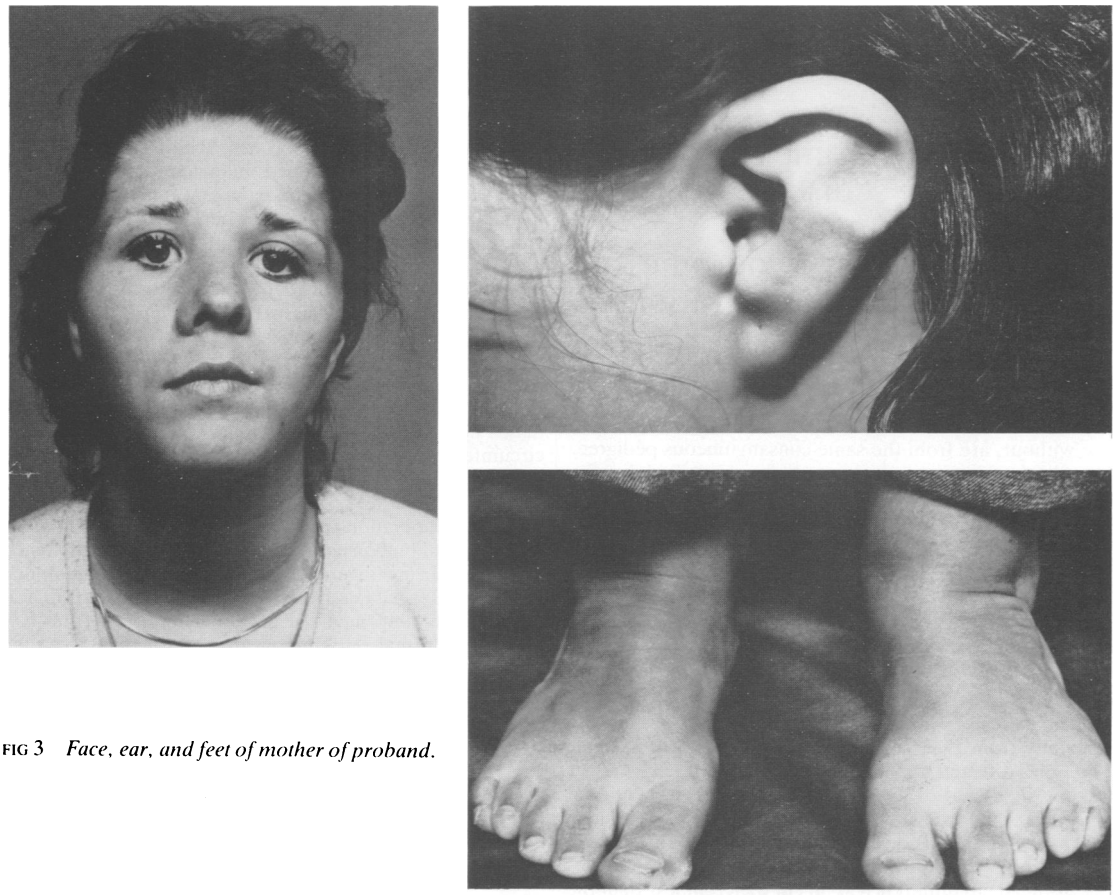

FIG 3 *Face, ear, and feet of mother of proband.*

the fourth and fifth toes and the unusual shaped pinnae which were present in the patient and her mother. The Apert syndrome[3] has complete symmetrical syndactyly of the hands and feet (minimally involving the second to fourth digits), the Saethre-Chotzen syndrome[4 5] has cutaneous syndactyly of the second and third fingers, and both the Greig cephalopolysyndactyly[6] and the Pfeiffer syndrome[7] have variable syndactyly, but neither of them shows unusual shaped pinnae. The pattern of syndactyly involving only the lateral toes, the nasal features, and the unusual shape of the pinnae in these patients appear to be a unique combination suggesting a new dominantly inherited syndrome. Variable expression was shown by the unilateral and less severe coronal synostosis in the mother, perhaps the maternal grandfather who was not examined, and the more severely affected brothers of the mother, who had hearing loss and mental retardation in spite of early craniectomies. It is important to distinguish this syndrome from other similar craniosynostosis syndromes which may carry a different prognosis or have a different pattern of inheritance.

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