Craniofrontonasal dysplasia

SIR,

In his review of craniofrontonasal dysplasia,¹ Dr Ian Young discussed the excess of affected females with this condition. Our experience has been similar. All five patients with craniofrontonasal dysplasia seen at this hospital have been female.

Two children had normal parents (one is shown in fig 1) and in a third child (fig 2), the parents looked normal but the father reported that he had had an unusually low frontal hairline as a child. The mother of this third child had two spontaneous miscarriages before the birth of her affected daughter. The fourth and fifth cases (fig 3) were an affected mother and daughter.

In addition to the features discussed in the review we found all our patients had thick, wiry hair. The

FIG 2 Two year old girl. Cranioplasty at three months for bicoronal synostosis.

FIG 1 Eight month old girl. Note the facial asymmetry, hypertelorism, and bifid nasal tip.

FIG 3 Mother and daughter with craniofrontonasal dysplasia. Note coarse, thick hair.

affected mother had had a cleft lip and palate repaired as an infant. One child had a diaphragmatic hernia. All were of normal intelligence.

Our cases confirm the female bias noted previously in craniofrontonasal dysplasia.

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Reference

- ¹ Young ID. Craniofrontonasal dysplasia. *J Med Genet* 1987;24: 193-6.

Ectrodactyly in sisters and half sisters

SIR,

In the April 1987 issue of your Journal (1987;24: 220-4), Mufti and Wood reported on two sisters and two half sisters with ectrodactyly. In addition, two of them had aplasia of the tibia. They suggested an autosomal recessive mode of inheritance in this family.

We think the reported family is probably an example of the aplasia of tibia-ectrodactyly syndrome, as was recently described by Majewski *et al.*¹ These authors described six new families with 35

affected persons, reviewed published reports on 27 sporadic cases and 99 familial cases, and showed that this disorder has great variability and markedly reduced penetrance.

Recently, we were able to investigate two cousins with this disorder. The first case had peromelia of the right ulna with agenesis of the radius, ectrodactyly of the left hand, a hypoplastic right femur and tibia with synostosis between the femur and fibula, and a hypoplastic left femur and aplasia of the left tibia (fig 1). The second case had postaxial hexadactyly of the right hand, syndactyly of the left third and fourth fingers, a normal left leg, and a hypoplastic femur, aplasia of the tibia, and a small foot with a hypoplastic hallux on the right side (fig 2). Their fathers are brothers. They were investigated physically and radiologically, but showed no anomaly. However, one of the sisters of the second case had mildly hypoplastic big toes on both sides which might be the mildest recognisable symptom of this disorder (fig 3).¹

We have tried to see whether a metacarpophalangeal profile analysis was useful in distinguishing relatives with clinical non-penetrance from unaffected relatives. In the above mentioned family, as well as in family 4 reported by Majewski *et al.*,¹ this analysis proved not to be useful.

In 1986, Lurie and Ilyina² published a comparable case, who had in addition distally 'bifurcated' femora. They thought this to be a separate entity and proposed the name Gollop-Wolfgang complex. A distally bifurcated femur may also be a component of the aplasia of tibia-ectrodactyly syndrome as was shown by Majewski *et al.*¹ However, as the distal femur, tibia, and foot ectrodactyly and the (developmentally homologous) distal humerus, radius, and hand ectrodactyly are probably on different developmental fields,³ and so are aetiologically non-specific and possibly heterogeneous, it remains

FIG 1 Radiological appearance of left hand of case 1.

FIG 2 Radiological appearance of both hands of case 2.