Recessively inherited brachydactyly type C

MICHAEL BARAITSER AND JOHN BURN

From the MRC Clinical Genetics Unit, Institute of Child Health, 30 Guilford Street, London WC1N 1EH, and The Hospital for Sick Children, Great Ormond Street, London WC1N 3JH.

SUMMARY We report a brother and sister, the children of unaffected consanguineous Iraqi parents, with type C brachydactyly.

The Bell classification of brachydactyly¹ divides this group of conditions into types A1 to A5, B, C, D, and E. Fitch² in 1979 suggested that types A4 to A5 be omitted because the latter is indistinguishable radiologically from type B, while type A4 is possibly a mild expression of A1. She suggested that type E should be further subdivided depending on which metacarpals are involved. The table summarises the classification. There have been numerous reports of isolated brachydactyly occurring as a familial trait; these reports have suggested an autosomal dominant pattern of inheritance.

We report two sibs with type C brachydactyly, whose parents were consanguineous and unaffected, suggesting autosomal recessive transmission in this family.

Case reports

The parents, first cousins, born in Iraq, were normal on clinical examination. Their first child had died, but clinical details were not available. Their surviving children, a girl of 7 years and a boy of 14 months, are described. The abnormal signs in both sibs were

confined to the hands and feet (figs 1-4). Both were of normal stature and development. In the hands, digits 1, 2, and 3 were short, resulting in a configuration in which the ring or 4th finger was the longest. In the feet, the same abnormalities were present. X-rays showed the shortening to be the result of abnormalities of the middle phalanges with relative sparing of the 4th. In the hands, the thumbs were short not only because of a short proximal phalanx, which is equivalent to the middle phalanx of the fingers, but also because the first metacarpals were short. The feet showed similar changes. In the older child, the proximal phalanges of the big toes were bifid.

Received for publication 29 September 1982.

TABLE Current classification of brachydactyly.

Types	Bones involved	Digits affected
A1	Middle phalanges	All can be affected; 3rd least affected making it longer than 4th
A2	Middle phalanges	Especially digit 2
A3	Middle phalanges	Especially digit 5
B	Middle and terminal phalanges	Digits 2-5
C	Middle phalanges and sometimes proximal. (Affected phalanges may be bifid and/or have radial projection.)	Most severe in digits 2-3 (4th is longest digit or equal to 3rd)
D	Metacarpal	Thumb
	Distal phalanx short and broad	Thumb
E	Short metacarpals	One or more digits

Adapted from Bell¹ and Fitch.²

FIG 1 Clinical and radiological appearance of the hands of the 7-year-old girl.

FIG 2 Clinical and radiological appearance of the feet of the 7-year-old girl.

FIG 3 Clinical and radiological appearance of the hands of the 14-month-old brother.

FIG 4 Clinical and radiological appearance of the feet of the 14-month-old brother.

Discussion

The Bell classification of brachydactyly is useful, but it is sometimes difficult to distinguish between types A1 and C. In type A1, the main findings are a broad hand with proportionate shortening of all the digits caused by short middle phalanges. In type C the shortening also affects the middle phalanges, especially of digits 2, 3, and 5, but Fitch² differentiates the two as follows.

In type A1 the proximal phalanx of the thumb is short, whereas in type C it is the metacarpal which is particularly involved. In addition, in type A1 the third middle phalanx is longer than the fourth, whereas in type C the fourth is not as severely affected as the third.

In the family reported here, the proximal phalanges of the thumb and big toe were short, as was the first metacarpal. The fourth middle phalanx is the least severely affected. Designation as type C is therefore appropriate.

The inheritance pattern of type C brachydactyly has, to date, been reported to be uniformly dominant, but the presence of this abnormality in two sibs whose unaffected parents are consanguineous strongly suggests recessive transmission in this family. A striking feature in the older child was the bilateral duplication of the first proximal phalanx with preaxial projection of the more proximal fragment, resulting in valgus deformity of the big toe. This feature is described in type C brachydactyly.²

On clinical examination, the disproportionate shortening of the big toe was reminiscent of fibrodysplasia ossificans progressiva. However, the x-ray appearance was not typical, and ectopic bone formation was not present.

We are grateful to Carol Reeves and Sadie Pottinger for preparing the illustrations and manuscript, respectively.

References

- 1 Bell J. On brachydactyly and symphalangism. *Treasury of Human Inheritance* 1951;5:1-30.
- 2 Fitch N. Classification and identification of inherited brachydactylies. *J Med Genet* 1979;16:36-44.

Requests for reprints to Dr J Burn, MRC Clinical Genetics Unit, Institute of Child Health, 30 Guilford Street, London WC1N 1EH.