Hereditary brachydactyly with nail dysplasia

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SUMMARY A family is described in which sometimes asymmetrical brachydactyly, with nail dysplasia of the involved fingers, has been transmitted through 6 generations affecting 6 males and 15 females. The thumbs and feet have been normal. This previously unreported condition is inherited as an autosomal disorder with full penetrance and variable expressivity, and causes only minor inconvenience.

The familial occurrence of shortening of the digits caused by the anomalous development of the contributing phalanges or metacarpals has long been recognised. Many examples of previously reported pedigrees were considered by Bell (1951), and the classification of the uncomplicated skeletal disorders she proposed has subsequently been followed by Temtamy and McKusick (1969) and more recently by Carter and Fairbank (1974). This classification has centred around the precise skeletal deformity and associated features sometimes encountered, including involvement of the feet and nail deformity. The family described below has a hitherto unreported combination of defects, comprising bilateral and sometimes asymmetrical shortening of the fingers, with normal thumbs and toes, and striking dysplasia of the nails of the affected digits.

The family

The proband (V.2) is a 22-year-old girl who presented with abnormalities of the fingers and nails (Fig. 1a and b). Deformities of all the distal phalanges were present, and in addition both ring fingers showed only a very small bone fragment in the intermediate phalanx. The right little finger showed fused distal and intermediate phalanges, in the left little finger the distal phalanx was absent, and the distal phalanges on the left middle and index fingers were fused with the intermediate phalanges. The thumbs, forearms, and feet were normal clinically and radiologically. The gross nail dysplasia of all the fingers and sparing the thumbs was apparent, with absence of the nails of the ring fingers and rudimentary nail structures on the other fingers. Physical examination of this otherwise healthy

![Fig. 1 Photograph (a) and radiograph (b) of hands of Case V.2.](http://jmg.bmj.com/)

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girl showed her to be normal, apart from minimal evidence of a surgically corrected cleft palate, which has not affected any other member of the family.

The pedigree of this Welsh family is shown in Fig. 2, from which can be traced hand deformities through 6 generations, with 6 affected males and 15 females; both males and females are carriers. Apart from the personally examined cases, affected members showed shortening of the fingers of both hands and nail dysplasia, but with marked variation in extent of involvement. In no patient were the thumbs or feet involved, and no other skeletal or other abnormality had been noted in the family. Four other members were personally examined:

III.6
In this 84-year-old woman, the only abnormality detected was symmetrical shortening of the index fingers of both hands, attributed to distinct shortening of the distal phalanges, and short, hypoplastic nails of these fingers. Radiological examination and photography were not feasible in this now senile woman.

IV.11
This 53-year-old woman had the following deformities (Fig. 3a and b): in the left hand, there was fusion of the intermediate and distal phalanges of the middle and ring fingers, and in the index and little fingers single phalanges replaced the intermediate and distal phalanges. In the right hand, the little finger had a short phalanx replacing the intermediate and distal phalanges; in the middle, ring, and index fingers there was fusion of a remnant of the terminal phalanges with short and deformed intermediate phalanges. Apart from the skeletal changes, the absence of the nails on the little and index fingers of the left hand and on all the fingers of the right hand was illustrated; the abnormal shortening of the nail on the left middle finger was evident, as were the normal thumbs with their nails.

V.I
The anomalies in this 25-year-old woman were symmetrical, with absent terminal phalanges of ring and little fingers, fusion of the terminal and intermediate phalanges of the index and middle fingers, the terminal phalanges being short (Fig. 4a and b). The nail deformities accompanying the finger abnormalities were also symmetrical.

VI
Fairly symmetrical digital abnormalities were detectable in this 2-year-old girl, and comprised absence of any fully formed terminal phalanx and deformed 'intermediate' phalanges, with those of both ring fingers being exceptionally short (Fig. 5a and b). Even at this early age, the severe dysplasia of the nails of all affected digits was evident.
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Fig. 3 Photograph (a) and radiograph (b) of hands of Case IV.11.

Fig. 4 Photograph (a) and radiograph (b) of hands of Case V.1.

Fig. 5 Photograph (a) and radiograph (b) of hands of Case VI.
Discussion

This family does not fall into any of the groups commonly recognised, though it bears some resemblance to the pedigrees comprised in Group B of Bell (1951) and the normal thumbs and feet in the affected members of the present family will be recalled. Apart from a few pedigrees cited by Bell (1951), the association of brachydactyly with dysplasia or absence of nails has also been reported in some rare families with more complex skeletal deformities (Bass, 1968; Cuevas-Sosa and Garcia-Segur, 1971), raising considerations of a link between nail formation and development of the digits. There does not appear to be a constant relation between abnormalities of development of finger and nail structures, however, and the mechanisms involved are at present unknown.

In common with other families suffering from similar disorders, the anomalies are inherited in an autosomal dominant pattern, with complete penetrance and variable expressivity. Interestingly, the features are sometimes asymmetrical. Fortunately the present family appears to have no other detectable abnormality, and the disorder remains a source of some embarrassment without functional disability.

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References


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