Brachydactyly and polydactyly with dermal ridge dissociation and ridge hypoplasia

SUMMARY A child with brachymesophalangy and postaxial postminimal polydactyly was found also to have dermal ridge dissociation and ridge hypoplasia. She was the second child of unaffected, unrelated parents and was born after a normal pregnancy and delivery. No previous report of a similar combination has been traced.

Brachydactyly may either occur as an isolated developmental defect, usually inherited in an autosomal dominant manner, or as part of a more complex malformation not confined to the extremities. The classification of isolated brachydactyly is based on that of Bell (1951), who recognised three types with brachymesophalangy (shortening of middle phalanges) alone, types A1, A2, and A3; three further forms of brachyphalangy, types B, C, and D (stubby thumbs); and a form with brachymetacarpal and brachymetatarsal type E. In 1970, Edwards and Gale described a mixed type combining brachymesophalangy, brachymetacarpal, brachymetatarsal, and camptodactyly. More recently, Christian et al. (1972) reported a new form of preaxial brachy-
normal nails, hair, and teeth. Her height was 188.9 cm, which is between the 25th and 50th centiles for her age. No evidence of any cardiac defect could be elicited.

Fig. 1 Pedigree of family.

Fig. 2 Photograph of hands of the patient.

Fig. 3 Photograph of patient's finger tips to show ridge pattern.

Fig. 4 Dermatoglyphic analysis of patient's hands.

DERMATOGLYPHS
There was a marked ridge hypoplasia on the fingers and over the thenar eminence and distal part of the palms. This peculiarity was also observed on the toes and distal part of the soles, including the thenar distal and hypothenar distal regions of the sole.

Ridge dissociation of the 'dotted' type also occurred on her fingertips. The dotted ridges were clearly defined at the margins of the fingertips (Fig. 3), but there was a gradual change of the dotted ridges from mild to more severe dissociation from the distal towards the proximal regions of the fingertips.

Fingerprint patterns and dental wax impressions of her fingertips showed ulnar loops on all ten remaining fingers. On both palms there was a digital triradius, proximal to the site from which the extra postaxial finger had been removed, and bilateral patterns on interdigital areas I and III; the former were associated with an e triradius (Fig. 4). Foot prints of the proband were not taken nor were her sole patterns recorded.

Her parents and sister had normally formed ridges.

RADIOGRAPHIC FINDINGS
X-rays of the hands (Fig. 5) showed that all the fingers had only two phalanges, a proximal and a terminal phalanx. In addition, there was a small round bone in place of the middle phalanx of the middle right finger and the index and middle left fingers. These bones could be regarded as severely reduced middle phalanges and they were associated with ulnar deviation of their terminal phalanges.
Separate capitate and hamate bones could be seen in each carpus. X-rays of the feet showed missing middle phalanges in the second, fourth, and fifth toes on both sides, with severely reduced middle phalanges in the third toes similar to those in the middle fingers. There was no abnormality in the x-rays of the forearms, pelvis, knees, and legs below the knee.

Discussion

Brachydactyly types A1–5, B, C, Edwards and Gale's type, and the types described by Bilginturan et al. (1973), Schott (1978), and Sillence (1978) all involve some degree of shortening or absence of the middle phalanges. None of them involves polydactyly or dermal ridge hypoplasia or ridge dissociation. Each of these types has features distinguishing it from that described here. Brachyphalangy and post-axial polydactyly are seen in chondroectodermal dysplasia (McKusick et al., 1964), but there the distal phalanges are more severely shortened than the middle ones. The normal terminal phalanges and absence of cardiac defect exclude this inherited disorder in the patient reported here.

There have been a number of reports of either extreme narrowing of ridges, or of their dissociation into short segments or dots, or even of total absence of ridges. Dissociated ridges may occur in any dermatoglyphic area of the hands and feet. They are uncommon in normal people but rather frequent in certain medical disorders. In severe dissociations, the ridges are broken into short segments covering the dermatoglyphic area in a disorganised manner so that, frequently, no consistent pattern is recognisable. In mild dissociations, the ridges are broken into short, often dot-like, segments, but the patterns, however, remain clearly definable. Ridge disturbances have been observed in phenotypically normal people of different races and ethnic groups. Furuya (1961) studied ridge dissociation in apparently healthy Japanese. He divided his cases of ridge dissociation into two groups depending upon the extent to which ridged skin was involved: (a) total dot-and-short ridge patterns; and (b) partial dot-and-short ridge patterns. Both types of ridge dissociation showed differences with regard to the areas in which they occurred, and also to the extent to which the epidermis was affected. He concluded that the two types of ridge dissociation were determined by separate autosomal genes, both producing dominant inheritance with incomplete penetrance.

A similar pattern of inheritance was described in a single American family with complete congenital absence of ridges in association with transient congenital milia, flexion contractures of digits, and webbing of toes (Baird, 1964). Mild degrees of ridge dissociation were found by Abel (1936) with a frequency of 1 in 200 among 4000 criminals. There have been several previous reports of non-familial ridge dissociation in malformations of the extremities (Abel, 1936; Schade, 1937; Grebe, 1940), but none had the specific malformations described in the case presented here. Furthermore, the marked ridge hypoplasia with ridge dissociation of the present case has not been previously reported as an innate character and could tentatively be defined as a type A6 in Bell's classification.

A sporadic abnormality in a girl, such as that reported here, could be the result of a fresh mutation of an autosomal dominant congenital defect, or of a non-genetic developmental error. It is impossible to distinguish between these alternatives and the case is reported in the hope of stimulating further reports from others that might do so.

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References

Case reports


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