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 brachydactyly with short thumbs and first toes angulated at the metacarpophalangeal or metatarsophalangeal joint. Bilginturan et al. (1973) described a new form of combined brachyphalangy, brachymetacarpal, and brachymetatarsy associated with hypertension. Other developmental disorders associated with brachydactyly that have been reported include joint dysplasia (Liebenberg, 1973), and cerebellar ataxia with nystagmus (Biemond, 1934). Since Bell's account, two new types of brachymesophalangy have been described, that of Tatemany (1966), type A4, and that of Bass (1968), type A5. Schott (1978) has also recently published an account of a family with a new form of type B brachydactyly (shortening of terminal and, to a lesser extent, middle phalanges), and Sillence (1978) has indicated further heterogeneity in type A1 brachydactyly. In Schott's family, hands, but not feet, were involved, the thumbs were spared, and there was an associated nail dysplasia of the affected fingers. The findings on the hands and feet of the five members of a kindred seen by Sillence were consistent with type A1 brachydactyly, but they also had tall normal stature, scoliosis, and club feet. None of these different types of brachydactyly involved polydactyly, dermal ridge dissociation, and ridge hypoplasia as was seen in the sporadic case reported here.

Case report

The patient was a girl born on 18.7.71. She was born at term after a normal pregnancy and delivery, with a birthweight of 3·97 kg. Her mother was 27 years old and her father 32 years at the time of her birth. A normal elder sister was born in June 1970. The patient was noted at birth to have bilateral small fleshy fingers with nails, attached by a narrow piece of skin to the proximal phalangeal region of the outer border of the fifth fingers. There was a similar postaxial toe on the right foot but not on the left. All three extra digits were removed shortly after birth.

She was seen, with her parents and sister, in 1973 for an opinion relating to her possible acceptance for immigration abroad. At that time, direct examination of her parents and sister showed no abnormality of hands or fingers and no history could be obtained of such abnormality in any further member of the family. There was no parental consanguinity (Fig. 1).

On examination, the patient showed small scars where her extra digits had been removed. The second to fifth fingers of both hands were slightly short, but her thumbs were of normal length (Fig. 2). There was some limitation of flexion of the distal joint of the right thumb. There was no further abnormality of the limbs other than in the digits and she had
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normal nails, hair, and teeth. Her height was 889 cm, which is between the 25th and 50th centiles for her age. No evidence of any cardiac defect could be elicited.

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.
Brachydactyty 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 40000 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


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