Congenital malformations of human dermatoglyphs

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David, T. J. (1973). Archives of Disease in Childhood, 48, 191. Congenital malformations of dermatoglyphs. A classification for congenital malformations of dermatoglyphs is presented, dividing them into ridge aplasia, ridge hypoplasia, ridge dissociation, ridges-off-the-end, and a combination of the last two. The medical and genetic significance of these are considered in the light both of previous published cases and of new material. Malformations of dermatoglyphs are important as physical signs in paediatric diagnosis.

While dermatoglyphs are becoming increasingly important in the study of congenital malformations and as physical signs in paediatric diagnosis, it is not often realized that dermatoglyphs can be malformed per se. Such epidermal ridge patterns are usually either pass unrecognized or are abandoned as 'freak' patterns (Cherrill, 1954). In this paper the commonest ridge malformations will be described and a classification presented, but no mention will be made of rare patterns (e.g. lateral pocket loops, composites, mutant loops) which are better described as 'rare' until it can be shown that they are indeed abnormal.

Common ridge malformations

1. Ridge aplasia. This is a rare malformation consisting of congenital absence of epidermal ridges over the entire palmar and plantar surfaces. The palmar and interphalangeal flexion creases remain normal, but there is a great excess of very small creases on the skin, which appear as 'white lines' on the fingers or palm prints. To the naked eye and on the actual fingerprints the skin gives an 'orangepeel' appearance which is characteristic but not by itself diagnostic. The palmar and plantar surfaces do not sweat. Examples of genuine ridge aplasia have been found in a family (Baird, 1964, 1968) when it appeared to be inherited as an autosomal dominant trait. The affected members of this kindred also had transient congenital milia and bilateral flexion contractions of some fingers and toes. However, a 4-year-old girl has been found at this hospital who, in addition to ridge aplasia, suffers from severe dermatological problems at sites distant to the palms and soles, the condition thought to be a generalized disorder of keratin production (David and Warin, 1973). Her parents have normal epidermal ridges and normal prints. Reports about 'absent fingerprints' tend to be poorly documented (Ludy, 1944; Rott, 1970a) or show ridges to be present though hypoplastic or dissociated (Cooke, 1962; Holt, 1964; McCann, 1969; Cummins, 1970a, b).

2. Ridge hypoplasia. In this condition the ridges are not absent but are reduced in height, and this is often combined with a great excess of 'white lines' on the prints. It can be inherited as an autosomal dominant trait (Furuhata et al., 1957; Basan, 1965). Congenital ridge hypoplasia in an individual is impossible to distinguish from the acquired condition of epidermal ridge atrophy. The latter is a partly reversible change found in extreme old age, in some people with mental subnormality (Matsukura, 1953; Yamashita, 1960), and in 90 to 95% of adults with coeliac disease (David, Ajdukiewicz, and Read, 1970). Ridge atrophy tends to be less severe than congenital ridge hypoplasia, and the former is an acquired condition which is not itself inherited (though the underlying coeliac disease may well be inherited). The ridge atrophy found in adults with coeliac disease partly improves when a gluten-free diet is instituted (David et al., 1970).

Congenital ridge hypoplasia is sometimes present in patients with chromosome abnormalities, particularly those with autosomal aneuploidy. Under these conditions the ridge hypoplasia is usually a patchy change most commonly involving the proximal regions of the palms and soles, and can make observation of the dermatoglyphs extremely difficult at a time when they may be most useful as physical signs. Though this kind of ridge hypoplasia is congenital, some improvement can take place by
Fig. 1.—Mild ridge dissociation in the left middle fingerprint, mainly affecting the area of the triradius as well as below the core of the pattern.

growth of the hypoplastic ridges. It is worth mentioning that ridge hypoplasia can occasionally be found on some palmar and plantar surfaces of congenitally deformed limbs.

3. Ridge dissociation. In this condition, the ridges instead of running neatly in more or less parallel lines, are broken up into short ridges which tend to be curved and are completely disorganized (Nettles, 1963; Cooke, 1960; Cummins, 1967; Safara, 1969). It is most commonly found on the thumbs and in the region of the t triradius on the palm. Whether the t triradius is at its normal position near the wrist or at a t’’ position in the centre of the palm makes no difference; it is still the commonest site for ridge dissociation on the palm. The distribution of ridge dissociation is absolutely characteristic on the fingers. The thumb is the commonest digit to be affected, followed by the index, middle, ring, and little fingers, the lesion becoming progressively less in this order. When the ridge dissociation is mild it always affects the region of the fingerprint just below the core of the pattern (Fig. 1) (Cummins, 1968, 1970c), and this is a good distinguishing feature. The lesion can be so mild as to affect only a small area around the t triradius, or so gross that the entire palmar and plantar surfaces are covered with dissociated ridges, with complete loss of ridge patterns as a consequence (Fig. 2). As in ridge aplasia, the palmar and interphalangeal flexion creases are normal.

Ridge dissociation is a heterogeneous condition. It can be inherited as an autosomal dominant trait (Furuya, 1961, 1967; Dodinval et al., 1971) or it can be sporadic (Furuhata and Kawashima, 1950). One family has been reported where ridge dissociation was associated with tapering fingers, painful fingertips, and a minor nail dystrophy (Dodinval, 1972). The suggestion that ridge dissociation is
often associated with tapering fingers and a nail dystrophy (Dodinval, 1971) is neither evident from the literature nor from the families with ridge dissociation studied in the south west of England by the author. Nevertheless, it is logical to look for other ectodermal defects, and ridge dissociation has been observed by the author in one patient with hypohidrotic ectodermal dysplasia.

True ridge dissociation is sometimes mistaken for an area of scarring, and vice versa. It has been said that ridge dissociation is present in 18% of schizophrenics (Raphael and Raphael, 1962), but both the published examples of ridge dissociation were typical of injuries which had healed by granulation, most probably burns (David, 1969). To distinguish between mild ridge dissociation only on the thumbs and scars healed by granulation is always difficult, and is left best to an experienced observer who has seen many examples of ridge dissociation. The principal differences are outlined in the Table.

Other studies of dermatoglyphs in schizophrenia have not revealed an increased incidence of ridge dissociation (Sank, 1968; Mellor, 1968; Zavala and Núñez, 1970; Rothhammer et al., 1971), and the author has had the opportunity to examine the palmar dermatoglyphs of 600 patients with schizophrenia kindly lent by Dr. A. Pauline Ridges in Liverpool. There was not one example of ridge dissociation.

Out of 430 patients with congenital heart disease
studied in Bristol, 2 have ridge dissociation. One has multiple cardiac abnormalities with no family history of congenital heart disease, and his parents and 4 sibs have normal prints. The other has a truncus arteriosus with atrial and ventricular septal defects as well as a hare lip. His only sib has a normal heart and normal prints, but his father has marked ridge dissociation and also has bilateral polythelia with a normal heart. Some of the father's relatives also have ridge dissociation, the distribution of affected members fitting with autosomal dominant inheritance. The author has been able to examine the prints of a pair of monozygotic twins who were concordant for a rare dental abnormality (of unestablished aetiology) as well as for ridge dissociation. There is no family history of the dental condition, and the parents do not have ridge dissociation. It is likely that these are chance associations, and the author has also seen ridge dissociations in several sporadic cases where there were no detectable birth defects.

Ridge dissociation of a slightly different type is often found in patients with chromosome abnormalities, and also some patients with the de Lange syndrome (Berg et al., 1970) (Fig. 3). The ridges are broken up regularly into dots and the patterns are obscured (Wolf et al., 1965). A mild appearance of 'dotted ridges', when the patterns remain quite clear (Fig. 4), is easily visible in the prints of at least 80% of patients with mongolism (trisomy-21), but oddly it is never included in complex dermatoglyphic discriminant functions for this diagnosis. Similar mild ridge dissociation has been reported in 7 out of 20 cases with trisomy-18 and in 12 out of 20 cases with D1-trisomy (Taylor, 1968). A more severe form of the 'dotted ridges' variant of ridge dissociation was reported in a trisomy-18/normal mosaic (Alter and Schuleenberg, 1967). The ridge dissociation found in patients with chromosome abnormalities is likely to be a nonspecific effect of abnormal growth in utero rather than a direct genetic effect. 'Dotted ridges' can be found in normal people when it can be a fully reversible change. It has been noted in some patients with cystic fibrosis (Schwanitz and Rott, 1970), and the author has seen it completely disappear over a period of 6 months in a 20-year-old girl with small intestinal pseudo-obstruction (Fig. 4).

T. J. David

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<th>TABLE</th>
<th>Differences between ridge dissociation and burn scars</th>
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<td></td>
<td>Ridge dissociation</td>
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<tr>
<td>Commonest digit</td>
<td>Thumb</td>
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<tr>
<td>Area of fingerprint pattern first affected</td>
<td>Below core</td>
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<tr>
<td>Proximal phalanges affected</td>
<td>Not described</td>
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<tr>
<td>Area of palm first affected</td>
<td>t triradius</td>
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<tr>
<td>Affected digits + normal thumbs</td>
<td>Probably never</td>
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<td>Toes affected</td>
<td>Common if fingers affected</td>
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<td>Incidence</td>
<td>? 1 in 8000</td>
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Fig. 3.—Right sole print of a boy with the de Lange syndrome, showing the 'dotted ridges' kind of ridge dissociation seen in this condition. Note that the lesion is worse proximally, and over the hallucal area the ridges are normal.
Congenital malformations of human dermatoglyphs

Fig. 4.—‘Dotted ridges’ in a 20-year-old girl with small intestinal pseudo-obstruction. Prints of both thumbs are shown. Note that the pattern is in no way obscured (compare with Fig. 3).

4. Ridges-off-the-end. This is now recognized to be part of the first purely dermatoglyphic syndrome to be discovered (David, 1971). The first family with this syndrome showed 7 distinct traits inherited by a single autosomal dominant gene. 3 further families with this syndrome have since been studied by the author, with similar but not identical findings to the first family. By far the most striking feature of the syndrome is that the fingertip ridges, instead of running transversely, are vertical and run vertically off the end of the fingertips (Fig. 5). Sometimes the ridges only show the ‘tendency’ to run off the end, and these so-called ‘cuspal’ patterns are always radial rather than ulnar on the ring and little fingers, and are often radial on other fingers too. (Bilateral radial loops on the ring and little fingers are excessively rare in people without this syndrome.) Palmar abnormalities include a t triradius greatly displaced distally, usually to the t’’ position, deep interdigital loops (usually in the IVth space) and a curious ‘crack’ in the ridges over the hypothenar eminence (Fig. 6). This rare syndrome is apparently unassociated with any diseases, though there is good reason to believe that the hair patterning on the head is abnormal in some cases. The

Fig. 5.—Left thumb print of a man with the ‘ridges-off-the-end’ syndrome. The ridges run more or less vertically off the end of the fingertip.
'ridges-off-the-end' syndrome is easily distinguishable from ridges running vertically off the end of a congenitally deformed digit.

5. Ridges-off-the-end and ridge dissociation. There appear to be only two known examples of this rather unique combination. One was unwittingly published as 'ridge hypoplasia' by Matsukura (1953). The other is a member of a Welsh family with ridge dissociation studied by the author. Mild ridge dissociation was segregating as an autosomal dominant in this family, but one member (whose parents were first cousins) had complete ridge dissociation of his palms and soles (his palm is shown in Fig. 2), with a combination of ridge dissociation and ridges-off-the-end on all his fingertips (Fig. 7). The family was intellectually dim but there was no evidence of associated disease.

Discussion
If congenital malformations of dermatoglyphs are carefully studied then they can easily be classified into at least 5 distinct groups as shown above. Vague terms such as 'ridge disruption' (Alter, 1967), 'bead-cord ridges' (Rott, 1970b), and 'pearl-ridges'...
Congenital malformations of human dermatoglyphs

The incidence of these malformations is as yet unknown, partly because none of them is common but mainly because they are usually not correctly recognized. Ridge hypoplasia secondary to a chromosome abnormality is the commonest of the malformations, followed by the heterogeneous group of people with ridge dissociation. Ridge aplasia is likely to be genuinely rare, or it would have caused trouble by now for identification purposes. The 'ridges-off-the-end' syndrome was first thought to be excessively rare, but already in 4 years 4 families with this condition have been found in the United Kingdom alone, and there are now good reasons for thinking that it is not so rare after all. It should be mentioned that Abel (1936) found ridge dissociation in 11 out of 8000 'people', some of them criminals and probably mostly males, and that Dodinval et al. (1971) found it in only 2 out of 500,000 people in Belgium. The former figure is derived from people who could hardly be called 'controls', and the latter is likely to be a gross underestimate since these prints were not systematically searched for ridge dissociation. Indeed the second case was only found because he was the son of the first case.

With the wider use of dermatoglyphs as physical signs in paediatric diagnosis, the classification of abnormalities in this paper should help to clarify the importance of congenital malformations of dermatoglyphs. An essential for their recognition is the careful recording of finger, palm, sole, and toe prints.

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REFERENCES


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