Dermatoglyphs in skeletal dysplasias

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Summary

Finger and palm print patterns (dermatoglyphs) are formed in very early pregnancy, at about the same time the limbs are developing, and their formation probably depends upon the surface contours of the palms and soles during morphogenesis. Dermatoglyphs are useful in skeletal dysplasias for the detection and study of syndactyly and symphalangism, for dating the onset and the dysplasias, for demonstrating and dating asymmetry, and sometimes for providing specific diagnostic features.

The purpose of this short paper is not systematically to list all the dermatoglyphic findings in various skeletal dysplasias, a list which would be as tedious to prepare as it would be to endure. Instead, it is proposed to outline the logical basis for examining the dermatoglyphs of patients with skeletal dysplasias, since the author believes that for those who are unfamiliar with the subject, the principles which underlie the dermatoglyphic changes are far more important than the precise dermatoglyphic details.

The skin of the human palms and soles possesses easily visible epidermal ridges. It is the patterns formed by these epidermal ridges that fascinate those who are interested in dermatoglyphs and baffle everyone else. These ridges are the site of sweat pore openings and sensory nerve endings. In small areas, epidermal ridges appear to run in parallel lines, and where three such areas meet a 'triradius' is said to be formed. The triradius is an important feature for the classification of fingerprint patterns, and on palms and soles triradii form the main dermatoglyphic landmarks.

There are four fundamental principles relevant to this paper:

1. Epidermal ridge patterns are formed in very early pregnancy, at about the same time the limbs are developing and probably for a few weeks later. Certainly these patterns are fully formed by about the fourteenth week post-conception.

2. It therefore follows that any influence, whether genetic or environmental or both, which will change or affect the formation of dermatoglyphic patterns must act before the fourteenth week.

3. Dermatoglyphic patterns appear to be dependent for their formation upon the surface contours of the palms and soles during morphogenesis of the limbs. Any structural abnormality of the underlying hand or foot which is present in early pregnancy must inevitably lead to abnormalities of the dermatoglyphic patterns. (The converse, that abnormal dermatoglyphs imply some abnormality of fetal limb contour is unproved and may well be untrue.)

4. How genes influence the formation of dermatoglyphic patterns is not understood. It is a useful working guide, however, to assume that single gene disorders will not affect the dermatoglyphs unless that gene has an early prenatal influence on developing limb formation.

These principles lead to certain simple immediate conclusions:

(a) Patients with obvious limb malformations (such as ectrodactyly) can be expected to have obviously abnormal dermatoglyphs. These are no more than a reflection of the underlying skeletal malformation. Thus, for example, the dermatoglyphic changes found in the pre-axial part of the hand in the Rubenstein–Taybi syndrome merely reflect the underlying skeletal abnormality, and are themselves of no significance. The statement that these dermatoglyphic changes 'suggest a chromosomal abnormality' (McKusick, 1975) can therefore be seen to be without foundation.

(b) Patients with diseases due to a single gene can be expected to have normal dermatoglyphs unless the gene affects early prenatal limb development. Thus, for example, the search for dermatoglyphic changes in Wilson's disease was predictably negative.

Having made these dogmatic exclusions, how can dermatoglyphs be relevant in skeletal dysplasias? The author believes that there are at least five situations where they can be helpful.

1. Syndactyly

It has been shown that dermatoglyphic changes
can be extremely sensitive indicators of syndactyly. These changes are not only present when syndactyly is visible, but can also be a manifestation of a tendency to soft tissue syndactyly when the syndactyly is not visible to the naked eye. Thus it has been shown that while some patients with the Poland anomaly have obvious cutaneous syndactyly, in others the only manifestations of syndactyly are the gross dermatoglyphic changes. Dermatoglyphs have also been used to trace the gene for type II syndactyly within families. How an environmental agent (in the case of the Poland anomaly) or a gene (in the case of the type II syndactyly) cause dermatoglyphic changes but leave the hand structurally unaffected is not yet clear.

(2) Symphalangism
Dermatoglyphs are sensitive indicators of symphalangism and can be used to demonstrate at birth minor forms of the defect which may not be radiologically manifest until adult life.

(3) Dating the onset of the dysplasias
In several skeletal dysplasias the onset of the abnormality of growth or development is not at all certain. In theory one might hope that those disorders manifesting themselves (partly by affecting the limbs) before the fourteenth week would leave their mark on the dermatoglyphs. From experience of cases referred to the Bristol Bone Dysplasias Registry this has sometimes been found to be of some theoretical interest, though of little or no practical value in diagnosis.

(4) Asymmetry
In Bristol, workers have shown dermatoglyphs to be of interest, firstly in demonstrating asymmetry and secondly in dating the onset of the asymmetry to being either before or after the fourteenth week.

(5) Providing specific diagnostic features
Dermatoglyphs may become useful in delineating a few skeletal dysplasias, although much more information is needed in this field. Where dermatoglyphs are useful is in suggesting the presence of certain other syndromes, in particular, those associated with an abnormal chromosomal complement.

In conclusion, the author believes that an examination of the dermatoglyphs has become part of a multidisciplinary assessment of any patient with a skeletal dysplasia. Recording the finger, palm and foot prints, involving dirty (but easily washable) black ink, a lot of mess and making pictures on white paper, nurses' uniforms and anything else to hand, is probably the only investigation which most children actually enjoy having done to them.

Reference