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Dermatoglyphic clues to developmental retardation

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Abstract

Embryological evidence indicates that whorls are associated with rounded localized digital pads at 30-40mm CRL. They may therefore be regarded as patterns reflecting greater cell proliferation in early skin morphogenesis. If this is so, variation in whorl proportions should be detectable in conditions of which growth retardation is characteristic. A study therefore was made of Down syndrome patients using four series with suitable controls available in the literature. Results generally support the hypothesis : there is considerable diminution in the proportion of whorls on the three radial digits, somewhat less marked on the fourth digit and still less marked on the fifth digit; most frequently there is even an increase on the fifth. There is no marked difference in the pattern of reduction between right and left hands and between the two sexes. The implications of the findings are discussed.

Résumé

Les observations embryologiques indiquent que, au stade 30–40 mm du foetus (longueur vertex-coccyx), les tourbillons sont associés à des pelotes arrondies circonscrites. C'est pourquoi ils peuvent être considérés comme des dessins reflétant une plus grande prolifération cellulaire au début de la morphogenèse de la peau. S'il en est ainsi dans les états caractérisés par un retard de croissance, on devrait pouvoir détecter un changement dans les proportions de tourbillons. C'est pourquoi nous avons étudié des sujets affectés du syndrome de Down (mongolisme) en utilisant 4 séries et leurs témoins respectifs, repris dans la littérature. Dans l'ensemble, les résultats corroborent l'hypothèse : on observe une forte diminution de la proportion de tourbillons sur les trois doigts radiaux, un peu moins marquée sur le quatrième et encore moins, sur le cinquième; le plus souvent, il y a même une augmentation sur le cinquième doigt. Dans le type de réduction, il ne se marque aucune différence entre les deux côtés ni entre les deux sexes. Les implications de ces résultats sont discutées.

The successive steps in the development of the arm, the hand, the leg and the foot, start from an early limb bud stage and show characteristic changes in shape and structure. Shaping, differentiation and the spatial arrangement of the different cells are the basic processes of these stages.

These basic processes depend on the presence of several controlling factors, the absence or incomplete expression of which will cause either the non-development of the limb and the associated structures, or severe malformations. Thus, in the earliest stages, these basic processes have been shown to depend on the presence of the apical ectodermal ridge of the limb bud; this ridge subsequently gives rise to the ridge ectoderm and maintains a high level of mitotic activity in the underlying mesoderm (for review see Hinchliffe and Johnson, 1980).

Another controlling factor is a species- and area-specific pattern of cell death. A well known example of the latter occurs in the process of interdigital finger separation, which involves both local anchorings of the apical ridge ectoderm by collagen fibrils and interdigital cell death. If cell death did not occur at all, the result would be complete syndactyly, or if only the soft tissues were involved, the complete webbing of the fingers.

Another factor controlling development is that of the timing and coordination of such processes, a factor which is closely related to the one concerning the direction of growth and development. All these processes have been shown to be delicately tissue- and location-specific. Regular expressions of these factors are required for the normal development of limbs and tissues. However, minor variations either in the extent of expression of such factors or in the timing of their onset, their rate or their duration details which would affect coordination and direction are quite compatible with normal development. They would, however, result in minor differences in the shape and size of the tissue structures.

From embryological studies there is evidence that the gradients of development for volar skin features in the developing hand are in a disto-proximal and radio-ulnar direction (Cummins, 1929; Würth, 1937; Fleischhauer and Horstmann, 1951), and that there is faster growth radially (Würth, 1937). In the process of this development pads appear first on fingers, and subsequently on palms.

So far, studies concerning the development of dermal and subsequently epidermal ridges have demonstrated that ridges first appear in areas corresponding to the highest pad elevation (Bonnevie,

1927,1929; Cummins, 1929; Penrose and Ohara, 1973; Okajima, 1975). As early as 1927, Bonnevie observed an interrelation between pad shape and size and the formation of pattern type. According to her findings, the whorl can be seen as one extreme of dermatoglyphic patterning not only in its morphological complexity, but also because of its association with most pronounced, generally symmetric embryonic pads. The pads correspond to sites of greatest cell proliferation, so that the whorls can be taken as indicators of the extreme of this process. It is therefore reasonable to expect that the outcome of variations in developmental growth processes will also be apparent in dermatoglyphic features, and this is indeed so in cases of malformations of the hands and feet. But what are the effects of minor variations which do not cause malformations? It may be best to consider as an example one of the most extensively studied syndromes from clinical genetics, i.e. trisomy 21.

The special dermatoglyphic features of trisomy 21 have long been known (Cummins, 1939). Indeed, taking them together, Ford Walker (1957) was able to devise a diagnostic procedure indicating the probability that a particular individual might be a mongoloid imbecile, which is dependent upon the ratio of the frequency of patterns in mongols, and in controls. This ratio may then be taken as an index of the probability. Similar differentiating procedures have been proposed by Reed *et al.* (1970), Borgaonkar *et al.* (1971), Bolling *et al.* (1971), Deckers *et al.* (1973), Rodewald *et al.* (1976). These procedures are essentially empirical, that is to say they are based on the observation of frequency differences in selected dermatoglyphic features rather than on deductions drawn from the implicitly different mechanisms in trisomics and normals which cause these features. Undoubtedly there are differences in patterns of growth and development. For example, there are differences of proportion between the trunk and lower limbs, between hand breadth and length, and between ear breadth and length, to mention only some of the grosser measurements. But there are also many differences in fine detail such as in the growth of the face, leading for example to the apparently increased interorbital distance and epicanthal fold.

In order to investigate the effects of such minor growth variations on ridged skin, it may be useful to look at dermatoglyphics from a developmental viewpoint. As an example for such an approach the frequency of whorls on each of the ten digits was chosen. The data come from the literature and include four large series of patients with trisomy 21, and four control series from the same populations (Tab.1).

	Right					Left						
	V	IV	III	II	I	I	II	III	IV	V	N	
HOLT	18.56	26.95	4.79	2.40	13.17	10.18	3.95	7.78	21.56	11.98	167	T21 ♂
1964	13.99	28.67	4.20	6.29	12.59	14.69	8.39	6.29	22.38	16.08	143	T21 ♀
HOLT	15.20	50.20	21.60	33.20	39.60	29.60	29.60	16.40	33.60	14.20	500	Cont ♂
1964	10.00	39.80	11.40	32.40	33.00	24.80	29.80	15.20	32.40	10.40	500	Cont ♀
LOESCH	24.3	32.4	10.8	7.2	23.4	18.9	13.5	8.1	27.9	19.8	111	T21 ♂
1974	18.4	26.3	3.5	2.6	15.9	17.5	8.8	7.9	30.7	12.4	114	T21 ♀
LOESCH	13.8	45.7	10.1	26.1	33.3	23.9	26.8	13.8	34.1	13.8	138	Cont ♂
1974	13.6	44.4	15.4	34.6	35.2	30.2	30.2	16.7	35.2	13.6	162	Cont ♀
VRYDAGH	20.00	31.67	5.00	3.33	21.67	15.00	1.67	5.00	13.33	16.67	60	T21 ♂
1975	12.28	24.56	5.26	3.51	17.54	21.05	7.02	5.26	21.05	10.53	57	T21 ♀
VRYDAGH	13.27	48.98	20.41	31.12	45.41	33.67	33.67	16.84	29.59	9.18	196	Cont ♂
1971	14.95	46.39	13.92	33.51	42.27	40.21	33.51	16.49	36.60	14.95	194	Cont ♀
MALKY	27.4	25.8	8.1	4.8	24.2	12.9	4.8	6.4	19.3	12.9	62	T21 ♂
1963	11.8	23.5	5.9	5.9	8.8	5.9	11.8	11.8	29.4	17.6	34	T21 ♀
MEYRING	16.9	50.2	20.2	32.6	48.0	35.7	31.2	18.4	35.4	11.1	2308	Cont ♂
1953	11.1	40.5	15.0	31.4	39.2	33.9	30.4	18.7	36.2	10.7	1673	Cont ♀

Table 1 : Percentages of whorls on the ten fingers of four male and four female series of patients with trisomy 21 (T21) and of suitable controls (Cont).

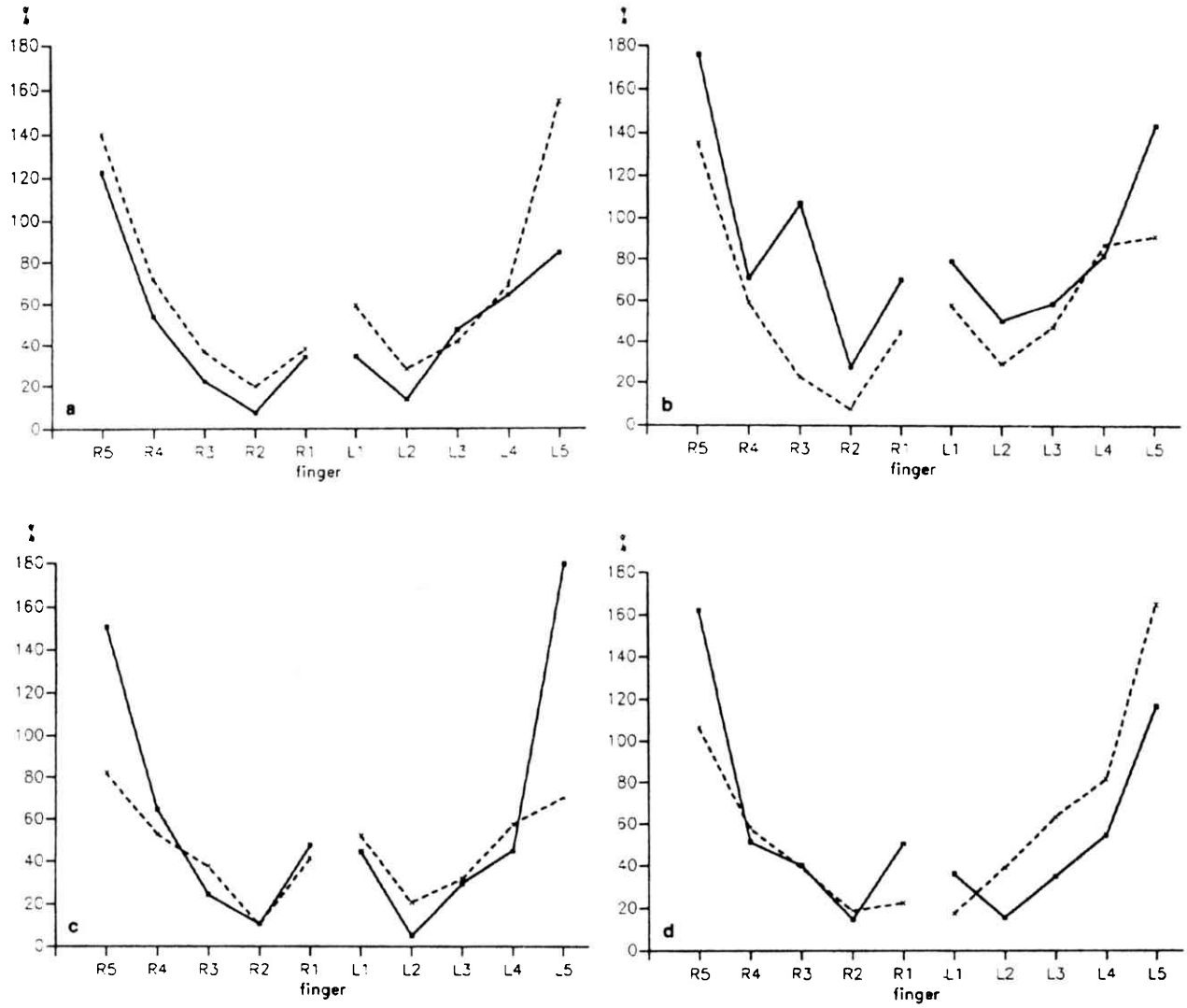

Fig. 1 : Ratio of whorl frequency between T21 and controls (=100 %)
a) Holt (1964), b) Loesch (1974), c) Vrydagh-Laoureux (1975), d) Malky (1963).

To demonstrate an effect of interference with normal growth processes for dermatoglyphics, it is necessary to have a base line against which comparisons can be made. Such a base line should be biological and represent the outcome of normal growth processes. For the purpose of the present analysis the frequency of whorls in normal individuals (controls) is taken as the reference point (100 %). Taking each finger separately, the frequency of whorls in trisomics is calculated as a percentage of this normal base line. A figure of 100 % therefore indicates the identity of whorl frequency in trisomics and normals, a figure greater than 100 % indicates a whorl excess on that finger in trisomics, and a figure of less than 100 % a deficit in trisomics. With one exception (third finger of the right hand in the male series of Loesch 1974), fingers one to four of the trisomic patients show a marked deficit of whorls on both sides (Tab. 1). The quantitative results are depicted by the whorl ratios in figure 1 and suggest that there is also a slower rate of growth with the formation of dermatoglyphics. But, in addition to this general assumption, more detailed deductions may be drawn from the differing behaviour of the single fingers. Thus, the lowest whorl ratios generally observed on the second fingers would point to the fact that this reduced rate of growth is the most evident at about the time of the onset of ridge formation. This would agree with Suzumori's (1980) observation of a dermal ridge development retarded by two or more weeks in fetuses with trisomy 21 compared with the parallel development in normal fetuses of the same gestation. The subsequent increase of whorl ratios in the two ulnar fingers of trisomics, which frequently culminates in an excess of whorls on the fifth finger, might be interpreted as an effect of longer duration of the process rather than an excess of growth with time. On account of the similarity of the diagrams plotted for

each of the four male and the four female trisomic samples the data of the males were pooled, as were those of the females (fig. 2). The pattern of reduction is quite similar on both hands and in both sexes.

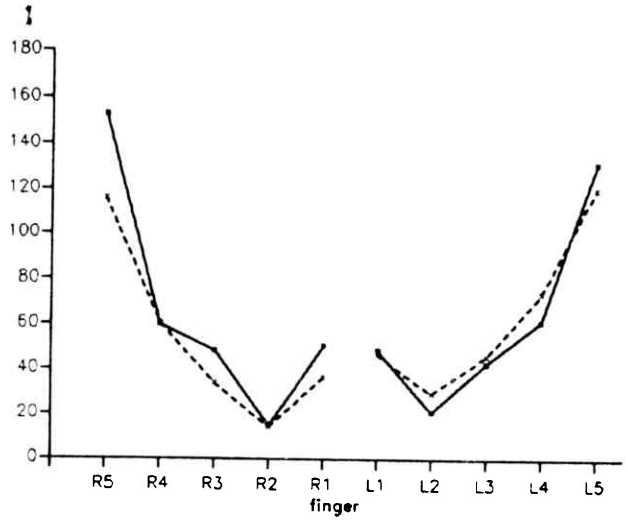

Fig. 2 : Ratio of whorl frequency between T21 and controls (=100 %) of the pooled data from the four series (see table 1). Males : — ; females : - - -

These observations rather strengthen the idea that it may be worth while to draw more attention to the developmental viewpoint of dermatoglyphics,and that such observations are not necessarily restricted to those made on fetuses and newborns.

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