

DERMATOGLYPHICS*

JAMES R. MILLER, Ph.D.

INTRODUCTION

About 20 years ago when I began my studies as a graduate student under Norma Ford Walker at the University of Toronto the literature on dermatoglyphics was sparse and could be gone through quickly. The book, *Fingerprints, Palms and Soles*, by Cummins and Midlo, published in 1943, was still the source of everything relevant in terms of history, fundamental biology, methods of formulation, etc. The section on dermatoglyphics and disease was pretty thin, and the only condition that was given more than a passing mention was mongolism. During the 1950s, probably the only relevant paper to appear was Walker's "The Use of Dermal Configurations in the Diagnosis of Mongolism" (1957a), which extended the knowledge of dermatoglyphics in this condition and attempted to quantify dermal configurations for use as a meaningful diagnostic tool. Its publication in 1957 was in the "pre-chromosome era," of course. With the publication explosion on human chromosomes in the 1960s, there was an incredible upswing in the number of reports on dermatoglyphics which stressed in particular their value in diagnosing various diseases. Although many of these diseases were determined by chromosomal aberrations, others resulted from the action of single genes or nongenetic factors or were of unknown etiology. As often happens when a particular field "takes off," numerous questionable and even completely erroneous reports appeared along with the relevant. It is not my intention to review this past decade critically but to limit my discussion to a few topics on the genetics of dermatoglyphics which I believe represent growing areas that need further investigation and that will be of practical and theoretical importance in the next decade.

SOME BASIC PRINCIPLES IN DEFINITION AND FORMULATION

Ridged skin occurs on the volar surfaces of the hands and feet of all primates. It also occurs on the distal part of the tail of some New World monkeys and on the paws of certain other mammals. (Discussion on comparative dermatoglyphics and their evolutionary significance can be found in Cummins and Midlo, 1943). The term "dermatoglyphics" (literally "skin-carvings") is used not only as a collective name for all features of ridged skin but also as a description of its study.

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* From the Division of Medical Genetics, Department of Paediatrics, University of British Columbia, Vancouver, British Columbia, Canada.

Although the ridged skin itself is formed in the early fetal period, the configurations are determined by morphologic events in the embryonic hand and foot. Like other features of limb development, the ridges form on the hands at a somewhat earlier period than on the feet. From the time the primary ridges have been formed midway through the fifth month, no further change is apparent either in the structure of the ridges or in the configurations they form. Some secondary ridge formation occurs up until the sixth month of gestation and there are changes in the size and relative position of certain features resulting from the normal growth processes of the hands and feet. Although once formed, the dermal configurations are resistant to environmental effects, they do reflect growth disturbances that occur before and during their development.

Technically, flexion creases on the palms and soles are not part of dermatoglyphics; however, the two are often considered together and some mention of flexion creases will be made later.

Methods of Study

In the older child and the adult the patterns can be seen with the naked eye, but in younger children and most infants some form of slight magnification is necessary for proper visualization. Observing and recording dermatoglyphic patterns is (or should be) a part of the routine physical examination of any child. Several methods for making a permanent record of dermatoglyphic patterns have been outlined by numerous authors (Walker, 1957b; Miller and Giroux, 1966; Uchida and Soltan, 1969). In general, the collection of readable prints is a simple matter in all but very young infants, newborns, and fetuses (Miller, 1968).

Formulation

To facilitate description and recording, certain rules have been established for pattern formulation and ridge counting (the latter is used as a measure of the distance between certain fixed points). However, no attempt will be made here to go into the specifics of these procedures which are presented in great detail by Cummins and Midlo (1943), Holt (1968), and Penrose (1968b). More recently, Penrose and Loesch in a series of reports (1969, 1970a, 1970b, 1971) have proposed a new type of classification based on topographical consideration which they believe will be more useful in genetic studies.

Digits. Three basic pattern types are found on the digits—the arch, the loop, and the whorl (Fig. 1). The type of pattern depends upon the number of triradii (a triradius is a point formed by the meeting of three different ridge fields). The arch,

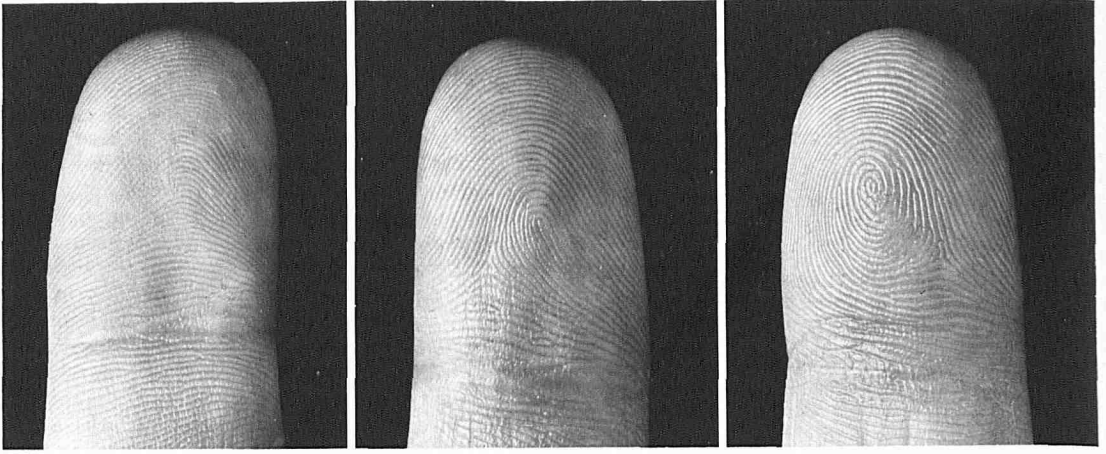


FIG. 1. Photographs of fingertips demonstrating the three basic digital patterns, from left to right: arch, loop, and whorl. (From Miller, J. R. and Giroux, J.: *Dermatoglyphics in pediatric practice*, *J. Pediatr.* 69:302-312, 1966.)

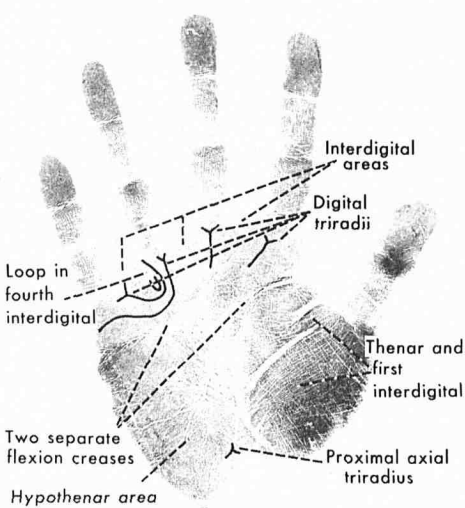


FIG. 2. Palm print of left hand showing main topographical areas. (From Miller, J. R. and Giroux, J.: *Dermatoglyphics in pediatric practice*, *J. Pediatr.* 69:302-312, 1966.)

the simplest configuration, does not have a triradius. The loop, designated ulnar or radial depending upon the margin of the hand to which it opens, has one triradius. The whorl, which usually forms a large concentric design, typically has two triradii associated with it. Many variants of these basic patterns and intermediate types occur quite frequently.

Palms. The main topographical areas of the palms are shown in Figure 2. Each area is a more or less distinct unit which in most palms is delineated by partial boundaries formed by triradii or by patterns in the form of loops or whorls. The axial triradius, which is normally located near the proximal margin of the palm between the thenar and

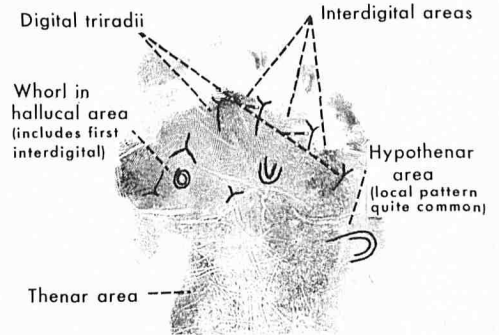


FIG. 3. Sole print of right foot showing the more important aspects of the plantar configurations. (From Miller, J. R. and Giroux, J.: *Dermatoglyphics in pediatric practice*, *J. Pediatr.* 69:302-312, 1966.)

hypothenar eminences, can occur at various positions distad. By describing the occurrence and nature of the patterns in the various areas indicated in Figure 2 and the position of the axial triradius, we can obtain a dermatoglyphic formulation for the palm.

Soles. The various topographical areas and the triradii of the soles are similar to those of the palms (Fig. 3). Only the more distal parts of the sole are shown since those are the areas where patterns occur most frequently.

GENETICS OF NORMAL PATTERNING

Dermatoglyphics present an intriguing challenge for genetic analysis since they constitute an admixture of qualitative and quantitative characteristics. Patterns can be described according to position and type. Such features as pattern size and the relative positions of triradii can be measured linearly or by ridge counting (the number of ridges that cut or touch a straight line between two fixed points, i.e., two triradii or a triradius and a core of

a loop or whorl). Although Sir Francis Galton was the first to suggest an hereditary basis for pattern types, the real pioneer in this field was the American biologist, H. H. Wilder, who in the early years of the century attempted by pedigree studies to demonstrate the important role of hereditary factors in ridge arrangements. Since this early work, many attempts have been made to analyze the genetics of such characteristics as pattern type and form. However, the results have been meager; and Holt, whose book (1968) constitutes the most comprehensive overview of the genetics of dermatoglyphics, states: "In fact, it can be said that in no case has the inheritance of a dermal ridge trait been unequivocally explained by single factor inheritance."

Because of this failure to elucidate the genetics of dermal configurations by qualitative methods, investigators such as Holt have attempted to substitute quantitative techniques. Using correlation analysis between relatives, Holt and others have provided evidence for genetic factors in the determination of such parameters as total finger ridge count (the sum of the ridge counts on the ten fingers of an individual).

In view of the very close agreement between the observed and theoretical correlations demonstrated in Table I, Holt concludes that total ridge count is an inherited metrical character that is controlled by the action of a number of perfectly additive genes and that environment plays a comparatively small part in its expression. In view of the remarkable similarity of total ridge counts in monozygotic twins, Slater et al. (1964) and others have developed statistical tables for the use of fingerprints in zygosity determination.

Using ridge counts as quantitative measurements, different studies have demonstrated that various dermal characteristics such as *atd* angle, *a-b* ridge count, and hallucal ridge count also have a strong underlying genetic component. However, in no character are the results as straightforward as those in the total ridge count. In some features, differences between parent-child and sib-sib cor-

relations suggest that single dominant or recessive genes are functioning, whereas a low monozygotic twin correlation suggests that nongenetic factors play a role in determining the position of the axial triradius.

DERMATOLYPHICS OF SOME GENETICALLY DETERMINED DISEASES OF THE HANDS AND FEET

Anomalous Development

Dermal configurations are obviously and ultimately determined by the normal development of the pentadactyl limb; hence any anomalous deviation from "normal" will be reflected to a greater or lesser degree in the dermatoglyphics. It is not surprising, for example, that individuals with the thalidomide syndrome have grossly distorted dermatoglyphic patterns. In some cases of anomalous limb development, the study of the dermal pattern leads to some valuable data about the fundamental biology of the skin and to some practical knowledge about the variable expressivity of single genes. The disease anonychia, which is an example of the first, is determined by a rare autosomal dominant gene. The nail is usually absent on the second and third fingers, often on the thumb, and very diminished on the fourth; the toes are similarly affected. In a study of the topology of ridged skin, Penrose (1965) examined the dermatoglyphics of members of a large family with this condition and observed that in the absence of the nail and nail-bed the ridged skin extended to the dorsal surface of the distal phalange and in some cases to the dorsal surface of the middle phalange. Apparently, then, this gene also affects those developmental events that determine the limitations of ridged skin. It has been my personal observation that, in general, individuals with hypoplastic nail development have ridged skin on the dorsum of the distal phalange.

Zygodactyly (syndactyly Type I—McKusick, 1971) is a condition in which the study of dermatoglyphics can help to detect the minor expression of

TABLE I

Correlations between relatives for total ridge count of fingers (based on data from Holt, 1968)

Relationship	Observed correlation coefficient	Theoretical correlation*
Parent-child	0.40	0.5
Mother-child	0.48	0.5
Father-child	0.49	0.5
Midparent-child	0.66	0.7
Sib-sib	0.50	0.5
Monozygotic twin-twin	0.95	1.0
Dizygotic twin-twin	0.49	0.5
Parent-parent	0.05	0.0

* "Parent-parent" based on random mating, all others on additive gene action.



FIG. 4. Print of area under toes 2 and 3 demonstrating mildest form of zygodactyly which may be expressed in dermal configurations only. Note single digital triradius with distal radiants embracing two digital area.

a gene. This condition also is determined by an autosomal dominant gene, and in its extreme form produces a total syndactyly between toes two and three. However, there is considerable variation in its expressivity, and sometimes the condition only involves webbing of the proximal parts of the digits. The dermal patterns on the sole reflect this fusion (Fig. 4) and may provide evidence for the mildest expression of the gene.

In addition to the descriptions by Holt (1968) several other authors have described features of other conditions involving anomalous limb development (Rosner and Aberfeld, 1970; Robinson et al., 1968; Smith and Berg, 1968). Mulvihill and Smith (1969) have examined how some of these features aid in the understanding of early developmental events.

Patternless Ridge Formation and Congenital Absence of Ridged Skin

Patternless ridge formation and the congenital absence of ridged skin seem to be extremely rare. Dysplasia of the ridge formations resulting in scattered short ridges or in ridges consisting of irregular dots is frequently observed in patients with Down's syndrome and in cases of the anomalous development of the limb described in the previous paragraph. However, this condition has been described as a familial disorder apparently determined by a single autosomal dominant gene. Most of the cases reported to date have been from Japan (see Holt, 1968, for references) but recently it has been described in a Belgian pedigree (Dodinval et al., 1971). This extreme dissociation of ridge formation presumably occurs during embryonic development, producing failure of the ridges to consolidate in a pattern formation.

The second condition, congenital absence of ridged skin, has been reported only once (Baird, 1964). This condition, which also appears to be determined by an autosomal dominant gene, was complete in all but two of the cases reported by Baird. The palms and soles in affected individuals have a paucity of sweat pores, and affected adults have calluses, fissures, and blisters on the palms and soles. Cummins (quoted by Holt) believes that the absence of the ridged skin in this family is due to a form of epidermolysis and that ridged skin was formed normally during embryogenesis but had been sloughed off.

DERMATOGLYPHIC FINDINGS NOT ASSOCIATED WITH SPECIFIC ANOMALIES OF HANDS AND FEET

Dermal configurations that are associated with diseases not characterized by abnormalities of hands and feet have received more attention than others. Potentially, the knowledge derived from such studies is important for proper diagnosis. However, these studies should be interpreted with caution since, despite several published lists of diseases with "abnormal" dermatoglyphics, too

few conditions have been studied to establish characteristic pattern frequencies. The problem is that very few, if any, patterns are "abnormal" since all are really variations or combinations of the basic three—the arch, the loop, and the whorl—plus open fields on the palms and soles. For significant results, the pattern frequencies of specific diseases must be compared with those of "normal" controls. But here again the problem is to determine what constitutes a normal control because other considerations such as sex and race must be evaluated. Dermatoglyphic data on an adequate control group, even a Caucasian sampling, have not been published. What reports we have all include different categories of control data, usually based on small samples collected under biased conditions, e.g., from university students, hospital patients, etc. This lack of adequate control data casts serious doubts on many of the "significant" findings reported in the literature during the past few years. Despite this problem, the fundamental reasoning underlying the use of dermatoglyphics in clinical studies is sound. Therefore, such studies should be pursued but with more awareness of the pitfalls than has been evident so far.

Down's Syndrome (Mongolism)

Cummins (1939) was the first to point out that certain dermatoglyphic features in Down's syndrome differed strikingly from those in controls. Walker and her students extended these studies and in 1957 she published her method for using dermal configurations in the diagnosis of Down's syndrome (Walker, 1957a). Her "index" was based on the comparison of specific pattern frequencies for each of 16 areas in a group of affected and non-affected individuals. The indices were in the form of histograms on a logarithmic scale; using this procedure, Walker diagnosed 70 percent of patients with Down's syndrome. Despite certain disturbing features about it, this technique contained all of the basic elements for the use of dermatoglyphics as an essential objective diagnostic tool.

Because of the extensive computation involved, Walker's method has not been widely used. Recently, however, Reed and his colleagues (1970) used discriminate analysis to develop what they call a "dermatoglyphic nomogram" for the diagnosis of Down's syndrome. Their populations consisted of 250 karyotypically proven cases of trisomic Down's syndrome and 332 controls with normal karyotypes. Pattern frequencies in 14 of 32 areas showed significant differences between the patients and the controls. However, only four areas—the right hallucal, the right *atd* angle, and the right and left index fingers—accounted for most of the observed variation between the two groups; hence the nomogram is based on these four.

Although several groups of investigators are

endeavoring to establish similar nomograms for other conditions, which will be discussed below, to my knowledge none are functional at this time.

Other Autosomal Conditions

After the discovery of the chromosomal etiology of Down's syndrome and the existence of the other trisomic states, the search for dermatoglyphic features that were common to the latter states began in earnest. Although the first report of the dermatoglyphic findings in the 18 and D_1 trisomic conditions was based on quite small samples, the observations have been repeatedly confirmed (Uchida et al., 1962). The most significant dermatoglyphic feature in the 18 trisomy is the high frequency of digital arches. Uchida and her colleagues confined their observations to the fingers, but subsequent studies have demonstrated the same high frequency of arch patterns on the toes of patients with 18 trisomy. About 50 percent of patients have simian creases and in general lack the loops and whorls; the characteristic dermatoglyphic features on the palmar and plantar surfaces are arches and open fields. Patients with the D_1 trisomy have extreme distal displacement of the axial triradius and most have bilateral simian creases. In addition, many have what seems to be a unique pattern in the hallux area of the sole, described by Uchida et al. (1962) as the "arch fibular-S pattern." Holt (1968) and Uchida and Soltan (1969) have reported in detail some of the dermatoglyphic findings in the autosomal trisomies and their potential value in clinical diagnoses.

Sex Chromosome Abnormalities

In general, the studies of the dermal configurations of patients with sex chromosomal aberrations have not been as rewarding as those of patients with autosomal aneuploidy. However, some relevant observations have been recorded.

Turner's syndrome. In individuals with the 45, X karyotype, there is a slight distal displacement of the axial triradius (Penrose, 1963; Uchida and Soltan, 1969) and an increased frequency of full and partial simian creases (Uchida and Soltan, 1969). However, the most striking dermatoglyphic feature is the high frequency of large patterns on the fingers and a consequent high total ridge count. The average total ridge count in most samples of Caucasian females is about 127, in patients with Turner's syndrome between 160 and 200.

Klinefelter's syndrome. Cushman and Soltan (1969) have published an analysis of the dermatoglyphics of 55 patients with 47, XXY Klinefelter's syndrome. In six of 20 dermatoglyphic features, the XXY patients resemble female more than male controls. For example, the mean total ridge count in these patients was about 118, which is lower than the count for females (about 127) and significantly lower than the count for males (about 147).

48,XXYY Syndrome. In individuals with the XXYY chromosomal constitution (a rare deviant of Klinefelter's syndrome), there is increased frequency of a rare hypothenar pattern, besides the dermatoglyphic features of the 47, XXY syndrome. This hypothenar pattern is characterised by an ulnar triradius together with a loop carpal, loop radial, or arch radial patterns (Uchida et al., 1964).

Impact of X and Y chromosomes on total ridge count. Alter (1965) and Penrose (1967 and 1968a) have discussed the significance of the above observations, especially the relation of X and Y chromosomes to total ridge count. The studies of Holt and others clearly demonstrated that total ridge count is a quantitative character under the control of autosomal polygenes. However, the genetic factors located on the sex chromosomes influence this character because the addition of X and Y chromosomes progressively reduces the total ridge count.

PROBLEMS INVOLVING THE MORPHOGENESIS OF VOLAR SKIN

Morphogenesis of Pattern Formation

To appreciate the potential biologic significance of dermatoglyphics, one must be aware of their ontogenesis. Unfortunately many of those pursuing this field today seem unaware of this fact. In 1943, Cummins and Midlo reviewed most of the early literature on the embryology of ridged skin. However, during the intervening 30 years only two reports relating directly to this matter have appeared (Hale, 1952; Mulvihill and Smith, 1969). The report of Mulvihill and Smith is especially important because it is the only one to deal with the ontogenesis of dermatoglyphics since the tremendous explosion of clinical interest in dermatoglyphics itself. Despite severe criticism by Holt (1970) I believe the contribution of Mulvihill and Smith would be important if only because it introduces new investigators in dermatoglyphics to some of the careful anatomic and embryologic studies of 30 to 40 years ago which have never been equaled. However, Mulvihill and Smith did more than this: in a forceful way, they pointed out that dermal configurations reflect embryonic events and depend upon the morphology of the hand and foot in general and of the embryonic volar pads in particular. Patterns observed postnatally are a function of the height and contour of the embryonic pads during the period of regression in early fetal life, when primary ridge formation is occurring. Patternless open fields, which usually occur on proximal parts of the sole and in the central part of the palm, represent areas where pad regression occurs rapidly and completely. Arches, loops, and whorls reflect the previous existence of low pads, pads of intermediate height (with asymmetry), and high pads, respectively. For example, the fact that patients with 18 trisomy syndrome have a high frequency of digital arches reflects the existence of low apical embryonic volar pads.

Evidence to support this general hypothesis can be derived from embryologic studies, phylogenetic considerations (Fig. 5), theoretical reasoning based on mathematical principles, and the study of dermal configurations in malformations of hands and feet. However, our knowledge of the developmental events that determine dermal configurations is still deficient. This is a reflection, to a large extent, of our general ignorance of human embryogenesis and early fetogenesis. The main difficulty in studying the development of the volar skin stems from the impossibility of visualizing the subtle continuous interaction between pad regression and the formation of the ridged dermis. Here is work for the enterprising embryologist, but surprisingly enough none to my knowledge has exploited the fact that certain marsupials, such as the opossum, possess ridged skin. In view of its precocious expulsion from the uterus, the marsupial would seem to be an ideal model for the study of many of the poorly understood and still controversial questions of the embryonic development of ridged skin. I do not believe that such studies are solely academic because, if the clinical value of dermatoglyphics is to be fully utilized, the embryonic events that affect them must be far better understood than at present.

The Flexion Creases of the Volar Skin

Technically, the flexion creases of the ridged skin are not part of dermatoglyphic studies. However, because of their intimate association, the two are invariably considered together, and clinical reports on dermatoglyphic findings usually record observations about a simian (or four-finger) crease or a single crease on the fifth finger. Therefore, the genesis of these creases should be mentioned briefly.

The flexion creases, which represent the point where the skin is attached to the underlying structures, are associated with localized deficiencies of ridge formation. A recent study of the fingers and palms of normal and abnormal hands by Popich and Smith (1970) has demonstrated that creases are secondary features that are related to the flexion movements of the hands of the late embryo and early fetus. Therefore, a single digital crease indicates a nonfunctional interphalangeal joint which may or may not be associated with a hypoplastic phalanx. Unfortunately, the developmental significance of the single upper palmar crease (simian crease) in relation to the underlying bony structure is still unexplained.

SPONTANEOUS PHENODEVIANTS AND TERATOGENIC RESPONSES

One of the most intriguing phenomena in mammalian development is the relationship between spontaneously occurring phenodeviants, which represent the extremes of developmental systems under polygenic control, and the anomalies produced by genetic or nongenetic factors. In experimental organisms such as the mouse, strain differences in the frequency of spontaneously occurring anomalies and of experimentally induced anomalies and the relationship of the two have been well documented (Dagg, 1966). For example, many studies in a number of species demonstrate the susceptibility of spontaneously occurring anomalies with a low frequency to the action of environmental teratogens. In man, of course, such studies are virtually impossible. However, dermatoglyphics may provide a tool for analyzing at least some of these developmental phenomena in man. Anthropologic literature contains ample evidence of racial differences in dermatoglyphic features (Cummins and Midlo, 1943). For the most part, these differences must reflect differences in the polygenic systems that seem to control "normal" dermal patterning. The trisomic conditions represent teratogenic states, each of which alters the normal control of the developing hand in a rather specific way. Attempts to develop dermatoglyphic nomograms (Reed et al., 1970) and other methods of topographical classifications (Penrose and Loesch, 1969, 1970a, 1970b, 1971) should yield practical information about the relation of the frequency of specific pattern types between control subjects and trisomic individuals. However, nomograms for



Fig. 5. Palm of baboon (*Papio papio*) demonstrating the association of extensive pattern formation with persistent pads in the interdigital areas.

TABLE II

Comparison of percentage frequencies of four dermatoglyphic traits in Japanese, Chinese, Canadian and English controls and patients with Down's syndrome*

Dermatoglyphic Feature	Laterality	Japanese		Chinese		Canadian		English	
		Control	Down's	Control	Down's	Control	Down's	Control	Down's
Significant distal displacement of axial triradius.	L	4.2	41.7	0.3	51.0	10.2	85.8	8.5	83.2
	R	4.0	42.4	1.3	43.1	13.3	84.4		
Arch tibial pattern in hallucal area.	L	7.2	78.4	7.0	90.6	0.5	46.6	0.2	50.4
	R	6.0	76.2	5.0	92.5	0.5	47.4		
Pattern in third interdigital area.	L	6.7	24.2	6.1	19.6	31.3	54.0	25.7	52.2
	R	16.8	52.5	29.3	49.0	55.5	85.4	48.2	83.9
Ulnar loop on second finger.	L	39.8	89.0	34.7	86.8	36.3	82.4	37.5	86.4
	R	34.9	94.0	33.8	98.1	31.1	82.3	35.5	92.2

* Data obtained from following source: Japanese: Shiono (1969), Chinese: Bryant et al. (1970), Canadian: Walker (1957), English: Fang (1950), Holt (1964), Penrose (1954) and Penrose and Smith (1966).

various racial groups should yield even more useful information about the developmental events. According to the available data, the nomograms, although essentially the same, have some subtle differences. The data in Table II compare certain dermatoglyphic features in control subjects and in patients with Down's syndrome in two "racial" groups. Of the four features considered, three (axial triradius, hallucal pattern, and pattern on the second digit) are used in the nomogram developed by Reed et al. (1970) and the fourth, the third interdigital pattern, was used by Walker (1957a) in the development of her index. Of the four dermatoglyphic features (right and left second digital patterns were considered separately by Reed et al.), the arch tibial pattern in the right hallucal area and the distal displacement of the right axial triradius provide the greatest discrimination score. In both the Caucasian and the Oriental populations represented in Table II, these features occur with a very low frequency in the normal controls. Of course, the sample is small—indeed, it is unique at the present time—but buttressed with what is known of the sensitivity of low frequency phenodeviants to teratogens in mice, it seems to suggest that certain of the dermatoglyphic features that occur with a very low frequency in normal subjects are sensitive to the action of teratogenic events.

CONCLUSIONS

To date, dermatoglyphic investigations in clinical studies have concentrated on comparing the pattern frequencies observed in certain disease states with those observed in normal individuals. Although such studies have yielded some useful data, they have also produced a large volume of material of questionable quality. As Mulvihill and Smith have pointed out, most studies compare a large number of variables, many of which are

undoubtedly highly correlated. Because each of these is treated as an independent variable, we get the false impression of an abundance of data that in fact does not exist. If enough statistical comparisons are made on such data, some significant differences are unearthed simply by chance. The trend towards the development of statistical procedures for distinguishing between normal and various disease states on the basis of objective criteria will undoubtedly discredit many of the questionable results that now inundate this field. In addition to providing valuable diagnostic tools, these discriminating tests should yield much valuable data on human developmental genetics.

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