

Dermatoglyphics in Medical Disorders

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Preface

The skin on the fingertips and palmar and plantar surfaces of man is not smooth. It is grooved by curious ridges, which form a variety of configurations. These ridge configurations have attracted the attention of laymen for millenia. They have also evoked the serious interest of scientists for more than three centuries. The anatomist Bidloo provided a description of ridge detail in the seventeenth century. Since then, additional information has been added by anthropologists, biologists, and geneticists. For the last century, the fact that each individual's ridge configurations are unique has been utilized as a means of personal identification especially by law enforcement officials. Widespread medical interest in epidermal ridges developed only in the last several decades when it became apparent that many patients with chromosomal aberrations had unusual ridge formations. Inspection of skin ridges, therefore, promised to provide a simple, inexpensive means for determining whether a given patient had a particular chromosomal defect. However, the promise was only partially fulfilled because of the inherent variability of skin ridge configurations. It was possible to draw conclusions about ridge abnormalities in groups of patients but not always in a given individual. Patients and clinicians became somewhat disenchanted with the clinical value of studying ridges. Nonetheless, considerable clinically useful information about groups of individuals with chromosomal defects has been discovered and therefore a knowledge of the types of deviations associated with various medical disorders can add appreciably to the diagnostic armamentarium of the clinician. Unusual

PREFACE

ridge configurations have been shown to exist not only in patients with chromosomal defects but also in patients with single-gene disorders and in some in whom the genetic basis of the disorder is unclear.

More than 30 years ago, a monograph on epidermal ridges was published by Cummins and Midlo (1943/1961). This classic provided interesting information on the historical development of the scientific study of epidermal ridges and invaluable advice on how to record and analyze epidermal ridge configurations. Cummins and Midlo (1926) also coined the name *dermatoglyphics* (derma = skin; glyphics = carvings) for the scientific study of ridges as well as the ridges themselves. This label has now gained universal acceptance. They also included some information on a number of medical disorders. However, their work precedes the burgeoning of cytogenetics that has occurred in the last several years and, therefore, the recent work on dermatoglyphics in medical disorders is not included in their monograph. Although several works on dermatoglyphics have appeared since publication of the Cummins and Midlo classic, none has treated the subject comprehensively.

The present monograph attempts to fill this gap. It aims to provide an illustrated guide to ridge analysis and also to bring together the widely scattered recent and older information on dermatoglyphic studies in medical disorders. The practicing physician may find much of value in this book that he can apply in his day-to-day work. At the same time, there are sufficient data to provide a reference resource to the medical research worker. Because genetic and anthropologic aspects of dermatoglyphics are dealt with adequately in other recent publications (Cummins and Midlo, 1943/1961; Martin and Saller, 1962; Holt, 1968; Loeffler, 1969), they are not discussed in any great detail in this book. Only sufficient information is given on the latter topics to provide a foundation to the serious medical investigator. The first part of the book gives information on how to record dermatoglyphics. The second part provides instruction in dermatoglyphic interpretation, concentrating on aspects that have medical relevance. The third part provides up-to-date summaries of information on dermatoglyphics in a variety of medical disorders. A section is also included on palmar and finger creases, although, strictly speaking, analysis of creases is outside the pale of dermatoglyphics. The medical disorders discussed are a selected rather than an exhaustive list because isolated case reports and questionable analyses of some medical disorders have been excluded. Only well-substantiated data are included. The information has been collected from both the English and the non-English literature so that dermato-

glyphic data that may not have been readily available heretofore are opened to a wider audience. For readers who require only summary data, tables with the salient conclusions are provided; for those who require more information, the text should prove useful.

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1 Embryogenesis and Genetics of Epidermal Ridges

Dermal ridge differentiation takes place early in fetal development. The resulting ridge configurations are genetically determined and influenced (or modified) by environmental forces. There is a paucity of knowledge concerning the developmental mechanism that determines ultimate epidermal ridge patterns but a relationship to the fetal volar pads clearly exists because ridge patterns form at the sites of these pads.

Fetal volar pads (Figure 1.1) are mound-shaped elevations of mesenchymal tissue situated above the proximal end of the most distal metacarpal bone on each finger, in each interdigital area, in the thenar and hypothenar areas of the palms and soles, and in the calcarea of the sole. Secondary fetal pads may be found in other areas, such as on the central palm or as pairs on the proximal phalanges (Mulvihill and Smith, 1969). The formation of these pads is first visible on the fingertips in the sixth to seventh week of embryonic development. The pads become very prominent during the subsequent several weeks, diminish again in the fifth month, and disappear completely in the sixth month. Within this period the dermal ridges coalesce into specific patterns, replacing the volar pads. It is believed that the presence of the volar pads as well as their size and position are, to a large extent, responsible for the configuration of papillary ridge patterns, as postulated by Bonnevie (1924). For example, small pads would result in a simple pattern (arch), whereas more prominent pads would tend to lead to the development of large and more complex systems of ridge configurations (loops and whorls). Simi-

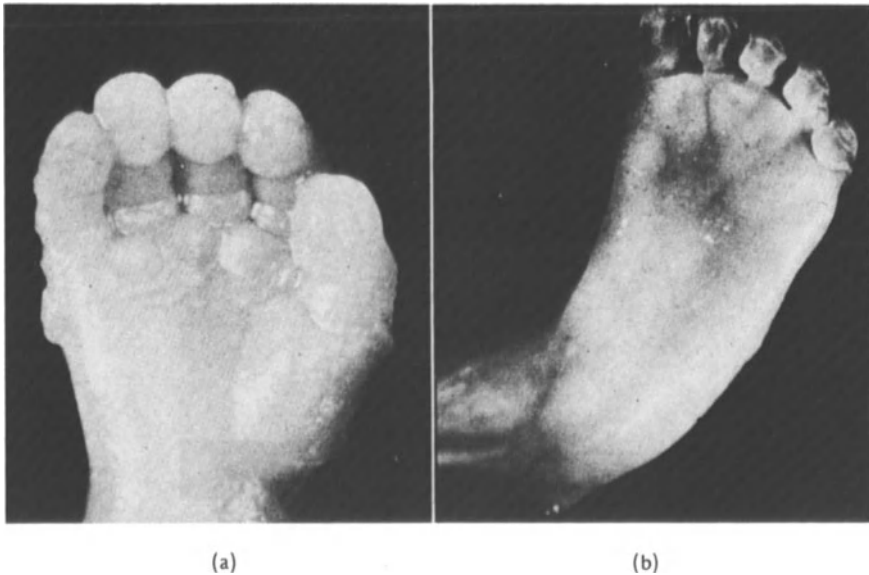


FIGURE 1.1 The hand (a) and foot (b) of a human embryo of approximately 70 days of age showing the fetal volar pads on the tips of the digits, in the interdigital areas, the thenar area of the palm, and the hallucal area of the sole.

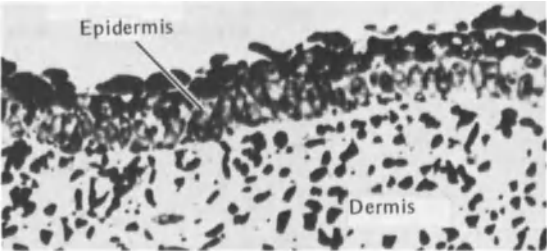
From Miller, J. R., and Giroux, J.: *Dermatoglyphics in pediatric practice. J. Pediatr.*, 69:302, 1966. Courtesy of the C. V. Mosby Company.

larly, fetal pads positioned symmetrically on the volar aspect of the fingertip would give rise to a pattern centered in the middle of the pattern area (whorl) and asymmetrical pads to a pattern asymmetrically oriented within the pattern area (loop, either ulnar or radial according to the position of the pad).

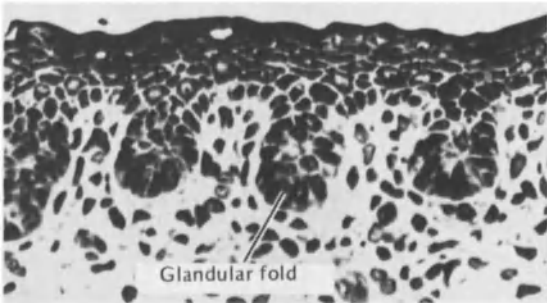
The embryogenesis of the papillary ridges has been studied extensively (Engel, 1856; Wilson, 1880; Kollman, 1883; Lewinski, 1883; Blaschko, 1887; Johnson, 1899; Bonnevie, 1924, 1927a, 1929, 1932; Cummins, 1929; Schaeuble, 1933; Abel, 1938; Gould, 1948; Hale, 1949, 1952; Blechschmidt, 1963). In recent studies, electron microscopy has been employed in an effort to investigate the developmental mechanism responsible for ridge formation and to describe disturbances of this process (Hirsch, 1971; Schweichel, 1971; Hirsch and Schweichel, 1973; Penrose and Ohara, 1973). Figure 1.2a shows a section of fetal skin at 9 weeks, prior to the development of ridge anlage. It has been established that the critical period of ridge formation begins in the fetus at approximately 70-mm crown-rump (C-R)

FIGURE 1.2 Skin development demonstrated on sections of fetal skin at 9 weeks (a), 16 weeks (b), and 23 weeks (c).

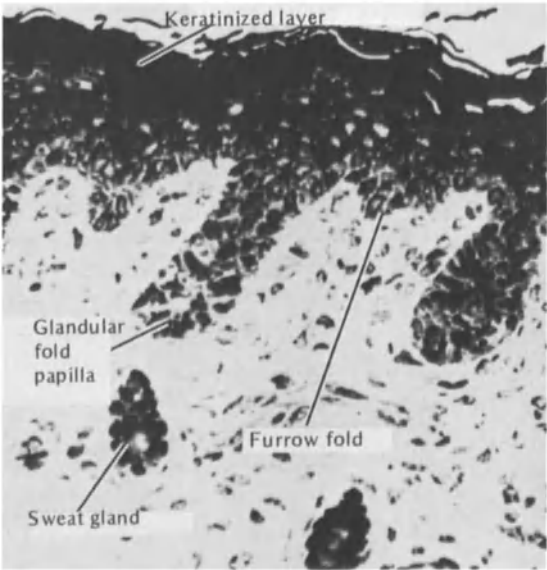
From Penrose, L. S., and Ohara, P. T.: The development of the epidermal ridges. *J. Med. Genet.*, 10:201, 1973. Courtesy of the *Journal of Medical Genetics*.



(a)



(b)



(c)

length, i.e., about 3 months of age, when the volar pads are near or just beyond their peak development. Whereas the outer surface of the epidermis remains smooth, an undulation can be observed in the basal layer of the epidermis. This shallow epidermal proliferation is seen in the fourth month as distinct, clearly defined folds of the lower layer of the stratum germinativum growing downward into the corium (Figure 1.2b). The corium, in turn, forms papillae projecting upward into the epidermis. These epidermal folds, later perceivable as glandular folds [or primary ridges according to Hale's nomenclature (1952)], form in a lateral-distal to medial-proximal direction on the fingertips. The center is initially free of folds. Subsequently, more and more folds form at the periphery of the pads and finally cover the entire pad surface (Hirsch and Schweichel, 1973). As growth continues, glandular folds divide at their tips and thus increase in number. After fold formation has been completed during the fifth month, the bulbous anlagen of sweat glands appear at points of deepest penetration of the folds into the corium. The anlagen grow as tubular epithelial cords into the connective tissue. At first, they are solid but they soon become hollow. Simultaneously, or perhaps slightly later, the glandular ducts begin to develop upward, reaching the surface of the epidermis during the sixth month (Hirsch and Schweichel, 1973). Sometime during this period, but without any strict temporal or spatial regularity, the furrow folds (or secondary ridges according to Hale, 1952; Figure 1.2c) appear between glandular folds (or primary ridges). Their development parallels that of the glandular folds except that sweat glands are confined to the latter. On the basis of the irregularity of the origin of the furrow folds as well as from observations in the literature, Hirsch and Schweichel (1973) have concluded that the furrow fold formation continues as a secondary phenomenon into the postnatal period and has no effect, or at most only a slight effect, on the formation of the papillary ridge patterns.

The epidermal ridge patterns are completed only after the sixth prenatal month, when the glandular folds are fully formed and after the sweat gland secretion and keratinization have begun. At this time, the configurations on the skin surface begin to reflect the underlying patterns. The surface epidermal furrows correspond to the furrow folds of the stratum germinativum and each epidermal ridge is formed above a glandular fold (Figure 1.3). The development of fetal skin structures, as observed by Schweichel (1971), is outlined in Table 1.1.

Ridge differentiation progresses from the apical pads proximally

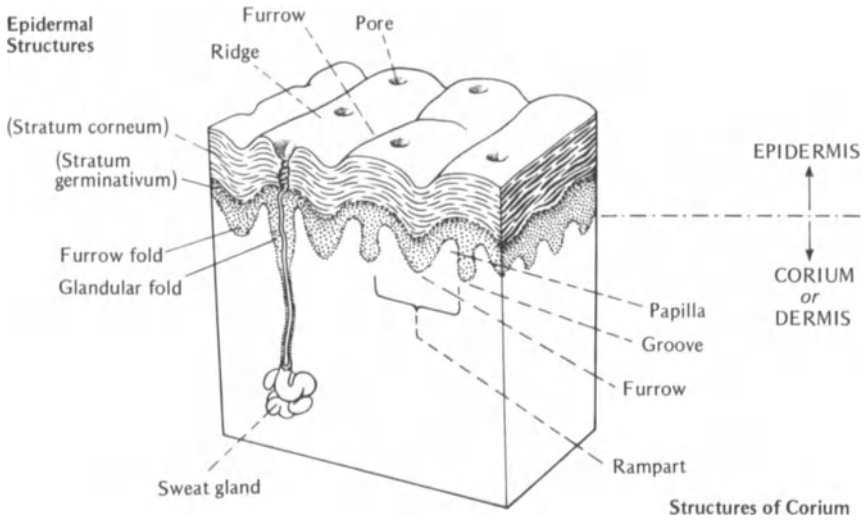


FIGURE 1.3 Diagrammatic representation of ridged skin with nomenclature applying to structures of epidermis and dermis.

From Penrose, L. S.: Memorandum on dermatoglyphic nomenclature. *Birth Defects*, 4(3):1, 1968. Courtesy of the National Foundation—March of Dimes.

and in a radioulnar or tibiofibular direction (Hale, 1952). The embryogenesis of the epidermal ridges on the feet is identical to that on the hands, except that each step occurs 2 or 3 weeks later.

Several hypotheses have been formulated concerning the forces that are responsible for the development of specific ridge patterns. Cummins (1926) speculated that the dermal ridge configurations were the result of physical and topographic growth forces. It is believed that the tensions and pressures in the skin during early embryogenesis determine the directions of the epidermal ridges. Bonnevie (1929) postulated that the fingertip patterns depended on the underlying arrangements of peripheral nerves. Penrose (1969) suggested that the ridges followed lines of greatest convexity in the embryonic epidermis. Hirsch and Schweichel (1973) have summarized present knowledge concerning the induction of the glandular folds and, in turn, the formation of the epidermal ridges. Based on previous observations and their own studies, they pointed out the regularity in the arrangement of the blood vessel–nerve pairs under the smooth epidermis–corium border which exists shortly before formation of the glandular folds. They postulated that the folds are induced

TABLE 1.1. Development of skin structure in the human fetus^a

	MONTH ^b						
	2	3	4	5	6	7	8
Crown-rump length (mm)	40	60	100	150	200	230	270
Blood vessels	++	++	++	++	++	++	++
Fetal pads	+	++	+	(+)	—	—	—
Nerves	(+)	+	++	++	++	+++	+++
Glandular folds	—	—	+	++	++	++	++
Furrow folds	—	—	—	(+)	+	+	+
Sweat glands	—	—	—	+	+	++	++
Epidermal ridges	—	—	—	—	(+)	+	++

^a According to Schweichel (1971).^b —, absence of the trait; (+), +, ++, +++, increasing degrees of development.

by the vessel–nerve pairs. The authors have illustrated the relationship between the nerves and epidermal ridges in abnormalities, such as aplasia of dermatoglyphic patterns when nerves fail to grow into the epithelium or in an abnormal ridge development associated with an abnormal nerve development. They list inadequate supply of oxygen to the tissues, deviations in the formation and distribution of sweat glands, disturbances in proliferation in the epithelial basal layer, and disturbances in keratinization of the epithelium as other factors that may influence epidermal ridge patterns. Even environmental factors, such as external pressure on the fetal pads and perhaps embryonic movements, particularly finger movement, can influence ridge formation.

Past research has demonstrated that the epidermal ridge patterns are under genetic influence. Galton (1892) and Wilder (1902, 1904) are the first to have studied the hereditary basis of dermal patterns, which has since been confirmed by numerous genetic studies. A high degree of similarity of dermatoglyphic traits has been found between monozygotic twins, whereas considerably less agreement exists between dizygotic twins. These observations have been utilized diagnostically in determining zygosity of twins (Siemens, 1927; Rife, 1933; Meyer-Heydenhagen, 1934; MacArthur, 1938; Essen-Möller, 1941; Geipel, 1941; Maynard Smith and Penrose, 1955; Nixon, 1956; Lamy *et al.*, 1957; Richter and Geisser, 1960; Slater, 1963; Allen, 1968; Parisi and Di Bacco, 1968; Hamilton *et al.*, 1969; Parisi *et al.*, 1970; Nylander, 1971). Because of the closer resemblance of dermatoglyphics among close relatives than among unrelated persons,

the possibility of using dermatoglyphic analysis as a complementary means in establishing paternity was suggested (Nürnberger, 1925; Bonnevie, 1927b; Müller and Ting, 1928; Müller, 1930; Harrasser, 1935; von Wehren, 1937, cited by Cummins and Midlo, 1961; Böhmer and Harren, 1939; Geipel, 1939).

The role of heredity in the establishment of the dermal patterns cannot be denied. However, the mode of inheritance of these ridge arrangements has not been clearly established. Studies of inheritance of the pattern sizes, direction, and shape often yield contradictory conclusions. Individual dermatoglyphic traits were claimed to be inherited as dominant, incompletely dominant, recessive, single gene or polygenic, with complete or incomplete penetrance and variable expression of the genes (Elderton, 1920; Bonnevie, 1924; Grüneberg, 1928; Newman, 1930; Müller, 1930, 1931; Weninger, 1935, 1947; Geipel, 1937; Weinand, 1937; Czik and Malan, 1938; Essen-Möller, 1941; Fang, 1949, 1950; de Wilde, 1953; Penrose, 1954; Wichmann, 1956; Bansal and Rife, 1962; Pons, 1963, 1964; Smith, 1964; Glanville, 1965).

Realizing the need for more objective means of dermatoglyphic analysis, many investigators have been exploiting the quantitative features of dermatoglyphics, such as counting individual ridges within a pattern or elsewhere between two well-defined points or using angular measurements. This quantitative approach has contributed appreciably to an understanding of the genetic influence on dermatoglyphics. A comprehensive review of genetics of the quantitative dermatoglyphic traits has been published by Holt (1968), whose own work represents a significant portion of research in this field.

At present, there is wide agreement that the heredity of most dermatoglyphic features conforms to a polygenic system, with individual genes contributing a small additive effect. Modern cytogenetic methods, which allow rather precise identification of chromosomes, are certain to be of great value in studying the correlations between individual chromosome aberrations and dermatoglyphic features and may lead to establishing the loci of genes that influence dermatoglyphics. However, a limitation to the precise genetic analysis of dermatoglyphics is the difficulty in delineating some features and reducing them to quantifiable characteristics. Many transitional features exist and too much latitude for subjective classification is still possible. Improvements in reliability of classification and more precise delineation of dermatoglyphic features will undoubtedly be followed by advances in an understanding of the importance of genetic factors in the development of epidermal ridge configurations.

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2 Methods of Recording Dermatoglyphics

Dermatoglyphics offer at least two major advantages as an aid to the diagnosis of medical disorders: (1) the epidermal ridge patterns on the hands and soles are fully developed at birth and, thereafter, remain unchanged for life; (2) scanning of the ridge patterns or recording their permanent impressions (i.e., prints) can be accomplished rapidly, inexpensively, and without any trauma to the patient.

A number of methods for recording dermatoglyphics exists. The methods vary in their requirements for equipment, time, and experience and in the quality of the prints produced.

Dermatoglyphic patterns are usually recognizable by the naked eye. A simple magnifying lens, preferably with a light source such as is found in an otoscope, helps greatly in scanning dermatoglyphics, especially in infants and small children whose patterns are very fine. The scan alone often gives the investigator sufficient data for most medical purposes but permanent impressions or prints are necessary for quantitative analyses of dermatoglyphics.

To enhance the quality of dermatoglyphic prints, it is necessary to remove sweat, oil, and dirt from the skin. This can be accomplished by washing the ridged areas with soap and water and with ethyl alcohol or ether.

Care must be taken to print the ridged areas completely. The ridges are primarily on the volar surface but also pass upward along the lateral margins of the fingers, palms, toes, and soles. Therefore, a print of only the volar surface may be incomplete and it is often necessary to roll the digits, palms, and soles to insure obtaining a

print of the whole pattern. Palm prints must include the area from the distal crease of the wrist to the metacarpal-phalangeal creases, and complete printing of both ulnar and radial sides of the ridged areas must be assured. Similarly, the heel and areas of the sole under the proximal portions of the toes must be included to avoid missing important details of the patterns. The quality of each print should be inspected as it is made so that in cases of possible technical defects an improved print can be taken immediately.

For those who use dermatoglyphics regularly, it is convenient to prepare standardized dermatoglyphic cards with spaces for entry of name and other identifying information. A space may be designated for each fingertip as well as for other pattern areas. When standardized cards are not used, it is wise to add the identification data to the print immediately after it is taken so that the specific digit or area and right and left are clearly marked. When only selected areas are printed, each must be identified at the time the impression is taken to avoid confusion and possible mistakes when the prints are analyzed.

A magnification lens of approximately four or five power helps in inspecting ridge details of printed areas. This strength of magnification is sufficient for most ordinary purposes, including counting of ridges. A low-power binocular microscope (eyepiece $6\times$, objective $0.7\times$) with a large field (25 mm) has been recommended for study of ridge detail (Holt, 1968). A needle or other object with a sharp point is needed for accurate ridge counting and in tracing the radiants.

Greater magnification may be required to study ridge detail in an aborted fetus or stillborn child. A technique for studying the dermal patterns in human fetuses has been described by Miller (1968), who has found the patterns discernible in fetuses from 90-mm C-R length to term. The observations were made under a dissecting microscope with a maximal magnification of $40\times$, although no magnification larger than 25 was needed. Problems in visualizing ridge detail were encountered in older fetuses because of the vernix caseosa and the thickness of the stratum corneum, but application of a commercial depilatory cream accompanied by vigorous rubbing with a dry tissue improved the quality in these specimens. In fetuses less than 4 months old but over 175-mm C-R length, bathing the surface is recommended to help bring out the ridge formation. Very fine ridges may be accentuated by a coloring agent, such as ink from felt pens.

Standard Methods

The methods described in the following section have been designed primarily to obtain prints of ridged skin for dermatoglyphic analysis. All methods described here are relatively easy to use, rapid, and inexpensive (with the exception of the photographic method, which requires an initial investment for the photographic instrument). However, the methods vary somewhat in the quality of the prints obtained. Any of these printing techniques may become a method of choice of an individual investigator, based on his preference of the features that each method offers.

INK METHODS

One of the best known and most widely used dermatoglyphic printing methods utilizes printer's ink and a good quality paper. The ink method gives very good results when used in adults and with cooperative children. It is the standard method of finger printing used by law enforcement agencies for identification purposes. The necessary equipment consists of printer's ink, a roller, a glass or metal inking slab, a sponge rubber pad, and good quality paper, preferably with a slightly glazed surface.

A small amount of ink is placed on the inking slab and spread with the roller into a thin, even film. The area to be printed is pressed against the slab, taking care that the whole area to be printed is covered with the ink. A firm surface is used under the sheet of paper on which the inked finger is pressed. However, impressions of the palms and soles obtained in this manner are often imperfect, showing interruptions of the epidermal patterns where the hollow of the palm or sole has not come into contact with the inked slab or the paper. To eliminate such irregularities, it is helpful to place a sponge rubber pressure pad under the paper on which the prints are made. The rubber pad is molded into concave portions of the hand and foot, which otherwise are difficult to print. Some operators prefer to roll the palm over a paper covered cylinder, such as a jar or bottle, to avoid imperfect printing of hollow areas.

Strong (1929) advocated use of a resilient surface for both inking and printing. In his method, a sheet of previously inked, paraffined paper is placed on a rubber pad and the ridged skin is brought into contact with the inked surface. The skin is then applied to a clean paper placed on the pad.

Purvis-Smith (1969) described a modified printing method using a roller with a rigid center and a plastic foam roller sleeve. A sheet of paper with a high-gloss surface facing outward is wrapped around the roller. The paper is fixed in place with an adhesive tape. The hand or foot is inked by pressing an inking plate against the skin and the print is taken by moving the roller from the wrist crease toward the fingertips in one smooth sweep.

When printer's ink is not available, an ordinary stamp pad in combination with shiny, smooth paper gives fair results.

The optimal amount of ink and pressure needed to obtain satisfactory prints is learned with a little practice. However, because these two factors are crucial for successful printing, the ink method is not suitable for use with uncooperative children and those with very fine ridges. Also some individuals object to being smeared with ink, which may soil clothes and is somewhat difficult to remove.

Commercial ink printing sets are available.¹ Such sets enjoy wide use on obstetrical services for identification of newborn infants. "Foot Printer" plates are covered with a special ink. The foot is placed on the inked pad and then is pressed on a special paper, provided with the set, that has a relatively hard and glossy surface. However, the quality of the prints usually obtained leaves much to be desired, as the task of printing is often assigned to hospital personnel unfamiliar with dermatoglyphics. The resulting prints are often incomplete or badly smudged.

For optimal results, the feet of the newborn should be clean and dry. Heavy inking should be avoided as it tends to smudge the fine details of the ridged skin. Although this method has been recommended by some experienced workers (Walker, 1957; Uchida and Soltan, 1963; Miller and Giroux, 1966; Purvis-Smith, 1969), the prints are not always of sufficiently good quality to allow accurate counting of the ridges.

INKLESS METHODS

The most common method for obtaining prints makes use of a commercially available patented solution and specially treated, sensitized paper.² The solution does not stain the skin, is nonirritating, and easily washes off with soap and water. The method is suitable for printing hands or feet with well-demarcated dermal patterns, such as those of older children and adults. However, owing to the combined difficulties caused by delicate skin ridges and poor cooperation, the

¹ Hollister, Inc., 211 E. Chicago Ave., Chicago, Illinois 60611.

² Faurot, Inc., 299 Broadway, New York, New York 10007.

technique meets with less success when used in infants or young children.

The inkless method was described in detail by Walker (1957). The area to be printed is rubbed well with a flannel pad dampened with a patented chemical solution. The treated skin is then pressed firmly onto the sensitized paper. It is helpful to place the sensitized paper on a sponge rubber pad when the print is taken. This allows the concave regions in the interdigital and central palmar areas to make contact with the paper. Care must be taken not to apply too much fluid or pressure or the resulting prints will be smudged. The amount of liquid, the pressure, and the duration of contact between palm and paper is learned from experience. According to Walker (1957) even small children can be printed using this method.

Freiberg (1901) described an inkless method using paper dampened with a saturated solution of tannic acid in 80 percent alcohol. The skin was painted with a solution of 50 parts of tincture of ferric chloride, 45 parts of 80 percent alcohol, and 5 parts of glycerin and pressed on paper. Cummins *et al.* (1929) obtained some excellent prints with the tannate method but results were not uniformly good. The method is not popular currently.

Several other techniques that can be listed as “inkless” methods have been described. They are not used for routine dermatoglyphic analysis but may be utilized if no other means of obtaining permanent prints is available. Bauder’s procedure, described by Cummins and Midlo (1961), makes use of a light machine oil, which is applied to the skin. The impression on paper is dusted with the black powder commonly used in treating latent prints and then sprayed with a fixing solution of 20 parts of alcohol, 2 parts of white shellac, and 1 part of sandarac gum.

MacArthur and Ford’s method, cited by Cummins and Midlo (1961) is based on the same principle as the machine oil technique. It makes use of a hand lotion or face cream, which is rubbed into the skin. The moist skin is then pressed lightly on mimeograph paper. Finely powdered lampblack is rolled repeatedly over the paper with a rocking motion and the ridge impressions stand out clearly. The print is then fixed from the back in a solution of resin in alcohol (30 g of resin to 1 liter of 95 percent alcohol).

TRANSPARENT ADHESIVE TAPE METHOD

Use of a transparent adhesive tape as a means of obtaining prints of dermatoglyphic patterns is not new. In 1948, Böök described such a method using a transparent cellulose tape and white or

pale yellow chalk. After the skin had been cleaned with ether, the fingertips were rubbed with chalk and a piece of tape was placed over the area with the adhesive side against the skin. Light pressure was applied and the strip was then carefully removed. The chalk dust that adhered to the tape recorded the ridge detail and the adhesive strip was examined under a magnifying glass. This procedure is similar to the indirect process employed by criminologists for "lifting" patterns off dusted smooth surfaces containing impressions of finger prints.

Cotterman (1951) modified this method, making it particularly useful for infants and small children. He painted India ink onto the entire palm or sole. After the ink had dried, strips of transparent adhesive tape were rolled onto the skin surface from one side to the other, insuring contact with the whole ridged area. The tape was then removed and transferred to paper for a permanent record. This method overcomes the difficulty usually encountered in printing the concave portion of the palm and the convexity of the foot, areas that are often poorly or only partially printed when using the methods previously described.

MacLennan's (1964) method was practically identical to that of Cotterman except that a standard ink was used instead of India ink. The slight advantage of Cotterman's method is that the India ink is easier to remove from the skin with soap and water.

A modification of the above methods that is based on the same principle involves use of dry pigment, either carbon paper or powdered graphite, and wide sheets of self-adhesive tape. The method was described by Aase and Lyons (1971). Ordinary carbon paper is used for adults and older children, whereas in newborn infants the best results are obtained with powdered graphite. In adults and older children, the carbon paper is rubbed lightly and evenly over dry skin surfaces. In newborns, the dried hand is dusted sparingly with a small amount of powdered graphite, using a powderpuff. Sommer and Gregg (1973) recommended a common oil pastel crayon as the pigment. In these methods, the print is produced by applying a dry pigment to the skin and lifting it off with tape.

A wide stick of graphite,³ used as a source of carbon powder, has been very satisfactory in our experience. The tape we prefer is 4-inch-wide Scotch Book Tape No. 845.⁴ It is wide enough to allow printing

³ Airco Speer Electrodes & Anodes, 4861 Packard Road, Niagara Falls, New York 14302.

⁴ Minnesota Mining and Manufacturing Company, St. Paul, Minnesota 55101.

of almost all palms and is easier to handle than wider sheets of adhesive tape. The graphite stick is rolled over the area to be printed. If necessary, a small amount of graphite powder can be loosened from the surface of the stick with a nail file. A strip of tape is then attached to the proximal part of the palm over the wrist crease and is smoothed down over the palm to the fingertips. All areas of the palm and fingers should be in contact with the adhesive side of the tape. The tape is then removed by peeling off from the wrist toward the fingertips. The carbon powder clearly shows the ridge patterns. The prints obtained are attached to a sheet of white or lightly colored paper and ridge patterns are seen through the transparent backing of the tape. In some cases, such as in young children and uncooperative patients, it may be easier to print each finger separately.

The powdered carbon-transparent tape method is very inexpensive, rapid, and easy to use with all types of patients, including newborns and individuals with malformed hands and feet that are otherwise difficult to print on flat, hard surfaces. Prints are invariably clear and not smudged. They can be preserved for an indefinite period of time. Moreover, the transparent tape containing the prints can be marked with a pen or a grease pencil, which can later be erased without destroying the print. It should be remembered that the print made with ink and the print "lifted" by transparent tape are mirror images of each other.

Sutarman (1971) has offered another modification of the adhesive tape method, substituting for the dry coloring pigment a solution of polyvinyl formal in ethylene dichloride that is painted or sprayed onto the skin. A thin film forms in 10 to 15 seconds and is easily peeled off by applying a strip of transparent vinyl adhesive tape. The tape, together with the adherent film, is then mounted on a slide for inspection under a microscope. Other plastic solutions may be used so long as they do not irritate the skin.

PHOTOGRAPHIC METHOD

Harrick (1962–1963) developed a photographic apparatus (manufactured as the Norelco Fingerprint Instrument⁵) for use especially with newborns and infants. The technique is based on the principle of frustrated total internal reflection which occurs when an object is pressed against a prism. The magnified image is photo-

⁵ North American Philips Company, Inc., Norelco Electronic Products, 100 E. 42nd Street, New York, New York 10017.

graphed by a polaroid camera. The procedure is clean, because no ink is required, and the clarity of the print is not disturbed by applying pressure. This is an important factor because, with ink methods, it is difficult to insure uniform, gentle pressure required for successful printing. The prints obtained by the instrument are very clear but, because only areas in direct contact with the rigid surface of a prism can be photographed, the method has only a limited value in dermatoglyphic analysis. Also, the equipment needed is relatively expensive.

Special Methods

The methods listed below are not widely used to obtain dermatoglyphic prints. However, they may have some advantages that the standard methods cannot offer, such as allowing the study of the correlation between the epidermal patterns and the underlying bone structures (radiodermatography), study of sweat pores (hygrophotography), or study of the spatial shape of the ridged skin areas (plastic mold method). These methods may be useful to the investigator interested in special, rather than standard, dermatoglyphic features.

HYGROPHOTOGRAPHY

Hygrophotography, a process by which an image can be obtained on a sensitized surface by the combined action of light and humidity, has been employed for dermatoglyphic purposes (Sivadjian, 1961, 1970). This method demonstrates epidermal patterns, as well as pores and the sites of active sweat glands. The technique is based on the fact that the sensitized surface of the hygrophotographic plate or film has the property of changing its color when exposed first to light and then to water or humidity. In using this technique, hygrophotographic film is first exposed to light and is allowed to darken. The subject then puts his hand or foot on the film for a period varying from a few seconds to a few minutes. The film reacts with the moisture from cutaneous perspiration. Subsequently, the print is developed as a negative, using bromide paper.

Obviously, this technique is more expensive and time consuming than the previously described printing methods, and it is not widely used for dermatoglyphic purposes although very good prints can be obtained. It has, however, a definite advantage in special studies, as

in those in which the activity of the sweat glands is of interest. According to Sivadjan (1961), sweat gland clusters are unique to each individual and so provide means of positive identification. This method can therefore be used in individuals with dermabrasions in which the ridges have been lost.

Another photographic method was devised by Mathews (cited by Cummins *et al.*, 1929). A developing solution (25 g sodium sulfide, 5 g sodium hydroxide, 2 g soluble starch, and 100 ml distilled water) is prepared. A blotting paper pad moistened with this mixture serves as the "inking slab." The part of the hand or sole to be printed is first pressed for a few seconds against the moist blotter and then applied against a sheet of photographic paper. The unfixed prints may be exposed to light for a short time without damage, but to achieve permanency the prints must be fixed, washed, and dried as in the usual photographic developing process. Montgomery (1925) considered this technique to be superior to the ink method, especially in dealing with infants. A slightly modified version of Mathews' method was described more recently by Arthur (1972).

In yet another photographic method (Schött, 1928, cited by Cummins and Midlo, 1961), photographic film was applied to skin on which lanolin was applied to cover the epidermal ridges. The film was then processed as in ordinary photography.

RADIODERMATOGRAPHY

Radiodermatography uses radiography to demonstrate dermatoglyphics (Poznanski *et al.*, 1969). The idea of employing x rays for the purpose of identifying epidermal ridge patterns is not a recent one. Valšik (1933, cited by Cummins and Midlo, 1961) used the x rays indirectly, making separate radiographs and paper prints. Radiopaque lead markers were used to mark the triradii on the radiographs.

Radiographic techniques involving use of contrast materials to fill the areas between the dermal ridges have been described by several investigators. Castellanos (1939) reviewed the early methods, mentioning Nelken (1920, cited by Castellanos, 1939), who combined radiography with the study of fingerprints as a means of identification (dactyloscopy). Credit was given to Bécclère for originating dactyloscopic radiography. Bécclère (1918) massaged bismuth carbonate, a radiopaque contrast material, into the skin surface after first treating the skin with lanolin. X rays taken afterwards revealed not only bone

structures but also dermatoglyphic patterns. Power (1921), however, did not find the use of bismuth salts satisfactory. He also tried and rejected barium and lead salts. However, he found commercial white oil paint, which probably had a lead base, "a perfect medium for rendering visible even the finest details of the skin." To avoid the troublesome washing of the skin after the procedure, he used a mixture of paint with paraffin, which can be removed as a solid shell.

Other materials used for radiodermatographic purposes include litharge (Barnes and McLachlan, 1943), zinc bromide solution in a rubber cot (Richards, 1956), and 325-mesh tantalum powder (Poznanski *et al.*, 1969; Garn *et al.*, 1970). Tantalum powder seems to be the most suitable material, especially for routine use in young children or mentally retarded individuals, because it is not only highly radiopaque but also nontoxic.

Radiodermatography is more complicated, time consuming, and less economical than ink-paper or graphite-tape methods. It has, however, a special use in studies correlating the dermal ridges and skin folds with underlying osseous structures. It can also be used in criminal dactyloscopy when decomposition and putrefaction of the body make it impossible to obtain the fingerprints by other methods.

PLASTIC MOLD

Although not widely used for conventional purposes, plastic mold or dental wax impressions of ridge patterns may be mentioned as another technique for studies of dermatoglyphics and sweat gland activity (Sutarman and Thomson, 1952; MacKinnon, 1964). It has been used in studies of dermatoglyphics in nonhuman primates (Tips *et al.*, 1964), whose irregular volar reliefs and uncooperativeness make them difficult subjects to print by other methods. Tips *et al.* (1964) have found it also the method of choice for human subjects in studies involving correlations between dermatoglyphics and hand or foot form or in dealing with malformations of which conventional techniques yield inadequate prints. Prepared mold mixture is spread over the dried volar surface in a proximal-distal direction and then massaged over the digits. After it has hardened 3–5 minutes, it is removed from the skin, ready for analysis. The molds are permanent and maintain the natural shape of printed areas.

Sands (1972) found the dental-impression alginate compounds a much less expensive molding material than the latex materials used by Tips *et al.* (1964). The alginate takes only 1 minute to set to a rubbery mold from which plaster of Paris casts may be made.

AUTOMATIC PATTERN RECOGNITION

In recent years, mechanical methods have been developed for automatic pattern recognition. The techniques are used for purposes of personal identification and also for analyzing dermatoglyphics for medical uses. In Trauring's (1963) method, ridge minutiae form the basis for the analysis. Three "reference" minutiae are identified manually on the skin and a number of "test minutiae" are selected. These are all stored in a computer which records the density pattern. The dermatoglyphic patterns that include these minutiae are projected onto a screen. An electronic device sensitive to light and dark contrasts is used to scan the projected image, thus "counting" the ridges. Once the "reference" density pattern is detected, a linear and rotational coordinate transformation is computed in relation to the arbitrarily selected reference minutiae. "Misplacements" of the pattern relative to the arbitrary point "origin" are therefore discounted. The system then proceeds to check whether in the pattern being tested the "test minutiae" are of the type that are in the stored pattern and whether they are correctly located relative to each other. Trauring's method was meant to serve as an automatic method for identification of persons and offers no special value in medical studies.

Many scientists have felt it desirable to substitute a more objective technique for the subjective clinical analysis of dermatoglyphics now in use. Grasselli (1969) recognized that a typical ridge pattern contains much redundancy and he attempted to make use of the redundant information. In his method, the fingertip pattern is divided into 320 squares, called *samples*. A "random-access flying-spot" scanner digitizes the pattern into 512×512 points in eight grey tones. The digitized points are electronically evaluated in each sample and the predominant ridge slope can be determined. The system allows for eight possible slopes (numbered 0 to 7). The "sampling matrix," i.e., the matrix showing the predominant slope in each sample, gives a fairly good picture of the fingertip pattern and eliminates all the fine structure (the "noise"). Pattern landmarks are easily identified as they yield very different slopes in neighboring samples. Critical points, such as triradii, can be more accurately located by refining the samples or by ridge-following algorithms. With a little "smoothing," a sketch of the pattern can then be drawn automatically or the digitized pattern can be evaluated automatically.

Hall *et al.* (1973) proposed another method potentially useful in medical applications of dermatoglyphic analysis. In this technique, fingerprints are obtained by standard ink methods. A flying-spot

scanner, together with a coherent optical processor, is used to obtain a spatial Fourier transform. An image dissector system digitizes this transform. The digital information obtained can then be used to extract diagnostic information automatically, using some predetermined rules or learning algorithms and digital pattern classification techniques. Although this method, as proposed, is offered for analysis of the fingertips only, it may be applied to the palms and soles as well.

The disadvantage of this method is that it requires a clear print with very good contrast for analysis by the scanner. Such prints are frequently not available. The prohibitive cost of the analyzing equipment is another drawback for the use of this method by individual investigators. However, this could be overcome by employing a central analytical station. Because of the fine resolution of the system and because the complete print, including all minutiae, is contained in the spatial Fourier transform, this method appears to allow investigators to resolve the question of whether the fine structure of a dermatoglyphic pattern contains any information of interest in medical disorders.

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CHAPTER 2: METHODS OF RECORDING DERMATOGLYPHICS

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3 Dermatoglyphic Pattern Configurations

Numerous investigators have proposed rules, principles, and definitions that allow reliable analyses of dermatoglyphics. The earlier proposals have been reviewed by Cummins and Midlo (1961). Their monograph served as the most comprehensive guide to dermatoglyphic analysis for more than three decades.¹ When a relationship was reported between unusual dermatoglyphic features and certain chromosomal abnormalities, interest in dermatoglyphic analysis increased rapidly. The widespread interest in this field and a need for a standardized dermatoglyphic terminology stimulated an international symposium. As a result, a “Memorandum on Dermatoglyphic Nomenclature” was published (Penrose, 1968). The memorandum listed and defined dermatoglyphic features, outlined accepted methods of dermatoglyphic analysis, and provided a nomenclature that was intended to serve as a universal standard. The more subtle aspects of dermatoglyphics, which were previously dealt with in detail by Cummins and Midlo (1961) were not included in the memorandum.

This chapter should provide the reader with sufficient knowledge to allow her or him to carry out a routine dermatoglyphic analysis without need to resort to other sources. For the most part, the terminology advocated by Cummins and Midlo (1961) and by Penrose (1968) has been used throughout this book but, when deemed desirable, minor modifications in their terminology have been in-

¹ First published in 1943 (Philadelphia, Blakiston).

roduced. Where dual or multiple methods of analysis or a choice of terminology has been available, a recommendation based on our experience or our preference has been offered.

Ridge Detail (Minutiae)

Although the epidermal ridges are viewed as more or less parallel lines within a limited area of the ridged skin, careful inspection of epidermal ridge patterns reveals numerous irregularities of direction, discontinuities, and branchings of individual ridges. These intricate details of ridge structure, termed *minutiae* by Galton (1892), are highly variable and their number, type, shape, and position are unique to the individual. The minutiae are therefore a valuable and reliable tool for personal identification. However, apart from identification purposes, minutiae are not known to be of any medical value. Although Ökrös (1958) suggested their use in cases of questionable paternity, other genetic studies of minutiae (Grüneberg, 1927; Newman, 1930; Steffens, 1965) lent very little support to this potential application. Evaluation of a possible diagnostic application of minutiae in Down syndrome (Loesch, 1973) was inconclusive. However, Loesch (1973) used only a very simplified analysis. A more sophisticated, objective, and automatic technique of pattern classification (see Chapter 2) might yet reveal some usefulness of minutiae in medical diagnosis.

Several classifications of minutiae have been proposed, ranging from those recognizing only very few (Loesch, 1973) to those including a large number (Steffens, 1965) of the types. Penrose (1968) proposed a classification based on the six common types of minutiae (Figure 3.1) described by Cummins and Midlo (1961), and added a seventh type, a *comb* (Figure 3.5F).

One of the more common minutia is the *island* or *point*, which consists of a very short ridge lying independent of other ridges. It has an approximately circular shape and bears only one sweat pore. A *short ridge* is another common type of minutia and contains from two to about five sweat gland pores. The *fork* represents a bifurcation of a ridge. Because of its shape, it is sometimes referred to as a Y formation. Two such branches of a bifurcated ridge may rejoin after a short course, forming an *enclosure* (lake, ring, eyelet). An abrupt termination of the ridge is called an *end*. A *comb* is a ridge formation in which three or more parallel ridges join another ridge almost at right angles to their direction of flow. The *interstitial lines*

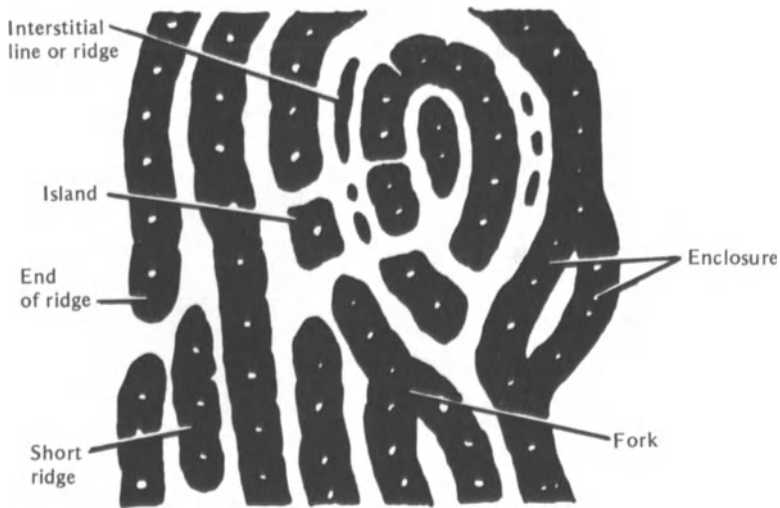


FIGURE 3.1 Nomenclature of minutiae.

From Penrose, L. S.: Memorandum on dermatoglyphic nomenclature. *Birth Defects*, 4(3):1, 1968. Courtesy of the National Foundation–March of Dimes.

(subsidiary ridges) are sometimes found in the furrows between the individual ridges. They are narrow, usually less than one-half of the width of ridges, and they characteristically lack sweat pores. They have also been called incipient, nascent, rudimentary, secondary, or vestigial ridges. However, these terms have been found to be inappropriate and misleading and their use is not recommended (Penrose, 1968). The interstitial lines are omitted in ridge counting.

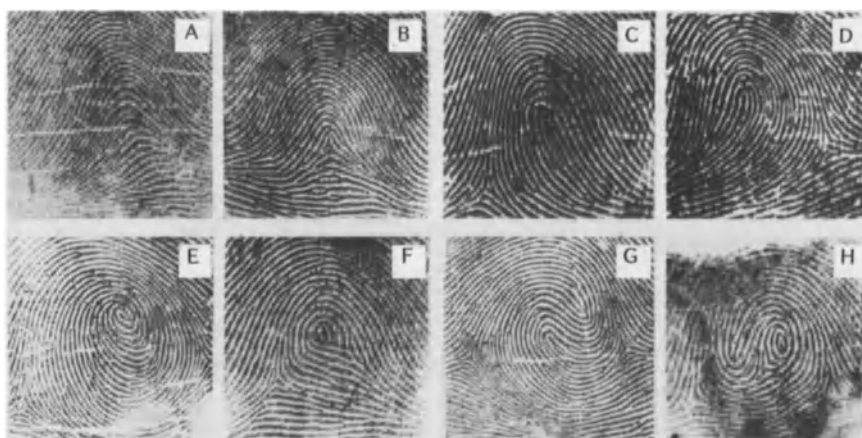
Pattern Configurations

FINGERS

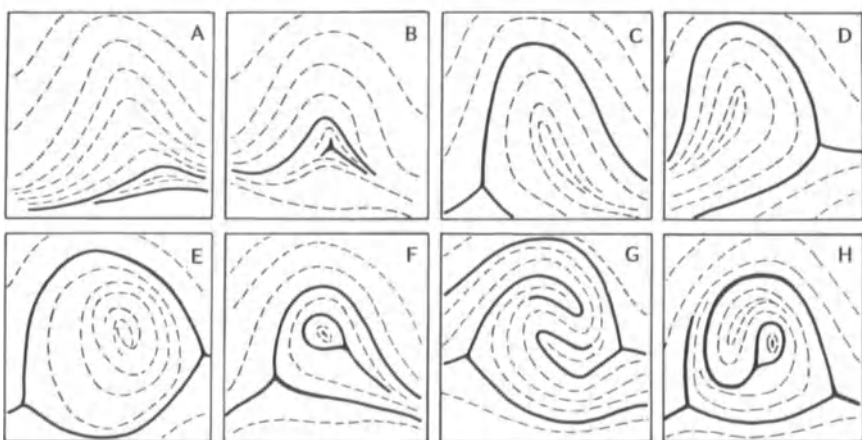
Fingertip pattern configurations

In 1892, Galton divided the ridge patterns on the distal phalanges of the fingertips into three groups: arches, loops, and whorls. Although numerous subclassifications have been subsequently offered, this simple classification is still recognized and used by the majority of investigators today (Figure 3.2).

The simplest pattern to be found on the fingertips is an *arch*. It is formed by a succession of more or less parallel ridges, which traverse



(a)



(b)

FIGURE 3.2 Types of fingertip patterns: actual prints (a) and schematic drawings with boldly traced type lines (b). A, simple arch; B, tented arch; C and D, loop (ulnar or radial); E, simple whorl; F, central pocket whorl; G, double loop whorl; H, accidental whorl.

From Alter, M.: Dermatoglyphic analysis as a diagnostic tool. *Medicine*, 46:35, 1966. Courtesy of the Williams & Wilkins Co., © 1966.

the pattern area and form a curve that is concave proximally. Sometimes the curve is gentle; at other times it swings more sharply so that it may also be designated as a low or high arch, respectively, if it is desirable to describe its sweep in more detail. The arch pattern is subdivided into two types. The *simple* (or *plain*) *arch* (A) is composed of ridges that cross the fingertip from one side to the other without recurving (Figure 3.2A). In a technical sense, such a ridge configuration is not a true pattern. If, however, the ridges meet at a point so that their smooth sweep is interrupted (Figure 3.2B), a *tented arch* (T or A¹)¹ is formed. The point of confluence is called a *triradius* because ridges usually radiate from this point in three different directions.

In the *tented arch*, the *triradius* is located near the midline axis of the distal phalanx. The distal radiant of the *triradius* usually points vertically toward the apex of the fingertip. Ridges passing over this radiant are abruptly elevated and form a tentlike pattern—hence the designation “*tented arch*.” Although the distal radiant usually terminates after only a short vertical course, it may occasionally re-curve sharply and point laterally or proximally. Such arches may simulate a loop (Figure 3.3, patterns 32 and 36) or even a much reduced whorl (Figure 3.3, pattern 34), as illustrated by Cummins and Midlo (1961), and careful attention to ridge details is required to classify the pattern correctly.

The most common pattern on the fingertip is a *loop*. In this configuration, a series of ridges enters the pattern area on one side of the digit, recurves abruptly, and leaves the pattern area on the same side (Figure 3.2C, and D). If the ridge opens on the ulnar side the resulting loop is termed an *ulnar loop* (U, L^u), whereas if it opens toward the radial margin it is called a *radial loop* (R, L^r). A loop has a single *triradius* or confluence point of ridges. The *triradius* is usually located laterally on the fingertip and always on the side where the loop is closed.

Loops may vary considerably in shape and size. They may be large or small, tall or short, vertically or horizontally oriented. Their size can be “measured” by counting the ridges, as is described later. Occasionally, “transitional” loops can be found which resemble whorls or complex patterns. For instance, pattern 17 in Figure 3.3 is a loop, although it resembles the more complicated double loop whorl (pattern 10), because one of its loops is degenerate; i.e., it

¹ The symbol T should be used for a *tented arch* and the symbol A^t should be reserved for an *arch tibial* to avoid possible confusion.

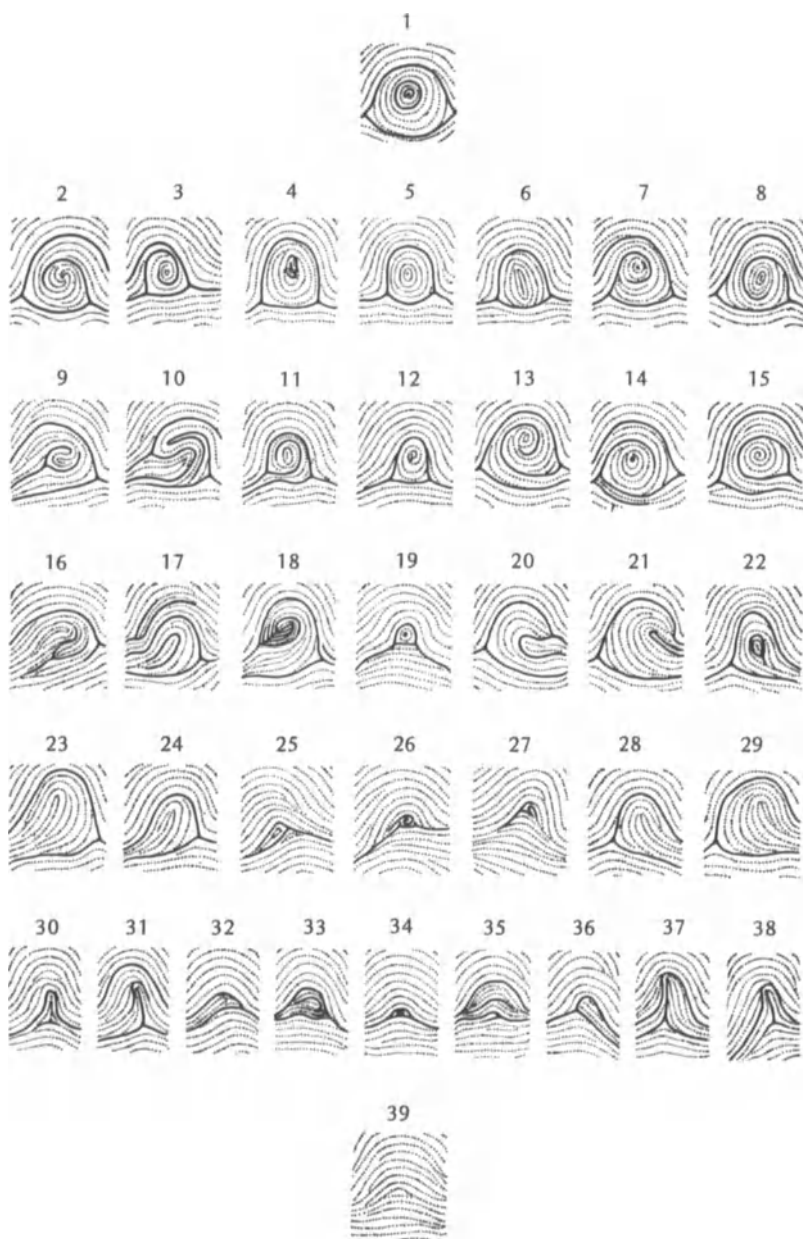


FIGURE 3.3 Various fingertip pattern types, including transitional patterns.

From Cummins, H. and Midlo, C.: *Finger Prints, Palms and Soles*. New York, Dover Publications, 1961. Courtesy of Mrs. Charles Midlo.

has a ridge arrangement simulating a loop but not containing any recurving ridges. Pattern 22 in Figure 3.3 is also a transitional loop in spite of its resemblance to a central pocket whorl (pattern 15) because of the absence of recurving ridges between the triradius and core.

A *whorl* (W) in Galton's classification is any ridge configuration with two or more triradii. One triradius is on the radial and the other on the ulnar side of the pattern. Henry (1937) limited the designation of "whorl" to those configurations having ridges that actually encircle a core. He called more complex patterns "composites" but most investigators use the Galton classification based on the number of triradii present (Penrose, 1968). The ridges in a *simple whorl* (Figure 3.2E) are commonly arranged as a succession of concentric rings or ellipses. Such patterns are described as *concentric whorls* (W^c , Figure 3.4A). Another configuration spirals around the core in either a clockwise or a counterclockwise direction. This pattern is called a *spiral whorl* (W^s , Figure 3.4B). Sometimes, both circles and ellipses or circles and spirals are present in the same pattern so that, for example, a whorl that is concentric near the core becomes a spiral toward the periphery of the pattern. The size of the whorl can vary considerably, as illustrated in the sequence of patterns 1, 5, 12, 19, and 26 in Figure 3.3. As in the loop, the size of the whorl is determined by means of a ridge count.

A *central pocket whorl* (W^p , Figure 3.2F) is a pattern containing a loop within which a smaller whorl is located. Central pockets are

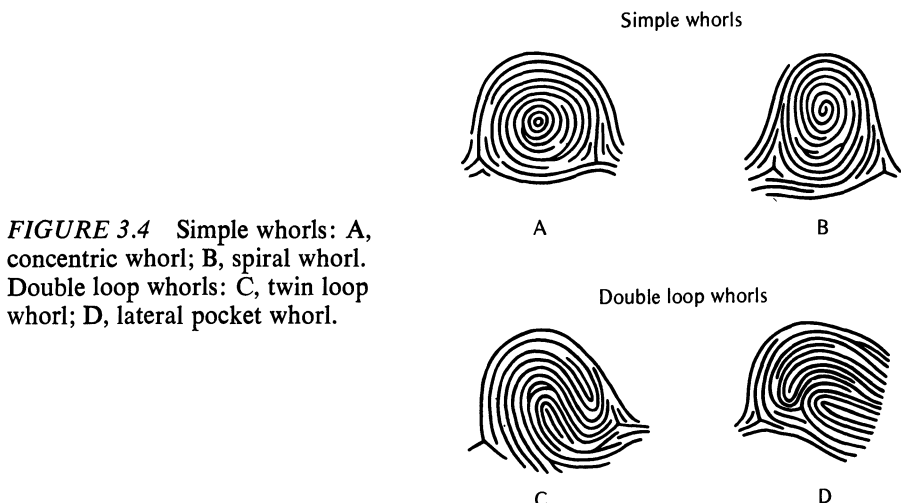


FIGURE 3.4 Simple whorls: A, concentric whorl; B, spiral whorl. Double loop whorls: C, twin loop whorl; D, lateral pocket whorl.

classified as ulnar or radial according to the side on which the outer loop opens.

Another type is composed of interlocking loops, which may form either a *lateral pocket* (W^p) or a *twin* (or *twinned*) *loop* (W^t) pattern. Each has two triradii and the two types of whorls are morphologically similar. However, in a twin loop whorl, the ridges emanating from each core open toward the opposite margin of the finger (Figure 3.2G and 3.4C) and the pattern cannot be designated as either ulnar or radial. In a lateral pocket loop whorl both ridges emanating from the core emerge on the same side of the pattern (Figure 3.4D.) The pattern can be described as a radial or ulnar subtype. The significance of separating these two varieties of loop whorls for medical diagnosis remains unproved and, therefore, they are ordinarily grouped together as a *double loop* (W^d).

Complex patterns, which cannot be classified as one of the above patterns, are called *accidentals* (W^{acc} , Acc, Figure 3.2H). They represent a combination of two or more configurations, such as a loop and a whorl, triple loops, and other unusual formations.

A number of more detailed classifications of fingertip patterns has been proposed that can be useful in very large series studied for anthropological purposes. However, in view of the great inherent variability of patterns, a very detailed subdivision of the pattern types is unlikely to be useful in the medical diagnosis of an individual case.

Dermatoglyphic landmarks

The three basic dermatoglyphic landmarks found on the fingertip patterns are the triradii, cores, and radiants.

A *triradius* is formed by the confluence of three ridge systems (Figure 3.5). The geometric center of the triradius is designated as a *triradial point*. Ideally, the triradial point is the meeting point of three ridges that form angles of approximately 120° with one another (Figure 3.5A). However, if the three ridges fail to meet, the triradial point can be represented by a very short, dotlike ridge called an *island* (Figure 3.5B) or by a ridge ending (Figure 3.5C), or it may lie on a ridge at the point nearest the center of the divergence of the three innermost ridges (Figure 3.5D). Sometimes, the triradial point does not lie on a ridge and is determined as the point where the three angles between the innermost ridges are each as near as possible to 120° (Figure 3.5E,F). The triradial point forms one terminus of the line along which ridges are counted (see p. 60). Unusually large patterns may have ridge configurations suggesting that a triradius would be formed were not the pattern interrupted by the presence of a fingernail or termination of the ridged skin of the digit. The tri-

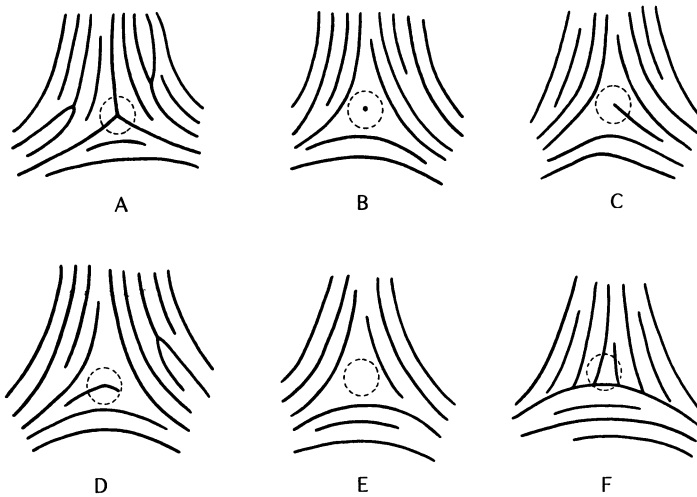


FIGURE 3.5 Diagrams of different types of ridge arrangement in the area of a triradius. The triradial point is the center of the dotted circle. See text for description.

Adapted from Penrose, L. S.: Memorandum on dermatoglyphic nomenclature *Birth Defects*, 4(3): 1, 1968.

radius in such cases is described as *extralimital* (Figure 3.6). Extralimital triradii are rarely encountered on the fingers or toes but are commonly observed in the hypothenar areas of the palms and the hallucal areas of the soles.

Another important landmark employed in ridge counting is a *core*, which is in the approximate center of the pattern. The core may be of different shapes. In a loop pattern, the core is usually represented by a straight, rodlike ridge or a series of two or more such parallel ridges over which other recurving ridges pass (Figure 3.7). If a straight ridge is absent in the center of the loop, the innermost recurving ridge is designated as a core. In a whorl, the core can appear as a dot or a short ridge (either straight or bent) or it can be shaped as a circle or an ellipse in the center of the pattern. In ridge counting, not the whole core but the *point of core* only is used. The point of core is at the distal tip of the straight line forming the core (Figure 3.7A). When the innermost recurving ridge contains no ending ridge, the point of core is placed on the shoulder of the loop farther from the triradial point (Figure 3.7B). The shoulders of a loop are the points at which the recurving ridge definitely curves. When an even number of rodlike ridges is present, the point of core is placed on the end of one of the two center ridges farther from the digital triradius (Figure 3.7C and F). If there are two straight ridges



FIGURE 3.6 Rolled print of a fingertip whorl pattern with “extralimital triradii.” No actual triradii are present, although the print extends beyond the area of ridged skin on both sides. Because of its unusually large size, the pattern extends beyond the distal interphalangeal flexion crease.

From Holt, S. B.: *The Genetics of Dermal Ridges*, 1968. Courtesy of Charles C Thomas, Publisher, Springfield, Illinois.

within the innermost recurving ridge, one of which does not rise as high as the shoulder of the loop, the tip of the other ridge is chosen as the point of core (Figure 3.7D). When an uneven number of rods makes up the middle of the pattern, the point of core is the tip of the central rodlike ridge (Figure 3.7E and G). The recurving ridge representing the core must have no appendage connected perpendicularly to its tip on the outside. In the presence of such an appendage, the loop is considered spoiled and the next loop outside is considered in locating the point of core (Figure 3.7H and I). Two recurving ridges side by side at the center of the pattern are treated as one loop with two rods within the recurve. The “rod” farther from the triradius is chosen as carrying the point of core (Figure 3.7J).

The *radiants* (*type lines*) are ridges that emanate from the triradius and enclose the pattern area. These ridges constitute the “skeletal” framework of the pattern area. In schematic drawings, the type lines alone are used to represent the pattern (Figure 3.2b). By following the ridges that originate in the triradius, the type lines can

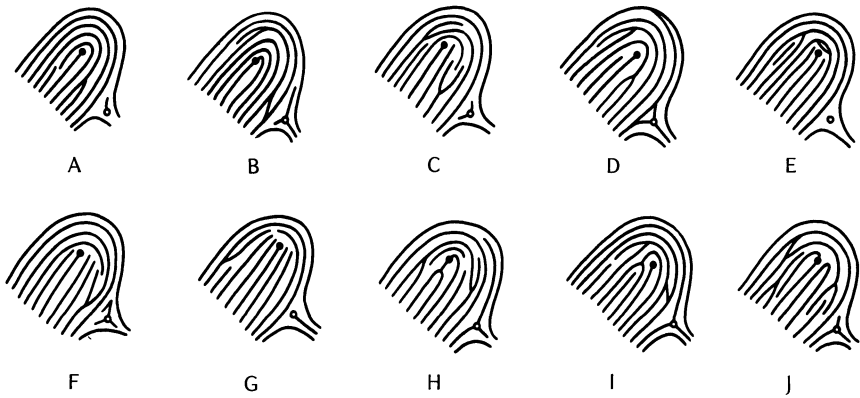


FIGURE 3.7 Diagrams of different types of ridge arrangement in the area of a core. The closed circle represents the point of core, the open circle is the triradial point. See text for description.

easily be traced. If a traced ridge forming a type line is interrupted, the tracing is made through the interruption. If there is no direct continuation of the ridge, the tracing is continued on an adjacent ridge farther from the interior of the pattern area. If the traced ridge is bifurcated, the tracing is followed on the peripheral branch of the fork.

Patterns of middle and proximal phalanges

Considerably less attention has been paid to the configurations on the middle and proximal phalanges than to the fingertip patterns. Although the presence of definite patterns on the middle and proximal phalanges was long appreciated (Whipple, 1904; Pinkus, 1927), a classification of these patterns was not offered until 1937 by Ploetz-Radmann. She described four basic and eight composite types of configurations (Figure 3.8), which were observed in a series of 200 Germans. All of these patterns have also been identified in a sample of 100 Chinese individuals by King (1939). Basu (1973) recognized several new pattern configurations—a vestigial loop, a double loop whorl, and a vestigial double loop—and there is a likelihood that other, as yet undescribed, patterns occur on the middle and proximal phalanges.

The potential clinical value of the patterns on these phalanges has not been well studied. However, they may be useful in discriminating between monozygotic and dizygotic twins (MacArthur, 1938) as well as in personal identification (Chatterjee, 1959; Kalyanasundaram, 1960; Singh, 1962). Studies in twins and biological families (Ploetz-

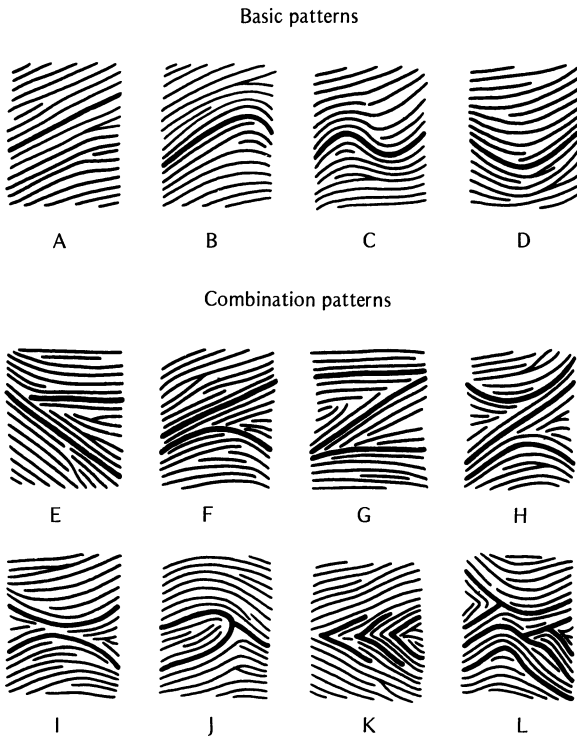


FIGURE 3.8 Types of ridge configurations of middle and proximal phalanges. Basic patterns: A, straight; B, hook; C, wave; D, arch. Combination patterns: E, angle; F, arch and angle; G, double arch; H, double arch and angle; I, double arch; J, closure; K, feather; L, accidental pattern.

According to Ploetz-Radmann, M.: Die Hautleistenmuster der unteren beiden Fingerglieder der menschlichen Hand. *Z. Morphol. Anthropol.*, 36:281, 1937.

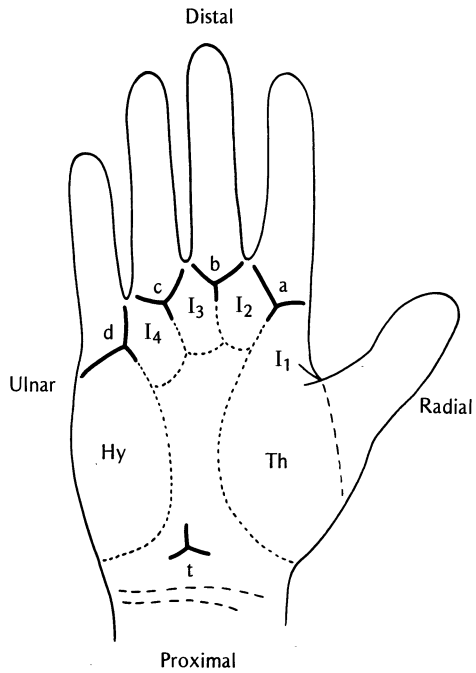
Radmann, 1937; Basu, 1968) showed that the ridge patterns of middle and proximal phalanges were hereditary, with apparently a polygenic mode of inheritance.

PALMS

Palmar pattern configurations

In order to carry out dermatoglyphic analyses that can be compared in different individuals, the palm has been divided into several anatomically defined areas. The areas approximate the sites of em-

FIGURE 3.9 Diagram of a palm showing the dermatoglyphic pattern areas. Th, thenar; Hy, hypothenar; I₁–I₄, first–fourth interdigital areas.



bryonic volar pads and include the thenar area, four interdigital areas, and the hypothenar area (Figure 3.9).

THENAR AND FIRST INTERDIGITAL AREAS. These two areas are closely related anatomically. In dermatoglyphic analyses they are usually considered as one area labeled *thenar/first interdigital* (Th/I₁). In most cases, there is no pattern in the Th/I₁ area but the ridges follow a mild curve around the base of the thumb. Sometimes the simple flow is disturbed by an area of abruptly disarranged ridges, which are oriented at an angle to the general direction of other ridges in the area. They do not form a true pattern. Hence, this configuration is called a *vestige* (Figure 3.10). A vestige or a true pattern can be present in either the thenar or the I₁ area or in each of the areas at the same time. Patterns, when present, are most often loops. Whorls are rarely encountered. A series of thenar, first interdigital, and Th/I₁ patterns is illustrated in Figure 3.11.

SECOND, THIRD, AND FOURTH INTERDIGITAL AREAS. These areas are found in the distal palm in the region of the heads of the metacarpal bones. Each interdigital area is bordered laterally by digital tri-radii. The digital tri-radii are almost always located proximal to the

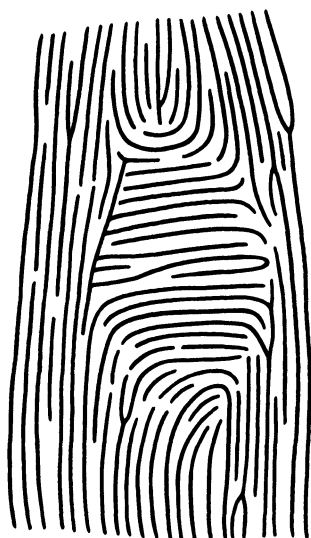
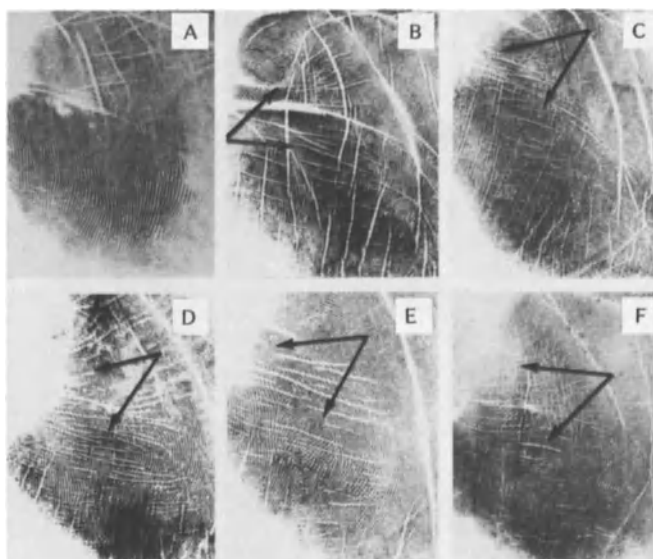


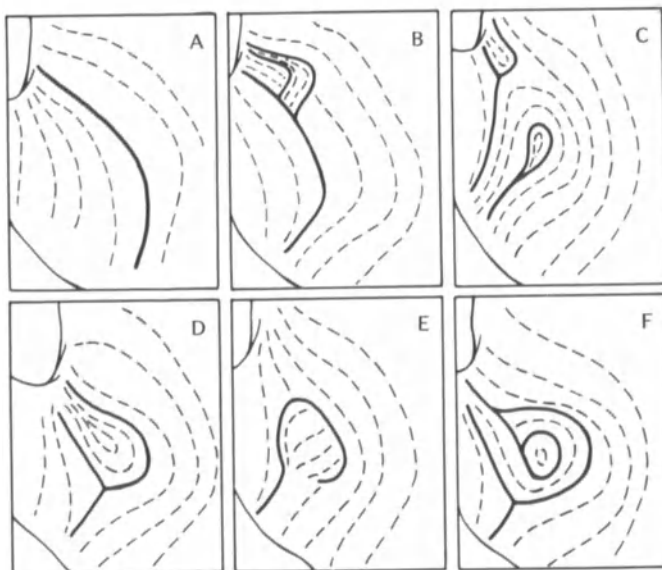
FIGURE 3.10 A common type of vestige in the thenar/first interdigital area.

base of digits II–V (Figure 3.9). Digital triradii are labeled *a*, *b*, *c*, and *d*, starting from the triradius located at the base of digit II and moving toward the triradius associated with digit V. The *second interdigital area* (I_2) lies between triradii *a* and *b*, the *third interdigital area* (I_3) between triradii *b* and *c*, and the *fourth interdigital area* (I_4) between triradii *c* and *d*. If a digital triradius is absent, the mid-point of the base of the corresponding digit can be used to separate the interdigital areas.

Configurations encountered in the interdigital regions are loops, whorls, vestiges, and open fields (Figure 3.12). *Loops* are the most common patterns found in the distal palm. Almost invariably, they open distally (L^d) into the nearest interdigital space (Figure 3.12B). Other exits of the loops (as indicated by the direction of the core) are exceedingly rare so that the interdigital pattern may be referred to as a loop without any additional specification. However, if more detailed notation is desired, the loops can be labeled L^d (distal), L^p (proximal), L^u (ulnar), and L^r (radial). Sometimes, a loop is accompanied by an accessory triradius, which is designated by using the letter *D* or *d* (Figure 3.12C). A loop without an accessory triradius can be labeled *L* or *l* (Figure 3.12B). Capital letters are reserved for large loops, whereas the lower case letters indicate small loops. A small loop has a ridge count of no more than six across its greatest width when counted in the long axis of the loop within the boundary of the main line enclosing the loop.



(a)



(b)

FIGURE 3.11 Types of thenar/first interdigital area patterns: Dermatoglyphic prints (a) and schematic drawings with boldly traced type lines (b). A, open field/open field; B, open field/vestige; C, loop/loop; D, loop/open field; E, vestige/open field; F, whorl/open field.

From Alter, M.: Dermatoglyphic analysis as a diagnostic tool. *Medicine*, 46:35, 1966. Courtesy of The Williams & Wilkins Co., © 1966.

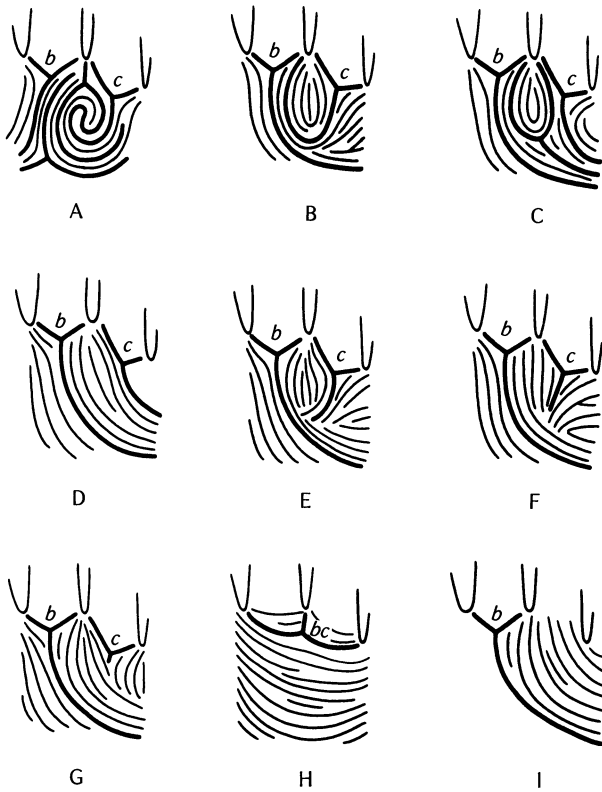


FIGURE 3.12 Diagrams of various ridge arrangements and digital triradii in the interdigital areas. A, whorl; B, loop without an accessory triradius; C, loop with an accessory triradius; D, open field; E, vestige with a tendency toward a loop; F, vestige without a tendency toward a loop, abortive main-line C (symbol X); G, open field, abortive main-line C (symbol x); H, interdigital triradius replacing two usual digital triradii; I, open field, missing digital triradius c.

Whorls (W) are found only rarely in the interdigital areas. When present, they are small and accompanied by accessory triradii.

Vestiges (V) are relatively common interdigital configurations. They do not represent true patterns but consist usually of a series of straight parallel or converging ridges having a direction different from the neighboring ridged areas. Vestiges give an impression of a ridge disarrangement. The vestiges can occur within a true pattern producing, for example, a loop-vestige (l^* or L^*).

Open fields (O) are the most common ridge configurations encountered in the distal palm. These are truly patternless areas,

formed by almost parallel ridges. Some increase in the number of ridges occurs where the interdigital area widens between the triradii. In addition to the more or less uniform increase in ridges usually found, a locally concentrated ridge *multiplication* (M) can occasionally be observed. This configuration is also considered an open field.

Occasionally, two ridge configurations can be present in the same interdigital area, which is then labeled by a double symbol, e.g., L/V. The configuration appearing on the radial side is recorded first. Cummins and Midlo (1961) advocated the use of this dual formulation even in some instances where only a single pattern or vestige was present in this same area, for example, when the pattern or vestige was off center or so small that it did not fill the interdigital area. The symbols used were O/L, V/O, etc.

True patterns are relatively rare in the I_2 area but are common in both I_3 and I_4 areas. Holt (1968) reported a strong negative correlation between patterns in I_3 and I_4 . This means that if a pattern is present in I_3 , I_4 is usually open and vice versa. I_2 and I_3 areas have higher frequencies of patterns on the right palm, whereas I_4 patterns occur more frequently on the left palm.

HYPOTHENAR AREA. True patterns are commonly present in the *hypothenar area* (Hy). The patterns are whorls, loops, and tented arches. Simple arches, open fields, vestiges, and ridge multiplications also occur (Figure 3.13). Sometimes one of the latter configurations coexists with a true pattern. True patterns are essentially of the same character as those encountered on the fingertips. Whorls (W) in the hypothenar area, however, have three triradii instead of two. The small letter *w* is sometimes introduced to describe a small whorl in the hypothenar area that has no more than eight ridges from the core to the nearest triradius. Similarly, a symbol *l* can be used for a loop with a ridge count of no more than eight ridges. Otherwise, a capital L is used to describe the loops. A superscript specifies the direction of opening of the loop, i.e., L^u (ulnar), L^r (radial), L^c (carpal). In the same manner, single and tented arches are labeled A^u , A^r , A^c or T^u , T^r , T^c , respectively, to indicate the direction of their concavity. Arches are the most frequent patterns in the hypothenar areas. Open fields, formed by a series of almost straight ridges, are exceedingly rare in this area and the letter O should be reserved for this configuration only. Usually the ridges curve to some degree forming an arch configuration. If two configurations are present in the hypothenar area,

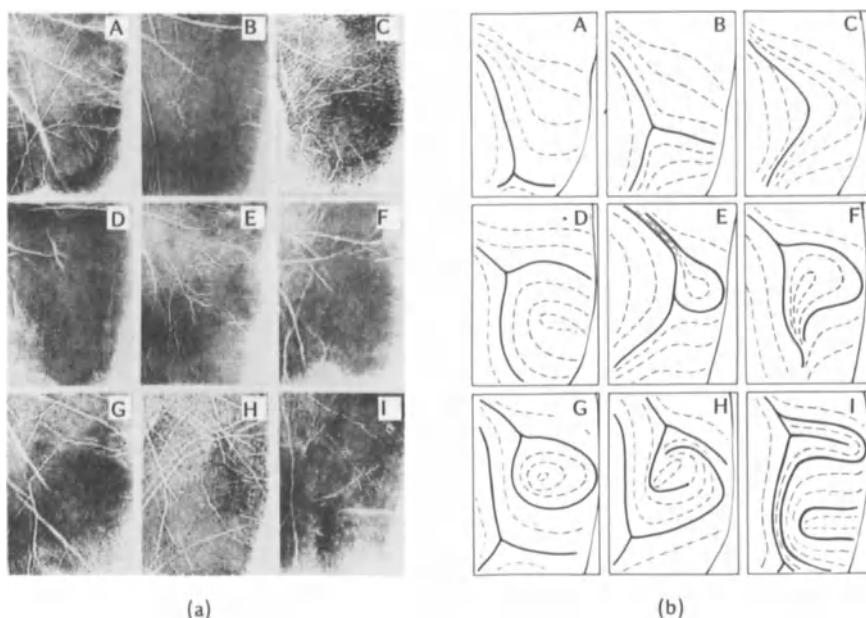


FIGURE 3.13 Some of the types of the hypothenar patterns: Dermatoglyphic prints (a) and schematic drawings (b). A, arch ulnar; B, arch ulnar/arch carpal; C, arch radial; D, loop ulnar; E, loop radial/arch carpal; F, loop carpal; G,H, whorl; I, S-pattern whorl.

From Alter, M.: Dermatoglyphic analysis as a diagnostic tool. *Medicine*, 46:35, 1966. Courtesy of The Williams & Wilkins Co., © 1966.

they can be expressed by a dual formulation, e.g., A^c/L^u , the distal configuration being written first. Various ridge configurations and their combinations, as found in the hypothenar area, are illustrated in Figure 3.14. The figure includes some minor modifications in the nomenclature of the pattern types introduced by Penrose (1968) in comparison with the diagrams originated by Cummins and Midlo (1961). Thus, Cummins and Midlo used the symbol S for a double loop hypothenar pattern designated as W^d in Figure 3.14. The complex pattern, formulated as T^r/L^u in Figure 3.14, was called a Y by Cummins and Midlo, who felt it was not classifiable as one of the other patterns.

The triradius or triradii close to the palmar axis are termed axial triradii (t). Symbols t , t' , and t'' are used to designate the position of these triradii in the proximal-distal direction on the palm. Axial triradii are discussed in detail later.

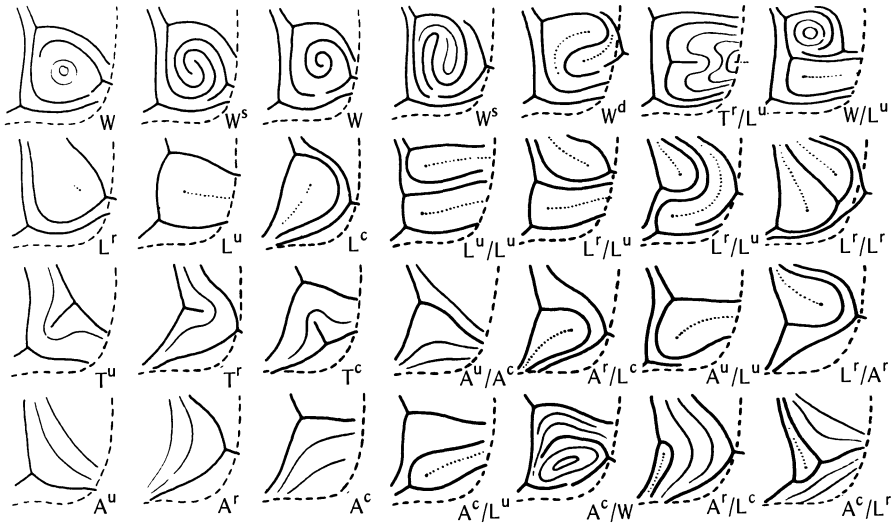


FIGURE 3.14 Various types of patterns in the hypothenar area. First three vertical columns represent primary types and the four right columns, derived types of patterns. W, whorl; L, loop; T, tented arch; A, simple arch; superscripts r, radial; u, ulnar; c, carpal; d, double loop pattern; s, spiral.

From Penrose, L. S.: Memorandum on dermatoglyphic nomenclature. *Birth Defects*, 4(3):1, 1968. Courtesy of the National Foundation–March of Dimes.

Palmar landmarks

The digital and axial triradii and the main line traced from each constitute important landmarks for dermatoglyphic analysis. *Digital triradii* were mentioned earlier in connection with the interdigital areas. Typically, there are four digital triradii in the distal portion of the palm (Figure 3.9). They are found in the metacarpal region at the base of digits II, III, IV, and V. Each triradius is normally associated with one digit. By convention, they are termed *a*, *b*, *c*, and *d*, proceeding in a radioulnar direction. The two distal radiants of each digital triradius run laterally to the nearest interdigital area subtending the digit concerned. The proximal radiant is typically directed toward the center of the palm. Traced along its whole course within the palmar area, it constitutes a palmar *main line*, one of the dermatoglyphic characteristics usually noted in a dermatoglyphic analysis. There are four main lines, each emanating from one of the digital triradii and labeled by capital letters A, B, C, and D (Figure 3.15) corresponding to the triradius having the same lower case letter.

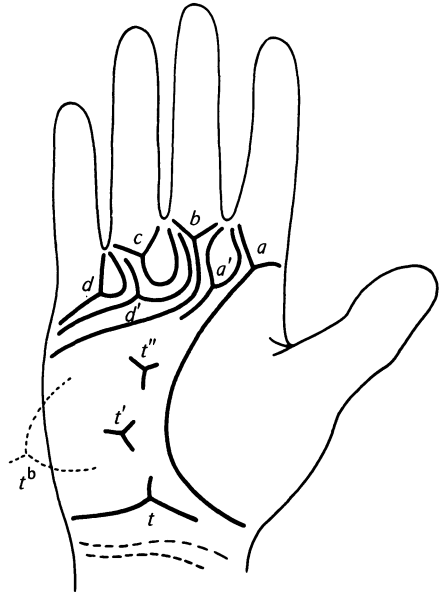
FIGURE 3.15 Palm with traced main lines A, B, C, D, and T and palmar formula 11.9.7.5'.13'-tt''t"-W.L./L.O.L.O.



Not infrequently a number of triradii other than four is found in the distal palm because a triradius may be missing, two triradii may be fused into a single triradius, or there may be an additional (accessory) triradius or triradii in some of the interdigital areas. A missing triradius is usually replaced by gently curving ridges and is almost invariably limited to the area of triradius *c* (Figure 3.12I). Rarely, a digital triradius other than *c* is missing. An *accessory triradius* can be observed sometimes in an interdigital area in connection with an interdigital pattern. The extra triradii are referred to as *a'*, *b'*, *c'*, and *d'*, according to the nearest digital triradius (Figure 3.16). A special case of a missing triradius is an *interdigital triradius*, which may subtend two or more digits. Such a triradius, lying in the center of an interdigital area is labeled in relation to the triradii it replaces, e.g., *bc* for a triradius in the third interdigital area, between the normally formed triradii *b* and *c* (Figure 3.12H). Because interdigital triradii are typically found in zygodactyly, an interdigital triradius is sometimes referred to as a “zygodactylous triradius.” However, because this triradius can also be found on hands without any traces of zygodactyly, the term “interdigital triradius” is preferred as a more accurate as well as more descriptive one.

The terminations of the main lines are assigned numbers distributed along the periphery of the palm in order to convey in-

FIGURE 3.16 Diagram demonstrating the nomenclature of palmar triradii: *a, b, c, d*, digital triradii; *a', d'*, accessory digital triradii; *t, t', t''*, axial triradii; *t^b*, extralimal border (ulnar) triradius.



formation about their course. Altogether, 15 numbers are used. Some of these numbers represent points (triradii *a, b, c, d, t*) and others indicate areas at the palmar margins (Figure 3.17A). The numbering starts in the proximal part of the thenar area and continues along toward the ulnar, distal, and radial borders of the palm. Table 3.1 lists the location and boundaries of each main-line terminus.

The system used by Cummins and Midlo (1961) differs from the one listed in Table 3.1 (according to Penrose, 1968) in that it includes only 14 points and areas and does not separate areas 13' from 13''. Penrose's (1968) system is therefore somewhat more detailed and should be adopted.

Occasionally, the main line is part of a hypothenar pattern and does not exit on the palmar margin but recurves toward the center of the palm. Such a line is assigned the number of the area where it most closely approaches the side of the palm and the letter *h* is attached to the number. This situation is practically limited to the main line that enters the L' or W hypothenar pattern and usually passes close to area 3 or 4. Hence it is called 3*h* or 4*h* (Figure 3.19D).

The terminations of the main lines, recorded in the order D, C, B, A, are used to express the main-line formula. Periods are used to separate the numerical symbols.

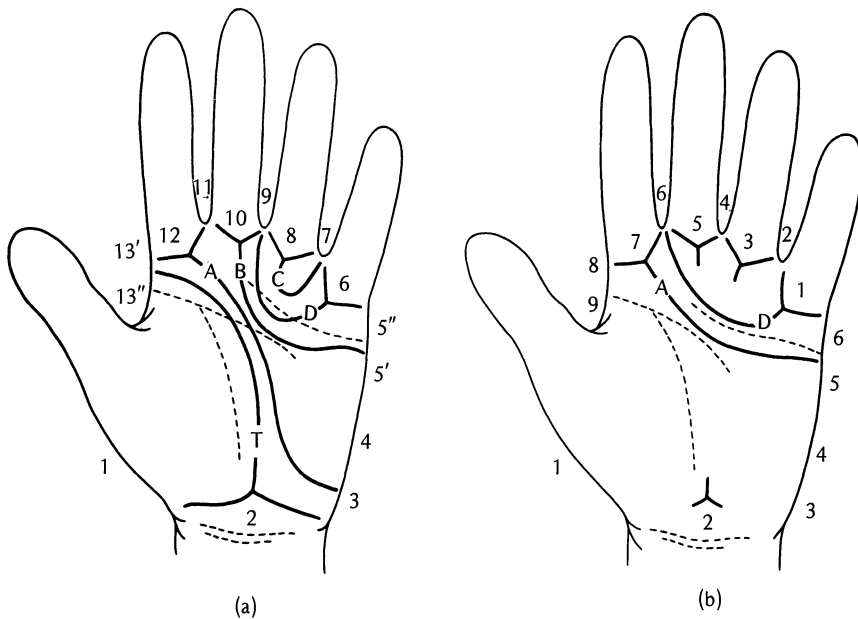


FIGURE 3.17 Numerical values used to designate termini of palmar main lines in the main-line formula (a) and in deriving the main-line index (b).

Sometimes, the termination of the distal radiant of the axial triradius *t*, the main-line T, is added to the main-line formula. If more than one axial triradius is present, the proximal one is used to trace the T line. An example of the main line formula 11.9.7.5'.13' is illustrated in Figure 3.15.

The main-line formula constitutes the first part of the palmar formula. It is followed by the position of the axial triradius or triradii and then by the symbols used for the palmar configuration areas in the following order: hypothenar, thenar/first interdigital area, second, third, and fourth interdigital areas. For example, the palmar formula of the palm in Figure 3.15 is 11.9.7.5'.13'-*t* *t'* *t''* - W.L/L.O.L.O.

The main line is traced in the same manner as the type line on the fingertips. However, certain rules must be observed in order to insure uniformity of the tracing in different dermatoglyphic analyses. The basic rules are as follows: (1) When it is hard to identify the ridge representing the proximal radiant of the triradius, the first ridge on the radial side of the triradius is arbitrarily chosen as the beginning of the main line. (2) If the ridge chosen as the main line

TABLE 3.1. Termini of the palmar main lines^a

NUMBER	AREA OR POINT OF MAIN-LINE TERMINUS
1	Proximal radial border of the thenar area and interval between this and <i>t</i>
2	Triradius <i>t</i>
3	Interval between <i>t</i> and the midpoint of the ulnar border of the hand from the distal wrist crease to the proximal crease of digit V
4	Midpoint between the distal wrist crease and the proximal crease of digit V on the ulnar border
5'	Interval between midpoint of ulnar border and ulnar termination of the distal transverse crease
5''	Interval between the ulnar termination of the distal transverse crease and that of the proximal crease of digit V
6	Triradius <i>d</i>
7	Distal edge of interdigital area IV
8	Triradius <i>c</i>
9	Distal edge of interdigital area III
10	Triradius <i>b</i>
11	Distal edge of interdigital area II
12	Triradius <i>a</i>
13'	Interval between distal edge of interdigital area I and radial termination of the radial longitudinal crease (thumb crease)
13''	Interval on radial border of the palm between the termination of the radial longitudinal crease and the base of the thumb

^a According to Penrose (1968).

is interrupted, the tracing continues on the ridge that lies most directly in line with the ridge originally used. In the absence of such a ridge, the tracing is transferred to an adjoining ridge with the closest linear relationship to the end of the original ridge. If such discrimination cannot be made, the main line is traced along the general course of the ridge flow. Where the ridges are curved, a ridge toward the concavity is used. When the ridges are relatively straight, the ridge toward the ulnar side or toward the distal margin is chosen as the main line. The tracing of the main lines under the above circumstances is illustrated in Figure 3.18. (3) The main line should be traced without anticipating its terminus in order to avoid bias. (4) Abrupt changes in the direction of ridges and imperfect formation of the ridges in the area of the flexion creases cause difficulties in tracing the main lines across these creases. Inadequate printing of the areas within the creases presents a similar problem. Therefore, special

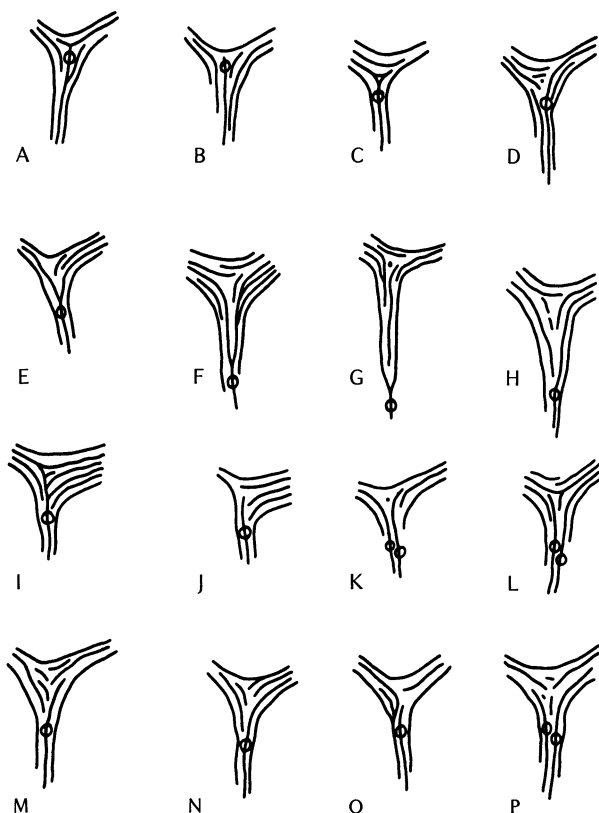


FIGURE 3.18 Diagrams illustrating tracing the main lines from the triradii. The proximal radiant is marked by a circle. Where two circles are present in the diagram, the ridge on the radial side is selected as the proximal radiant.

From Cummins, H., and Midlo, C.: *Finger Prints, Palms and Soles*. New York, Dover Publications, 1961. Courtesy of Mrs. Charles Midlo.

care is required to print the ridged area to avoid any artifactual disruption of ridge continuity. If the main-line ridge is interrupted, it is traced across the crease empirically, onto the ridge that seems to be in closest direct linear relationship with the original ridge.

Certain complications may arise in tracing the main lines. Cummins and Midlo (1961) describe special circumstances that do not conform to the previously discussed rules, offering guidelines for dealing with the following unusual situations:

1. Double formulations. These are of two types: a dual and an alternative formulation. In the *dual formulation* there are actually two terminations of a given main line, which are expressed in the

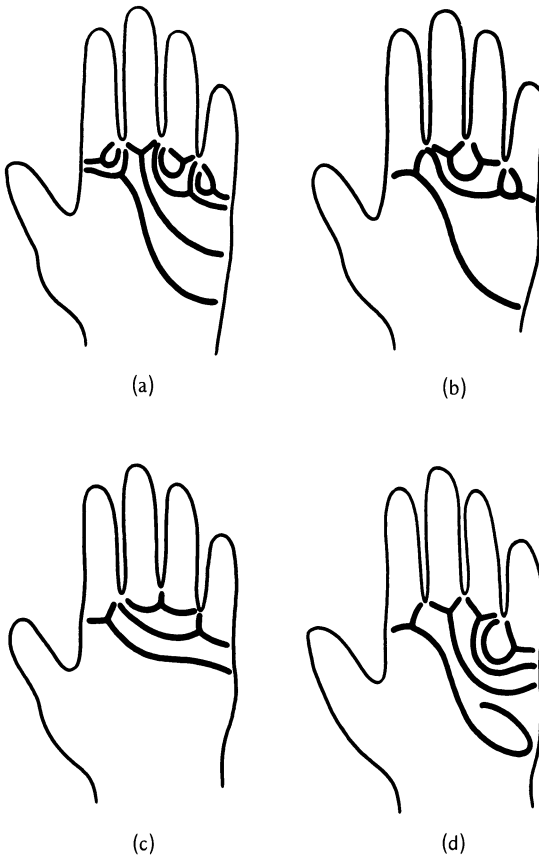


FIGURE 3.19 Special situations in tracing of the main lines.
 (a) Main-lines A and D are traced from triradii *a* and *d* as well as from accessory triradii *a'* and *d'*. Main-line formula is 7-9.9.4.11-3. (b) Dual formulation of main-line D and mutual fusion of main-lines C and B. Main-line formula is 11/7.10.8.3. (c) Interdigital triradius *bc* replaces triradii *b* and *c*. Main-line formula is 11.0*id*0.5'. (d) Main-line A does not exit on the ulnar palmar border but recurves. Main-line formula is 7.5''.5'.3*h*.

main-line formula by two symbols separated by a slant line with the larger number recorded first, e.g., 11/7 (Figure 3.19b). The *alternative formulation*, where the main line termination cannot be clearly determined, is listed in parentheses after the more definite termination, as for example 11(10).

2. Absence of a digital triradius. The absence of a digital triradius and, therefore, of a corresponding main line is noted by the symbol *O*.

3. Abortive main lines. The term “abortive” main line is reserved for situations where only a short segment of the usual main line is present. Sometimes only the triradius is present but the distal radiant is so short that it is almost nonexistent. A symbol for this situation is *x* (Figure 3.12G). More frequently, there is a short distal radiant emerging from the triradial point; however, it is rodlike and ends abruptly in recurving ridges. The configuration gives the impression of a tented arch. This is expressed by the symbol *X* (Figure 3.12F).
4. Mutual fusion of main lines. Occasionally, a main line can be traced into the proximal radiant of another digital triradius. Each of the main lines is then given the number belonging to the triradius into which the appropriate line is directed (Figure 3.19b).
5. Tracing main lines from accessory triradii. The main-line formula represents a descriptive record of ridge courses in the palm. It is therefore important that the formula also include the course of the radiants emanating from the accessory triradii. Graphically, this situation is expressed by noting the radiants of both regular and accessory triradii, separated by a dash, such as 11–7, with the larger number being written first (Figure 3.19a).
6. Interdigital triradii. In the occasional cases of an interdigital triradius replacing two usual digital triradii, a special symbol is used in the main-line formula. For example, if a triradius *bc* is present in the third interdigital area where the individual triradii *b* and *c* are missing, this is reflected in the formula as *OidO* where the two *O*’s represent the missing triradii and the symbol *id* the occurrence of an interdigital triradius (Figure 3.19c).

Apart from the digital triradii, there is another important triradius, the axial triradius (*t*), which is found on practically all normally developed hands. It occurs usually very near the proximal palmar margin, superficial to the wrist bones near the axis of the fourth metacarpal bone. The position of this triradius is subject to considerable variation, particularly in the proximal–distal direction along the axis of the fourth metacarpal bone and, to a lesser degree, in the ulnar–radial direction of the axial triradius. Not infrequently there is more than one axial triradius on the palm. All of them are included in proximal–distal order in the palmar formula, separated by dashes from the preceding main-line formula and the following pattern formula.

Different symbols are used to describe the position of an axial triradius (Figure 3.16). The symbol *t* is reserved for axial triradii found in the proximal region of the palm, near the wrist crease. This

is sometimes referred to as the "normal" or proximal position. A triradius situated near the center of the palm is termed t'' and called distal. The symbol t' expresses the intermediate position of the triradius, i.e., between t and t'' . It is therefore usually labeled an "intermediate triradius" and sometimes, because of its distal shifts from the proximal position, it is referred to as a distal triradius, similar to t'' . An extremely distally displaced triradius, such as is occasionally found distally to the proximal transverse crease, can be termed t''' .

Obviously, the interpretation of the position of the axial triradius is to some extent subjective and therefore not very exact. Because of the frequency with which a displaced axial triradius occurs in medical disorders, an effort to make a more satisfactory discrimination between different positions of axial triradii is desirable and several methods have been proposed. Because no reliable direct method has been found, the position of the axial triradius is generally described indirectly, by measuring the angle formed by lines connecting the axial triradius and the digital triradii a and d (the *atd* angle). Another way used to express the position of an axial triradius more exactly than is possible by means of observation is to measure the axial t distance, i.e., the ratio between the palm's length and the distance between the triradius and the distal wrist crease. Methods of measuring both the *atd* angle and the axial t distance are discussed later.

Lateral displacement of the axial triradius can be expressed by specifying the direction of the displacement. For example, a triradius shifted toward the radial side can be called t^r . More frequently, the observed displacement is toward the ulnar border and is labeled t^u , or in an extreme case, a triradius found in the hypothenar region very near the ulnar edge of the palm, the triradius can be labelled as t^b (border triradius; Penrose, 1968). Such a triradius may even be *extralimital*, i.e., absent in the area of the ridged skin but had the ridges continued they would have met in a triradius (Figure 3.16). Because there is no definite anatomical landmark to allow a distinction between the ulnar and border triradii, all triradii shifted toward the ulnar side of the palm can be termed t^u . These triradii are mostly connected with W, L^r, and A^r patterns in the hypothenar area.

TOES

The types of patterns encountered on the distal phalanges of the toes are essentially the same as those of the fingertips. They are analyzed in a comparable way and the symbols used are identical

to those described for the fingertips. The only difference is in the designation of loops, which are called loop fibular (L') and loop tibial (L'), rather than ulnar and radial as used on the fingers. However, there are differences in the pattern type frequencies between the fingers and toes, the toes showing considerably more arches and fewer whorls than the fingertips in the same group of individuals.

SOLES

Plantar pattern configurations

The pattern areas on the soles are to a large extent analogous to those on the palms (Figure 3.20). However, the *thenar area* on the tibial side of the sole is greatly elongated in comparison with the palmar thenar area, and it is, therefore, divided into a *proximal* and a *distal* section. There are four plantar interdigital areas, labeled

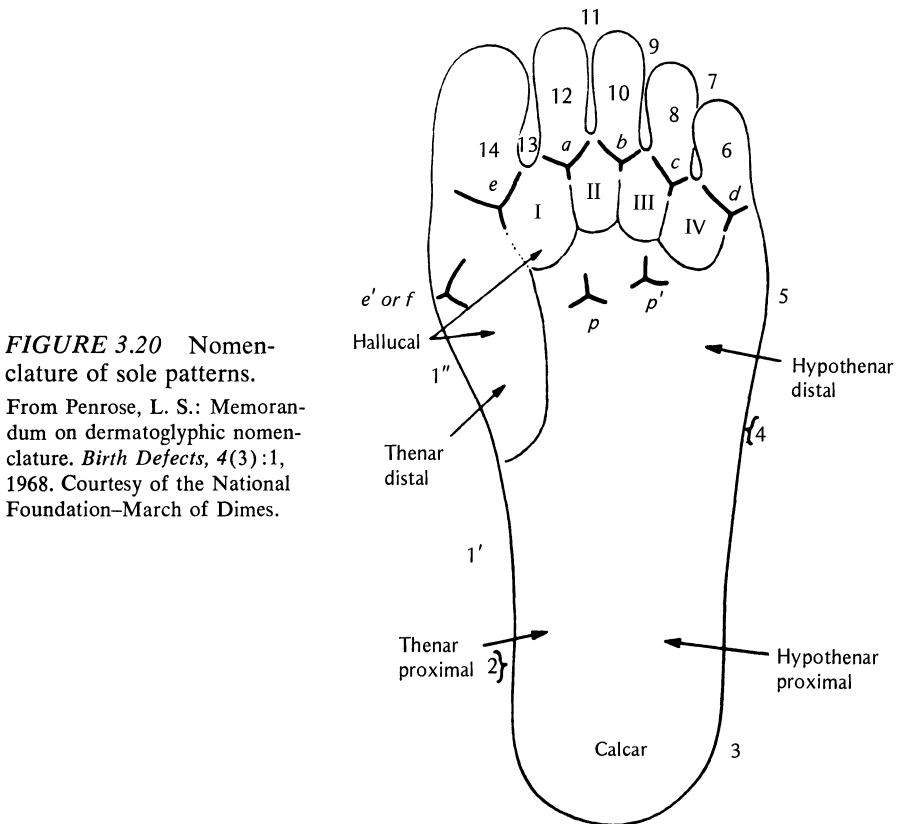


FIGURE 3.20 Nomenclature of sole patterns.
From Penrose, L. S.: Memorandum on dermatoglyphic nomenclature. *Birth Defects*, 4(3):1, 1968. Courtesy of the National Foundation-March of Dimes.

I–IV in a tibiofibular direction. The distal thenar and first interdigital areas are combined and referred to as the *hallucal area*. The long *hypothenar area* (on the fibular side of the sole) is also divided into proximal and distal sections. A region of the sole that does not have an analog in the palm is the *calcar area*, which refers to ridged skin covering the heel.

A special effort must be made to obtain complete sole prints. Because the ridged areas extend well beyond the plantar surface and the toe patterns are often large and sometimes shifted laterally, many printing techniques yield very unsatisfactory results. Minor malformations of the toes caused by wearing shoes represent another obstacle in printing. The difficulties can be easily overcome by employing the method that uses a wide adhesive tape and graphite or other suitable pigments. Each toe print can be taken separately, making sure that its whole pattern area is printed.

Dermatoglyphic configurations encountered on the soles are basically like those on the palms; i.e., they include whorls, loops, arches, and open fields. Superscripts d, p, f, and t are used to identify the distal, proximal, fibular, and tibial directions, respectively, of the patterns. The same rules for designating the pattern openings apply in both the palms and soles. Therefore, only special situations not encountered on the palm are discussed in the following paragraphs.

The *hallucal area* (the distal thenar and the first interdigital areas combined) covers the tibial area of the ball of the foot. Pattern identification is usually not difficult. The only discrepancy encountered is in the terminology used for simple *arches*, which are sometimes termed *open fields* with the symbol O (Cummins and Midlo, 1961). Although simple arches are not true patterns, labeling of hallucal arches as open fields without specification of their direction might obscure important information, particularly in view of recent knowledge concerning the role of hallucal arches in specific medical disorders (such as the arch fibular pattern in trisomy 13). The term “open field” should be reserved for situations where a series of ridges does not curve but follows a more or less straight line. Some of the hallucal patterns are illustrated in Figure 3.21.

Interdigital areas are homologous to those found on the palms. Because the first interdigital area belongs morphologically to the hallucal area, there are usually only three interdigital areas identified on the distal sole, labeled usually II, III, and IV. (The symbol I is used for the first interdigital area when its differentiation from the hallucal area is desirable.) These symbols allow easy distinction

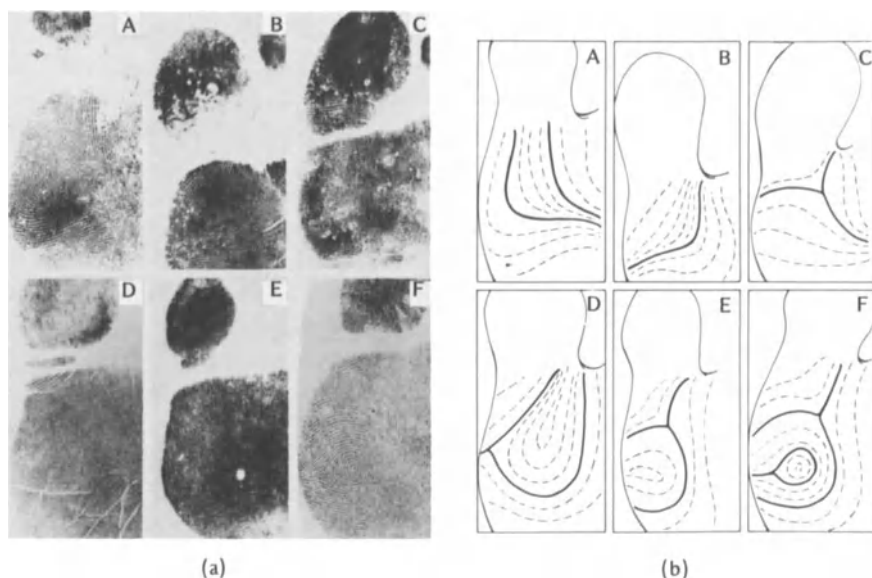


FIGURE 3.21 Some of the types of the hallucal patterns: Dermatoglyphic prints (a) and schematic drawings (b). A, arch fibular; B, arch tibial; C, arch proximal; D, loop distal; E, loop tibial; F, whorl.

From Alter, M.: Dermatoglyphic analysis as a diagnostic tool. *Medicine*, 46:35, 1966. Courtesy of The Williams & Wilkins Co., © 1966.

from the palmar interdigital areas, which are labeled I₁–I₄ throughout this book. Areas II–IV are bordered laterally by plantar digital triradii *a* and *b*, *b* and *c*, and *c* and *d*, respectively.

Configurations found in the interdigital areas are comparable to the homologous palmar patterns. However, the shapes of the plantar patterns are usually more elongated than those on the palms and the patterns are often tortuous. Therefore, the loops are classified according to the initial exit directions of their cores instead of by their final exits, using symbols L^d, L^p, L^t, and L^f. Vestiges (V) and open fields (O), as defined previously, are relatively common.

The hypothenar area covers the fibular side of the sole between the interdigital areas and the heel. It is conventionally divided into a proximal and a distal part. True patterns may be found in both, in either, or in neither of the parts. Ridge configurations are described by dual formulations, with the distal configuration recorded first (e.g., L^t/O). Sometimes, there is a single pattern centered intermediately, which is then expressed by a single formulation in

the plantar formula. A single formulation is also used where there is a lack of patterns in both areas, i.e., when an open field consisting of more or less parallel ridges covers the whole hypothenar area.

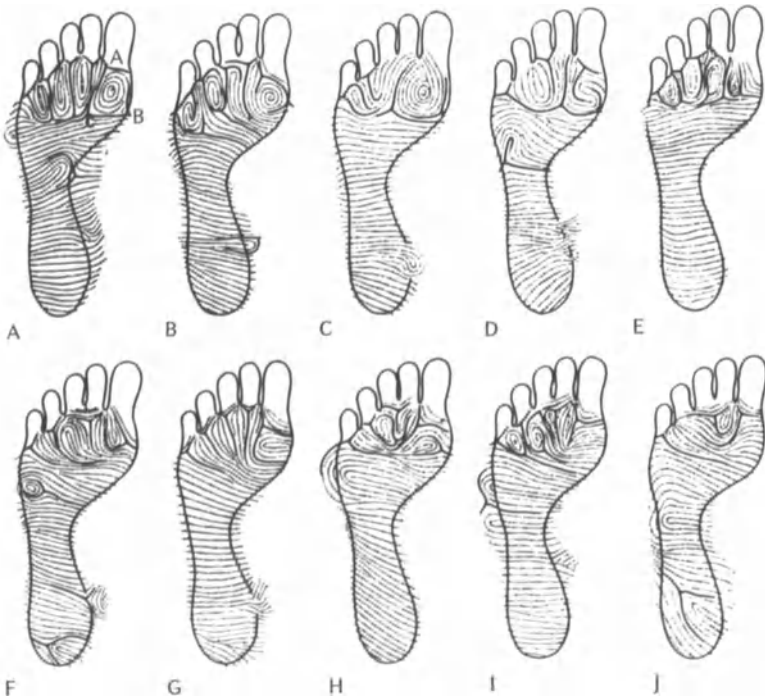
The *proximal thenar area* should not cause classification difficulties. The same symbols as those used on the other areas of the sole are used to describe dermal configurations present in this area.

FIGURE 3.22 Diagrams of ridge arrangements on the soles with plantar formulas:

- | | |
|--|--|
| A, W.L ^d .L ^d .L ^t .L ^t /L ^t .O.V.; | F, L ^d .V.L ^p .O.W/V.L ^t .L ^t ; |
| B, W.L ^p .W.W.V.O.L ^t ; | G, L ^t .O.O.O.O.V.V.; |
| C, W sm .O.L ^d .L ^p .V.O.L ^t ; | H, L ^t .V.W.O.L ^t /O.O.O.; |
| D, W ^{d1} .O.L ^d .+IV.L ^t .O.V/V.; | I, T ^d .L ^p .W.W.L ^t /L ^t .O.V.; |
| E, W ^{cp} .W.W.L ^d .V/O.O.O.; | J, O.L ^d .O.O.L ^t .L ^t .O. |

Wsm, seam; a whorl with an abrupt interruption of the concentric circuit, with a number of ridges ending in a perpendicular relation to a part of the ridge system. Other symbols explained elsewhere in the text.

From Cummins, H., and Midlo, C.: *Finger Prints, Palms and Soles*. New York, Dover Publications, 1961. Courtesy of Mrs. Charles Midlo.



The *calcar area* occupying the heel of the foot is usually patternless (i.e., an open field is present). True patterns rarely occur. When found, they are described by symbols already explained.

There is much less information in the literature on plantar patterns than on palmar configurations. Several factors are responsible for this, e.g., difficulties encountered in obtaining complete prints by most printing methods and a greater inconvenience to subjects in having their soles rather than their palms and fingers printed. Moreover, some investigators have suspected that the foot ridge patterns contain relatively little information of medical significance.

A complete *plantar formula*, an analog to the palmar formula described earlier, is used infrequently. It lists the configurations of all eight plantar areas in the following order: hallucal, interdigital II, interdigital III, interdigital IV, distal hypothenar, proximal hypothenar, calcar, and proximal thenar. Figure 3.22 illustrates examples of plantar formulas.

Plantar landmarks

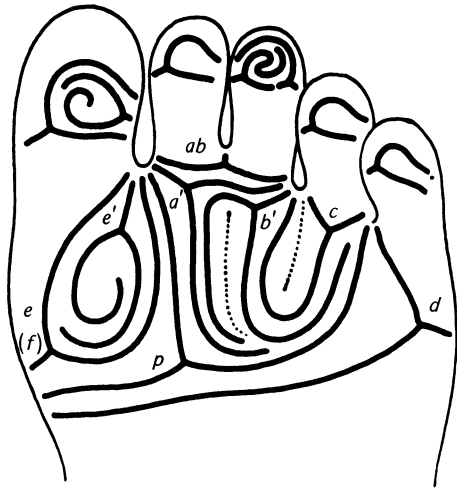
Typically, there are five *digital triradii* in the distal region of the sole. Four of them are comparable to those found in the palms. They are labeled *a*, *b*, *c*, and *d* in the tibiofibular direction, each of them located proximal to each of the digits II–V. The fifth triradius, labeled *e*, located in the vicinity of the base of digit I (great toe), does not have an analog in the palm. Depending on the type of configuration, an additional triradius may be present in the hallucal area. It is labeled *e'* or *f*. Both symbols are still used. In order to make the data comparable, a uniform labeling of the additional triradius must be observed. In the pair *e–e'*, the *e* triradius is the one more closely associated with the great toe. If symbols *e* and *f* are used, *e* occupies a relatively distal position, whereas *f* is near the tibial border. Therefore, triradius *f* may be identical with triradius *e* in some instances and with triradius *e'* in others (Figures 3.20 and 3.23).

Frequently, one or more triradii labeled *p* (proximal) are observed proximal to the hallucal and interdigital regions, near the junction of the hallucal and second interdigital areas. Symbols *p'* and *p''* are used to designate a degree of deviation of the *p* triradius toward the fibular side of the sole (*p''* being the closest to the fibular border).

As on the palms, a plantar digital triradius may be missing or two triradii may be replaced by one interdigital triradius in an intermediate position. Unlike palms, however, plantar interdigital

FIGURE 3.23 Distal plantar area with patterns, nomenclature of triradii, and traced main lines.

From Penrose, L. S.: Memorandum on dermatoglyphic nomenclature. *Birth Defects*, 4(3):1, 1968. Courtesy of the National Foundation–March of Dimes.



triradii are common, mostly represented by the fusion of the *a* and *b* triradii into an *ab* triradius (Figure 3.23).

Main lines can be traced from the proximal radiants of triradii *a*, *b*, *c*, *d*, and *e* according to the rules used for tracing the palmar main lines. The areas and points of the main-line exits and their numerical designations are to a large extent analogous to those of the palms. They are marked in Figure 3.20. This classification (Penrose, 1968) has some minor changes, compared to an earlier classification (Wilder, 1903) in adding number 2 and omitting number 16 (previously assigned to the *p* triradius).

There are several technical problems in tracing main lines in the soles. Apart from printing difficulties, some areas described by the same numerical symbol are too large (because of a lack of usable anatomic landmarks) to allow a precise description of the main-line exit.

Quantitative Analysis

Many dermatoglyphic characteristics can be described quantitatively, e.g., by counting the number of triradii or ridges within a pattern and measuring distances or angles between specified points.

PATTERN INTENSITY

Pattern intensity refers to the complexity of ridge configurations. It can be expressed by counting the number of triradii present. A digit can have pattern intensity 0–3, according to the number of

triradii. The simple arch, which lacks a triradius, is assigned number 0, whereas the tented arch and the loop have intensity 1, as each of them has one triradius. Similarly, the pattern intensity of the palm or sole can be expressed as the sum of all triradii present. Interdigital and extralimal triradii are included in the triradial count.

RIDGE COUNTING

Ridge counting is used to indicate the pattern size. It is primarily utilized on fingertips and toes as a way of expressing the distance between digital triradii or the ridge density in a given area.

Finger and toe ridge counts

The counting is done along a straight line connecting the triradial point to the point of core (Figure 3.24). The ridges containing the point of core and the triradial point are both excluded from the count. Otherwise, every ridge crossing the line is counted, including a ridge that terminates just after crossing the line. However, a ridge terminating just before touching the line is not counted. If the ridge bifurcates before or on meeting the line, two ridges are counted. Interstitial lines are not counted.

Whorls that possess two triradii and at least one point of core allow two different counts to be made, one from each triradius. Each count is made along a line drawn between the triradial point and the nearer point of core. The rule to be observed is that the straight lines used for counting the ridges must not cross one another. They should be chosen so that they both cross the ridged areas as nearly as possible at right angles. The two counts can be specified as *radial* and *ulnar* counts. It must be remembered that a radial loop has an ulnar triradius from which the count is made, whereas an ulnar loop has a radial count. When both counts are given, the first is the radial count and the second is the ulnar count. Patterns with three triradii offer three possible ridge counts. Customarily, ridge counts are recorded after the symbol of a pattern on the appropriate fingertip, either in parentheses after the symbol, e.g., W⁴¹ (17/11), or in the line below the one containing the symbols of the fingertip patterns. Usually, the symbols and ridge counts are recorded in order, beginning with the little finger of the left hand and continuing to the thumb. The digits of the right hand are entered in order starting with the thumb and proceeding toward the little finger. This represents the order in which the fingertip impressions are recorded when

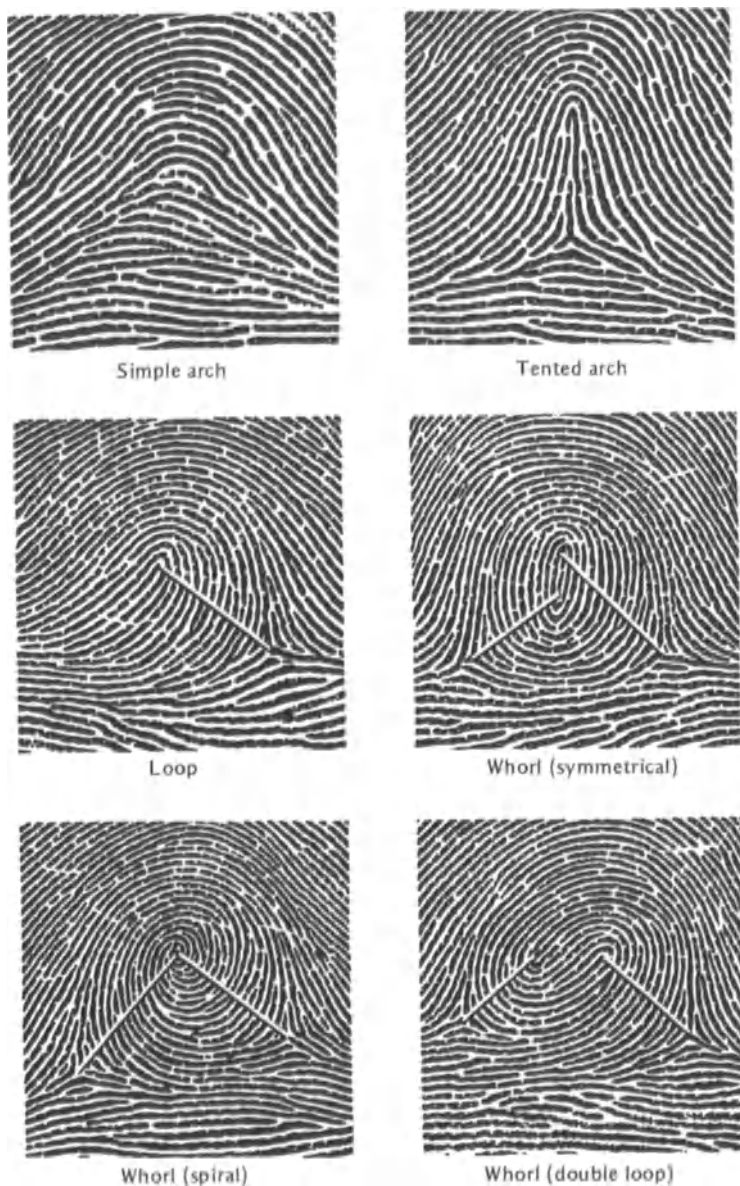


FIGURE 3.24 Ridge counting in various fingertip pattern types. The counting is done along the straight lines connecting the core and the triradius.

From Holt, S. B.: *The Genetics of Dermal Ridges*, 1968. Courtesy of Charles C Thomas, Publisher, Springfield, Illinois.

the popular “ink” methods are employed. Therefore, caution must be exercised when recording the ridge counts of the patterns printed by “lifting” methods, such as those using adhesive tape. In the latter instances, the entries are made in a counterclockwise direction from the left little finger to the right little finger in order to record the patterns as in methods using ink.

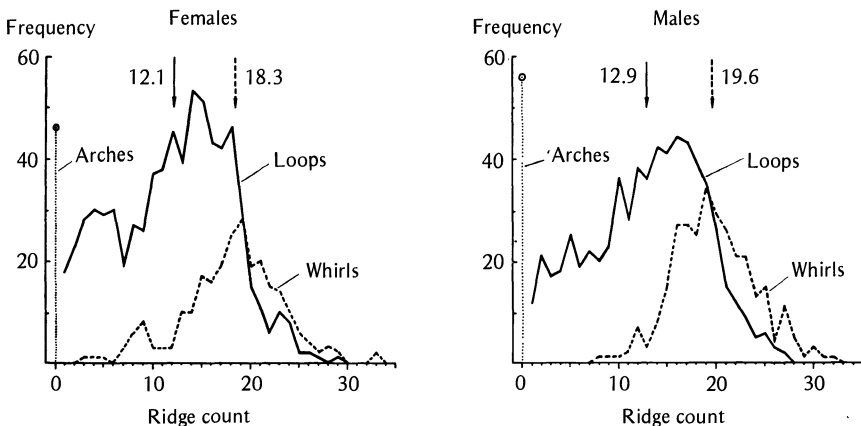
Because the ridge counts are used to express the pattern size, only the largest count is scored in a pattern with more than 1 possible count. Both simple and tented arches have 0 counts.

A *total finger ridge count* (TFRC) represents the sum of the ridge counts of all ten fingers, where only the larger count is used on those digits with more than one ridge count. An *absolute finger ridge count* (AFRC) is the sum of the ridge counts from all the separate triradii on the fingers. The TFRC and the AFRC are of course the same if no whorls are present on the fingertips. The TFRC expresses the size of a pattern, whereas the AFRC reflects the pattern size as well as the pattern intensity, which depends on the pattern type. Using the AFRC, therefore, a small whorl and a large loop may have a similar or equal ridge count.

To some extent, the ridge count reflects the pattern type (Holt, 1961). A ridge count of 0, of course, implies presence of a simple or tented arch. A low ridge count usually corresponds to a small

FIGURE 3.25 The relation between finger ridge count and pattern type. The mean ridge counts for loops and whorls in each sex are indicated by arrows. Data based on 100 unrelated males and 100 unrelated females.

From Holt, S. B.: *The Genetics of Dermal Ridges*, 1968. Courtesy of Charles C Thomas, Publisher, Springfield, Illinois.



loop, although occasionally also to a small whorl. A high count is more likely to indicate a whorl; however, large loops are also frequent. Holt (1961) illustrated the relation between ridge count and pattern type. In her sample, the mean ridge count of loops was considerably lower than that of whorls in both males and females (Figure 3.25).

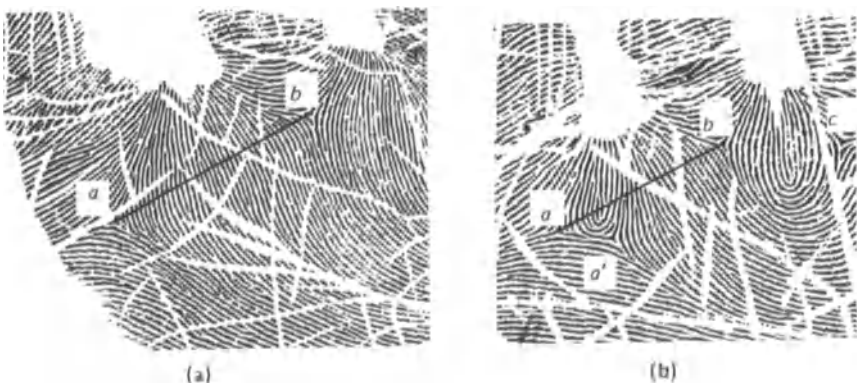
Ridge counts of the digital areas of the palms

Ridges are often counted between two digital triradii. The ridge count most frequently obtained is between triradii *a* and *b* and is referred to as the *a-b* ridge count. Counting is carried out along a straight line connecting both triradial points. The count excludes the ridges forming the triradii. Otherwise, the counting is done according to the same principles as apply in ridge counting on the digits. When an accessory triradius *a'* is present in the second interdigital area, counting is still done from the *a* triradius, which is invariably the more radial of the two (Figure 3.26).

Counting ridges between *b* and *c* and between *c* and *d* triradii is sometimes difficult because of the direction of some ridges forming the interdigital patterns. These ridges may lie almost parallel instead of perpendicular to the line of counting and are therefore not crossed by the line between triradii. The *b-c* and *c-d* ridge counts are rarely used in dermatoglyphic analysis for medical purposes.

FIGURE 3.26 Method of ascertaining the *a-b* ridge count (a) with a single triradius *a*; (b) with an accessory triradius *a'* in addition to triradius *a*.

From Holt, S. B.: *The Genetics of Dermal Ridges*, 1968. Courtesy of Charles C Thomas, Publisher, Springfield, Illinois.



Ridge counting in patterns lacking triradii

Once the point of core is located, counting ridges in technically good prints is quite straightforward. However, very large patterns may extend beyond the limits of the ridged skin areas, and the triradii can be absent. Such triradii are called "extralimal." The site of an extralimal triradius can be approximated from the direction of ridge flow. The resulting ridge count, which is always high, can be only estimated. This approximate count is reported by listing the count number with a question mark or a ~ sign in front of the number.

On fingertips, patterns with extralimal triradii are almost always large whorls and a triradius on either the ulnar or the radial side, or both sides, may be missing. Patterns with extralimal triradii are very rare on the fingertips but are relatively common on the palms and soles.

Estimation of the ridge count on missing or mutilated fingertips

A missing or extensively scarred fingertip with undecipherable ridges presents another problem in ridge counting. In order to salvage the information contained in the TFRC, the ridge count on such a finger can be estimated. Using the high correlations between the corresponding digits of the left and right hands as justification (Holt, 1968), one may simply assign the same value as found on the homologous digit of the other hand to the missing or mutilated fingertip. This is, of course, only a rough approximation.

Smith (cited by Holt, 1968) proposed a more sophisticated and more accurate method for estimating the probable ridge count of the missing fingertip. He considered the values of the other nine available digits and calculated a value for the remaining finger using a regression line based on the mean values of the fingertip ridge counts on each digit of an apparently healthy British population. There are, however, two serious objections to the application of this method in individuals with medical disorders. First, the mean values for the fingertip ridge counts of a British population (Holt, 1968) may not necessarily apply to the population from which the individual with the medical disorder has been drawn or even to other Caucasian populations. Second, a patient with a medical disorder may differ considerably in his ridge count from healthy controls on which the mean values are based. Therefore, a regression line based on normal populations may not be appropriate for individuals with a medical disorder.

POSITION OF THE AXIAL TRIRADIUS

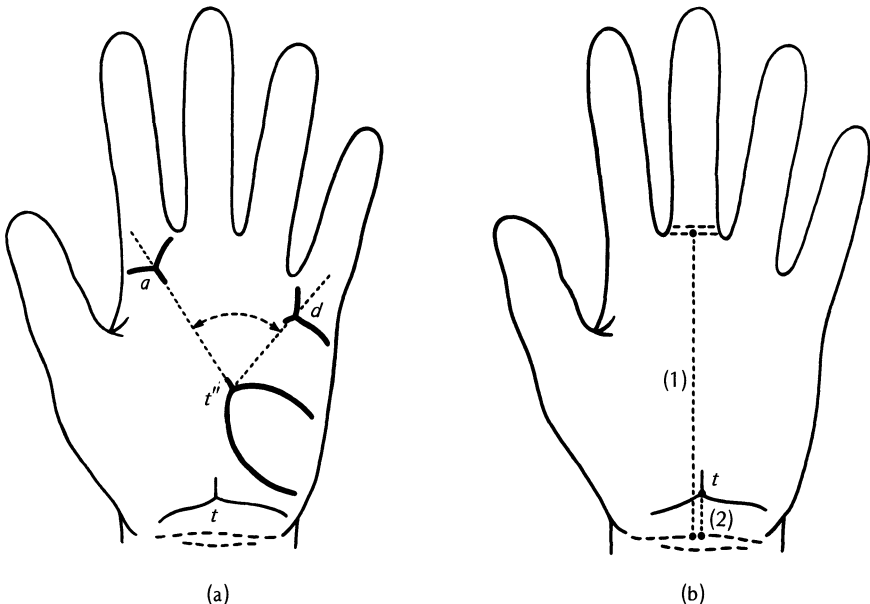
In order to eliminate subjective vagueness in interpreting the position of the t triradii, several quantitative methods have been suggested.

atd angle

Perhaps the most widely used method is based on the *atd* angle. This angle is formed by lines drawn from the digital triradius a to the axial triradius and from this triradius to the digital triradius d . The more distal the position of t , the larger the *atd* angle.

Some palms, especially those with a pattern in the hypothenar area, may have more than one axial triradius. In such cases, it has been customary to record the widest *atd* angle, i.e., the angle emanating from the most distal axial triradius (Figure 3.27a). This is, however, a questionable practice, because it does not allow a distinction between a t triradius actually displaced distally and the presence of additional axial triradii. It has been suggested that terms be introduced to distinguish between a single triradius in a distal position and the presence of multiple triradii wherein at least one is

FIGURE 3.27 Methods of ascertaining the position of the axial triradius on the palm. (a) Measuring the maximal *atd* angle; (b) measuring the distal deviation of the axial triradius.



in a distal position. Wright *et al.* (1972) and Mavalwala (1973) used the terms *proximal medial triradius (pmt)* and *distal medial triradius (dmt)*. In their terminology, the *dmt* applies to the triradius in a distal position, associated with a pattern when a *pmt* is also present. A single axial triradius (which is not a part of a hypothenar pattern) in a distal position is considered as an elevated *pmt* rather than a *dmt*.

In palms with more than one axial triradius, the *atd* angle originating from each axial triradius should be measured.

Sometimes, accessory *a'* or *d'* triradii are present on the palm. Penrose (1949) originally suggested that these triradii be neglected in measuring the *atd* angle. Later (Penrose, 1954–1955), however, he advocated that the most radial *a* triradius and the most ulnar *d* triradius be used as the starting points of the measurement. These triradii provide the widest *atd* angle obtainable on a given palm.

There are several disadvantages to using the *atd* angle as a dermatoglyphic parameter. The most important shortcoming is that the *atd* angle tends to decrease with age because the palm grows more in length than in breadth (Penrose, 1954–1955). This problem can be at least partially overcome by always noting the age of the subject or by introducing an age correction. A second objection is that the size of the angle is affected by the amount of spreading of the fingers when the patterns are printed. The pressure exerted while the palm is printed also can affect the *atd* angle. Printing the palms with the fingers fully adducted may eliminate this problem but tends to yield less satisfactory distal palmar prints with paper printing techniques. In such cases, it is advisable to obtain another set of prints with fingers abducted.

The numerical values of the *atd* angles have been employed in determining the axial triradius positions, i.e., to distinguish between *t* and *t'*, *t'* and *t''*. The limiting values used to represent each position have been determined arbitrarily by different investigators based on their experience. For example, Penrose (1954–1955) suggested that an angle less than 45° be designated as *t*, angles between 45° and 56° as *t'*, and any larger angles as *t''*. Geipel (1961) considered 61° rather than 56° as representing the cutoff point for a *t''* designation. Sánchez Cascos (1965) used an *atd* angle of 71° or more to establish the *t''* position, and Preus and Fraser (1972) advocated angles of over 63° to represent the *t''* position. The discrepancies among experienced investigators are therefore considerable and indicate the need for objective criteria in describing the position of the axial triradius.

Part of the discrepancy concerning conversions of numerical values of the *atd* angle into positional descriptions of the *t* triradius may result from the relationship between the shape of the hand and the size of the *atd* angle. The same angle that in a short, broad palm is associated clearly with a proximal triradius *t* may be associated with an elevated triradius in a long, narrow palm. Measuring the maximal *atd* angle enabled Penrose (1949, 1954–1955) to differentiate Down syndrome, where patients generally have broad and short palms, from phenotypically normal individuals. However, it is inappropriate to assume that a wide *atd* angle is invariably associated with a distal axial triradius and one cannot automatically assign a *t* position on the basis of the *atd* angle.

The *atd* angle does not express the magnitude of any radial or ulnar deviation of the axial triradius. For example, the *atd* angle measured from an ulnar triradius found in the middle of the palmar ulnar margin can yield the same angle as that measured from a proximal axial triradius in its more usual position. Sánchez Cascos (1965) attempted to account for a lateral deviation of the axial triradius by measuring the *adt* angle. He offered the limit of 86° or more for an ulnar triradius, between 76° and 85° for a “middle” triradius, and 75° or less for a radial triradius. However, these numbers are as arbitrary as those proposed for describing the *t* position by measuring the *atd* angle. David (1971) proposed a formula to correct the *atd* angle for any lateral deviation. The correction of the *atd* angle in the long axis of the palm is based on assumptions that the line *a–d* is at right angles to the long axis of the palm and that the long axis of the palm bisects the *a–d* line. The formula is as follows:

$$\tan T = \frac{\sin atd}{2 \sin dat \times \sin adt}$$

where *T* is one half of the corrected *atd* angle. The angles *atd*, *adt*, and *dat* are measured on the palm, using the most proximal *t* triradius present on the palm. Although the formula introduces a certain improvement in designating the position of one *atd* angle, it cannot overcome the variations of the size of the *atd* angle with age and with shape of the palm.

MEASUREMENT OF DISTAL DEVIATION

Another method proposed for determining the position of the axial triradius (Walker, 1957) uses the ratio between the length of the palm and the length of the distance between the wrist crease

and the axial triradius. The vertical distance between the most distal wrist crease and the most proximal crease of the third digit is measured (Figure 3.27b). The distance between the axial triradius and the distal wrist crease is also measured and expressed as a percentage of the length of the palm. According to Walker, 14.9 percent or less corresponds to the axial triradius in the t position, between 15 and 39.9 percent to the t' , and 40 or more percent to t'' . Penrose (1968) introduced a change in this method, advocating the use of the most proximal crease of the fourth digit instead of the third and using 14 percent or less as a limit for the t triradius. Because the usual position of the axial triradius is considerably closer to the vertical axis drawn from the fourth rather than the third digit and the wrist crease, Penrose's method is preferred. When the triradius is deviated laterally from the axis, its position is determined by a line drawn perpendicular to the axis. Measurement of the percentage distance is less age dependent than the atd angle and it is not influenced by the shape of the palm or by variation of the position of landmarks, such as the a and d triradii. It therefore offers more information about the position of the axial triradius than does the atd angle. However, the method does not reflect the lateral deviation of the axial triradius. Moreover, the wrist and metacarpophalangeal creases do not constitute precise points from which to measure longitudinal distance.

Sharma (1962, 1963) proposed a modification of Walker's method, leading to what he called the " t index." He drew the longitudinal axis of the palm print between the most distal wrist crease and the most proximal metacarpophalangeal crease on the digit in vertical alignment with the proximal axial triradius. The longitudinal axis was then divided into six equal parts. An axial triradius lying in the first, second, third, or fourth segment would then be designated t , t' , t'' , or t''' , respectively. A triradius on the line between segments was given the more distal designation. The method does not really represent an improvement over Walker's method and it is not widely used.

Ridge counting

Ridge counting between the triradii d and t has been proposed as yet another means of describing the position of the axial triradius. Penrose (1968) specified the most distal hypothenar triradius as the point from which the count is made. David (1971) mentioned the most medial triradius d as the other limit. The resulting number is smaller when the t is distally placed than when it is in a proximal position. The advantage of this method is that the $d-t$ ridge count is

constant in an individual and independent of age. However, there are numerous disadvantages in this method. Apart from being rather tedious and time consuming, the method gives somewhat inaccurate results because the line along which the counting is done crosses the distal and sometimes even the proximal transverse flexion creases, where the ridges tend to be interrupted. Presence of a hypothenar pattern causes another difficulty because the line of counting may cross the ridges at angles far from a right angle and the same ridge may be crossed more than once. Also, the ridge breadth is not constant so that the $d-t$ ridge count reflects ridge breadth as well as the position of t . This method is rarely used to record the position of the t triradius.

Breadth ratio

Breadth ratio is another method occasionally used to determine the t position. It is based on a measurement of the perpendicular distance from t to a line drawn between the a and d triradii. This measurement is expressed as a ratio of the $a-d$ distance. Although it allows for age variation, it does not allow for variations in the palm breadth or for the displacement of the digital triradii.

The position of the axial triradius has been considered to be of great importance and has been used as a valuable dermatoglyphic trait in individuals with various medical disorders. In view of the above-discussed objections to each of the methods employed for determining the position of the axial triradius, it seems possible that misinterpretations have been introduced. Therefore, the use of the most distal triradius, without discrimination as to the presence or absence of the proximal t triradius, is a serious omission that may invalidate many reports of an abnormal atd angle in association with various disorders. These characteristics may account for some part of the significant differences found in the position of the axial t between "normal" and "abnormal" individuals.

In order to evaluate the t position as a diagnostic trait, uniform methods for designating the position of t must be used. It is advisable to express the position of the axial triradius using several designations: t , t' , t'' , based on subjective estimates; the width of the atd angle; and the ratio of axial t distance to total length of the palm along an axial line.

MAIN-LINE INDEX

The main-line formula, as previously described, serves as an indication of the general direction of palmar ridge flow. Cummins (1936) observed that the termini of two of the main lines, A and

D, alone may adequately reflect the ridge direction. From this observation he proposed a *main-line index* based on the sum of the two numbers corresponding to the exits of main lines A and D. The numerical values assigned to these exits are distributed in the same way as those used for the main-line formula exits except that the numbers 1–8 substitute for main-line formula numbers 6–13 (or number 8 for 13' and number 9 for 13'') (Figure 3.17b). Exit 5'' is scored as 6 in the main-line index. The resulting value gives an estimate of palmar ridge transversality that may be of clinical significance. A low value for the index indicates vertical ridge alignment, whereas a high value reflects a tendency for the palmar ridge direction to be horizontal. The main-line index is recorded for each palm separately.

Dermatoglyphic Topology

Single dermatoglyphic traits can be readily determined and used to compare the two hands, different individuals, and various ethnic groups, as well as healthy individuals and individuals with medical disorders. However, the complex of configurations present on the whole surface of the palms or soles is much more difficult to use in comparisons, particularly because of the great variability of the pattern details and the difficulties in reducing them into statistically acceptable units. Penrose (1965) suggested a topological approach to this problem. Topological classification (Penrose and Loesch, 1969, 1970a) is based on a description of all loops and the enumeration of all the triradii. Each whorl is rated as the equivalent of two loops, and arches, vestiges, and other ridge configurations, which are not considered as true patterns, are neglected. Extralimital ones are included in counting the triradii. The resulting pattern intensity, as measured by the number of loops on the normal palm or sole, is four less than the number of triradii (Penrose, 1965). All loops and triradii, with the exception of digital triradii, are recorded in a formula. The authors of topological dermatoglyphic classification offered a "dermatoglyphic dictionary" of 131 palmar (Penrose and Loesch, 1970a) and 196 plantar (Penrose and Loesch, 1969) formulas based on a sample of normal males and females of European origin (250 individuals of each sex for the palms and 150 of each sex for the soles). From this "dictionary," the frequencies of individual dermatoglyphic features and of their combinations may be obtained and used as control data in clinical and genetic studies.

Because of the limited number of individuals whose dermatoglyphics form the dictionary, some normal but less common combinations of dermatoglyphic patterns are not included in the list. Moreover, the frequencies of dermatoglyphic configurations, as they appear in the dictionary, are based on individuals whose specific place of European origin was not indicated and therefore the formulas may not be representative of ethnic groups from all regions.

TOPOLOGICAL CLASSIFICATION OF PALMAR DERMATOGLYPHICS

For classification purposes, the palm is divided into five configurational areas, more or less equivalent to those used in traditional dermatoglyphic analysis. The thenar and first interdigital areas are considered as one area, labeled I. Areas II and IV are somewhat larger (Figure 3.28) than their analogs in previously described classifications (Figure 3.9). Area V, the hypothenar area, is labeled H.

Loops are classified as *peripheral* and *central*, instead of distal and proximal. The cores of peripheral loops (labeled I, II, III, IV, and H) point away from the center of the palm, whereas the cores of central loops (labeled $\hat{I}$, $\hat{II}$, $\hat{IV}$, and $\hat{H}$) point toward the center of the palm (Figure 3.29a). There are three loops that cannot

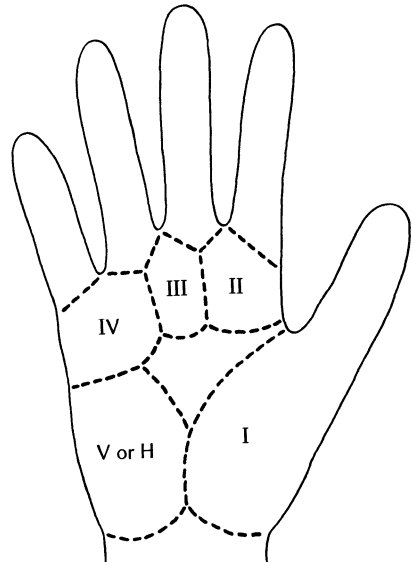


FIGURE 3.28 Configurational areas on the palm as applied in topological nomenclature.

According to Penrose, L. S., and Loesch, D.: Topological classification of palmar dermatoglyphics. *J. Ment. Defic. Res.*, 14:111, 1970.

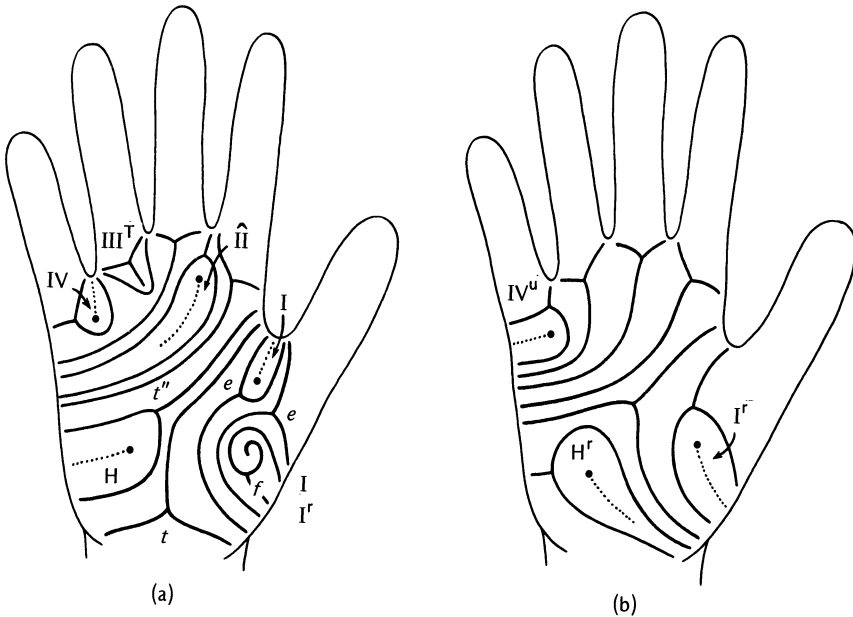


FIGURE 3.29 Examples of topological nomenclature. (a) Palm with a central loop ($\hat{I}$), peripheral loops (I, IV, H), tented loop (III^T), and a radial loop (I^r). Topological formula of the palm is $I I I^r \hat{I} III^T IV H e e f t t'' 4 (4)$; (b) Palm with loops requiring separate classification (IV^u , H^r and I^r).

Adapted from Penrose, L. S., and Loesch, D.: Topological classification of palmar dermatoglyphics. *J. Ment. Defic. Res.*, 14:111, 1970.

be fitted easily into the above classifications and have to be described specifically. They are (1) a peripheral loop in the fourth interdigital area, the core of which is on the ulnar border of the palm (IV^u); (2) a loop in the hypothenar area with its direction and exit toward the radial side of the wrist (H^r); and (3) a peripheral loop in area I, exiting on the radial (thenar) border (I^r). All three of these loops are illustrated in Figure 3.29b.

The term *tented loop* was used by Penrose and Loesch (1970a) to describe ridge configurations in the hypothenar area, which are considered as *tented arches* (T^c , T^r , T^u) in traditional dermatoglyphic classification (Cummins and Midlo, 1961). Similarly, interdigital configurations formed by a flow of ridges around an abortive main line, which traditionally are not regarded as true patterns, are termed “tented loops” in the topological classification (Figure 3.29a). The superscript T (e.g., III^T) is used for these interdigital tented “loops” usually only when it is not possible to

assign a direction to the loop core. Otherwise, the pattern is simply classified according to the configurational area in which it is found. Symbols used in topological classification of the loops are listed in Table 3.2.

Palmar triradii are also part of the proposed formula. Digital triradii are not specified separately but their number is noted at the end of the formula. However, the presence of interdigital (zygodactylous) triradii is recorded. The symbol *z* is used to describe the interdigital triradius *ab* (fused digital triradii *a* and *b*), and *z'* and *z''* substitute for the symbols *bc* and *cd*, respectively. All axial triradii are recorded, using the usual symbols *t*, *t'*, *t''*, *t'''*. A distinction is made between a border triradius (*t^b*), which either is found near the ulnar border of the palm or is extralimal, and an ulnar triradius (*t^u*), situated near the center of the hypothenar area.

TABLE 3.2. Symbols used in topological classification of loops on the palms and soles^a

SYMBOL	AREA	PALM	SOLE
I, II, III, IV	Interdigital	Peripheral	Distal
$\hat{I}$, $\hat{II}$, $\hat{III}$, $\hat{IV}$	Interdigital	Central	Proximal
I^r	First interdigital	Radial	—
$\hat{I}$	Hallucal or thenar distal	—	Proximal (or tibial)
I^l	Hallucal or thenar distal	—	Fibular
II^T , III^T , IV^T	Interdigital	Peripheral tented	—
IV^u	Fourth interdigital	Peripheral ulnar	—
V	Hypothenar distal or central	—	Fibular
$\hat{V}$	Hypothenar distal	—	Tibial
H	Hypothenar	Peripheral	—
$\hat{H}$	Hypothenar	Central	—
H^r	Hypothenar	Radial	—
T^c	Hypothenar	Tented, core pointing carpally	—
T^r	Hypothenar	Tented, core pointing radially	—
T^u	Hypothenar	Tented, core pointing towards ulnar side of palm	—

^a According to Penrose and Loesch (1969, 1970a).

CHAPTER 3: DERMATOGLYPHIC PATTERN CONFIGURATIONS

TABLE 3.3. Symbols used in topological classification of triradii on the palms and soles^a

SYMBOL	PALM	SOLE
<i>e</i>	Triradius in distal part of area I Triradius in central or proximal part of area I	Triradius close to the base of the hallux but can deviate as far as the center of the hallucal area, to which it is always distal Triradius close to the thenar distal border of the sole but can deviate as far as the center of the hallucal area
<i>t, t', t'', t'''</i>	Axial triradii as traditionally described	—
<i>t^b</i>	Border triradius (including extralimital triradii)	—
<i>t^r</i>	Triradius deviated to radial side of palm	—
<i>t^u</i>	Triradius near the center of hypothenar area	—
<i>g</i>	—	Triradius on the fibular side of the thenar distal area, often proximal to the D line; i.e., its position is too tibial and too proximal to be satisfactorily described as <i>p</i>
<i>h</i>	—	Triradius proximal to <i>d</i> in the hypothenar distal area
<i>p</i>	—	Triradius situated proximally between areas I and II, roughly on the axis of digit II
<i>p'</i>	—	Proximal triradius between areas II and III, roughly on the axis of digit III
<i>p''</i>	—	Proximal triradius between areas III and IV, roughly on the axis of digit IV
<i>z</i>	Interdigital (zygodactylous) triradius—short form for fused <i>ab</i>	Same as the palmar symbol
<i>z'</i>	Interdigital (zygodactylous) triradius—short form for fused <i>bc</i>	Same as the palmar symbol
<i>z''</i>	Interdigital (zygodactylous) triradius—short form for fused <i>cd</i>	Same as the palmar symbol

^a According to Penrose and Loesch (1969, 1970a).

The symbol t' is reserved for the rare axial triradius that is shifted radially from its usual position on the palm.

New symbols are introduced for triradii that are part of the patterns in area I. These are triradii e , which lie in the distal half of the area and f , found in the proximal part of the area (Figure 3.29a). Symbols of all triradii used in topological classification are listed in Table 3.3.

The formula of an analyzed palm is written in the following order:

1. Loops according to the numerical order of the areas (i.e., I, II, III, IV, H); peripheral loops are recorded before central and separately specified loops.
2. Triradii in alphabetical order ($a-d$), followed by the number of unspecified interdigital triradii.
3. At the end of the formula, if so desired, the exit of main-line A, which has some diagnostic significance, can be added in parentheses.

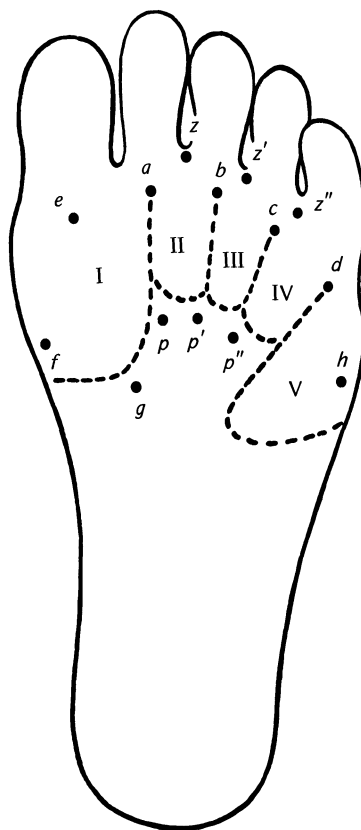
For example, Figure 3.29a shows a drawing of a palm with formula $I I I' \hat{I} \hat{I} III^t IV H e e f t t'' 4 (4)$. Because only dermatoglyphic elements present on the palm are recorded, a formula of a palm may be as brief as $t4$ in case of a palm free of any patterns and having only an axial triradius in the usual, proximal position and four digital triradii, none of which is zygodactylous.

TOPOLOGICAL CLASSIFICATION OF PLANTAR DERMATOGLYPHICS

Classification of the dermatoglyphics on the sole is based on the same principles as the above-described classification of palmar dermatoglyphics. Symbols applied to the appropriate loops and triradii are listed in Tables 3.2 and 3.3, respectively. Only the patterns in the distal part of the sole are analyzed and recorded (Figure 3.30). The interdigital regions are labeled I, II, III, IV, and V, with V reserved for the hypothenar distal area. Loops are specified according to the general directions of their cores into distal (I, II, III, IV) and proximal ($\hat{I}$, $\hat{II}$, $\hat{III}$, $\hat{IV}$), or, in some cases, fibular (I'). In the hallucal area, three types of loops are recognized (I, $\hat{I}$, and I'). $\hat{I}$ is used as a term equivalent to I^t (loop tibial). In the area V, V^t is equivalent to V and V^t to $\hat{V}$.

FIGURE 3.30 Configurational areas and triradii on the sole as applied in topological classification.

According to Penrose, L. S., and Loesch, D.: Dermatoglyphic sole patterns: a new attempt at classification. *Hum. Biol.*, 41:427, 1969.



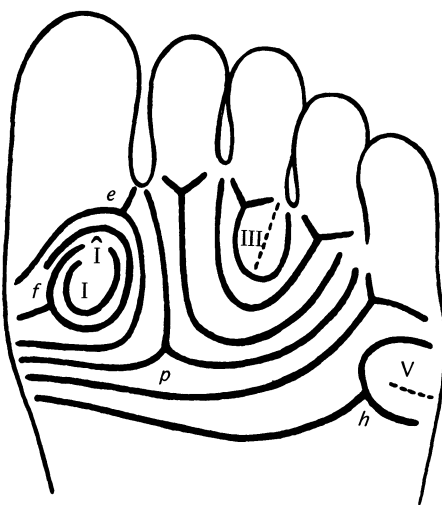
As on the palms, the digital triradii, including accessory triradii, are not separately listed unless they are zygodactylous. The triradii near the tibial border of the sole are labeled *e* and *f*; those close to the fibular edge of the sole are named *h*; and centrally placed, proximal triradii are named *g* or *p* (Figure 3.30).

The formula is written in the same manner as the palmar formula, i.e., (1) loops in numerical order of the areas, with distal preceding proximal and fibular loops within a given area; (2) triradii in alphabetical order, followed by the number of unspecified digital triradii. Figure 3.31 illustrates a sole with the formula $I \hat{I} III V e f h p 4$.

Penrose and Loesch (1969, 1970a,b) applied their topological classification to samples of patients with various chromosomal aberrations in order to demonstrate the usefulness of their method in diagnosis. The results of their studies are discussed in Chapter 6, dealing with dermatoglyphic features in specific medical disorders.

FIGURE 3.31 Distal part of a sole with topological classification $\hat{\Pi}$ III V $e f h p 4$.

Adapted from Penrose, L. S., and Loesch, D.: Dermatoglyphic sole patterns: a new attempt at classification. *Hum. Biol.*, 41:427 1969.



Frequency of Dermatoglyphic Traits in Normal Populations

BILATERAL SYMMETRY

Similar to other parts of the human body, the palmar and plantar epidermal ridge patterns of the same individual are never exactly identical, although there are tendencies toward symmetry between the homologous left and right areas. The differences between the left and right side involve the pattern types, ridge counts, and ridge breadth.

On the fingertips, ridges are usually finer and quantitative values tend to be lower on the left hand. Statistically, whorls and radial loops tend to be more frequent on the fingertips of the right hand whereas arches and ulnar loops are found more often on the left hand (Table 3.4). Although the fingertip patterns of the hands of an individual are not nearly mirror images, there is a strong trend to share the same pattern type on the homologous fingertips of the same person. Between 74 and 82 percent of the homologous pairs of fingers in population samples of various ethnic groups have been found to bear the same fingertip pattern type (Henckel, 1933; Dankmeijer, 1938; Table 3.5). About 20–30 percent of individuals exhibit complete symmetry of the pattern types on their fingertips. The frequencies of numbers of pairs of homologous fingers with the identical fingertip patterns per individual are shown in Table 3.6.

Analysis of the symmetry of pattern types reveals an uneven dis-

TABLE 3.4. Percentage frequencies of the major pattern types on the fingertips of 400 phenotypically normal North American individuals

	DIGIT	ARCH		RADIAL LOOP		ULNAR LOOP		WHORL	
		LEFT	RIGHT	LEFT	RIGHT	LEFT	RIGHT	LEFT	RIGHT
Males (<i>n</i> = 200)	I	5.5	3.0	0.0	0.5	65.0	50.5	29.5	46.0
	II	15.5	17.5	18.5	24.0	36.0	27.0	30.0	31.5
	III	12.5	7.5	1.5	1.5	68.0	71.0	18.0	20.0
	IV	2.0	2.0	3.0	1.5	64.0	47.0	31.0	49.5
	V	0.0	1.0	0.0	0.0	87.0	83.0	13.0	16.0
	Total	6.7		5.0		59.9		28.4	
Females (<i>n</i> = 200)	I	7.5	4.5	0.0	0.0	63.5	63.0	29.0	32.5
	II	21.0	16.0	16.5	15.5	38.0	37.0	24.5	31.5
	III	16.5	10.0	2.0	0.5	66.0	76.0	15.5	13.5
	IV	4.5	4.0	1.0	1.5	65.5	58.0	29.0	36.5
	V	5.5	3.0	0.0	1.0	81.5	81.0	13.0	15.0
	Total	9.2		3.8		63.0		24.0	

tribution of identical patterns on individual digits, with a consistent trend toward the highest degree of symmetry on the fifth digit and the lowest on the second digit (Table 3.5). The differences in frequencies of identical patterns on single digits may be at least partially explained by the varying frequency of pattern types on these fingers (Cummins and Midlo, 1961; Table 3.7), for it is known that

TABLE 3.5. Pairs of homologous fingers with an identical fingertip pattern type bilaterally

DIGIT	MALES (<i>n</i> = 200)		FEMALES (<i>n</i> = 200)		TOTAL (<i>n</i> = 400)	
	NUMBER OF PAIRS	PERCENT	NUMBER OF PAIRS	PERCENT	NUMBER OF PAIRS	PERCENT
I	147	73.5	155	77.5	302	75.5
II	101	50.5	117	58.5	218	54.4
III	148	74.0	157	78.5	305	76.2
IV	145	72.5	154	77.0	299	74.8
V	176	88.0	176	88.0	352	88.0
Total	717	71.7	759	75.9	1476	73.8

TABLE 3.6. Percentage of homologous right and left finger pairs with an identical pattern type

NUMBER OF PAIRS						POPULATION	SOURCE
0	1	2	3	4	5		
0.1	1.1	7.5	22.5	40.4	28.5	Dutch, Javanese	Henckel (1933)
0.0	2.2	9.2	28.1	38.3	22.2	North American Caucasian	Schaumann and Alter (unpublished)

TABLE 3.7. Order of decreasing frequency of various fingertip pattern types on individual fingers

PATTERN TYPE	FINGER					POPULATION
Whorl	I	IV	II	III	V	British ^a
	IV	I	II	III	V	German ^b
	IV =	I	II	III	V	Dutch ^c
	IV	I	II	III	V	North American Caucasians ^d
	IV	I	II	III	V	Japanese ^e
	I	IV	II	V	III	Javanese ^c
	I	IV	II	V	III	Pygmies ^c
	I	IV	II	III	V	Liberian Negroes ^c
Loop	V	III	IV	I	II	British ^a
	V	III	II	I	IV	German ^b
	V	III	IV	I	II	Dutch ^c
	V	III	I	IV	II	North American Caucasians ^d
	V	III	II	I	IV	Japanese ^e
	III	V	II	IV	I	Javanese ^c
	V	IV	III	II	I	Pygmies ^c
	V	III	IV	II	I	Liberian Negroes ^c
Arch	II	III	I	IV	V	British ^a
	II	III	I	IV	V	German ^b
	II	III	I	IV	V	Dutch ^c
	II	III	I	IV	V	North American Caucasians ^d
	II	III	I	IV	V	Japanese ^e
	II	III	I	IV	V	Javanese ^c
	II	III	I	V	IV	Pygmies ^c
	II	I	III	IV	V	Liberian Negroes ^c

^a Cummins and Midlo (1943).

^b Siegle (1951).

^c Dankmeijer (1938).

^d Schaumann and Alter (unpublished).

^e Hasebe, cited by Martin and Saller (1962).

each pattern type exhibits a different level of bilateral symmetry, highest for loops and lowest for arches (Dankmeijer, 1938).

On palms, the patterns in hypothenar, second, and third interdigital areas are more frequent on right hands, whereas thenar and fourth interdigital areas show patterns more often on left hands (Table 3.8).

Palmar main lines show certain variation between the left and right palms. The general ridge direction, as indicated by the courses of main lines, tends to be more transverse on the right. For example, line A on the right terminates more often in the distal part of the ulnar margin and line D tends to exit more radially than the homologous lines on the left. Main-line C is more frequently abortive or absent on the left palms (Table 3.9) and line T terminates in a position closer to the thumb on the right palms.

The sole patterns (Table 3.10) exhibit more symmetry than the palmar patterns. Left hallual and second interdigital areas tend to have slightly higher pattern intensities than the right ones, whereas in other areas the reversed situation or approximately equal frequencies of patterns are observed.

Arches and whorls occur more frequently on the left toes than on the right. Whereas arches also show a left-sided preponderance on the fingers, whorls on the fingers have a right-sided preponderance. Tibial loops are more commonly found on the left toes, whereas the homologous radial loops on the fingers occur more often on the right.

TABLE 3.8. Percentage frequencies of the palmar patterns^a of 400 phenotypically normal North American individuals

	MALES (<i>n</i> = 200)			FEMALES (<i>n</i> = 200)		
	LEFT	RIGHT	TOTAL	LEFT	RIGHT	TOTAL
Thenar/I ₁						
Thenar only	5.5	4.0	4.8	8.0	3.0	5.5
I ₁ only	2.5	0.5	1.5	2.5	0.0	1.2
Both thenar and I ₁	2.0	0.5	1.2	0.0	0.0	0.0
I ₂	1.5	4.5	3.0	1.0	3.0	2.0
I ₃	30.5	50.5	40.5	22.5	47.0	34.8
I ₄	50.5	44.5	47.5	47.5	41.0	44.2
Hypothenar	35.0	37.5	36.2	32.5	36.0	34.2

^a Only true patterns considered; open fields and vestiges omitted.

TABLE 3.9. Percentage frequencies of various terminations of the palmar main lines in phenotypically normal North Americans

MAIN LINE	TERMINATION ^{a, b}	MALES (<i>n</i> = 200)			FEMALES (<i>n</i> = 200)		
		LEFT	RIGHT	TOTAL	LEFT	RIGHT	TOTAL
A	1	5.4	2.4	3.9	5.0	1.0	3.0
	2	7.4	2.4	4.9	7.4	1.5	4.4
	3	57.4	40.9	49.0	55.4	42.0	48.6
	4	15.4	25.2	20.4	15.8	23.4	19.7
	5'	12.9	22.4	17.7	14.9	26.3	20.6
	5''	0.5	1.9	1.2	0.5	3.4	2.0
	11	1.0	4.8	2.9	1.0	2.4	1.7
B	3	1.0	0.5	0.8	0.0	0.0	0.0
	5'	21.5	5.0	13.2	17.5	6.5	12.0
	5''	44.0	35.0	39.5	40.0	30.5	35.2
	7	33.5	56.0	44.8	42.5	61.0	51.8
	8	0.0	1.0	0.5	0.0	0.0	0.0
	9	0.0	2.5	1.2	0.0	2.0	1.0
C	5'	1.0	0.0	0.5	0.0	0.0	0.0
	5''	13.5	8.5	11.0	18.0	6.0	12.0
	6	1.0	0.5	0.8	1.0	1.5	1.2
	7	31.0	24.0	27.5	28.5	28.0	28.2
	8	4.0	2.0	3.0	2.0	3.0	2.5
	9	29.0	50.0	39.5	26.0	45.0	35.5
	10	0.0	0.5	0.2	0.0	0.0	0.0
	11	0.0	3.0	1.5	0.0	1.5	0.8
	X	12.0	6.5	9.2	12.5	7.0	9.8
D	O	8.5	5.0	6.8	12.0	8.0	10.0
	5''	0.0	0.0	0.0	0.5	0.0	0.2
	7	19.5	10.5	15.0	26.0	13.0	19.5
	8	1.0	1.5	1.2	0.5	2.5	1.5
	9	47.0	28.5	37.8	36.0	27.5	31.8
	10	4.0	3.0	3.5	0.5	0.5	0.5
	11	28.5	56.5	42.5	36.5	56.5	46.5

^a Only those termination points are listed in which the main lines were actually found to exit. For explanation of numbers designating the termination areas see Figure 3.17a.

^b X, abortive main line; O, missing main line.

TABLE 3.10. Percentage frequencies of the pattern types on the great toes and hallucal areas of phenotypically normal North Americans

AREA	PATTERN ^a	MALES (<i>n</i> = 200)			FEMALES (<i>n</i> = 200)		
		LEFT	RIGHT	TOTAL	LEFT	RIGHT	TOTAL
Great toe	A	8.0	6.0	7.0	8.0	7.0	7.5
	L ^f	72.5	81.5	77.0	79.5	84.0	81.8
	L ^t	7.0	4.0	5.5	4.5	3.0	3.7
	W	12.5	8.5	10.5	8.0	6.0	7.0
Hallucal area	A	7.0	6.5	6.8	8.5	10.0	9.2
	L ^d	47.0	54.0	50.5	52.5	60.0	56.3
	L ^t	13.5	9.5	11.5	8.0	8.0	8.0
	W	32.5	30.0	31.2	31.0	22.0	26.5

^a A, arch; L^f, loop fibular; L^t, loop tibial; L^d, loop distal; W, whorl.

SEX DIFFERENCES IN DERMATOGLYPHICS

Certain differences exist in both the qualitative and the quantitative dermatoglyphic traits between males and females. Females have narrower ridges than males. This tendency is correlated to some extent with the smaller size of the female hand. However, even in males and females with the same hand sizes, females tend to have a smaller ridge breadth (Cummins and Midlo, 1961).

On the fingertips, females have generally lower frequencies of whorls and radial loops but a higher number of arches (Table 3.4). This tendency toward simpler patterns in females is practically universal in population samples of all ethnic groups. Pattern type frequencies on the toes exhibit a trend similar to those on the fingers.

TABLE 3.11. Mean values of total finger ridge count, *a-b* ridge count, and *atd* angle in phenotypically normal North Americans

	MALES (<i>n</i> = 200) MEAN ± S.D.	FEMALES (<i>n</i> = 200) MEAN ± S.D.
TFRC	142.9 ± 49.6	120.4 ± 52.3
Summed <i>atd</i> angle		
Proximal	81.2 ± 9.5	83.4 ± 12.2
Distal	85.3 ± 13.5	87.6 ± 16.9
Summed <i>a-b</i> ridge count	84.0 ± 9.8	81.4 ± 10.6

TABLE 3.12. The percentage ranges and means of the fingertip pattern frequencies in different racial groups^a

ETHNIC GROUP	N ^b	FINGERTIP PATTERN TYPE							
		WHORLS		ULNAR LOOPS		RADIAL LOOPS		ARCHES	
		RANGE	MEAN	RANGE	MEAN	RANGE	MEAN	RANGE	MEAN
Caucasians	112	26-49	35.4	50-66	55.6	4-7	4.3	2-9	4.3
Negroes	88	15-42	27.4	53-74	61.4	1-4	2.6	2-18	8.8
American Indians	76	32-58	42.6	37-58	49.4	1-5	3.1	1-9	5.0
Oriental	55	36-56	46.7	39-57	48.1	1-4	3.0	1-5	1.8
Australasians	60	33-78	52.7	22-64	44.9	0-3	1.1	0-4	1.4
Asian Indians	7	33-56	42.6	33-59	51.8	1-4	2.2	1-7	3.4

^a According to Plato (1973).

^b Number of population samples summarized in the table.

TABLE 3.13. The percentage ranges and means of the palmar pattern frequencies in different racial groups^a

ETHNIC GROUP	N ^b	PALMAR PATTERN AREA											
		HYPOTHENAR		THENAR/ I_1		I_2		I_3		I_4		MEAN	MEAN
		RANGE	MEAN	RANGE	MEAN	RANGE	MEAN	RANGE	MEAN	RANGE	MEAN		
Caucasians	43	21-52	34.5	6-20	10.1	0-11	4.7	25-58	43.3	37-68	54.4		
Negroes	32	15-32	24.9	9-42	19.3	4-29	13.8	25-58	39.0	68-91	81.4		
American Indians	46	5-28	13.8	11-55	33.2	0-5	1.4	8-56	25.2	43-80	66.8		
Oriental	17	9-30	20.1	5-12	9.9	1-5	2.5	7-24	16.3	55-84	69.5		
Australasians	44	10-47	23.4	10-48	22.6	0-15	5.3	8-45	25.8	55-89	71.2		
Asian Indians	7	15-35	26.6	3-21	11.6	2-6	4.0	51-53	52.4	49-61	55.2		

^a According to Plato (1973).

^b Number of population samples summarized in the table.

Palmar patterns exhibit some sex differences within comparable groups of individuals. Most frequently, females have higher pattern frequencies in the hypothenar and fourth interdigital areas but lower pattern frequencies in thenar/first interdigital and second and third interdigital areas. The trends, however, are far from universal for all populations. Table 3.8 lists the palmar pattern frequencies of a sample of phenotypically normal North American individuals.

The total finger ridge count, which is influenced by the number and type of sex chromosomes, is lower in females than in males (Table 3.11).

RACIAL DIFFERENCES IN DERMATOGLYPHICS

Different ethnic groups exhibit highly significant variations in the frequency of dermatoglyphic configurations. The differences are not limited to the major racial groups but extend to relatively small ethnic groups and even small population isolates. Tables 3.12 and 3.13 illustrate the high variability of pattern type frequencies within and between racial groups of apparently healthy samples. Therefore, it is important in any study of dermatoglyphics to use an appropriate, ethnically matched control group.

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4 Congenital Malformations of Dermatoglyphics

Anomalies of the ridged skin are occasionally encountered on the volar aspects of human hands and feet. They may result from destruction of dermal patterns caused by trauma, such as burns, mutilations, or skin diseases, or from inborn aberrations of the papillary ridge patterns. The latter are of special interest because they may be hereditary and some are typical of specific medical disorders.

Congenitally imperfect ridge formation is a result of disturbances in embryologic development during the period of ridge differentiation. Abel (1936) attributed the defects to some irregularities in tension and pressure within the epidermal tissue occurring between the second and fourth fetal months. Hirsch and Schweichel (1973) suggested that pattern anomalies could be secondary to nerve growth disturbances: aplasia caused by failure of nerves to grow into the epithelium, dysplasia caused by both quantitative and qualitative deviations of the subepithelial nerve branches from the norm, and ridge distortion might be attributable to a disturbance in the spatial arrangement of nerves. Because the factors causing pattern disturbances can only influence the areas of epidermis where ridges have not yet differentiated, the period of embryogenesis during which the disruptive factors have been active can be deduced from the type of damage in the dermatoglyphic patterns. Areas of aberrant ridge formation can be of variable extent, from minute patches to the entire surface of ridged skin of the fingers, palms, toes, and soles.

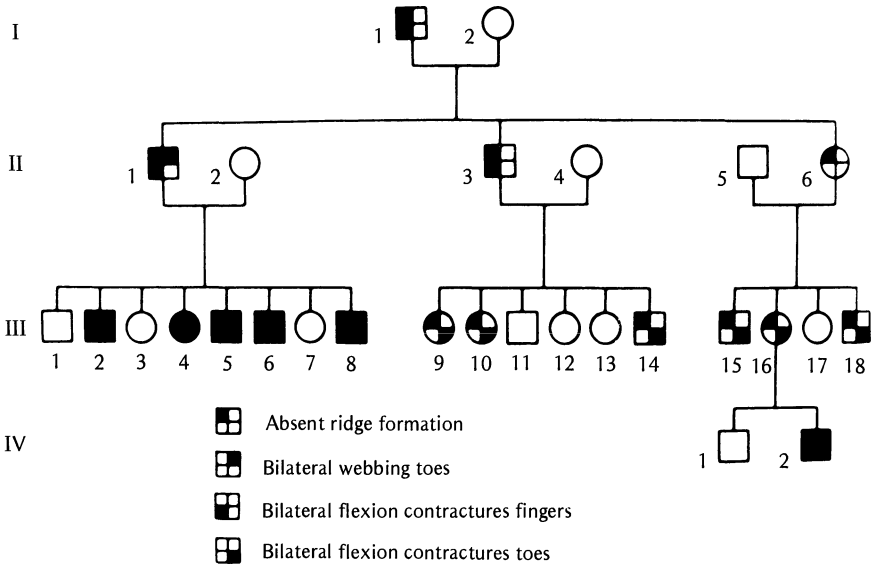
Considerable differences exist in the appearance of the anomalous

epidermal surface. The epidermal ridges are either lacking completely or appear as dots and short segments, sometimes distributed in a haphazard manner so that no coherent dermatoglyphic pattern can be distinguished. These disturbances represent several different entities. There is, however, no generally accepted terminology that allows a clear distinction between the various epidermal ridge disorders. The following classification of the congenital malformations of dermatoglyphics is therefore suggested: (1) ridge aplasia, (2) ridge hypoplasia, (3) ridge dissociation, and (4) ridges-off-the-end.

Ridge Aplasia

Ridge aplasia is a very rare congenital malformation of epidermal ridges characterized by an absence of the ridges over the entire volar surfaces of the hands and feet. Baird (1964, 1968) described an American kindred in which 16 of 28 members in four generations showed a complete absence of dermatoglyphics (Figure 4.1). Since then, another child in the fourth generation has been

FIGURE 4.1 Pedigree of a family with congenital ridge aplasia.
From Baird, H. W.: Absence of fingertips in four generations. *Lancet*, 2:1250, 1968. Courtesy of *Lancet*.

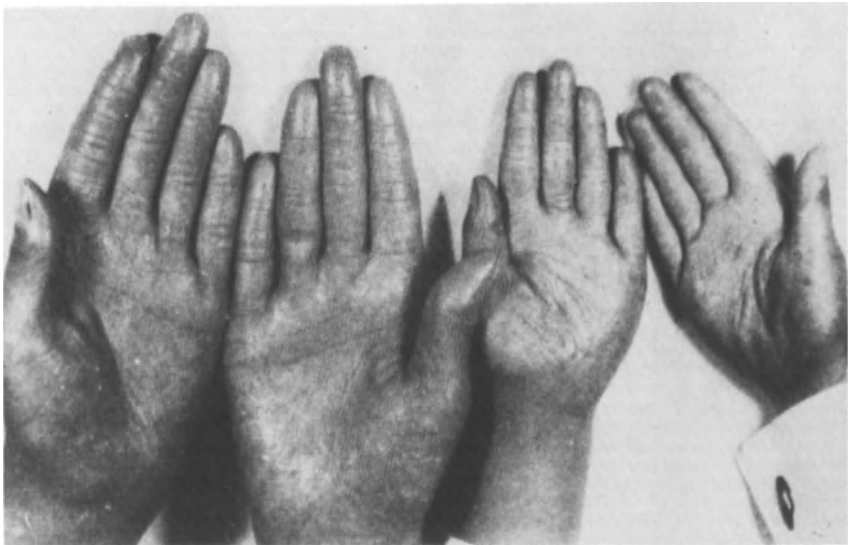


born with absent epidermal ridge patterns (Baird, personal communication 1975). The defect was apparently part of an autosomal dominantly inherited syndrome. Affected members of the family had transient congenital milia and some also had bilateral flexion contractures of the fingers and toes. The volar skin of affected infants was smooth, thin, and red, similar in appearance to the dorsal hand surface of normal individuals except for the absence of hair. Later, calluses developed on the palms and soles, making the skin thick and hard. Sweating in the affected areas was virtually absent. Although the skin biopsy revealed normal sweat glands and sweat ducts in the dermis, the ducts did not penetrate the epidermis and no sweat pore openings were found on the skin surface. The skin of both hands and feet was covered by an excessive number of fine creases. No other ectodermal defects were present in any members of the kindred.

Similar findings were reported by Basan (1965) in eight of 17 individuals in three generations of one kindred. Members of the family affected by ectodermal dysplasia had missing palmar and

FIGURE 4.2 Volar aspects of the hands of a mother and her 4-year-old son showing stretched skin lacking epidermal ridge patterns.

From Basan, M.: Ektodermale Dysplasie. *Arch. Klin. Exp. Dermatol.*, 222:546, 1965. Courtesy of J. F. Bergmann Verlagsbuchhandlung.



plantar epidermal patterns, abnormal nails, and simian creases. The volar skin was described as dry, rough, stretched, parchmentlike, and covered with numerous creases (Figures 4.2 and 4.3). Two of the affected individuals showed a tendency toward formation of epidermal ridges that consisted of short segments (ridge dissociation).

Rott (1971) described two additional patients with ridge aplasia. Both individuals had medical disorders. The first suffered from progeria and the other had hydrocephalus, cleft palate, a suspected heart defect, stenosis of the tear ducts, absent nipples, nail anomalies, and syndactyly of the fingers and the toes. The latter patient exhibited barely discernible epidermal ridges on the fingertips and soles.

David (1973a) observed yet another patient with ridge aplasia

FIGURE 4.3 Dermatoglyphic prints of the hand in proband (a) and his mother (b) with ridge aplasia. Note the high number of secondary palmar creases and a single transverse palmar crease on the mother's palm.

From Basan, M.: Ektodermale Dysplasie. *Arch. Klin. Exp. Dermatol.*, 222:546, 1965. Courtesy of J. F. Bergmann Verlagsbuchhandlung.



and severe dermatologic problems at sites other than the palms and soles. Earlier, Ludy (1944) presented a patient with absent fingerprints who suffered from hyperkeratosis. His two sons, a daughter, and a granddaughter also lacked dermatoglyphic patterns, although they did not have hyperkeratosis. Because no documentation or more detailed descriptions of the epidermal defect has been offered, it is impossible to judge whether the anomaly in this kindred indeed represents a ridge aplasia or a different type of congenital malformation of the ridges, such as a severe ridge dissociation.

Presence of both dissociated and missing ridges on the fingertips of an individual with a severe hand malformation was reported by Cooke (1955). Three of the proband's digits were patternless but identifiable ridge segments could be seen. The remaining fingers, which were grossly reduced in size, had very smooth skin in place of normally ridged epidermis.

Several other individuals with "absent fingerprints" have been reported but the subjects in fact invariably possessed dissociated or hypoplastic rather than absent ridges.

Ridge Hypoplasia

Ridge hypoplasia is a condition in which the epidermal ridges are reduced in height, giving a "worn-off" appearance. The areas of hypoplastic ridges are usually covered with an excessive number of fine secondary creases (white lines) which further obscure the patterns and make dermatoglyphic analysis extremely difficult. As noted by David (1973a), *congenital ridge hypoplasia* is impossible to distinguish from *acquired ridge atrophy*. The latter condition is sometimes observed in very old age, where it is caused by generalized thinning of the skin. Ridge atrophy was also found in the majority of a series of adult patients with celiac disease (David *et al.*, 1970). The atrophy was noted to improve as the general condition of the patients improved. Atrophy of ridges also follows interruption of the nerve supplying the ridged area. Verbov (1970) reported ridge flattening as a common finding in hypohidrotic ectodermal dysplasia. Cummins (1967) mentioned a marked decrease of ridge height as a result of certain occupations, such as those connected with handling of rough objects or immersion of hands in liquids for long periods. These changes are readily reversible after contact with the disturbing materials is discontinued.

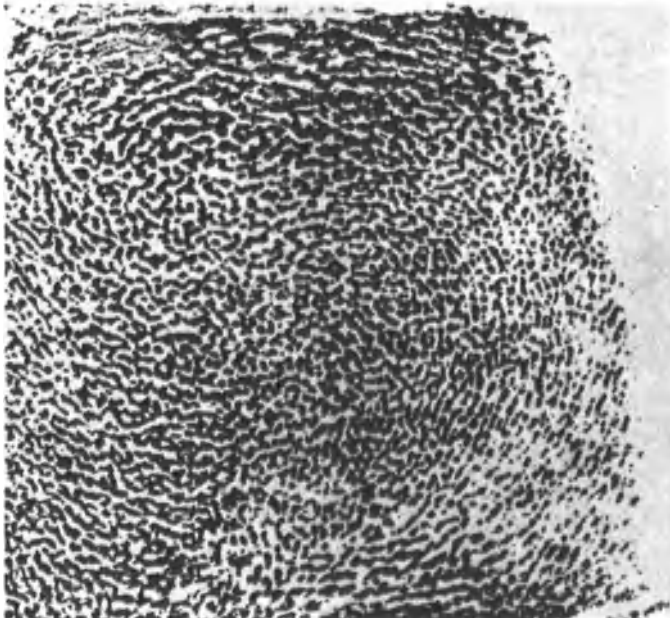
Ridge Dissociation

Ridge dissociation refers to a heterogeneous group of ridge dysplasias. In phenotypically more severe dissociations, the ridges are broken into short and often curved segments that cover the pattern areas in a chaotic, disorganized way (Figure 4.4) instead of forming smooth, more or less parallel, continuous lines. In less pronounced dissociation, the patterns remain clearly identifiable but the ridges that form them are broken into very short, usually dotlike segments (Figure 4.5). Such dissociations are often referred to as “dotted ridges,” “strings of pearls,” etc.

Dissociated ridges are rare among phenotypically normal individuals but relatively frequent in certain medical disorders. They can be found sporadically or inherited as an autosomal dominant trait (Abel, 1944; Cooke, 1962; Cummins, 1970a, David, 1973a, Dodinval, 1972; Dodinval *et al.*, 1971; Furuhashi *et al.*, 1957;

FIGURE 4.4 A thumb print of an individual with ridge dissociation. No coherent pattern is recognizable.

From Goddard, C. H.: Hands of mystery. *Finger Print and Identification Magazine*, 31:4, 1950. Courtesy of the Institute of Applied Science.



Furuya, 1961; Goddard, 1950; Grebe, 1940; Okamoto, 1958). Aberrant ridges have been observed in healthy individuals of different races and ethnic groups (Cooke, 1960, 1962; Cummins, 1968, 1970a,b; Furuhashi and Kuwashima, 1950; Furuya, 1961; Goddard, 1950; Nettles, 1963; Okamoto, 1958; Safara, 1969). Ridge disturbances may occur in any dermatoglyphic area. The involvement of epidermal areas exhibiting dissociated ridges varies greatly from minimal lesions affecting only very small areas within a pattern (Cummins, 1968, 1970b; Nettles, 1963; Safara, 1969; Figure 4.6) to the total volar surface of the fingers, palms, toes, and soles (Cooke, 1950, 1962; Cummins 1970a; Dodinval, 1972; Dodinval *et al.*, 1971; Goddard, 1950).

A relationship between ridge dissociation and individual fingertips was reported by Abel (1936), who found the frequency of affected fingers to decrease in the following order: thumb, index, ring, middle, and little finger (I, II, IV, III, V). In his study, the thumb was involved in each case of ridge dissociation. However, Furuya (1961) reported the order of decreasing frequency of involved fin-

FIGURE 4.5 Distal part of a palm print of a patient with Down syndrome showing ridge dissociation of the “dotted ridge” type.

From Rott, H. D.: *Hautleistenstörungen bei verschiedenen Krankheiten*. In Hirsch, W. (Ed.): *Hautleisten und Krankheiten*, Berlin, 1971. Courtesy of Freie Universität Berlin.



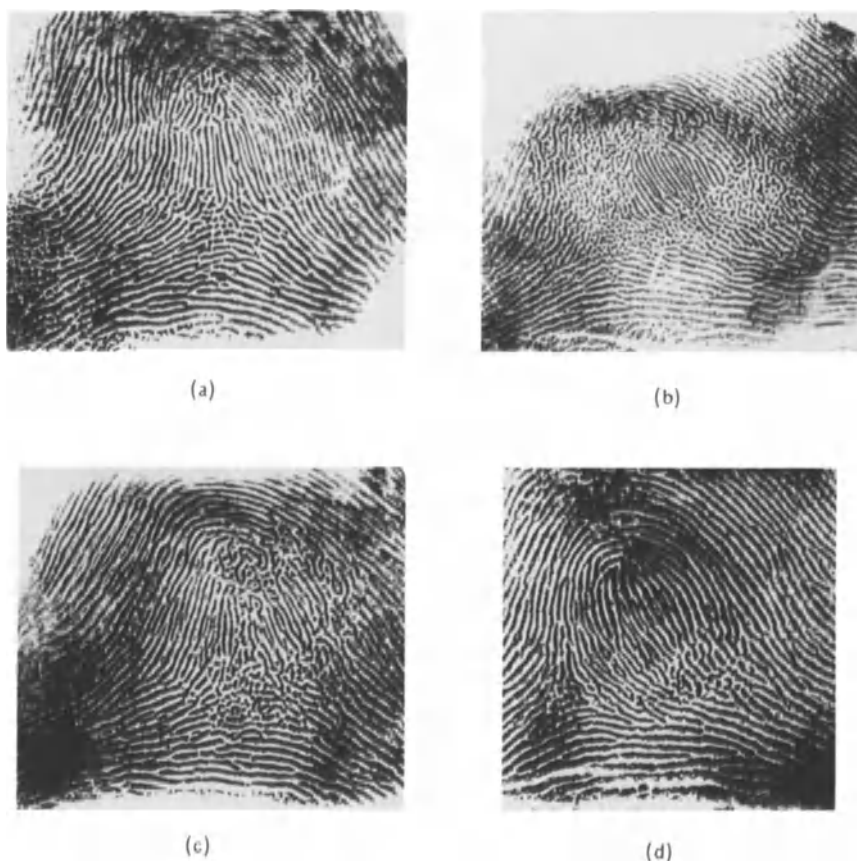


FIGURE 4.6 Ridge dissociation involving various parts of the fingertip areas.

From Abel, W.: Über Störungen der Papillarmuster. *Z. Morphol. Anthropol.*, 36:1, 1936. Courtesy of E. Schweizerbartsche Verlagsbuchhandlung (Nägele u. Obermiller).

gertips in a normal Japanese population as III, I-II, IV, V. In mentally deficient Japanese, the sequence was V, II, III-IV, I.

Furuya (1961) investigated all known cases of ridge dissociation in apparently healthy Japanese individuals. He divided the probands into two groups according to the extent of the involved epidermis: (1) total and (2) partial dot-and-short-ridge patterns. According to the published illustrations, the individuals with the total dot-and-short-ridge patterns showed involvement of the entire surface of the ridged skin of the fingertips and a variable extent of dissociation on the middle and proximal phalanges, palms, toes, and soles. Par-

tial dot-and-short-ridge patterns were limited to certain areas of the fingertips only. Apart from the fingertips, some affected persons in the latter group also exhibited ridge dissociation on some areas of the palms, toes, and soles. Both types of ridge dissociation seemed to be controlled by an autosomal dominant gene with incomplete penetrance. Because none of the kindreds investigated was found to display both types of the abnormality, Furuya concluded that the two types of ridge dissociation were apparently genetically independent of each other, each being controlled by a different gene.

Dodinval *et al.* (1971) reported a large family with ridge dissociation associated with tapering fingers, nail dystrophy, and painful chapping of the fingertips. The anomaly was transmitted as an autosomal dominant trait with complete penetrance and variable expressivity.

Although rare in normal populations, ridge dissociation is found with increased frequency in individuals suffering from various medical disorders. Abel (1936) pointed out the tendency of aberrant ridges to occur in persons with diseases and malformations, such as albinism, oxycephaly, malformations of the extremities (e.g., polydactyly, syndactyly, oligodactyly, perodactyly), spina bifida, deaf-mutism, familial amaurotic idiocy, and mental deficiency. His observations were confirmed by other investigators, notably in persons with limb malformations (Schade, 1937; Grebe, 1940), unspecified mental retardation (Matsukura *et al.*, 1957, cited by Furuya, 1961), and various chromosomal aberrations, such as trisomies 13, 18, and 21; deletion of the short arm of chromosome 4; and the de Lange syndrome (see Chapter 6). Ridge dissociation was also reported in ectodermal defects, such as hypohidrotic ectodermal dysplasia (Verbov, 1970; David, 1973a) and keratosis follicularis (Rott, 1971).

Although the appearance of dissociated ridges can vary considerably, no specific subtype of this epidermal anomaly is known to be associated with any particular medical disorder. This may result from a lack of descriptive information in the majority of reports. In spite of numerous reports of "hypoplastic" or "poorly formed" ridges in such defects as trisomy 21, 18, and 13; deletions of the short arm of chromosome 4; or the de Lange syndrome, the illustrations of the ridged skin in these reports show what should be termed a ridge dissociation rather than ridge hypoplasia. Usually, the dissociation is of the "dotted ridges" type with relatively clear and easily recognizable patterns. However, more severely dissociated ridges are not exceptional.

The appearance of the disturbed ridges can show substantial transition across a palm, a sole, or even a single pattern area. Figure 4.7 shows a gradual change from mildly dissociated ridges with a recognizable pattern toward more severe ridge disturbances, where the pattern is totally obscured.

Ridge dissociation can sometimes be mistaken for scarring and vice versa. Raphael and Raphael (1962) observed ridge dissociation in 18 percent of patients suffering from schizophrenia. However, Beckman and Norring (1963), who found distorted, “worn down” ridges among schizophrenics with about the same frequency as Raphael and Raphael (1962), concluded that the increase of ridge dissociation in schizophrenics might be explained as a result of injuries rather than as a sign of developmental disturbance. They hypothesized that psychotic individuals showed skin injuries more often than healthy persons. David (1969) supported this observation on the grounds that Raphael and Raphael’s (1962) illustrated examples of ridge dissociation were typical of injuries healed by granulation tissue. Subsequent studies of dermatoglyphics in schizo-

FIGURE 4.7 Part of a sole print in a patient with deletion of a short arm of chromosome 4. Note the transition from mild to severe ridge dissociation from right to left.

From Wolf, U.: Defizienz an den kurzen Armen eines Chromosoms Nr. 4. *Humangenetik*, 1:397, 1965. Courtesy of Springer Verlag.

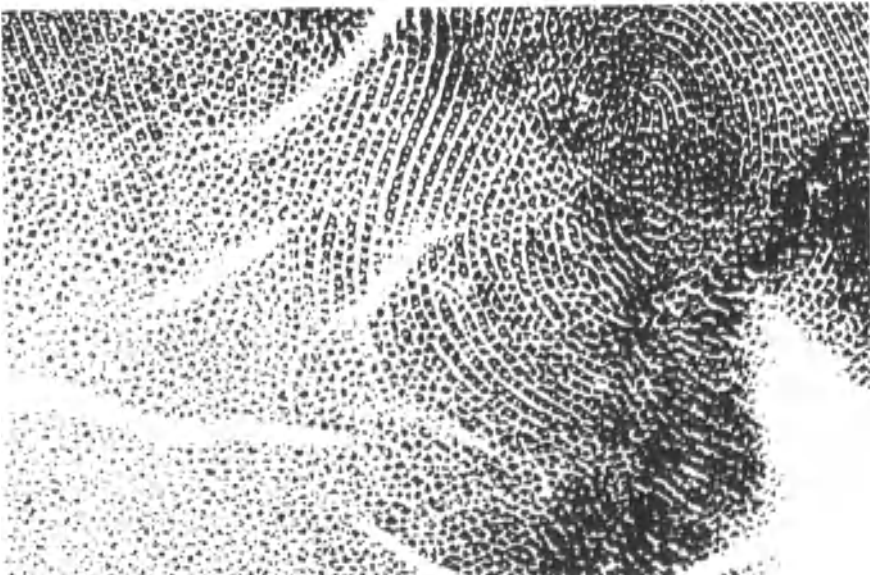
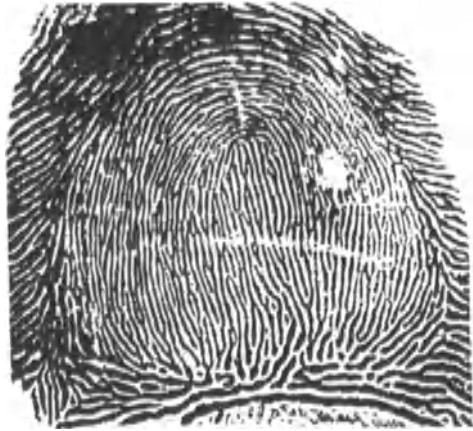


FIGURE 4.8 Aberrant ridge formation on a fingertip showing an extreme narrowing and abnormal arrangement of ridges.

From Abel, W.: Über Störungen der Papillarmuster. *Z. Morphol. Anthropol.*, 36:1, 1936. Courtesy of E. Schweizerbartsche Verlagsbuchhandlung (Nägele u. Obermiller).



phrenia (Mellor, 1968; Sank, 1968; Zavala and Nuñez, 1970, Rothhammer *et al.*, 1971; David, 1973a) failed to confirm the reports of an increased frequency of ridge dissociation.

Ridge dissociation apparently may sometimes be reversible. David (1973a) reported the complete disappearance of "dotted ridges" over a period of 6 months in a girl with small intestinal obstruction.

In rare cases, aberrant formation of epidermal ridges may involve breadth and direction of ridges. Figure 4.8 illustrates a fingertip with an extreme narrowing of abnormally oriented ridges in the pattern area.

"Ridges-off-the-end"

"Ridges-off-the-end" represents another anomaly of the epidermal ridges. Instead of running transversely and flowing toward the opposite side of the pattern, these ridges are vertical and run off the end of the fingertip. Ridges with such an upward course were reported in several otherwise apparently normal individuals (Cooke, 1958). David (1971) coined the term "ridges-off-the-end syndrome" (ROES). ROES is inherited as an autosomal dominant trait and is not associated with any medical disorder. Sometimes the fingertip ridges of affected individuals do not actually run off the end of the fingertip but merely tend to do so. These patterns are always radial rather than ulnar on the fourth and fifth digits and often on other digits too. Other dermatoglyphic anomalies of ROES include a tendency for the fingertip patterns partly to cross the distal interphalangeal crease, with the triradius occurring at or proximal

to the crease. In addition, there is a considerable distal displacement of the axial triradius to a t'' or t''' position; interdigital patterns, usually in the I_4 area, extending more proximally than usual; and a peculiar vertical "crack" in the ridges over the hypothenar eminence.

Subsequently, David (1973b) delineated another dermatoglyphic syndrome, which he proposed to name the Nelson syndrome after the propositus. The syndrome, which also seemed to be inherited as an autosomal dominant trait, shared some of the palmar features of ROES, such as deep interdigital loops, a distally displaced axial triradius, and a vertical crack in the hypothenar area. The abnormality of the ridges on the fingertips, however, was minor, consisting of pointed loops on some of the fingertip patterns. There were no "ridges-off-the-end," nor was there an enlargement of the fingertip patterns proximally to the distal phalangeal crease.

"Ridges-off-the-end" has been reported to be present simultaneously with dissociated ridges on all the fingertips of one member of a family with an autosomal dominant mild ridge dissociation (David, 1973c).

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5 Flexion Creases

Flexion creases (Figure 5.1) represent the location of firmer attachment of the skin to underlying structures. Although they differ in origin from the epidermal ridges and do not belong strictly to the dermatoglyphic system, palmar creases are usually included in routine dermatoglyphic analysis because their alterations may be of diagnostic value in a variety of medical disorders.

Embryology of Flexion Creases

Flexion creases are formed during early intrauterine life and, therefore, they can be influenced by factors causing aberrant development of the embryo. The first major palmar crease to appear on the embryonic palm is the radial longitudinal crease that borders the thenar eminence. It is followed in development by the proximal and distal horizontal creases (Pösch, 1925). The creases are first seen on the radial side of the palm and extend in an ulnar and distal-proximal direction.

The creases were believed to result from flexion of the fetal skin. Schaeuble (1933), however, found that the thenar crease was already present in embryos of 27 mm (C-R) length, i.e., at about 7 weeks gestation. The proximal and distal transverse flexion creases were observed in 40 mm C-R embryos (about 9 weeks gestation). These observations were confirmed by Würth (1937), who found that the flexion creases develop during the second and third em-

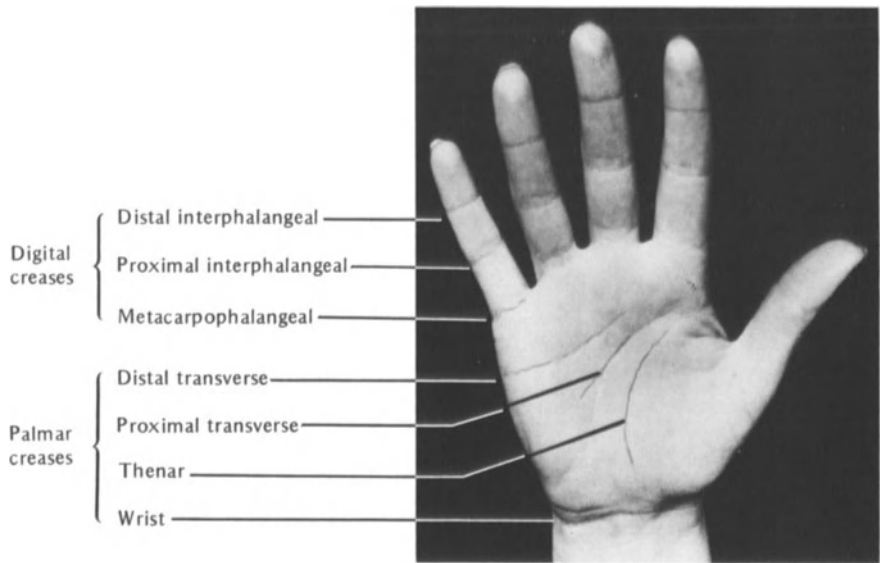


FIGURE 5.1 Nomenclature of hand creases.

bryonal month. Onset of spontaneous hand movements has not been observed in embryos of less than 11.5 weeks gestation (Humphrey, 1964), i.e., after the flexion creases are formed. Würth (1937) concluded that the flexion creases develop independently of palmar and finger movement. Moreover, he found no relationship between the embryonal flexion creases and the development of the underlying bones and muscles of the hand. These findings, however, do not exclude the possibility that the creases “anticipate” future hand movements. The theory that palmar and digital creases are secondary features determined by the form and particularly by the function of the developing hand instead of having a primary genetic determination was supported by studies of creases in malformed hands (Popich and Smith, 1970). Hand malformations are associated with typical alterations of the palmar creases. However, even the abnormal creases reflect the folding movement of the anomalous hand (Figures 5.6, 5.7, and 5.8).

Popich and Smith (1970) explained the spatial relationships of main palmar flexion creases as follows: The thenar crease is the consequence of oppositional function of the thumb and thenar muscle pad; the distal transverse crease follows the underlying sloping alignment of the third to fifth metacarpal–phalangeal joints; the proximal transverse crease is influenced on its radial aspect by flexion of the second metacarpal–phalangeal joint.

Classification of Palmar Flexion Creases

Human palms are covered to a greater or lesser degree by creases of different length, depth, and direction. This complicates the crease classification. Numerous attempts have been made to classify the creases either separately or in groups of increasing complexity (Féré, 1900; Pösch, 1925, H. Debrunner, 1952, 1957; I. M. Debrunner, 1957; Wendt, 1958; Leiber, 1960; Alter, 1970).

Loeffler (1969) divided the palmar creases into three groups: major, minor, and secondary (*Hauptfurchen*, *Nebenfurchen*, and *Sekundärfurchen*).

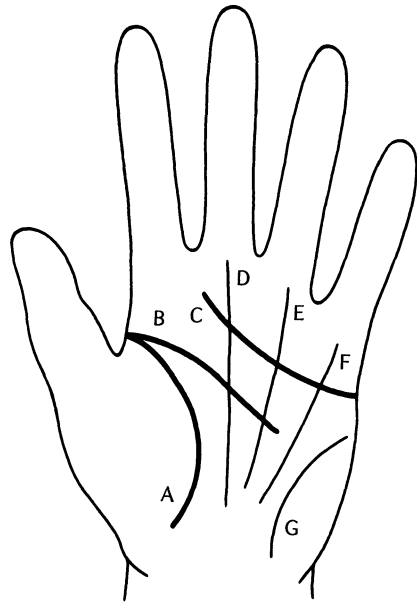
MAJOR CREASES

Most investigators have been concerned particularly with three major creases: the radial longitudinal crease, the proximal transverse, and the distal transverse crease (Figure 5.2). The *radial longitudinal crease*, commonly called the thenar, thumb, or vertical crease, is the curved crease encircling the thenar eminence and ending at the radial side of the hand somewhat above the distal wrist crease. It is the deepest crease in this area when the thumb is opposed. The *proximal transverse* (or proximal horizontal) *crease* is found usually just distal to the middle of the palm and its radial end is either fused with or shifted distally from the thenar crease. The proximal transverse crease sweeps from the radial border of the palm in a gentle, proximally concave bow across the palm and ends usually at the medial border of the hypothenar eminence. The *distal transverse* (or distal horizontal) *crease* is located between the proximal crease and the heads of the underlying metacarpal bones. Starting ordinarily in the space between the index and middle fingers, the crease curves gently proximally, ending on the ulnar edge of the palm.

The three major palmar flexion creases and a crease in the axis of the middle finger may form a crude letter "M" on the palmar surface. Leiber (1960) proposed a detailed classification of the major palmar creases based on (1) the type of the M figure (i.e., open, closed, intermediate, and special form), (2) the distance between the proximal and distal transverse creases, and (3) the relationship of the length of the proximal and the distal transverse creases.

Variations in the course and appearance of the major palmar flexion creases in a normal population were described by Alter

FIGURE 5.2 Schematic representation of the location of main palmar flexion creases. A, thenar crease; B, proximal transverse crease; C, distal transverse crease; D, middle finger crease; E, ring finger crease; F, little finger crease; G, hypothenar crease.



(1970). He measured differences in distances between proximal and distal transverse creases and noted the frequency of double thenar creases, accessory creases, and abnormalities of the flow of the creases, including broken, forked, or branched creases and cascade configurations of the lines. He also noted the frequencies of unusually short and long transverse creases.

Sometimes the proximal and distal transverse creases are replaced by or joined into one single crease that traverses the whole palm (Figure 5.3). This *single transverse flexion crease* is usually referred to as a *simian crease* or *line*. This designation is inappropriate as it does not represent the characteristic palmar crease configuration among apes and monkeys. Only the pavian monkey shows this crease, whereas gorilla, orangutan, and chimpanzee have several transverse palmar creases. However, the term “simian crease” has gained acceptance through its widespread use and for the sake of brevity it is used throughout this book. Several other terms used to designate the single palmar crease (or line) include single upper palmar crease, single transverse fold, and four-finger line.

Tillner (1953) has hypothesized that the single transverse crease is formed from either the proximal or the distal crease alone, rather than from both transverse creases. Leiber (1960), however, considered the single transverse crease as a fusion product of the two normal transverse creases because of either a reduction in the length



FIGURE 5.3 Single transverse flexion crease.

of one of them or a progressive reduction in the distance between them. Sometimes the proximal and distal transverse creases approach each other, do not actually touch, but are joined by a bridging crease (Figure 5.4). They may also touch and one or both may extend beyond the point of confluence as branches. The bridged or branched single-crease configurations are called “incomplete,” “bridged,” “transitional,” or “simian variant” creases. Some investigators have failed to separate simian and simian variant types, referring to both as simian creases. The high variability in appearance of simian creases often leads to confusion and is also at least partially responsible for the differences in the frequencies of this crease cited in various samples drawn from healthy populations. The reported frequencies vary between about 1 and 15 percent in control populations but may be considerably higher in groups of individuals

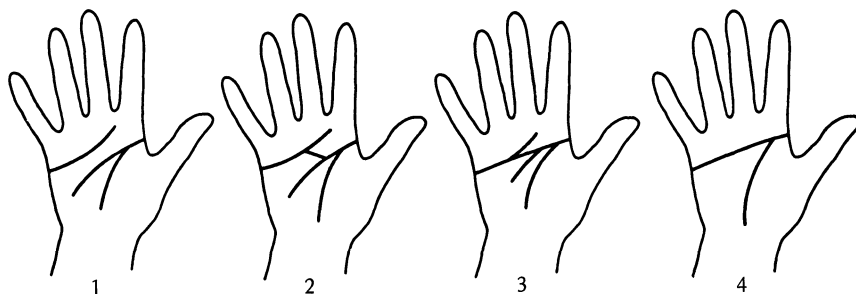


FIGURE 5.4 Schematic drawing of major palmar flexion creases illustrating a transition between normal creases and a single palmar crease (simian line). (1) Normal flexion creases (proximal and distal transverse creases separated); (2) transitional type 1 (proximal and distal creases connected by a bridging crease); (3) transitional type 2 (fusion of the transverse creases with branching proximal and distal segments, incomplete single palmar crease); (4) single flexion crease.

From Beckman, L., Gustavson, K. H., and Norring, A.: Finger and palm dermal ridge patterns in normal and mongoloid individuals (the Down syndrome). *Acta Genet.*, 12:20, 1962. Courtesy of S. Karger AG, Basel.

with developmental defects. The simian crease, therefore, may have medical diagnostic value.

Several variants of the single transverse crease are illustrated in Figure 5.5. According to Leiber's (1960) scheme, the transverse crease in variants (A) and (E) represents a shortened proximal crease and lengthened distal crease whereas variants (B) and (D) are transverse creases that have approximated each other, forming a bridged (or incomplete) single transverse crease. Variant (B) shows closer approximation of the joined creases than variant (D). In (G) there is a lengthened proximal and shortened distal transverse crease. Variant (C) shows a fusion of the distal transverse crease with a lengthened proximal transverse crease (Sydney line). In (F) there is approximation of creases that appear to be the distal and unusually sloped proximal transverse crease; the additional horizontal crease resembles a segment of the proximal transverse crease in its usual slope.

Popich and Smith (1970) attempted to account for the embryonic development of a single transverse crease on the basis of the location and nature of the early fetal interdigital pads. According to their hypothesis, the transverse palmar crease is influenced by both the sloping alignment of the third to fifth metacarpal-phalangeal joints and the nature of the fetal interdigital pads at the time of crease development.

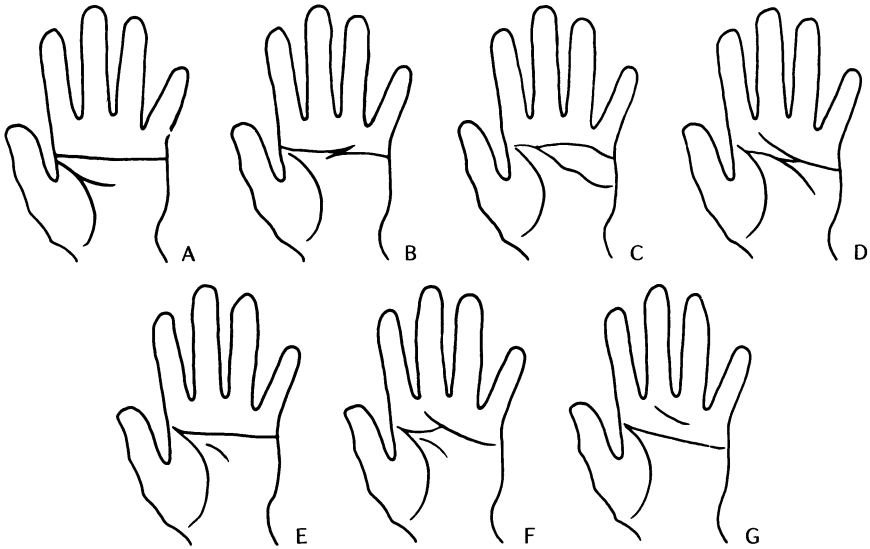
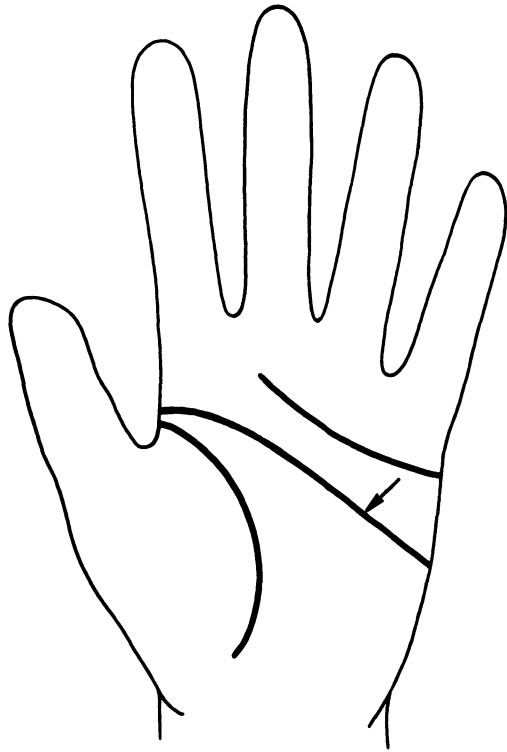


FIGURE 5.5 Variations of the single transverse crease. See text for details.

From Johnson, C. F., and Opitz, E.: Unusual palm creases and unusual children. *Clin. Pediatr.* (Philadelphia), 12:101, 1973. Courtesy of The J. B. Lippincott Co.

Variation in appearance of the proximal transverse crease have been noted for centuries by the cheiromancers but they have been studied only recently in relation to medical disorders. One variation is called a *Sydney line*, after the city in Australia where it was observed by Purvis-Smith and Menser (1968). This line represents a proximal transverse crease that extends beyond the hypothenar eminence to the ulnar margin of the palm (Purvis-Smith, 1972; Figure 5.6). The distal transverse crease persists and appears normal. This extended proximal crease was described earlier by Erne (1953, cited by Weninger, 1971) and corresponds to the special form 1 of Weninger and Navratil (1957). In Leiber's (1960) classification of main palmar creases, the Sydney line is not considered to be a significant abnormality. Later, however, an increased frequency of the Sydney line was reported in individuals with Down syndrome (Purvis-Smith, 1972), congenital rubella (Purvis-Smith and Menser, 1968; Purvis-Smith *et al.*, 1969), and leukemia (Menser and Purvis-Smith, 1969). The Sydney line was the most frequently seen palmar crease aberration in a series of children with delayed development, learning difficulties, or minor behavioral problems (Johnson and Opitz, 1973). Considerable variation in

FIGURE 5.6 Schematic drawing of a Sydney line.



frequency of the Sydney line in specific disorders has been reported by different authors, which may be explained, at least in part, by nonstandardized definitions and occasional erroneous classification of this line. Purvis-Smith (1972) considered a Sydney line to be any configuration in which “the proximal palmar crease extends toward the ulnar margin of the palm past the midline axis of the fifth finger.” Wertelecki *et al.* (1973) used somewhat stricter criteria and labeled an elongated transverse palmar flexion crease as a Sydney line only if it reached the ulnar margin of the palm. If the line failed to reach the ulnar margin even by only a few millimeters, they referred to the configuration as an aberrant Sydney line. Wertelecki’s *et al.* (1973) classification modifies the Purvis-Smith (1972) definition without differing greatly from it. In order to avoid occasional misinterpretations of the Sydney line, such as classifying it as a simian crease, it should be stressed again that in the Sydney line configuration the distal palmar crease is present on the palm, separate from the lengthened proximal crease.

Variants of the Sydney line which were observed by Johnson and

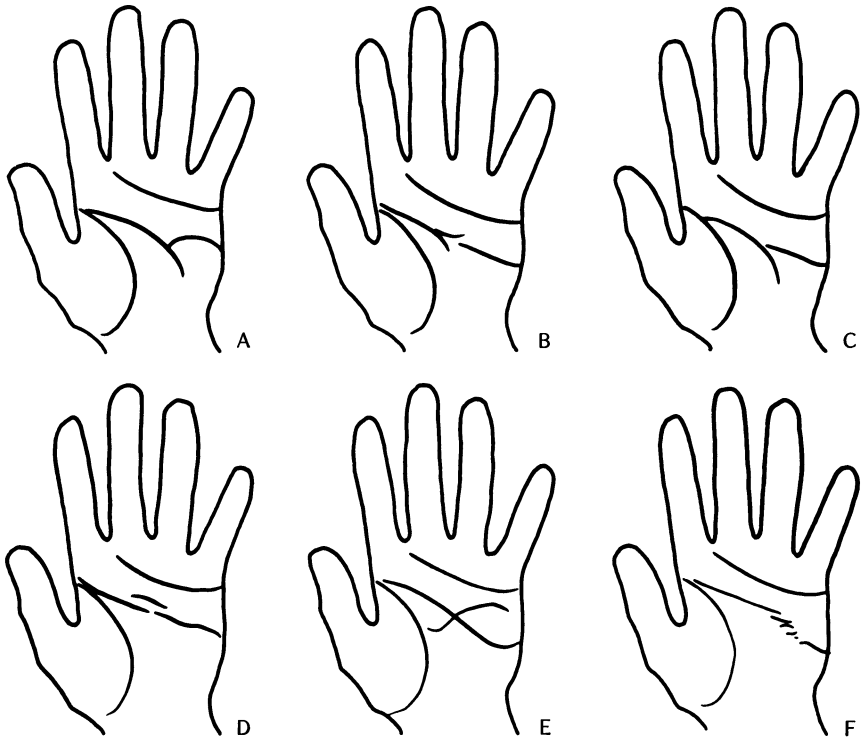


FIGURE 5.7 Variations of the Sydney line. See text for details.

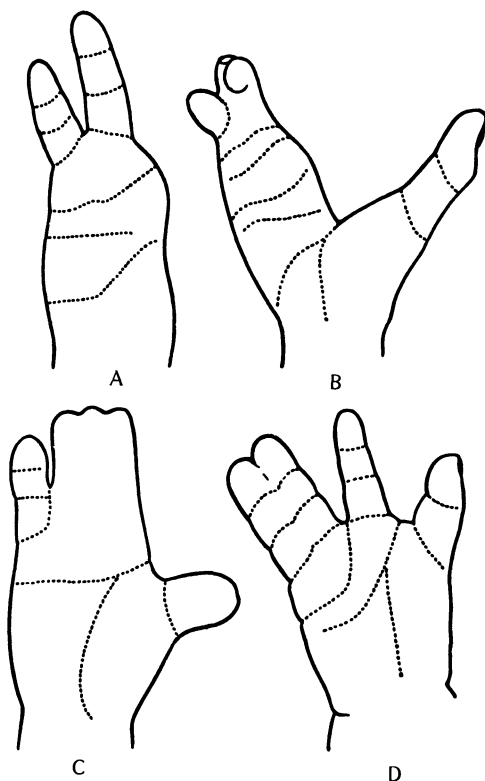
From Johnson, C. F., and Opitz, E.: Unusual palm creases and unusual children. *Clin. Pediatr. (Philadelphia)*, 12:101, 1973. Courtesy of The J. B. Lippincott Co.

Opitz (1973) are illustrated in Figure 5.7. They include (A) arching, which resembles variation (C); in both variants there is a single branch extending from the proximal transverse crease, whereas in variant (B) a double branch occurs. In variation (D) the crease is interrupted and a short horizontal crease is present above the gap. In variation (E) a third horizontal crease is present in addition to the distal and lengthened proximal creases. This extra crease, which crosses the Sydney line, has a very unusual appearance. In variation (F) the proximal transverse crease is interrupted, cascading to the ulnar border of the palm.

As mentioned earlier, hand malformations are associated with typical alterations of the palmar creases that relate to the flexion movement of the involved hand (Figures 5.8 and 5.9). The relationship between the anomalous hand and the formation of flexion creases is illustrated by an association between the thumb and the

FIGURE 5.8 Grossly altered palmar creases, relative to the abnormal planes of flexion in malformed hands. A, hypoplasia and oligodactyly; B, ectrodactyly; C, syndactyly; D, syndactyly and oligodactyly.

From Popich, G. A., and Smith, D. W.: The genesis and significance of digital and palmar hand creases: Preliminary report. *J. Pediatr.*, 77:1017, 1970. Courtesy of The C. V. Mosby Company.



thenar crease (Popich and Smith, 1970; Holt, 1972). In individuals with an absent, rudimentary, or hypoplastic functionless thumb, the thenar crease is missing (Figure 5.9A, B, and C). Hypoplastic thumbs with limited function are associated with poorly developed thenar creases (Figure 5.9D). An altered position of the thumb results in a thenar crease that is similarly altered in position. For example, a proximally placed thumb is accompanied by a proximally placed thenar crease (Figure 5.9E).

MINOR CREASES

In addition to the major creases just described, several minor creases are often present on the palm. Their presence, prominence, length, and shape are more variable than those of the major creases. Loeffler (1969) divided the minor creases into four groups:

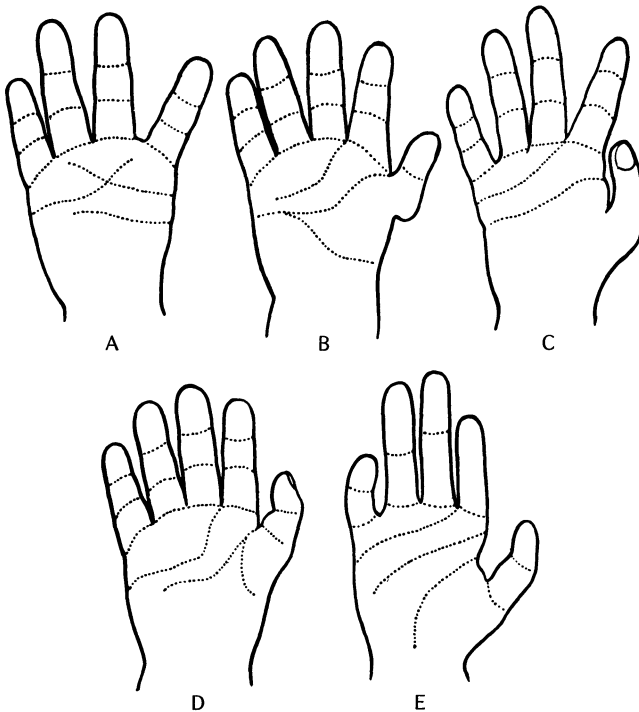
1. Three longitudinal creases, when present, run from the central part of the wrist toward the third, fourth, and fifth digits. Ac-

cordingly, they are referred to as the *middle, ring, and little finger creases* (Figure 5.2).

2. The *accessory distal crease* (*Zweifingerfurche*) may occasionally be found under the third and fourth digits, beyond the distal transverse crease.
3. "*E lines*" may be located at the distal ulnar edge of the palm between the origin of the distal transverse crease and the metacarpophalangeal crease of the fifth finger. There are usually three such parallel lines. They resemble an E when viewed on the right hand, whereas on the left hand, the mirror image of an E may be seen.
4. A *hypothenar crease* occurs occasionally in the hypothenar eminence, running in a proximal–distal direction concave toward the ulnar side of the palm.

FIGURE 5.9 Thenar crease in malformed hands. A, aplasia of the thumbs; B, rudimentary thumb with no oppositional function; C, hypoplastic functionless thumb; D, hypoplastic thumb with limited function; E, proximally placed thumb.

From Popich, G. A., and Smith, D. W.: The genesis and significance of digital and palmar hand creases: Preliminary report. *J. Pediatr.*, 77:1017, 1970. Courtesy of The C. V. Mosby Company.



SECONDARY CREASES

The term “secondary crease” was introduced by H. Debrunner (1957) and I. M. Debrunner (1957) to describe any visible palmar creases other than major and minor creases. Their number, length, depth, and direction vary widely in different individuals. Unlike major and minor creases, secondary creases may vary with age and sex.

A system for quantitative evaluation of palmar creases, based on both primary and secondary creases, was devised by H. Debrunner (1952). He outlined five degrees of intensity of palmar lines, varying between situations in which only the major creases were present (type I) to those in which the palm was heavily traversed by minor and secondary as well as major creases (type V).

OTHER HAND CREASES

Phalangeal creases

The thumb normally has a single phalangeal flexion crease, whereas all other fingers have a proximal and distal flexion crease, one at each interphalangeal joint. Occasionally, one of the two interphalangeal creases is missing on a finger, most frequently the little finger. Presence of a single digital crease is associated with abnormally short phalanges or with finger flexion deformities (Uchida and Soltan, 1963; Popich and Smith, 1970). A single digital flexion crease of the fifth finger was described by Penrose (1931) as characteristic of Down syndrome. However, a single phalangeal crease of the fifth finger is also frequently present in trisomy 13 and trisomy 18 and occasionally in other medical disorders. Creases related to joints reflect the flexion movements, as can be best illustrated in malformed hands with structural and functional changes in the digits (Popich and Smith, 1970). For example, the digital creases are absent over nonfunctional joints (Figure 5.10A, B, and E), and the creases related to joints with limited flexion tend to be indistinct. Syndactylous digits, which function as a unit, share a continuous crease over their volar surface (Figure 5.10B and C). If an extra functional joint is present, as in triphalangeal thumb, an extra digital crease is present (Figure 5.10D).

THE EXTRA DIGITAL CREASE. Occasionally, an additional transverse crease can be found just beyond the distal interphalangeal flexion crease (Figure 5.11) of one or more digits, most frequently

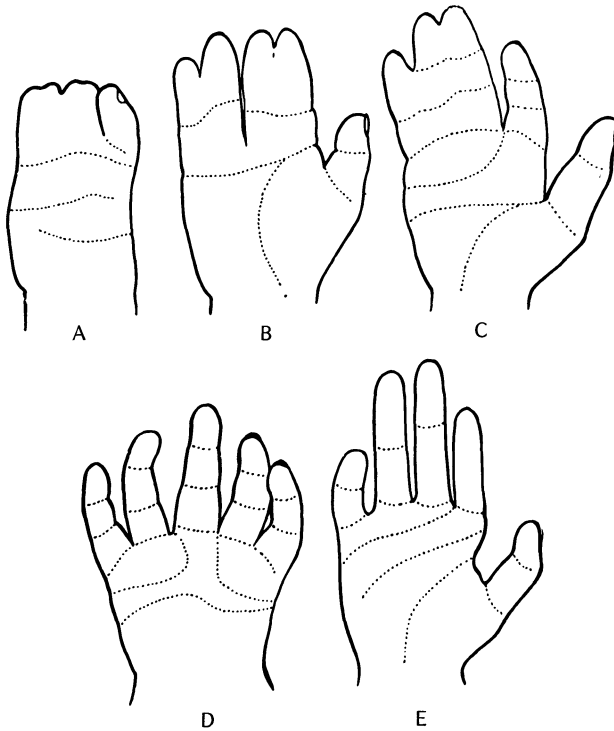


FIGURE 5.10 Digital creases in malformed hands. A, syndactyly with no digital flexion; B, syndactyly with flexion only at the proximal interphalangeal joints; C, syndactyly with flexion possible at both proximal and distal interphalangeal joints; D, triphalangeal thumb; E, hypoplasia of phalanges and lack of flexion at some interphalangeal joints.

From Popich, G. A. and Smith, D. W.: The genesis and significance of digital and palmar hand creases: Preliminary report. *J. Pediatr.*, 77:1017, 1970. Courtesy of The C. V. Mosby Company.

on the middle finger. This extra crease is present at birth and persists during life. DeJong and Platou (1967) distinguished this crease from accessory distal interphalangeal creases, white lines, and scars and described the characteristics by which the extra crease can be recognized.

Two or more epidermal ridges must separate the extra crease from the usual distal interphalangeal flexion crease; the two creases must not connect. Characteristics of the separating epidermal ridges must be similar to those over the adjacent distal phalanx. The extra crease must disrupt the local ridge configuration or there must be a localized deficiency of ridge formation.

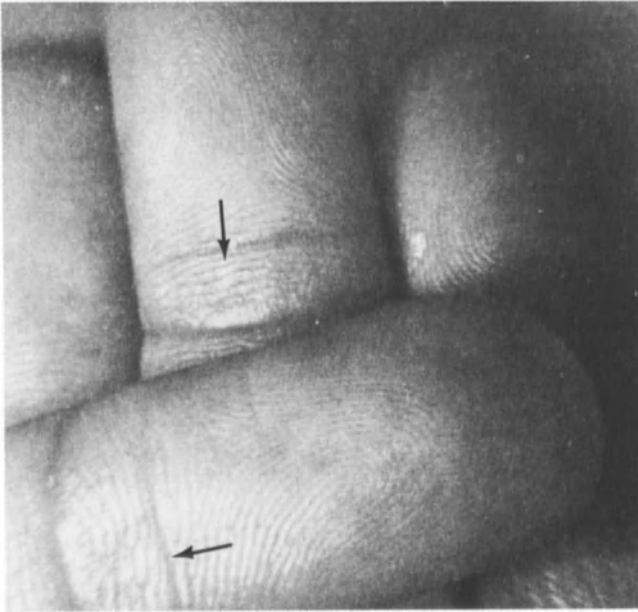


FIGURE 5.11 The extra transverse digital crease beyond the regular distal interphalangeal flexion crease.

From Zizmor, J.: The extra transverse digital crease: a skin sign found in sickle cell disease. *Cutis*, 11:447, 1973. Courtesy of Dunn-Donnelley Publishing Corp.

The extra digital crease may have significance in relation to specific medical disorders. DeJong and Platou (1967) found a higher frequency of occurrence of this crease among Negro patients with sickle cell hemoglobinopathy (38 percent) than in healthy Negro individuals (18 percent). In addition, whereas six of the 33 patients possessed the extra crease bilaterally, none of the controls had an extra digital crease on both hands. Zizmor (1973) found an even more marked difference in the frequency of the extra crease comparing patients with sickle cell disease (90 percent) and controls (10 percent). The crease may therefore prove to be a diagnostically useful trait in newborns evaluated for sickle cell disease, particularly if it is present bilaterally on the middle fingers.

Metacarpophalangeal creases

Configurations and morphologic features of metacarpophalangeal creases, which divide proximal phalanges from the palm region,

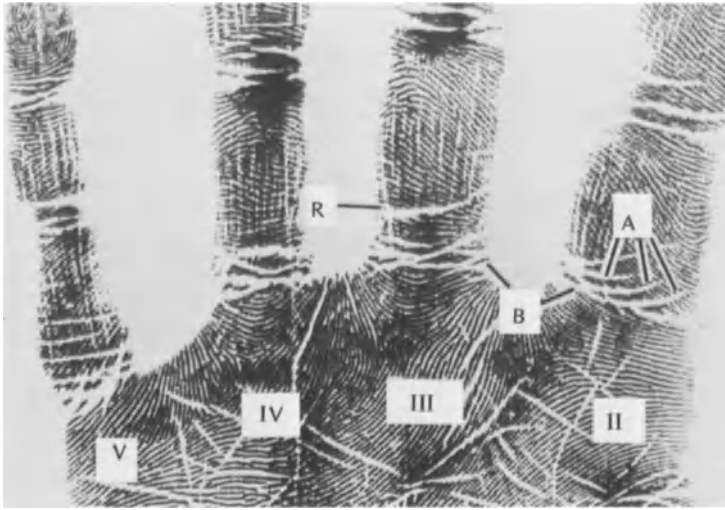


FIGURE 5.12 Metacarpophalangeal creases. A, accessory creases; B, boundary crease; R, ring crease.

From Okajima, M.: A dermatoglyphic study of metacarpophalangeal creases. *Am. J. Phys. Anthropol.*, 24:371, 1966. Courtesy of The Wistar Press.

were described by Okajima (1966), who also suggested their use for the diagnosis of zygoty (Okajima, 1967). He observed the tendency of metacarpophalangeal creases to show characteristic patterns in each digit and introduced a classification using the terms “boundary,” “ring,” and “accessory” creases (Figure 5.12). The boundary crease is the main crease which divides the phalanx from the palm. Sometimes an additional crease is present distal to the boundary crease which looks as if a ring has been worn (hence the name “ring” crease). The ring crease is generally found only on the middle and ring finger. Accessory creases are shorter and may be separate from or branch out from the boundary crease. They are usually parallel to each other, being located distally from the main (boundary) crease and slanting toward it.

Wrist creases

There are usually two creases present in the wrist area. They are generally referred to as the proximal and distal wrist (or bracelet) creases. The distal coincides approximately with the limit of ridged skin.

Plantar Flexion Creases

Because the skin creases on the volar aspects of the feet develop in late intrauterine life, their presence has been used as a helpful clinical sign for determining gestational age. They have been considered the single most reliable physical index of maturity (Usher *et al.*, 1966). Plantar skin creases of the newborn are assessed by noting the creases that persist when the skin of the sole is stretched from the toes to the heel (Farr *et al.*, 1966). At birth, the plantar creases are deep furrows. The finer superficial lines, which often appear even in premature infants after the first day or two of life, are not considered as they bear no relationship to gestational age.

In infants of less than 34 weeks gestation, there are no visible plantar creases. Between 34 and 36 weeks one or two transverse skin creases appear on the anterior part of the sole, whereas the posterior two-thirds of the sole remains smooth. By 37 and 38 weeks of gestation, more creases appear, covering the anterior two thirds of the sole. In full-term infants of 39 weeks or more, the entire sole is covered by a complex series of creases of varying length coursing in various directions (Figure 5.13a,b,c).

Damoulaki-Sfakianaki *et al.* (1972) found significant differences between Caucasian and Negro full-term, newborn infants in the length of the sole that was covered with creases. On the average, Caucasians had 83 percent (range 71–92 percent) and Negroes had 71 percent (range 21–85 percent) of the sole length covered by creases. According to this finding, lack of sole creases cannot be used to distinguish between the premature and full-term American Negro infants. In contrast to the study of Damoulaki-Sfakianaki *et al.* (1972), Parkin (1971) did not observe significant differences in plantar skin creases between black infants of Uganda and white infants born in Great Britain.

Apart from their use in assessing gestational age, sole flexion creases have little known clinical significance. However, the sole creases have been investigated less intensively than those of the palms. One of the reasons for the lack of information on sole creases may be that they become rather inconspicuous after early childhood. Persistence of the creases after the second or third year of life is rare. Because the embryonal development of the foot closely resembles that of the hand, it may be assumed that the plantar creases are formed between the embryonal pads (Schenk and Patzer, 1959). Based on the analysis of the feet of 100 newborns,

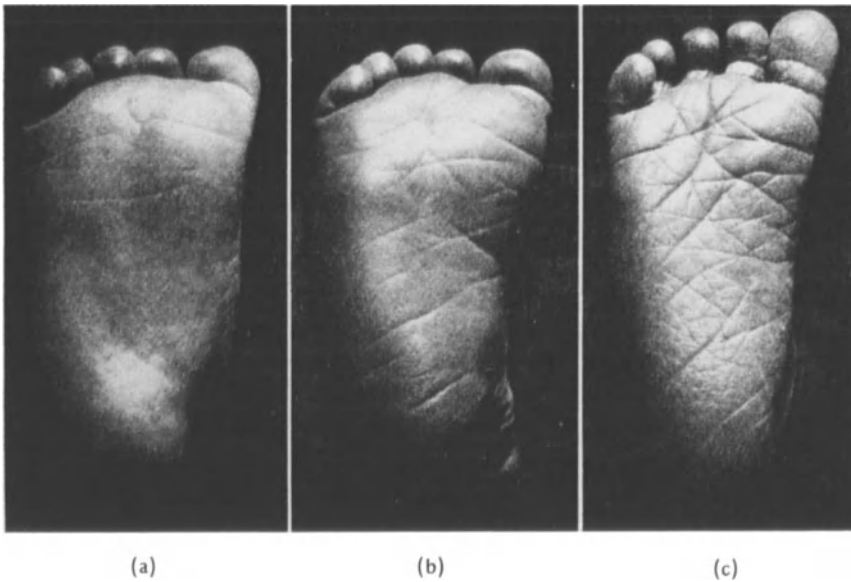


FIGURE 5.13 Infant sole at different gestational ages.
(a) Smooth posterior three-quarters of the sole (36 weeks).
(b) Some creases over the whole sole surface (38 weeks).
(c) Complex series of creases over the whole length of the sole (40 weeks).

From Usher, R., McLean, F., and Scott, K. E.: Judgment of fetal age. II. Clinical significance of gestational age and an objective method for its assessment. *Pediatr. Clin. North Am.*, 13:835, 1966. Courtesy of W. B. Saunders.

Schenk (1958, cited by Schenk and Patzer, 1959) proposed a morphologic scheme of the plantar creases based on nine major creases (Figure 5.14). According to this scheme, the creases are defined as follows: Crease 1 originates under the second toe and runs almost straight in the direction of the heel. Crease 2 begins between the first and second toes or under the second toe and continues arching toward the fibular side of the sole. Crease 3 starts between the second and third toes and runs obliquely toward the fibular side. Crease 3 can merge with crease 8 or cross it. The crossing is relatively rare because crease 3 is usually short. Crease 4 originates under the third toe and bows toward the tibial border. Crease 4 crosses crease 2, forming a forklike figure with it that is characteristic for newborns. Crease 5 starts between the fourth and fifth toes and runs toward the center of the sole, varying greatly in its length. In almost half of the feet on which this crease is present,

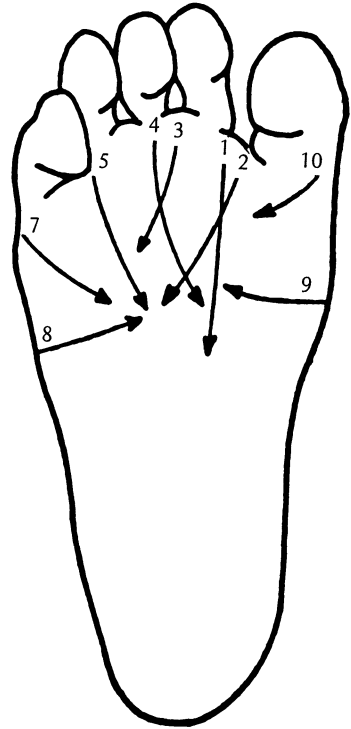


FIGURE 5.14 Schematic drawing of the main plantar creases in a newborn.

From Schenk, H., and Patzer, H.: *Das Fussfurchenbild des Neugeborenen. Kinderärztl. Prax.*, 27:169, 1959.
Courtesy of Georg Thieme Verlag.

it forms a fork. The branches of this fork can diverge toward the opposite borders of the sole. Crease 5 can cross crease 2 or 8 or even creases 1 and 9 if these are well formed. Crease 6 was not designated. Crease 7 originates between the fifth toe and the fibular border of the sole and runs obliquely or almost straight toward the center of the sole. Crease 8 runs from the fibular border toward the center of the distal planta. Crease 9 begins at the tibial border of the thenar area and runs toward the center of the distal planta. Crease 10 originates in the area between the great toe and the tibial border of the sole. Crease 10 can meet and merge with crease 8, creating an impression that crease 8 runs across the distal sole under the great toe. Not all creases are necessarily present on each sole of the newborn. The frequencies of individual creases are listed in Table 5.1.

Hirsch (1968) saw a resemblance between a transverse crease formed by fusion of creases 8 and 9 and the simian crease of the palm.

Nakiela (1972) based his observations of the plantar flexion creases on footprints of 1160 adult male Poles and 500 feet of in-

TABLE 5.1. Frequencies of plantar creases on the soles of 100 newborns^a

CREASE	NUMBER	PERCENT
1	189	94.5
2	200	100.0
3	69	34.5
4	198	99.0
5	159	79.5
7	45	22.5
8	200	100.0
9	200	100.0
10	73	36.5

^a According to Schenk and Patzer (1959).

dividuals of both sexes and ages varying between 3 months and 80 years. In order to analyze the data topologically, the author used a scheme that divided the footprint horizontally into several areas (Figure 5.15a). In adults, creases were present only on the arch of the foot. No creases were observed on the heel and more extended to the external border of the foot. There was marked variation of the creases, which allowed only relative regularities to be reported. The sole creases on the right and left were not symmetrical. The most frequent creases found in the Polish sample were those designated 1 and 2 by the author (Figure 5.15b). They were found in 38 and 34 percent of the soles, respectively. Crease 1 was described as beginning at the medial border of the foot below the thenar eminence of the great toe, running parallel to line x (see Figure 5.15a) but never extending further than the axis of the second toe. Furrow 2 originated at the medial border of the foot at the boundary between the heel and arch and continued, usually obliquely, toward the external side of the foot. Both creases were sometimes observed to be double and crease 2 even triple or quadruple.

In neonates and small children, a transverse crease was usually observed. This crease (Figure 5.15c) was more or less horizontal, extending from the thenar eminence to the external border of the foot. Short creases running across the thenar eminence and creases under the hypothenar eminence were sometimes present. These creases were never encountered in adults.

Although the creases ran in various directions in relation to the long axis of the foot, longitudinal and oblique creases predominated.

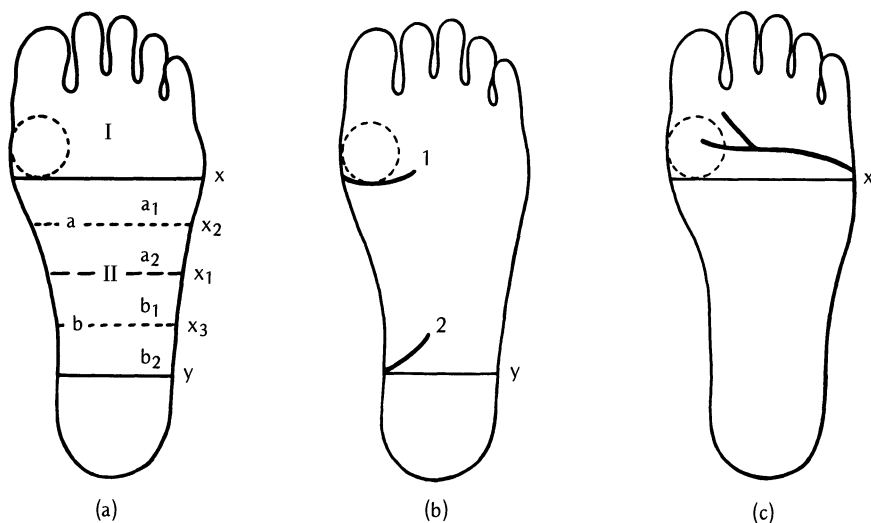


FIGURE 5.15 Schematic drawing of the most frequent sole creases. (a) Diagram of a sole with orientative lines and areas. (b) Diagram of creases 1 and 2. (c) Diagram of transverse crease most frequently found in young children.

According to Nakiela, J.: Flexion furrows of the feet in Poles. *Folia Morphol. (Warsz.)*, 31:301, 1972.

In 7.5 percent of the adult footprints examined, the furrows were entirely absent. There was a marked individual variation in the flexion creases with regard to course, frequency, and arrangement. No relationship was found between the crease arrangement and sex.

Unusual plantar creases may have some clinical significance. Penrose (1963) pointed out the presence of a marked crease between the first and second toes of patients with Down syndrome. This crease is sometimes referred to as a “sandal crease.” Such a crease was also observed in a majority of patients with the Rubinstein–Taybi syndrome who were investigated for this trait (Rubinstein, 1969; Filippi, 1972). Several deep furrows on the palms or soles are characteristic for the trisomy 8 syndrome (Lejeune *et al.*, 1969). However, they have also occasionally been reported in other chromosomal disorders.

White Lines

A variable number of shallow grooves of different length, width, and direction can often be observed on the fingertips (Figure 5.16). The grooves, which are called *white lines* (derived from their ap-



FIGURE 5.16 White lines on two fingertips.

pearance on dermatoglyphic prints) cross the epidermal pattern areas of the fingertips in various directions, independently of the direction of the papillary ridges. The white lines are probably caused by skin buckling rather than skin flexion. They are easily differentiated from scars (Figure 5.17) or wrinkles, such as appear after prolonged immersion of the fingers in water. The epidermal ridges continue through the furrow of the white line. On a histologic section, the glandular folds and furrows underlying the white lines do not show any interruptions in their structure at the border of epidermis and corium but they can be traced to the stratum lucidum, which flattens along their course (Wendt, 1955, 1956). If the epidermis is damaged down to the prickle cell layer and the basal layer is undamaged, the epidermis is capable of regenerating white

FIGURE 5.17 A deep cut healed in a scar which disrupts epidermal ridge pattern.



lines as well as papillary lines (Mierzecki and Miklaszewska, 1968).

Originally, the white lines were studied for possible use in personal identification. However, they are not permanent features of the skin (Domingues, 1931, 1933; Lerich, 1933). Existing white lines can disappear partially or completely and new ones can appear. However, white lines are considerably more persistent features than the wrinkles acquired in prolonged contact with water or cracking of the skin caused by weak chemical irritants.

Solth and Wendt (1958) investigated the relationship between white lines of the fingertips and the finer (secondary) palmar creases. Using H. Debrunner's (1952) classification, they found that 42.1 percent of persons without white lines had little or no creasing. In contrast, 46.1 percent of individuals with white lines had many palmar creases. These positive correlations between white line and crease frequency seem to indicate that the variability in expression and intensity of these skin traits has a common origin.

According to most observers (Aznar, 1933; Reyna Almandos, 1937; Wendt, 1955; Mierzecki and Miklaszewska, 1968), the incidence of white lines increases, in general, with age. Wendt (1952) found that among 171 children between 2 and 6 years of age, only 5.6 percent showed white lines. Moreover, in children no significant difference was observed between the sexes. White lines in children were virtually limited to the thumb.

There is no widespread agreement about the frequency of white lines in normal populations. Cherrill (1950) found these lines in approximately 11 to 12 percent "of a large number of prints" of apparently normal persons ranging in age from 16 to 75 years. However, his material was derived from police files and could have been made up predominantly of males. Frequencies of individuals with white lines observed by ourselves and other authors are listed in Table 5.2. Females generally showed a higher frequency of white lines than males. Some of the variability in frequency may have been caused by differences in definition of a white line and in printing techniques. We listed all white lines of even one ridge breadth because even these short white lines showed up clearly with our transparent tape and graphite printing technique (see Chapter 2). Moreover, we were able to discern white lines on lateral margins of the digits because our prints were well rolled.

Among all the fingers that had white lines, slightly more than half were found on the left hand (Wendt, 1955; Mierzecki and Miklaszewska, 1968). Some fingers seemed to be more likely to have white lines than others. Cherrill (1950) observed that white

TABLE 5.2. Frequencies of white lines in normal populations

POPULATION	AGE OF SUBJECTS (YEARS)	NUMBER OF SUBJECTS	PERCENT WITH WHITE LINES			SOURCE
			MALES	FEMALES	TOTAL	
German	2-63	1883	22	39	30	Wendt (1952)
German	2-63	2787	20	38	28	Wendt (1955)
Polish	7-70	150	32	40	36	Mierzecki and Miklaszewska (1968)
North American	22-63	400	93	98	96	Schaumann and Alter (unpublished)

lines occurred mainly on the third, fourth, and fifth digits of the left hand. If absent on these fingers, the white lines were rarely present on the other fingers. Wendt (1955) found the highest frequency of lines on the fourth digits. The ranking in order of decreasing frequency was IV, III, V, I, II. In our material, this order was V, IV, III, I, II in both males and females.

The cause of white lines is not well understood. Exogenous factors, such as extended periods of immersion of hands in water, have been suggested as being important (Aznar, 1933; Macaggi, 1935; Wendt, 1955). The higher frequency of white lines among women, particularly housewives, seems to support this hypothesis. Wendt (1952) found white lines in 89.5 percent of housewives, which was a strikingly higher percentage than in any other occupational group.

Wendt (1955) and Mierzecki and Miklaszewska (1968) considered somatic habitus of the individual to be related to the development of white lines. While only 8.1 percent of the persons of athletic build showed the white lines, 20.0 percent of pyknics and 27.9 percent of leptosomes showed this trait (Wendt, 1955).

Hereditary factors have also been investigated. Wendt (1955, 1956) observed white lines in 43.3 percent of the offspring in families where both parents showed these lines and only in 14.0 percent of children whose parents both lacked the white lines. In families where only one parent had the white lines, 21.8 percent of children were found to have this trait. Among monozygotic (MZ) twins, 95.5 percent were concordant for white lines, compared to 61.7 percent of like-sex dizygotic (DZ) twins (Wendt, 1956); 14.3 percent of MZ but no DZ twin pairs showed similarity in position of

white lines on at least two fingers; and 41.1 percent of MZ and 17.0 percent of DZ pairs shared practically an identical number of white lines.

Cherrill (1950) suggested a pathologic cause for the occurrence of the white lines, connecting their presence with the occurrence of a disease or with the stage of a disease, e.g., epilepsy. However, Wendt (1952) did not confirm Cherrill's results. Hirsch and Recke (1971) reported an increase in the frequency of white lines among 26 patients suffering from various skin diseases. Verification is needed because only a few patients with each of the different skin disorders were included in their study. According to David *et al.* (1970), white lines disappear in active celiac disease with severe ridge atrophy and reappear when the celiac disease is controlled and the ridges begin to grow again.

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6 Medical Disorders with Associated Dermatoglyphic Abnormalities

Among the very large number of reports describing dermatoglyphics in medical disorders, only a few are well substantiated and based on large enough series to justify the conclusion that the dermatoglyphics are abnormal. Isolated case reports, patients in whom the diagnosis was in doubt and disorders for which there is no consensus among observers that the dermatoglyphics are indeed unusual were not included in this chapter. Because different chromosomal aberrations might exert independent effects on dermatoglyphics, most reports of patients with more than one chromosomal defect, such as patients with translocations, were excluded. The disorders included represent homogeneous groups, insofar as possible, and unless stated otherwise the dermatoglyphic data reflect pooled information from all available sources on such homogeneous groups.

Congenital Malformations of Hands and Feet

Because ridge differentiation is closely associated with embryogenesis of the limbs, the most obviously abnormal dermatoglyphics can be expected to be found in individuals with malformed hands and feet. Indeed, all anomalous extremities show an unusual epidermal ridge arrangement with the magnitude of ridge abnormality roughly proportional to the skeletal defects. However, independent of the degree of limb distortion, certain dermatoglyphic topological rules still pertain. A mathematical relationship between the number

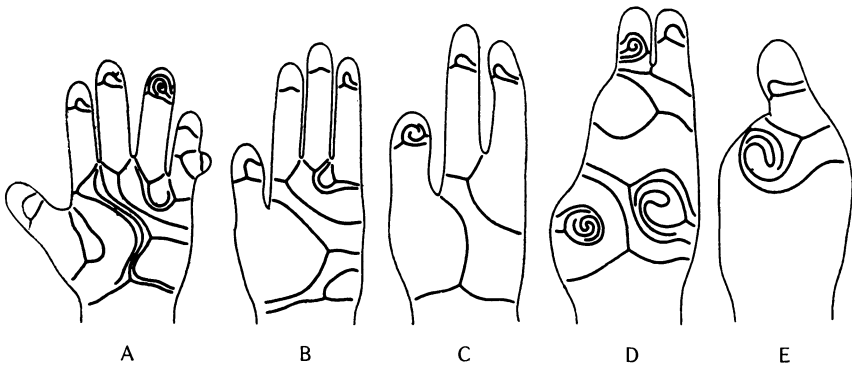


FIGURE 6.1 Diagrams of various malformed hands illustrating the relationship between the number of triradii, loops and digits ($T + 1 = L + D$). A, $13 + 1 = 8 + 6$; B, $7 + 1 = 4 + 4$; C, $6 + 1 = 4 + 3$; D, $9 + 1 = 8 + 2$; E, $3 + 1 = 3 + 1$.

From Penrose, L. S.: Dermatoglyphic topology. *Nature*, 205:544, 1965.
Courtesy of *Nature*.

of loops and triradii also remains intact even in gross degrees of congenitally deformed hands and feet, as illustrated in Figure 6.1. This relationship has been expressed by Penrose (1965) in the equation

$$T + 1 = L + D$$

where T is the number of triradii, L the number of loops, and D the number of digits. The formula is applicable to the hands and feet regardless of the number of digits present. The principles of dermatoglyphic topology (Penrose, 1965) as outlined in Chapter 3 must be observed. For example, each whorl is considered as two loops and the extralimital triradii are included in counting the triradii.

However, Dieker and Opitz (1969, p. 76) have pointed out that the formula does not apply in zygodactyly because “variable degrees of severity of this defect may reduce the number of triradii shared by the two affected adjacent digits to zero.”

THALIDOMIDE EMBRYOPATHY

Thalidomide is an agent with a profound teratogenic effect on limb development. The finding of grossly distorted epidermal ridge patterns, therefore, is hardly surprising. In a large study (Pfeiffer and Schulte zu Berge, 1964) of children with phocomelia resulting from thalidomide embryopathy, many unusual dermatoglyphic features were observed. Among the most striking were absence of the

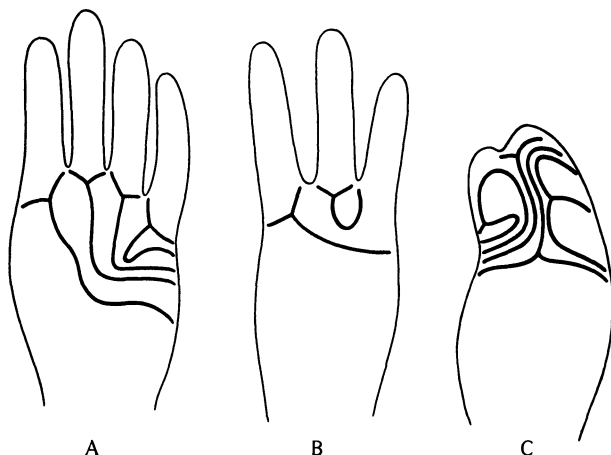


FIGURE 6.2 Schematic drawings representing different degrees of severity of hand malformations in thalidomide embryopathy.

A and B according to Pfeiffer, R. and Schulte zu Berge, U.: *Z. Mensch. Vererb. Konstitutionsl.*, 37:677, 1964; C according to Holt, S. B.: *J. Med. Genet.*, 9:448, 1972.

axial triradii accompanied by transverse flow of the palmar ridges, shifted and doubled digital triradii, abnormal course of the main lines, and ridge dissociation. Palmar flexion creases were also often found to be abnormal. The thenar crease was frequently missing or reduced in size and a tendency toward a single transverse crease was noted. The latter finding confirmed an earlier observation by Davies and Smallpeice (1963), who reported single transverse creases as a predominant feature on the palms of the thalidomide-damaged infants. Some of the dermatoglyphic features associated with hand malformations caused by thalidomide embryopathy are illustrated in Figure 6.2.

ABSENCE OR HYPOPLASIA OF THE THUMBS

Absent or hypoplastic thumbs occur occasionally as isolated anomalies but usually they are associated with other congenital malformations and syndromes, such as Holt–Oram syndrome, thalidomide embryopathy, Fanconi’s anemia, trisomy 18, and ring-D chromosome. Whether the absence of the thumb is the only anomaly or whether it is associated with any of a variety of other congenital anomalies, characteristic dermatoglyphic abnormalities are present. The ridge configurations in the proximal palm are dis-

torted, the ridges run transversely across the palm, and the axial triradius and the thenar crease are missing (Holt, 1968, 1972; Herrmann *et al.*, 1969; Guanti *et al.*, 1971). Hypoplastic thumbs are associated with less marked dermatoglyphic variations, according to the degree of hypoplasia. The axial triradius may be shifted from its usual position and, depending on the mobility of the phalangeal and metacarpophalangeal joints, the appropriate flexion creases may be rudimentary or missing.

Dermatoglyphics may present a clue as to whether the triphalangeal thumb is indeed a thumb or a duplicated index finger associated with absence of the thumb. The clue is provided by the presence or absence of an extra digital triradius and the main line originating from it. This triradius, referred to as an "extra *a*" by Temtamy and McKusick (1969), is present only if the questionable digit is the index finger.

TRIPHALANGY OF THE THUMBS

The triphalangeal thumb is usually thin and fingerlike and is frequently displaced distally to the level of other digits. The thenar eminence tends to be hypoplastic and shifted distally. Triphalangy of the thumbs is rarely observed as an isolated phenotypical defect. More often, it is found in such conditions as Holt–Oram syndrome (Holmes, 1965; Gall *et al.*, 1966; Massumi and Nutter, 1966; Poznanski *et al.*, 1971; Kaufman *et al.*, 1974) and thalidomide embryopathy (Lenz *et al.*, 1964; Pfeiffer and Schulte zu Berge, 1964) and occasionally in congenital hypoplastic anemia (Aase and Smith, 1969; Murphy and Lubin, 1972; Jones and Thompson, 1973), trisomy 13 (Poznanski *et al.*, 1971), and miscellaneous other abnormalities.

Triphalangeal thumbs are associated with distorted palmar dermatoglyphics (Bellelli, 1939a; Aase and Smith, 1969; Qazi and Smithwick, 1970; Jones and Thompson, 1973). The epidermal ridges tend to be transversely aligned across the palm and the axial triradii are either absent or displaced distally and radially. Single transverse palmar creases have been observed (Murphy and Lubin, 1972). There is a paucity of data concerning the fingertips. Radial loops, rarely found on digits other than the second in normal individuals, have been reported on the triphalangeal thumbs (Bellelli, 1939a). Jones and Thompson (1973) found three radial loops on the fingertips of a patient with triphalangeal thumbs and congenital hypoplastic anemia. In another patient with multiple anomalies, in-

cluding triphalangeal thumbs and great toes, hypoplasia of nails and teeth, deaf-mutism, convulsive disorder, and mental retardation (Qazi and Smithwick, 1970), simple arches were observed on all ten fingertips and nine toes. The patient had whorls on both thenar palmar areas.

HOLT-ORAM SYNDROME

Holt-Oram syndrome is a disorder consisting of skeletal and cardiovascular abnormalities. Skeletal anomalies include abnormal, usually hypoplastic, triphalangeal or even missing thumbs and occasionally hypoplasia of the radius or peromelia. Hypoplasia of the clavicles and narrow shoulders are frequently present. Among the cardiovascular defects, atrial and ventricular septal defects are most common.

TABLE 6.1. Dermatoglyphics in Holt-Oram syndrome

TRAIT	NUMBER	PERCENT
Fingertip patterns		
Arch	5	2.0
Radial loop	18	7.2
Ulnar loop	98	39.2
Whorl	113	45.2
Missing or unknown	16	6.4
Palmar patterns ^a		
Thenar/I ₁	0	0.0
I ₂	3	12.5
I ₃	4	18.2
I ₄	9	37.5
Hypothenar	16	53.3
Axial triradius ^a		
Proximal (normal)	11	16.2
Distally displaced	32	47.0
Absent	25	36.8
Thenar crease ^a		
Normal	15	53.6
Rudimentary	5	17.8
Absent	8	28.6
Single transverse palmar crease ^a	24	60.0

^a Number of palms with the trait.

Dermatoglyphics of the patients are obviously abnormal (Table 6.1). Among the unusual features are those associated with missing, hypoplastic, or triphalangeal thumbs, such as axial triradii that are missing or distally and radially shifted, a missing or rudimentary thenar crease, and transversely oriented epidermal ridges on the palms. In addition, pooling of the dermatoglyphic data revealed unusual frequencies of fingertip pattern types. Whorls were considerably increased in frequency, whereas ulnar loops were correspondingly decreased. The TFRC was generally increased, with a mean value of approximately 175.¹ A family reported by Sánchez Cascos (1967) with a low TFRC was an exception. However, the unaffected father had a TFRC of 47, the lowest of the whole family, whereas the mean TFRC of the four family members with the Holt–Oram syndrome was approximately 107.¹

A majority of the palms of affected individuals showed abnormal distal palmar creases, mostly a single transverse crease in its full or transitional forms.

Dermatoglyphic data (as listed in Table 6.1) were pooled from the following sources: Chang (1967), Emerit *et al.* (1965, 1968), Holmes (1965), Massumi and Nutter (1966), Rosner and Aberfeld (1970), Rybak *et al.* (1971), Sánchez Cascos (1967) and our own unpublished data.

ANONYCHIA

Anonychia is characterized by nail anomalies that usually include complete absence of the nail on the index and middle fingers, presence of minute portions of the thumbnail, and a less diminished nail on the ring finger. The nail of the little finger is usually normal. Anonychia on the toes parallels the anonychia of corresponding fingers. Anonychia can be associated with ectrodactyly, syndactyly, or polydactyly.

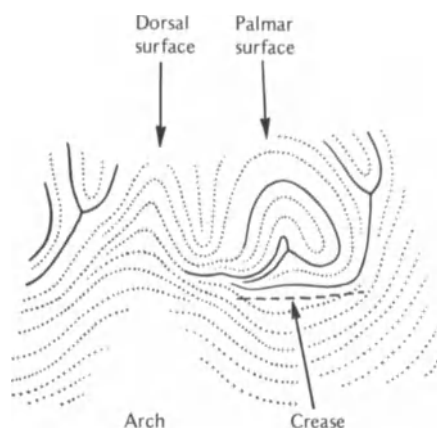
With absence or hypoplasia of the nails, the ridges extend over the dorsal area normally covered by the nail (Penrose, 1965). This is well illustrated on a rolled print of an affected finger (Figure 6.3). The ridged skin can cover not only the complete surface of the distal phalanx but in some fingers even the whole surface of the middle phalanx (Holt, 1968).

A similar but lesser degree of extension of the ridges is found in

¹ An exact mean TFRC could not be calculated because of the missing fingers in some individuals; an estimation has been used for the missing digits.

FIGURE 6.3 A rolled fingertip in anonychia. Print was rolled more than 360° so that the pattern begins to recur.

From Holt, S. B.: *The Genetics of Dermal Ridges*, 1968. Courtesy of Charles C Thomas, Publisher, Springfield, Illinois.



apical dystrophy (brachydactyly type B) and in the nail–patella syndrome (onycho-osteodysplasia), an integral part of which is the dysplasia of the nails of the thumb and index fingers.

DISTAL PHALANGEAL HYPOPLASIA

Hypoplasia of the distal phalanges is rarely observed as an isolated anomaly. More frequently, it is associated with other phenotypic disorders and syndromes, such as brachydactyly, onycho-osteodystrophy, pseudohypoparathyroidism (Albright osteodystrophy), the hand–foot–uterus syndrome, Sorsby syndrome (macula coloboma and brachydactyly), and others. A common dermatoglyphic finding in patients with distal phalangeal hypoplasia is a high proportion of arches on fingertips and toes whether the abnormality appears as a single defect or is associated with multiple clinical anomalies. Finding an arch pattern on all ten fingertips is not exceptional (Jongbloet and van Kempen, 1968; Qazi and Smithwick, 1970; Walbaum *et al.*, 1970; Rüdiger *et al.*, 1971; Robinow and Johnson, 1972; Cantwell, 1975). A close association between hypoplastic distal phalanges and a high incidence of arches and low ridge-count loops was pointed out by Robinow and Johnson (1972). They found arch patterns on 70 percent of the fingertips of individuals with distal phalangeal hypoplasia. None of the patients had any fingertip whorls. The authors offered a hypothesis that the hypoplastic phalangeal skeleton was associated with underdevelopment of the digital pads during embryogenesis when the dermal ridges were developing.

BRACHYDACTYLY

Brachydactyly is a condition characterized by reduced length of the fingers and toes because of shortening of any of their bony components, the metacarpals or phalanges. There are several types of brachydactyly, named according to specific shortening of certain parts of the digits (Table 6.2).

In type A brachydactyly, the middle phalanges are short, rudimentary, and sometimes fused with the terminal phalanges. Proximal phalanges of the thumbs and great toes are short. Penrose and Holt (1966) reported unusual dermatoglyphic features in a family of seven members who were affected with this type of brachydactyly (Table 6.3). Hoefnagel and Gerald (1966) had described

TABLE 6.2. Classification of brachydactyly

TYPE OF BRACHYDACTYLY	FINGER AND TOE INVOLVEMENT
A ₁	All the middle phalanges are rudimentary and sometimes fused with the terminal phalanges; proximal phalanges of the thumbs and big toes are short
A ₂	The second fingers and second toes are short, other digits more or less normal; the end of the second finger usually deviates radially
A ₃	Short middle phalanx of the fifth digit
A ₄	Short middle phalanx of the second and fifth digits; if the fourth digit is affected, it usually has an abnormally shaped middle phalanx that results in the radial deviation of the distal phalanx
A ₅	Absent middle phalanges of digits II to V with nail dysplasia; terminal phalanx of the thumb duplicated
B	Middle phalanges short, terminal phalanges rudimentary or absent; the thumbs and big toes usually deformed; symphalangism
C	Middle phalanges and some metacarpals shortened; deformed middle and proximal phalanges of the second and third digits; the fourth digit may be essentially normal, i.e., the longest digit
D	“Stub thumbs”; short and broad terminal phalanges of thumbs and great toes
E	Mainly shortening of the metacarpals and metatarsals

TABLE 6.3. Dermatoglyphic traits in a family with brachydactyly^a

TRAIT	AFFECTED MEMBERS		UNAFFECTED MEMBERS	
	NUMBER	VALUE	NUMBER	VALUE
Fingertip patterns				
Arch	61	87.1%	11	22.0%
Radial loop	5	7.1%	1	2.0%
Ulnar loop	4	5.7%	36	72.0%
Whorl	0	0.0%	2	4.0%
Mean TFRC				
Males	4	19.5	3	87.7
Females	3	3.0	2	61.0
Total	7	12.4	5	77.0
Mean summed <i>a-b</i> ridge count	7	98.0	5	100.0
Mean summed <i>atd</i> angle	7	111.4°	5	94.6°

^a According to Penrose and Holt (1966).

the kindred clinically. On the fingertips, most of the patterns in the brachydactylous individuals were arches, whereas ulnar loops were markedly diminished in frequency and no whorls were found. All five radial loops were observed on thumbs of the affected individuals. This is a highly unusual finding in controls and was not present in any of the unaffected family members. A tibial loop on the great toe is the equivalent of a radial loop on the thumb. It was found six times on the great toes (43 percent of all patterns) of the individuals with brachydactyly but in none of the normal family members. The mean TFRC was extremely low even in comparison with the already low TFRC of the unaffected individuals. The mean *atd* angle was markedly higher in the brachydactylous individuals compared to the normal members of the kindred. The mean *a-b* ridge count was increased in both affected and unaffected individuals. On the feet, the persons with brachydactyly showed an increased number of whorls on the toes and a somewhat increased pattern intensity on the ball area of the sole.

An increased frequency of fingertip arches was observed in a family with the hand-foot-uterus syndrome (Stern *et al.*, 1970). The excess in arches was not as pronounced as in the kindred reported by Penrose and Holt (1966). Part of this syndrome is brachymesophalangy with clinodactyly of the fifth fingers (type A₃ brachydactyly) and short first metacarpals. On the fingertips, 40

percent of all patterns were arches and the rest of the patterns were small, thin loops. The TFRC was drastically reduced, with a mean TFRC of 23.6 in three affected males. The person with the most affected fingers also had the most abnormal fingertip dermatoglyphics, consisting of nine arches and a TFRC of 3. The unaffected family members had a TFRC and a distribution of fingertip patterns, including fingertip whorls, like that of controls. All eight affected, but only one of five unaffected, members of the kindred had a distally displaced and ulnar shifted axial triradius. The thenar eminence was hypoplastic and usually without a pattern. Similarly, the hypothenar area was usually patternless. On the basis of dermatoglyphics alone, the authors encountered no difficulty in deciding whether they were dealing with an affected or an unaffected individual.

Unusual dermatoglyphics were common in type B brachydactyly. In this anomaly, the middle phalanges are short and the distal phalanges rudimentary or absent. The thumbs and great toes are usually deformed, often broad and flat with nails sometimes split and doubled. The nails are often absent or rudimentary but may be normal in the least affected fingers. Symphalangism is frequent. This type of brachydactyly is sometimes called ectrodactyly, apical dys trophy, or, if in association with syndactyly, a symbrachydactyly.

Abnormally arranged epidermal ridges accompany the anomalous terminal phalanges (MacArthur and McCullough, 1932; Degenhardt and Geipel, 1954; Fuhrmann *et al.*, 1965; Battle *et al.*, 1973). The fingertip patterns are displaced to the extreme end of the finger. The triradii are usually missing. If present, they are shifted distally to the tip of the finger. On fingers with absent or reduced nails, the epidermal ridges are usually arranged in concentric circles with no triradii (Figure 6.4A), the ridged skin extending over the area normally occupied by the nail. The longer fingers, particularly those with at least hypoplastic nails, show transitions toward more normal patterns. The broadened thumbs may have an apical triradius at the tip, as seen sometimes on the broad thumbs in the Rubinstein-Taybi syndrome. Apart from these dermatoglyphic anomalies, which are obviously associated with the finger malformations, several other unusual dermatoglyphic traits have been reported. In a kindred of nine affected and two unaffected individuals (Degenhardt and Geipel, 1954), there was an increase in both arches and whorls on the fingertips, whereas the ulnar loops were considerably diminished in frequency among the affected members of the family. Fingertip pattern distribution in the

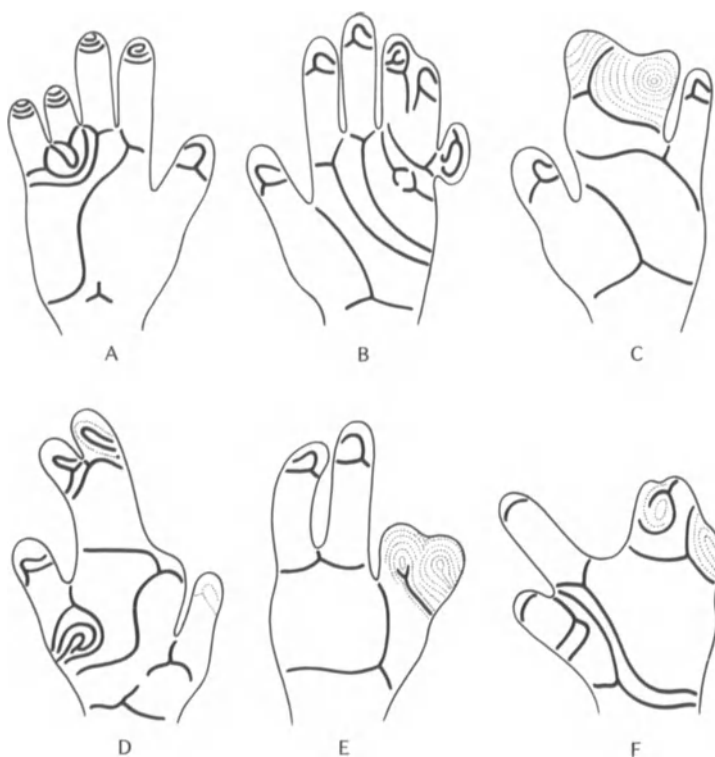


FIGURE 6.4 Dermatoglyphics in various hand malformations.

A, Brachydactyly with shortened middle phalanges and rudimentary or absent distal phalanges; B, syndactyly of digits IV and V with a polydactylous digit V; C, syndactyly of digits II-IV; D, grossly malformed hand in the de Lange syndrome with partial syndactyly of digits II and III, missing digit IV and abnormally positioned digit V; E, duplication of the thumb and absence of two digits, apparently II and III; F, grossly malformed hand with coexisting syndactyly, polydactyly and brachydactyly.

A according to Battle, H. I., *et al.*: *Ann. Hum. Genet.*, 36:415, 1973; B, C, E, and F according to Cummins, H.: *Am. J. Anat.*, 38:89, 1926; D according to Berg, J. M., *et al.*: *J. Med. Genet.*, 4:184, 1967.

normal members was not unusual. In another family (Fuhrmann *et al.*, 1965), an increased number of fingertip whorls and a high TFRC were found in a proband and his affected mother when compared to controls and the unaffected father. Palmar dermatoglyphic findings were not unusual. The sole prints of the mother and son were similar. Each had four whorls and six fibular loops on the toes

and whorls bilaterally on hallucal areas. In a large kindred of 18 affected and 34 normal individuals (Battle *et al.*, 1973), a strikingly high frequency of thenar/first interdigital patterns was observed among the affected family members. Nearly 56 percent of their palms had large patterns in both the thenar and the first interdigital area of each palm. In general, the thenar/first interdigital patterns occurred in subjects with broad or bifid thumbs, an association observed also in the Rubinstein-Taybi syndrome. Normal members of this brachydactylous kindred had only small, vestigial patterns without triradii in the thenar/I₁ area. The affected individuals frequently had single transverse palmar creases but did not differ from normal in the position of the axial triradius.

Brachydactyly type C involves the middle phalanges and some metacarpals. The middle and proximal phalanges of the second and third digits are usually deformed, whereas the fourth digit may be essentially normal, becoming the longest digit. A kindred with this type of brachydactyly and abnormal dermatoglyphics was reported by Robinson *et al.* (1968). Among ten affected members of the family, there were four radial loops on fingers other than the second (three on the third and one on the fourth). No displaced radial loop patterns were found on the fingertips of the 12 unaffected individuals. The thenar/I₁ patterns were more frequent among the brachydactylous individuals than among their healthy relatives. Otherwise, no striking differences were found on the palms of the subjects investigated.

In pseudohypoparathyroidism (Albright osteodystrophy) the shortness of the digits is usually caused by shortened metacarpals and metatarsals, resembling brachydactyly type E. However, phalangeal bones may also be short. Forbes (1964) reported dermatoglyphic findings in 19 patients with pseudohypoparathyroidism and pseudopseudohypoparathyroidism. There was some increase of fingertip arches (12.5 percent of the fingertip patterns) and hypothernar patterns (48 percent). Distal axial triradii were present in 56 percent of the palms and the mean *atd* angle was 55°. Only one full and several transitional simian creases were observed.

CAMPTODACTYLY

Camptodactyly is a condition of permanent flexion of one or both interphalangeal joints. Dermatoglyphics in two syndromes with multiple anomalies including camptodactyly were reported by Good-

man *et al.* (1972). The first syndrome consisted of camptodactyly with muscular hypoplasia, skeletal dysplasia, and abnormal skin creases which occurred in two siblings; two other siblings and the parents in this family were normal. The affected siblings had many fingertip whorls that extended distally on the terminal phalanges, whereas the triradii of these whorls were on the middle or even proximal portions of the digits. The TFRC was very high in both affected individuals (271 and 350), about twice as high as in their healthy relatives. The *a-b* ridge count was increased in the male proband but somewhat decreased in his affected sister. The maximal *atd* angles were considerably increased. The patients displayed vertically oriented main lines and the D line either was aborted or exited on the ulnar edge of the palm. There was considerable distortion of the epidermal ridges. The digital creases over the permanently flexed joints on the fingers and toes were absent. A large number of coarse, horizontally oriented secondary creases covered the palms and soles. There was an increased number of whorls on the toes of the patients but none occurred on the toes of the healthy siblings. Dermatoglyphics therefore provided a means of distinguishing between the affected and unaffected members of the family.

The second syndrome described by Goodman *et al.* (1972) consisted of camptodactyly with fibrous tissue hyperplasia and skeletal dysplasia. Unlike the first syndrome, no dermatoglyphic abnormalities were observed among the members of this family.

SYNDACTYLY

The phenotypic expressions of syndactyly vary greatly from minor skin webbing to complete cutaneous fusion of the fingers or toes with or without the fusion of phalangeal and even metacarpal and metatarsal bones. Syndactyly can be found as an isolated congenital malformation but often it constitutes a part of another syndrome, such as Apert syndrome (acrocephalosyndactyly type I), Carpenter syndrome (acrocephalopolysyndactyly), de Lange syndrome, oculodontoosseous syndrome, oral-facial-digital syndrome, and Smith-Lemli-Opitz syndrome, among others.

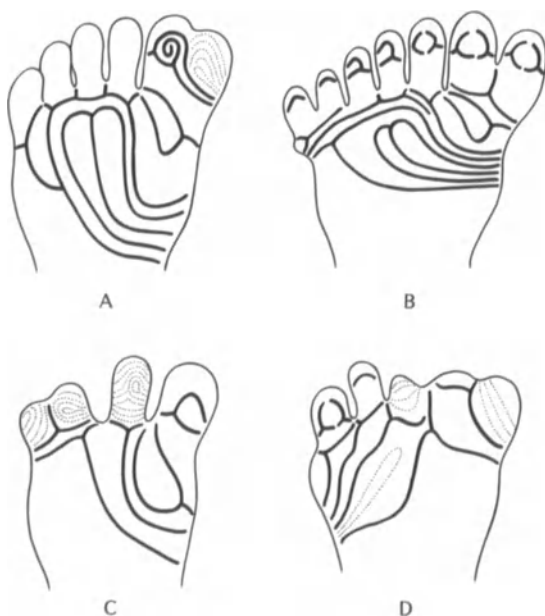
Dermatoglyphics of the syndactylous digits present an excellent source of material for the study of relationships between embryogenesis of the hands and feet and the epidermal ridges covering their volar aspects. Numerous studies have been devoted to this

association (Cummins and Sicomo, 1923; Koenner, 1933; Bellelli, 1939b; Grebe, 1940; Dankmeijer and Waltman, 1947; Temtamy and McKusick, 1969).

The palmar area proximal to the webbing between the digits is covered with transversely aligned ridges and an interdigital triradius generally underlies the web (Figures 6.4E and 6.5A and C). This triradius replaces two digital triradii, which are present normally. In cases of zygodactyly of the third and fourth fingers, therefore, the usual triradii *b* and *c* are missing and are replaced by an interdigital triradius *bc*, which lies between and can be considered a fusion of the normal *b* and *c* triradii. This interdigital triradius may be shifted distally in the more severe degrees of webbing (Figure 6.4D). The fingertip patterns vary with the degree of syndactyly from relatively normal frequencies to highly aberrant ones. Figure 6.4B represents a hand with combined syndactyly and polydactyly. The fingertip patterns of the syndactylous digits four and five are present but the direction of the type lines is aberrant. The palmar configuration proximal to the fused digits is also abnormal. Figure 6.4C represents a hand with complete syndactyly of digits II, III, and IV. There is only one fingertip pattern, a whorl, on the fourth digit, and the fingertip configuration of digits II and III is unlike any normal digital configuration. There is an interdigital triradius situated be-

FIGURE 6.5 Dermatoglyphics in various foot malformations. A, Duplication of the thumb with syndactyly of the thumb complex; B, polydactyly with supernumerary digits I and V and a low mound at the base of the last digit indicating a possibly abortive eighth digit; C, oligodactyly with apparently missing digit III and syndactyly of digits IV and V; D, grossly malformed foot with multiple anomalies.

According to Cummins, H.: *Am. J. Anat.*, 38:89, 1926.



tween the fused digits and the fifth digit. Otherwise, there are no digital triradii or any patterns on the palm. Figure 6.4D illustrates the grossly malformed hand of a patient with the de Lange syndrome. It includes partial syndactyly of digits II and III with an interdigital triradius shifted distally toward the fingertips. An interdigital triradius is also present on the hand in Figure 6.4E, although no detectable syndactyly of the involved fingers is present. Because the interdigital triradii indicate a close developmental relationship between adjacent digits, presence of such a triradius can be considered as a minor manifestation of zygodactyly.

POLYDACTYLY

Extra digits on the hand or foot are associated with dermatoglyphic aberrations. Extra digits that are attached to other digits (Figure 6.5A) may have two patterns, one for each potential fingertip, or the pattern on one digit may extend to the extra digit. A well-developed supernumerary digit has its own fingertip pattern (Figure 6.4B). Cummins (1926) observed a relationship between the degree of development of the duplicated digits and the type of associated fingertip pattern. He found that an imperfectly developed extra digit or a digit joined to its neighbor has a pattern different from the adjacent digit (Figure 6.5A). A completely duplicated and separated extra digit tends to have the same type of pattern as the digit it duplicates. For example, the duplicated great toe and the little toe in Figure 6.5B share the same type of pattern as their equivalents. An extra triradius that may represent an abortive eighth digit may also be noted in Figure 6.5B near a low mound at the base of the last toe. An extra digital triradius is generally found at the base of the supernumerary digit. The extra triradius may represent the only evidence of an extra digit, such as a pedunculated postminimus that has been surgically removed or has fallen off spontaneously (Cummins, 1932). The extra triradius therefore constitutes an unmistakable stigma of polydactyly and so can be of clinical significance.

OTHER GROSS HAND AND FOOT MALFORMATIONS

Because epidermal ridges reflect the embryonic development of the hands and feet, such gross malformations of the distal parts of the limbs as ectrodactyly or peromelia are accompanied by obvious dermatoglyphic aberrations (Cummins, 1923; Bindseil and Grimm,

1942; MacKenzie and Penrose, 1951; Degenhardt and Geipel, 1952; Schretlen and Hustinx, 1965; Dignan *et al.*, 1967; Hall *et al.*, 1969). The extent of dermatoglyphic alterations can be great, depending on the severity of the hand or foot malformations. Usual dermatoglyphic features, such as digital triradii and interdigital patterns, may be so disturbed that they cannot be identified. Figures 6.4F and 6.5D illustrate bizarre ridge arrangement on a grossly malformed hand and foot of an acephalic “monster” (Cummins, 1923). The lower limb malformations consist of coexistent osseous syndactyly, polydactyly, and brachydactyly. In spite of the abnormal epidermal configurations, ridge directions conform to the anomalous molding of the hand and foot structures.

Dermatoglyphics generally do not play an important part in clinical diagnosis of hand and foot abnormalities as these can be more precisely identified by other means such as an x-ray examination. However, understanding the relationship between embryonic development and ridge formation is necessary for a meaningful interpretation of the significance of dermatoglyphics in limb malformations and these relationships may apply to other medical disorders as well. Use of dermatoglyphics to diagnose hand and foot malformations may reveal even minor abnormalities that may otherwise escape attention.

Autosomal Trisomies

TRISOMY 21 (DOWN SYNDROME)

Down syndrome is a relatively common syndrome of multiple congenital malformations. In the majority of cases, the syndrome is caused by trisomy of chromosome 21, but about 6 percent of the patients show either a translocation or a mosaic trisomy involving chromosome 21. There is a striking similarity in the physical appearance of the patients. Their facial features are usually flattened, with oblique palpebral fissures, flat occiput, brachycephaly, small nose, depressed nasal bridge, speckled iris, and epicanthal folds. The mouth is usually held open and reveals a large furrowed tongue. The palate is commonly short and narrow and dental abnormalities are observed in most cases. The ears are often dysplastic and the neck is short and broad. The limbs are characteristically short. The hands are broad and square and the fingers are short. The fifth digit is often incurved (clinodactyly) with an especially short middle phalanx. The latter trait is often accompanied by a single digital

flexion crease. A single transverse flexion crease is found frequently on the palms. On the feet, there is often an increased space between the first and second toes and frequently a deep plantar furrow is observed in this area. The patients are almost without exception mentally retarded although the degree of retardation varies. They very often show laxity of joint movement and hypotonia. Congenital heart disease is a frequent feature of the syndrome.

Almost four decades ago, long before the chromosomal basis of Down syndrome was established, Cummins (1936, 1939) pointed out characteristic differences in dermatoglyphic features in patients with Down syndrome compared to the normal population. These original findings have since been confirmed by many investigators, even in patients of different racial and ethnic stock, and additional dermatoglyphic abnormalities have been identified (Figure 6.6).

A marked increase of ulnar loops on the fingertips is virtually a constant feature of the dermatoglyphics in Down syndrome (Table 6.4). The loops tend to be vertically oriented and L shaped. Frequently, ulnar loops are found on all ten fingertips of patients (Table 6.5). The elevated frequency of ulnar loops is associated with a decrease of whorls, arches, and radial loops. Moreover, when radial loops are present, they tend to be shifted from the index finger, where they are normally found with highest frequency, to the fourth or fifth digit (Table 6.6). The usual sex differences in frequency of the fingertip patterns are much less apparent between males and females with Down syndrome.

The total finger ridge count is lower and its variability is smaller than in the normal population (Table 6.7). The tendency toward decreased dermatoglyphic variability in the ridge counts is also common for other dermatoglyphic traits in Down syndrome (Cummins, 1939).

The mean values of the *a-b* ridge count do not appear to differ significantly from those of controls. Although Fang (1949) reported significantly lower mean *a-b* ridge counts in Down syndrome patients than in controls, his finding was not confirmed in a larger series (Holt, 1968a; Table 6.8). Shiono and Kadowaki (1971) found the mean *a-b* ridge counts of Japanese patients with Down syndrome slightly lower than in controls, but the differences were not significant.

The hypothenar area shows an increased frequency of patterns. Many of the patterns are large and often terminate distally by a high axial triradius. Walker (1958) observed hypothenar patterns in approximately 85 percent of the North American patients with Down

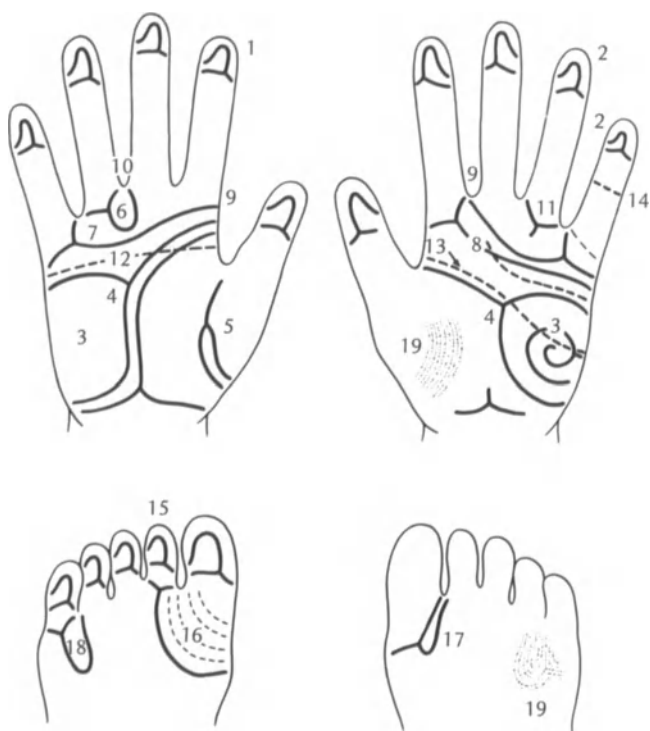


FIGURE 6.6 Dermatoglyphic features of Down syndrome.

(1) Ulnar loops considerably increased in frequency, often found on all ten fingertips, loops often high and L shaped. (2) Radial loops shifted to fourth and fifth digits. (3) Large hypothenar patterns, predominantly of the ulnar type, associated with (4). (4) Distal axial triradius, high maximal *atd* angle. (5) Thenar patterns decreased in frequency, size and complexity. (6) I_3 patterns increased in frequency. (7) I_4 patterns decreased in frequency. (8) Transverse alignment of main lines and ridges in the distal palm (high main-line index). (9) D line terminating in I_2 area or even on the radial border of the palm. (10) I_3 loop formed by a recurve of the C line. (11) Absent and abortive C lines increased in frequency. (12) Single transverse palmar creases increased in frequency. (13) Sydney lines increased in frequency. (14) Single interphalangeal crease on the fifth digit. (15) Fibular loops increased in frequency. (16) Arch tibial configuration in the hallual area markedly increased in frequency (very rare in controls). (17) Distal loops in the hallual area mostly small (in controls, predominantly large loops). (18) Distal loops in area IV increased in frequency. (19) Ridge dissociation.

TABLE 6.4. Percent of each fingertip pattern type in Down syndrome and controls

POPULATION	PATTERN TYPE ^a								SOURCE
	WHORL		ULNAR LOOP		RADIAL LOOP		ARCH		
	D	C	D	C	D	C	D	C	
Canadian	20.1	28.4	75.1	61.2	2.5	4.9	2.3	5.6	Walker (1957)
Swedish	12.1	21.4	84.5	66.3	1.5	5.6	1.9	6.7	Beckman <i>et al.</i> (1962)
British	12.7	26.1	82.8	63.5	1.8	5.4	2.7	5.0	Holt (1964)
Italian	12.6	33.1	81.0	57.2	5.2	3.5	1.1	6.2	Giovannucci and Bartolozzi (1968)
Polish	13.6	35.5	85.2	56.4	0.8	4.2	0.4	3.6	Gebala <i>et al.</i> (1969)
Polish	17.0	29.4	80.2	61.8	2.0	4.5	0.6	4.4	Zajackowska (1969)
Japanese	20.8	48.3	76.3	46.7	2.1	2.7	0.8	2.3	Matsui <i>et al.</i> (1966)
Japanese	26.1	42.7	70.6	52.2	1.9	2.8	1.4	2.3	Fujita (1969)
Japanese	21.6	47.5	74.2	47.5	3.6	3.3	0.6	1.7	Shiono <i>et al.</i> (1969)
Chinese	18.3	46.6	77.0	49.1	3.6	2.3	1.1	2.1	Bryant <i>et al.</i> (1970)

^a D, Down syndrome; C, controls.

TABLE 6.5. Percent of individuals with ten ulnar loops on the fingertips in Down syndrome and controls

POPULATION	ULNAR LOOPS ONLY ON FINGERTIPS		SOURCE
	DOWN	CONTROLS	
British	34.5	5.5	Holt (1964)
Japanese	29.0	5.7	Shiono <i>et al.</i> (1969)

TABLE 6.6. Percent of radial loops on individual fingertips in Down syndrome and controls

POPULATION GROUP ^a	DIGIT										SOURCE	
	1		2		3		4		5			
	LEFT	RIGHT	LEFT	RIGHT	LEFT	RIGHT	LEFT	RIGHT	LEFT	RIGHT		
Canadian	D	0.6	0.1	2.3	1.7	1.7	1.1	5.1	5.7	2.9	4.6	Walker (1957)
	C	0.8	0.8	19.4	20.3	2.6	3.4	0.9	0.3	0.1	0.3	
British	D	0.4	0.3	2.8	0.3	1.0	0.7	5.2	4.5	1.4	1.3	Holt (1964)
	C	0.2	0.1	23.1	21.4	4.3	2.8	0.7	0.6	0.1	0.3	
British	D	0.0	0.0	3.1	0.5	0.0	0.0	2.1	7.7	1.0	1.6	Smith <i>et al.</i>
	C	0.0	0.0	18.5	19.0	1.0	1.5	1.0	0.0	1.5	1.0	(1966)
Italian	D	0.0	0.0	4.4	3.7	1.5	0.7	22.2	16.3	2.2	0.7	Giovannucci and
	C	0.0	0.0	14.4	14.0	2.6	1.2	0.8	1.3	0.4	0.5	Bartolozzi (1968)
Japanese	D	1.0	0.0	0.0	0.0	0.0	0.0	8.0	2.0	3.0	3.0	Fujita (1969)
	C	0.7	0.4	11.7	9.9	2.1	0.9	0.6	0.9	0.3	0.5	
Japanese	D	1.0	0.0	2.0	0.0	0.0	1.0	11.0	6.0	9.0	6.0	Shiono <i>et al.</i>
	C	0.2	0.0	12.6	16.0	1.4	1.8	0.5	0.7	0.0	0.0	(1969)
Chinese	D	0.0	1.9	1.9	0.0	0.0	0.0	9.4	7.6	9.4	5.7	Bryant <i>et al.</i>
	C	0.6	0.3	7.3	11.2	1.0	0.6	0.3	0.3	0.3	0.6	(1970)
Indian	D	0.0	0.0	0.0	0.0	0.0	0.0	5.7	2.9	0.0	0.0	Saksena <i>et al.</i>
												(1966)

^a D, Down syndrome; C, controls.

TABLE 6.7. Mean total finger ridge count in Down syndrome and controls

POPULATION	MALES		FEMALES		SOURCE
	DOWN	CONTROLS	DOWN	CONTROLS	
British	130.3	145.2	124.4	127.0	Holt (1963)
Italian	93.8	135.0	92.2	127.4	Giovannucci and Bartolozzi (1968)

syndrome, compared to only 12 percent among controls. Other investigators (Table 6.9) reported less striking but nevertheless significant differences in frequencies of hypothenar patterns between Down syndrome patients and controls. Most of the patterns in the hypothenar area of the patients were of the ulnar type (Table 6.10), whereas radial loops, which were the predominant hypothenar patterns in controls, were extremely diminished in frequency among patients with Down syndrome (Erne, 1953, cited by Loeffler, 1969; Plato *et al.*, 1973).

A distally displaced axial triradius, frequently associated with a hypothenar pattern, is a typical dermatoglyphic trait in Down syndrome. Cummins (1939) found a high axial triradius (t'') in 72 percent of his patients, which was approximately eight times more frequent than in normal individuals. Similarly, increased frequency of t'' triradii was observed by other investigators (Table 6.11). Although the frequency of t'' triradii was lower among Japanese and Chinese patients than among Caucasian individuals with Down syndrome, the ratio of t'' frequency between Japanese or Chinese patients and controls resembled that of the Caucasian series. Giovannucci and Bartolozzi (1968) noticed that the high axial triradii in their controls were always displaced toward the ulnar side, rather than toward the median line as observed in patients with Down

TABLE 6.8. Mean summed $a-b$ ridge count in Down syndrome and controls

POPULATION	DOWN SYNDROME		CONTROLS		SOURCE
	MALES	FEMALES	MALES	FEMALES	
British	76.9	79.2	83.0	83.0	Fang (1949)
British	85.4	85.6	85.5	84.9	Holt (1970)
Japanese	71.6	68.8	72.3	70.6	Shiono and Kadowaki (1971)

TABLE 6.9. Percent of palms with a hypothenar pattern in Down syndrome and controls

POPULATION	DOWN SYNDROME		CONTROLS		SOURCE
	LEFT	RIGHT	LEFT	RIGHT	
Swedish	58.6	61.5	32.0	35.2	Beckman <i>et al.</i> (1962)
Italian	69.8	78.8	—	—	Dallapiccola and Ricci (1967)
Japanese	33.0	31.0	3.0	2.1	Shiono <i>et al.</i> (1969)
North American	55.6	61.1	25.9	33.3	Plato <i>et al.</i> (1973)

syndrome. They found the t'' alone on the palm (i.e., without t or t') only in 5.2 or 2.4 percent of Down patients and controls, respectively, and therefore suggested that the term “accessory t'' ” rather than “a distal displacement of the axial triradius” described this unusual dermatoglyphic trait more appropriately.

As a result of the high axial triradius, significantly increased maximal *atd* angles are commonly found (Table 6.12). Penrose (1954) suggested that only 12 percent of the patients with Down syndrome would be diagnosed incorrectly if the width of the *atd* angle were used as the sole means of diagnosis.

In contrast to the increased frequency of hypothenar patterns, the thenar patterns in Down syndrome were decreased in frequency, size, and complexity (Table 6.13). The frequency of third interdigital area patterns on the palms of Down syndrome patients has been consistently found to be significantly higher than in comparable controls (Table 6.14). In keeping with the findings in normal

TABLE 6.10. Percent of ulnar loops among all palmar hypothenar patterns in Down syndrome and controls

POPULATION	DOWN SYNDROME	CONTROLS	SOURCE
Italian, Swiss	60	5	Erne (1953), cited by Loeffler (1969)
Italian	87.2	—	Dallapiccola and Ricci (1967)

TABLE 6.11. Percent of distally displaced axial triradii (t'') in Down syndrome and controls

POPULATION	DISTAL AXIAL TRIRADIUS (t'')		SOURCE
	DOWN	CONTROLS	
American Negro	47.6	2.6	Walker <i>et al.</i> (1963)
Canadian	90.0	—	Soltan and Clearwater (1965)
Canadian	81.5	11.8	Walker and Johnson (1965)
Italian	81.2	6.5	
Italian	87.0	8.3	Giovannucci and Bartolozzi (1968)
Japanese	46.3	4.3	Matsui <i>et al.</i> (1966)
Japanese	44.5	4.1	Shiono <i>et al.</i> (1969)
Chinese	58.8	1.6	Bryant <i>et al.</i> (1970)

series, there is a higher frequency of patterns in right than in left I_3 areas. Although the frequencies of I_3 patterns in Oriental patients with Down syndrome were lower than those of Caucasians with this syndrome, they were still significantly higher than those of controls (Table 6.14).

In the fourth interdigital area, pattern frequency was decreased as compared with controls, although the actual percentages of I_4 patterns varied in different studies (Table 6.15). As in controls, the left palms of patients with Down syndrome show a consistently higher frequency of patterns than the right palms.

At least several unusual dermatoglyphic traits in Down syndrome can be attributed to the unusual shape of the hands, which are characteristically broad and short. One such feature is the transverse alignment of ridges over the distal palmar area (Cummins, 1936), which was reflected by an increased value of the main-line index (Cummins, 1939; Giovannucci and Bartolozzi, 1968). In the majority of palms of Down syndrome patients, the D line terminated in the second interdigital area and the I_3 loop was formed by a recurve of the C line (Holt, 1970). Occasionally the D-line

TABLE 6.12. Percent of palms with maximal *atd* angle of 57° or larger in Down syndrome and controls

POPULATION	AGE GROUP (YEARS)	DOWN SYNDROME		CONTROLS		SOURCE
		MALES	FEMALES	MALES	FEMALES	
British	0-4	92.0	84.2	10.7	20.3	Penrose (1954)
	5-14	83.9	80.9	8.9	10.6	
	15+	79.6	81.2	6.9	7.7	
Swedish	2-4.5	78.2	75.5	25.8	21.2	Beckman <i>et al.</i> (1962)
Indian	0-13	100.0	100.0	—	—	Saksena <i>et al.</i> (1966)

termination could be traced to the radial border of the hand (Table 6.16), which is an extremely rare termination in normal individuals. Plato *et al.* (1973) demonstrated the transverse alignment of ridges in the distal region of the palm by tracing the terminations of the C line to the radial or ulnar side of the palm. They have found the radial/ulnar ratio, which indicates the degree of transverseness, to be ten times higher in patients with Down syndrome than in comparable controls. Other unusual C-line terminations, including an abortive or missing C line (*X*, *x*, *O*), have been found, respectively, in 25.5 and 7.2 percent of patients with Down syndrome (Cummins, 1939), in keeping with results in normal individuals.

TABLE 6.13. Percent of palms with a thenar pattern in Down syndrome and controls

POPULATION	DOWN SYNDROME		CONTROLS		SOURCE
	LEFT	RIGHT	LEFT	RIGHT	
Swedish	1.0	2.9	11.7	5.5	Beckman <i>et al.</i> (1962)
Italian	1.8	0.9	—	—	Dallapiccola and Ricci (1967)
British	4.0	0.7	9.3	6.0	Berg (1968)
North American	2.6	1.7	13.4	5.6	Plato <i>et al.</i> (1973)

TABLE 6.14. Percent of true patterns in the third interdigital area of the palms in Down syndrome and controls

POPULATION	DOWN SYNDROME		CONTROLS		SOURCE
	LEFT	RIGHT	LEFT	RIGHT	
British	52.2	83.9	25.7	48.2	Fang (1950)
Swedish	56.7	80.8	20.3	60.2	Beckman <i>et al.</i> (1962)
Canadian	54.0	85.4	31.3	55.5	Walker and Johnson (1965)
North American	60.2	79.1	25.7	46.3	Plato <i>et al.</i> (1973)
Italian	70.8	87.5	26.0	51.3	Walker and Johnson (1965)
Italian	65.1	80.6	25.2	43.0	Giovannucci and Bartolozzi (1968)
Indian	54.2	51.4	—	—	Saksena <i>et al.</i> (1966)
Japanese	24.0	43.8	3.0	13.9	Matsui <i>et al.</i> (1966)
Japanese	24.2	52.5	6.7	16.8	Shiono <i>et al.</i> (1969)
Chinese	19.6	49.0	6.1	29.3	Bryant <i>et al.</i> (1970)

TABLE 6.15. Percent of true patterns in the fourth interdigital area of the palms in Down syndrome and controls

POPULATION	DOWN SYNDROME		CONTROLS		SOURCE
	LEFT	RIGHT	LEFT	RIGHT	
North American	29.2	13.8	—	—	Cummins (1939)
Swedish	10.6	6.7	44.5	36.7	Beckman <i>et al.</i> (1962)
Japanese	30.3	23.2	56.7	50.0	Shiono <i>et al.</i> (1969)
North American	16.3	10.0	59.4	45.6	Plato <i>et al.</i> (1973)

TABLE 6.16. Percent of palms with a radial exit of main-line D in Down syndrome and controls

POPULATION	RADIAL EXIT OF D LINE		SOURCE
	DOWN SYNDROME	CONTROLS	
German	3.5	—	Geipel (1961)
Italian	3.9	—	Dallapiccola and Ricci (1967)
British	3.0	0.5	Holt (1970)

However, in other series (Table 6.17), absent C triradii and abortive C lines were found in considerably higher frequencies in patients with Down syndrome than in controls.

A single transverse flexion crease occurred on at least one palm in about one-half of all individuals with Down syndrome. The differences in the frequency of simian lines in various series of Down syndrome patients were, at least to some degree, attributable to the fact that some investigators counted only complete simian creases, whereas others also included transitional types in their calculations. When transitional patterns were included, the simian line frequency was usually considerably higher (Table 6.18).

Recently, increased frequencies of Sydney lines have been reported in Down syndrome (Table 6.19). However, the excess of Sydney lines in patients compared to controls was not so striking as the excess of simian lines.

A single interphalangeal crease on the fifth finger was associated with aplasia or hypoplasia of the middle phalanx in Down syndrome. The single crease is virtually never observed in normal

TABLE 6.17. Percent of palms with an abortive or missing main-line C in Down syndrome and controls

POPULATION	DOWN SYNDROME		CONTROLS		SOURCE
	LEFT	RIGHT	LEFT	RIGHT	
Italian	48.6	13.0	—	—	Dallapiccola and Ricci (1967)
Japanese	48.5	22.1	14.1	10.4	Shiono <i>et al.</i> (1969)
Chinese	54.9	17.7	15.0	14.3	Bryant <i>et al.</i> (1970)

TABLE 6.18. Percent of palms with a single transverse palmar flexion crease in Down syndrome and controls

POPULATION	SIMIAN CREASE ^a						SOURCE
	COMPLETE		TRANSITIONAL		TOTAL		
	D	C	D	C	D	C	
Swedish	28.8	1.6	30.3	5.5	59.1	7.1	Beckman <i>et al.</i> (1962)
Canadian	40.0	—	—	—	—	—	Soltan and Clearwater (1965)
Italian	48.6	—	—	—	—	—	Dallapiccola and Ricci (1967)
Italian	53.3	1.0	32.6	3.7	85.9	4.6	Giovannucci and Bartolozzi (1968)
North American	26.4	3.6	4.9	1.1	31.3	4.7	Plato <i>et al.</i> (1973)
Indian	51.4	—	—	—	—	—	Saksena <i>et al.</i> (1966)
Japanese	48.0	9.8	—	—	—	—	Shiono <i>et al.</i> (1969)
Chinese	41.5	9.9	—	—	—	—	Bryant <i>et al.</i> (1970)

^a D, Down syndrome; C, controls.

individuals but occurs in approximately 20–30 percent of patients (Table 6.20). Lower frequencies of single interphalangeal creases were reported in Indian patients by Saksena *et al.* (1966), and among Chinese patients with Down syndrome a single crease was observed in about 38 percent (Bryant *et al.*, 1970), more often in male (43.8 percent) than in female (28.6 percent) patients.

On the toes of patients, the frequency of whorls was significantly decreased, whereas fibular loops were found to be increased compared to controls (Table 6.21). Therefore, the loops on the toes, like those on the fingertips, exhibited a tendency to open toward the smallest digit. However, unlike on the fingertips, where radial loops were decreased in frequency, the tibial loops on the soles were slightly more frequent in Down syndrome than in controls, particularly on the toes, where these patterns are normally rare. Usually tibial loops are found on the great toes. In Down syndrome, the frequency was diminished. Unlike the palmar pattern intensity,

TABLE 6.19. Percent of Sydney line in Down syndrome and controls

POPULATION	TYPE OF LINE	DOWN SYNDROME		CONTROLS		SOURCE
		MALES	FEMALES	MALES	FEMALES	
Australian	Complete	11	21	10	8	Purvis-Smith (1972)
North American	Complete	15.2	26.6	6.5	7.9	Plato <i>et al.</i> (1973)
	Aberrant	18.6	16.7	11.4	12.3	
	Total	37.9	51.7	19.7	21.1	

the overall plantar pattern intensity was decreased because of the reduced frequency of patterns in all but the fourth interdigital area. Absence of true patterns in the hallucal area of Down syndrome patients was common. An arch tibial, which is a very rare pattern in normal Caucasian individuals, was the predominant hallucal ridge configuration in patients with Down syndrome (Table 6.22). Walker (1958) observed that when patterns other than tibial arches occurred in the hallucal area of the patients, these were nearly always small distal loops with a ridge count of 20 or less, whereas in a normal series the loop distal pattern tended to have a higher ridge count. Small distal loops in the hallucal area were

TABLE 6.20. Percent with a single interphalangeal crease on the fifth digit in Down syndrome

POPULATION	SINGLE CREASE	SOURCE
British	20.0	Penrose (1931)
Canadian	20.0	Uchida and Soltan (1963)
Canadian	27.0	Soltan and Clearwater (1965)
Swedish	10.8	Hall (1964)
Italian	20.2	Dallapiccola and Ricci (1967)
Indian	8.6	Saksena <i>et al.</i> (1966)
Japanese	28.0	Shiono <i>et al.</i> (1969)
Chinese	37.7	Bryant <i>et al.</i> (1970)

TABLE 6.21. Percent of each toe pattern type in Down syndrome and controls^a

DIGIT	PATTERN TYPE ^b							
	WHORL		LOOP FIBULAR		LOOP TIBIAL		ARCH	
	D	C	D	C	D	C	D	C
1	26.4	13.0	68.2	74.0	3.9	7.5	1.6	5.5
2	1.2	20.2	80.2	73.3	7.0	0.7	11.7	5.8
3	11.3	55.2	71.6	40.4	3.2	0.4	14.0	4.1
4	0.4	14.1	80.8	70.9	4.0	0.7	14.8	14.4
5	0.0	0.4	50.2	46.9	1.2	1.4	48.6	51.4
Total	7.8	20.5	70.2	61.1	3.8	2.1	18.1	16.2

^a According to Smith *et al.* (1966).^b D, Down syndrome; C, controls.

found in about a third of the patients compared to only 10–12 percent of controls (Table 6.22). Large distal loops with ridge counts higher than 20 were found in about 13 percent of patients compared to about 41 percent of controls (Walker, 1957). The frequencies of both small and large distal loops reported by Smith (1964) were in very good agreement with those of Walker.

In the fourth interdigital plantar area, distal loops, and therefore the *p''* triradii, occurred much more frequently in patients with Down syndrome (Table 6.23) than in controls. Walker (1945, personal communication to L. S. Penrose) found them about twice as frequent as in control individuals. Similarly, Smith (1964) observed this pattern on one or both feet in three times as many male patients and five times as many female patients as in controls. The frequency of *I*₄ loop distal patterns was increased considerably in Down syndrome (50 percent) compared to controls (12 percent). True plantar patterns also occurred considerably more often in Chinese female, but not male, patients than in controls (Bryant *et al.*, 1970). Zygodactylous triradii hardly ever occurred in patients with Down syndrome and were absent even when webbing of the toes was noticeable (Penrose and Loesch, 1970).

Dermal ridges on the palms and soles of patients with Down syndrome were often badly formed, giving a characteristic appearance of “dotted ridges” or “strings of pearls” (*P* lines). The degree of ridge malformation varied greatly from very mild cases, with an involvement of small palmar or plantar areas, to severe forms, where most of the ridged skin showed these ill-formed patterns.

TABLE 6.22. Percent of arch tibial and small loop distal patterns in the plantar hallucal areas in Down syndrome and controls

PATTERN	POPULATION	DOWN SYNDROME		CONTROLS		SOURCE
		LEFT	RIGHT	LEFT	RIGHT	
Arch tibial	British	53.8	46.9	0.0	0.3	Smith (1964)
	Canadian	55.0	57.0	—	—	Soltan and Clearwater (1965)
	Canadian	46.6	47.4	0.3	0.3	Walker and Johnson (1965)
	Italian	54.2	33.3	0.0	0.0	Walker and Johnson (1965)
	Indian	59.1	60.0	—	—	Saksena <i>et al.</i> (1966)
	Japanese	87.7		4.6		Matsui <i>et al.</i> (1966)
	Japanese	78.4	76.2	7.2	6.0	Shiono <i>et al.</i> (1969)
	Chinese	94.3		9.3		Bryant <i>et al.</i> (1970)
Small loop distal	British	30.3	33.8	9.9	12.3	Smith (1964)
	Canadian	33.0	34.0	—	—	Soltan and Clearwater (1965)
	Canadian	33.8	31.2	10.0	13.3	Walker and Johnson (1965)
	Italian	33.3	62.5	6.4	16.7	Walker and Johnson (1965)
	Indian	32.3	31.4	—	—	Saksena <i>et al.</i> (1966)

Geipel (1964) found that only about 8 percent of the patients had no P lines, whereas over 50 percent showed dissociation of ridges predominantly, i.e., covering a great part of the palms. The latter was observed in only 4 percent of controls. Dissociated ridges were observed in the interdigital, hypothenar, and central palmar areas in about 75 percent of the patients and in the thenar area in about 62 percent.

After D/G and G/G translocations were discovered capable of producing Down syndrome, comparative analyses of dermatoglyph-

TABLE 6.23. Percent of true patterns in the fourth interdigital plantar areas in Down syndrome and controls

POPULATION	MALES		FEMALES		TOTAL		SOURCE
	DOWN	CON-TROLS	DOWN	CON-TROLS	DOWN	CON-TROLS	
British	55.3	16.5	37.1	7.6	48.1	12.5	Smith (1964)
Chinese	12.9	13.0	20.0	3.0	15.7	8.1	Bryant <i>et al.</i> (1970)

ics were carried out in an effort to determine whether there were any possible differences in dermatoglyphic characteristics between the trisomic and translocation types of Down syndrome. Dermatoglyphics in the translocation types were found to resemble closely those of trisomy cases (Penrose and Delhanty, 1961; Walker *et al.*, 1963; Gibson and Pozsonyi, 1965; Soltan and Clearwater, 1965; Dallapiccola and Ricci, 1967; Rosner and Ong, 1967). Essentially no differences in dermatoglyphics were discovered that could be utilized in distinguishing the two types of Down syndrome.

Mosaicism has been demonstrated to influence the dermal patterns of Down syndrome. Dermatoglyphic traits of mosaic patients showed a tendency to be intermediate between nonmosaic patients with Down syndrome and controls (Penrose, 1965; Penrose and Smith, 1966; Dallapiccola and Ricci, 1967; Polani and Polani, 1969).

A long time before karyotyping of Down syndrome became feasible, investigators were exploiting the possibilities of diagnostic use of dermatoglyphics. Utilizing only two dermatoglyphic traits that occurred with high frequency in Down syndrome (transverse alignment of ridges in the distal palmar area and large hypothenar patterns associated with distally displaced axial triradius), Cummins and Platon (1946) were able to diagnose correctly almost 90 percent of the individuals determined to have Down syndrome by other clinical means. Their results were confirmed in a larger study (Cummins *et al.*, 1950). Later, several diagnostic indices were proposed based on the relative frequencies of dermal patterns (Walker, 1957, 1958; Beckman *et al.*, 1965; von Greyerz-Gloor *et al.*, 1969; Reed *et al.*, 1970; Borgaonkar *et al.*, 1971; Bolling *et al.*, 1971; Deckers *et al.*, 1973a,b,c). A particularly simple and rapid test uses a dermatoglyphic nomogram (Borgaonkar *et al.*, 1967, cited by Reed *et al.*, 1970) based on the four pattern areas that account for most

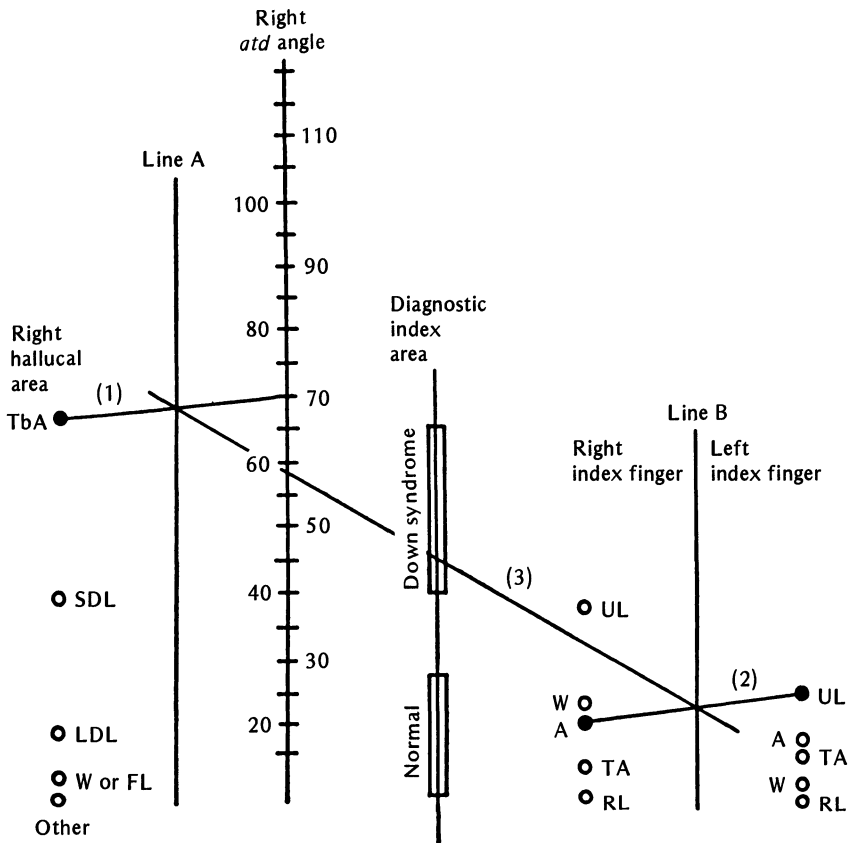


FIGURE 6.7 Dermatoglyphic nomogram for diagnosing Down syndrome.

From Reed, T. E., *et al.*: Dermatoglyphic nomogram for the diagnosis of Down's syndrome. *J. Pediatr.*, 77:1024, 1970. Copyright 1970, Indiana University Foundation, all rights reserved.

of the total dermatoglyphic variation between Down syndrome and control subjects (Figure 6.7). In addition, the recent index scores are very accurate, classifying incorrectly only a minor portion of cases tested. In spite of the high degree of accuracy, dermatoglyphic analysis can only supplement and not replace more precise means of diagnosis, such as karyotyping.

TRISOMY 18

Patients with trisomy 18 show developmental and mental retardation. A single umbilical artery is found in most of the patients and is a useful diagnostic sign at the birth of an affected infant.

Difficulty in feeding with poor sucking occurs and the infant fails to thrive. Congenital heart defects are common and, in a majority of patients, the skull is elongated, with a prominent occiput. The ears are malformed or low set, the palatal arch is narrow, and micrognathia is present. There is a short neck, short sternum, small pelvis, and hip abduction is limited. Hypotonia is present early but, in time, the infant becomes hypertonic. Inguinal or umbilical hernia, renal malformations, and cryptorchidism are frequent findings.

Flexion deformities of the hands, particularly overlapping fingers, are characteristic. The thumb is usually retroflexible or distally implanted. Partial syndactyly of the fingers, a single crease on the fifth digit and a single transverse palmar crease are frequently observed in patients with trisomy 18. The feet are often in a calcaneovalgus position and the big toe is short and dorsiflexed in most of the patients. Finger and toe nails are frequently hypoplastic.

Considering only patients with the full trisomy 18, the most typical dermatoglyphic finding was a strikingly high frequency of arches on the fingertips (Figure 6.8). Arches accounted for over 90 percent of all fingertip patterns of these patients, more than 40 percent of whom had ten digital arches. Fewer than six arches is very unusual in full trisomy 18, although it is sometimes observed in mosaic, translocation, or partial trisomy 18. Ulnar loops and whorls were found markedly diminished in frequency. Radial loops were only slightly reduced in frequency but tended to be displaced from their usual site on the fingertips so that 62 percent were found on the thumbs; the remaining ones were usually on the third, fourth, or fifth fingers. Only very rarely was the radial loop observed on the second digit, i.e., the digit that bears the majority of radial loops in normal individuals. A distal axial triradius was present in 53 percent of palms. The *atd* angle was often increased because of the distally displaced axial triradius into the *t'* or *t''* position. This increase, however, was generally not so great as in trisomy 13 or in Down syndrome.

The main-line A termination in the thenar area of the palm, first noted in trisomy 18 by Penrose in 1969, was found in 26 percent of the palms, considerably more often than in normal individuals.

The frequency of patterns in the third and fourth interdigital area was decreased.

As on the palms, there was an extreme paucity of patterns on the soles. The plantar pattern intensity was even lower than that observed in trisomies 13 and 21. In the hallucal area, all types of

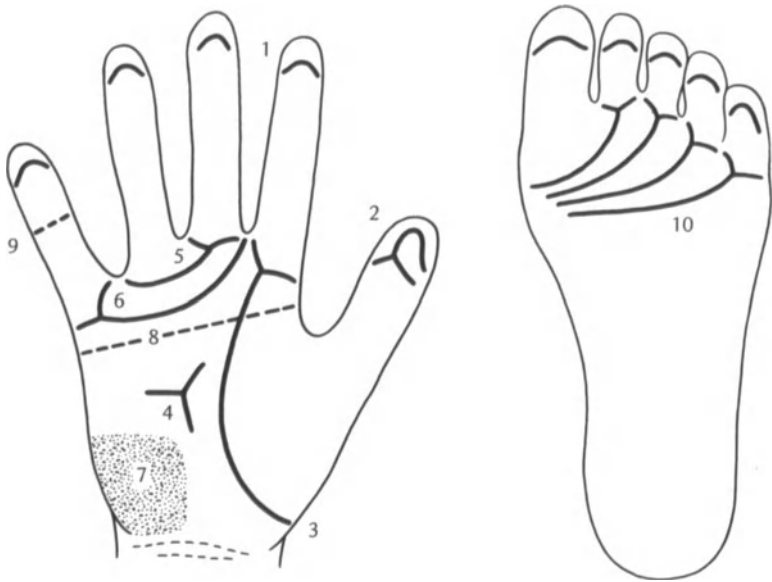


FIGURE 6.8 Dermatoglyphic features of trisomy 18. (1) Arches considerably increased in frequency. (2) Radial loops on digits other than second, particularly on the thumb. (3) Radial exit of main-line A. (4) Distal axial triradius. (5) I_5 patterns decreased in frequency. (6) I_4 patterns decreased in frequency. (7) Ridge dissociation. (8) Single transverse palmar crease increased in frequency. (9) Single flexion crease on the fifth digit. (10) Pattern intensity on soles extremely reduced.

true patterns were reduced as compared with controls. There was a considerable reduction in frequency of the proximal loops in the second interdigital area ($\hat{II}$) and of distal loops in the third interdigital area (III) with comparable decrease of triradii p' (Penrose and Loesch, 1970).

Hypoplasia or absence of dermal ridges has been reported in at least eight patients with trisomy 18 (Habedank, 1964; Weber *et al.*, 1964; Bartolozzi *et al.*, 1965; Lafourcade *et al.*, 1965; Ross, 1968; Taylor, 1968; Summitt, 1969; Rott *et al.*, 1970), including a case of mosaic trisomy 18/normal (Alter and Schulenberg, 1967). The degree of involvement varies between ridge hypoplasia of individual fingertips to complete absence of patterns of all fingertips, palms, and soles.

Flexion creases on the palms of trisomy 18 patients are often abnormal. About 75 percent of the individuals show a simian crease, usually bilaterally. A single flexion crease on the fingers,

usually the fifth digit, is also characteristic of trisomy 18. Approximately 80 percent of the individuals in whom flexion creases were reported had a single flexion crease on the fifth finger, usually bilaterally. In several instances, a missing distal crease was observed on the third or fourth digit, and even on all digits, and in some instances both distal and proximal digital flexion creases were missing.

Characteristic dermatoglyphic features of trisomy 18 are summarized in Figure 6.8 and Table 6.24. Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Baughan *et al.* (1969), Bottriaux *et al.* (1973), Chrysostomidou *et al.* (1971), Emanuel *et al.* (1970), Finley *et al.* (1963), Fluge *et al.* (1973), Geiser and Schindler (1968), Hecht *et al.* (1963), Hook *et al.* (1965), Lewis (1964), Miller *et al.* (1965), Müller *et al.* (1972), Passarge *et al.* (1966), Schepens

TABLE 6.24. Dermatoglyphic features in trisomy 18

FEATURE	NUMBER	VALUE
Fingertip patterns		
Arch	643	88.1%
Radial loop	24	3.3%
Ulnar loop	40	5.5%
Whorl	4	0.5%
Unknown	19	2.6%
Radial loops on individual digits		
First	13	61.9%
Second	1	4.8%
Third	3	14.3%
Fourth	1	4.8%
Fifth	3	14.3%
Mean <i>atd</i> angle	36	74.8°
Distal axial <i>t</i> ^a	48	53.3%
Interdigital patterns ^a		
I ₃	17	28.3%
I ₄	17	29.3%
Thenar exit of main-line A ^a	11	26.2%
Single transverse crease ^b	40	75.5%
Single crease on fifth digit ^b	34	79.1%

^a Number of palms with the trait.

^b Number of individuals with the trait.

et al. (1967), Scherz (1966), Shibata *et al.* (1973), Surana *et al.* (1972), Townes *et al.* (1962), Turner *et al.* (1964), Uchida *et al.* (1962), Walbaum *et al.* (1966), Weber and Sparkes (1970), Weichsel and Luzzatti (1965), Wolf *et al.* (1965), Zellweger *et al.* (1964), and our own unpublished data.

TRISOMY 13

Characteristic features of trisomy 13 include microcephaly, microphthalmos, cleft lip and cleft palate, low-set and malformed ears, capillary hemangiomata, polydactyly, retroflexible thumbs, long hyperconvex nails, and talipes equinovarus. Other clinical findings include those frequently encountered also in trisomy 18 syndrome, such as developmental retardation, failure to thrive, feeding difficulties, hypotonia, jitteriness and apneic spells, ocular hypertelorism, strabismus, epicanthal folds, presumptive deafness, micrognathia, short neck, extra skin on the nape, flexion deformity of fingers, inguinal or umbilical hernia, congenital heart disease, and undescended testes.

An unusual frequency of fingertip pattern types was found among patients with trisomy 13 reported in the literature. Whereas arches and radial loops were considerably increased in frequency, ulnar loops and whorls were decreased. Only 28 percent of radial loops were found on the second digit. The remaining radial loops were almost equally divided among the remaining digits.

The axial triradius was usually displaced distally to the center of the palm (t'' or t'''), which resulted in very wide *atd* angles. Only about 6 percent of palms showed an axial triradius in the t position. The mean summed *atd* angle was even higher than the mean values of *atd* in Down syndrome and trisomy 18. Radial displacement of digital triradius a , often observed in trisomy 13, also contributed somewhat to the widening of the *atd* angle and tended to increase the $a-b$ ridge count. A radial exit of the A line was found in more than 71 percent of palms of individuals with trisomy 13.

Penrose (1966) observed a marked increase of patterns in the thenar areas of the patients with 13 trisomy. In the pooled data, the frequency of patterns in the third interdigital area was markedly increased, with most of the individuals showing the trait bilaterally. Fourth interdigital patterns, in contrast, were diminished in frequency.

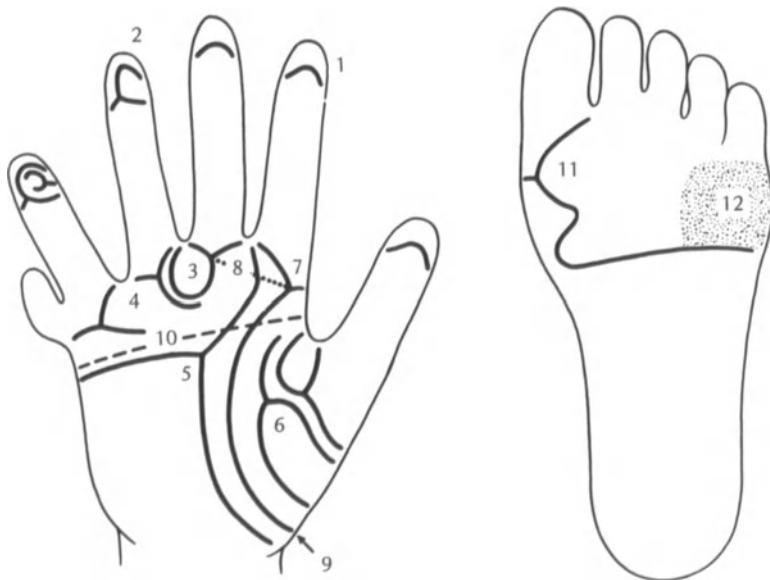
Approximately 60 percent of the palms showed a single trans-

verse flexion crease and a single crease on the fifth digits has been observed.

In contrast to the palms, where the pattern intensity was increased, the intensity of plantar patterns was greatly reduced. In the hallucal area of the soles, arch fibular or the arch fibular S pattern (Figure 6.9), which were first reported as characteristic for trisomy 13 by Uchida *et al.* (1962), were the most common patterns. The loop tibial and loop distal patterns followed with about equal frequencies. There was a deficit of proximal loops in the second interdigital area ($\hat{\text{II}}$) and of distal loops in the fourth interdigital area (IV), associated with a diminished frequency of triradii p , p'' , z , and z' (Penrose and Loesch, 1970).

Ridge dissociation was found in some cases, involving either small or large areas of epidermis on palms and soles. Patients with

FIGURE 6.9 Dermatoglyphic features of trisomy 13. (1) Arches increased in frequency. (2) Radial loops frequently on digits other than the second. (3) I_a patterns increased in frequency. (4) I_t patterns decreased in frequency. (5) Axial triradius extremely distal. (6) Thenar patterns increased in frequency. (7) Triradius a displaced radially. (8) a - b ridge count increased; associated with (7). (9) Radial exit of main-line A. (10) Single transverse flexion crease markedly increased in frequency. (11) Arch fibular and arch fibular S pattern (pictured here) frequent. (12) Ridge dissociation.



no visible patterns on both the hands and feet were also reported (Taylor, 1968).

Dermatoglyphic features characteristic for the trisomy 13 are summarized in Figure 6.9 and Table 6.25. Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Chen *et al.* (1971), Conen and Erkman (1966), Conen *et al.* (1962), Conen *et al.* (1966), Emberger *et al.* (1972),

TABLE 6.25. Dermatoglyphic features in trisomy 13

FEATURE	NUMBER	VALUE
Fingertip patterns		
Arch	59	22.7%
Radial loop	37	14.2%
Ulnar loop	99	38.1%
Whorl	45	17.3%
Unknown	20	7.7%
Radial loops on individual digits		
First	4	16.0%
Second	7	28.0%
Third	4	16.0%
Fourth	7	28.0%
Fifth	3	12.0%
Mean <i>a-b</i> ridge count	25	47.4
Mean <i>atd</i> angle	38	94.4°
Distal axial <i>r</i> ^a	79	94.0%
Interdigital patterns ^a		
I ₃	28	70.0%
I ₄	12	30.0%
Thenar exit of main-line A ^a	25	71.4%
Single transverse crease ^a	47	59.5%
Hallucal patterns		
Arch fibular, arch fibular		
S pattern	29	40.3%
Arch tibial	2	2.8%
Arch proximal	2	2.8%
Loop tibial	17	23.6%
Loop distal	15	20.8%
Loop fibular	1	1.4%
Whorl	6	8.3%

^a Number of palms with the trait.

Lee *et al.* (1966), Lubs *et al.* (1961), Marden and Yunis (1967), Marshall *et al.* (1964), Parrington and Edwards (1971), Smith *et al.* (1963), Stone *et al.* (1966), Taylor *et al.* (1970), Therman *et al.* (1961), Walzer *et al.* (1966), Yunis and Hook (1966), and our own unpublished data.

TRISOMY 8 MOSAICISM

In 1961, Jacobs *et al.* added C group trisomy to the three other trisomic conditions (trisomies 13, 18, and 21) then known to be compatible with life. Reports of at least 38 patients with an extra chromosome belonging to the C group have since appeared in the literature. Almost all of these patients were mosaics with a normal cell line as well as a trisomic cell line. In several of the more recently reported cases, the supernumerary chromosome was identified by banding techniques as chromosome 8. A comparison of phenotypic abnormalities of patients with chromosome 8 trisomy and those with unidentified trisomy C suggests that the majority if not all of the latter cases may also involve chromosome 8 (Bijlsma *et al.*, 1972; Malpuech *et al.*, 1972).

Clinically, individuals with trisomy 8 mosaicism show psychomotor retardation, an elongated and slender trunk with narrow shoulders and pelvis; a large and sometimes deformed head with a prominent forehead; strabismus; agenesis of the corpus callosum; abnormal nose; large, low-set, and malformed ears; micrognathia; short neck; restricted articular function; bone dysplasia, sometimes with extra ribs and vertebrae; hypertonicity; deep palmar and plantar creases; nail dysplasia; absent patellae; heart defects; renal anomalies; and frequent upper respiratory infections.

A summary of available dermatoglyphic data on patients with trisomy 8 mosaicism (Schaumann *et al.*, 1974) pointed out striking similarities of several unusual dermatoglyphic variables among some of the patients (Figure 6.10). These include an increased frequency of arch patterns on the fingertips, low TFRC, high palmar and plantar pattern intensity, a simian crease, and bilateral arches on the great toes. Arches were found to be the most frequent fingertip patterns, and the frequency of both ulnar loops and whorls was considerably decreased.

The occurrence of both arches and whorls on the fingertips of the same individual, as pointed out by Penrose (1972), was observed in four of eight reported cases of trisomy 8.

The TFRC was decreased in all individuals where this informa-

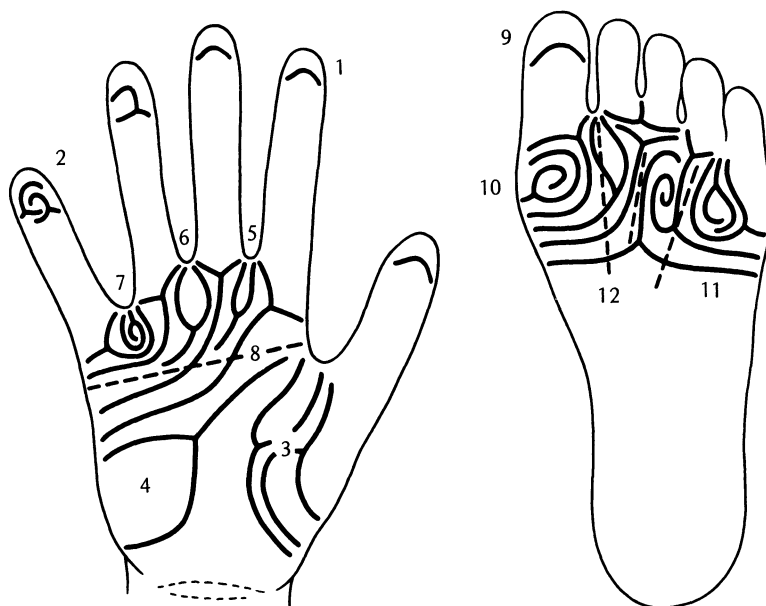


FIGURE 6.10 Dermatoglyphic features of trisomy 8 mosaicism. (1) Arches increased in frequency. (2) Whorls decreased in frequency but often present on the same hand together with arches. (3) Thenar patterns increased in frequency. (4) Hypothenar patterns increased in frequency. (5) I_2 patterns increased in frequency. (6) I_3 patterns increased in frequency. (7) I_1 patterns increased in frequency. (8) Single transverse palmar crease increased in frequency. (9) Arches on great toes increased in frequency. (10) Whorls in the hallux area increased in frequency. (11) Plantar pattern intensity considerably increased. (12) Deep furrows on palms and soles.

tion was available. Pattern intensity on both the palms and the soles was very high, expressed by the presence of patterns in the thenar, hypothenar, and interdigital areas of the palms and several whorls in the hallux and interdigital areas of the soles. Among the viable trisomies, the high plantar pattern intensity may be a characteristic feature only of trisomy 8. In patients with trisomies 13, 18, and 21, the pattern intensity on the soles was found to be markedly decreased (Penrose and Loesch, 1970). Arches on both great toes were observed in all three cases where foot patterns were noted. This trait is found in only about 4 percent of phenotypically normal individuals.

Dermatoglyphic data of patients with unspecified C trisomies seem to support the conclusion, based on similarities in clinical

signs and symptoms, that most of them probably represent trisomy of chromosome 8. Even when considered as a group, these patients shared certain dermatoglyphic characteristics with patients with trisomy 8 (Table 6.26), including a high frequency of fingertip arches, low TFRC, presence of both arches and whorls on different fingertips of the same individual, frequent distal displacement of axial triradii, high pattern intensity on palms, and frequent presence of a simian crease, usually unilaterally. In the hallucal areas, whorls formed a majority of the patterns and arches were found to be the predominant pattern type on the great toes, similar to trisomy 8 patients.

Deep palmar and plantar skin furrows represent a distinct feature of trisomy 8 (Lejeune *et al.*, 1969). They were noticed in five of 11 patients with trisomy 8, as well as in 11 of 15 unspecified C trisomy patients. However, they may have been present in infancy in some other of these patients, gradually disappearing with advancing

TABLE 6.26. Dermatoglyphics in trisomy 8 and other unidentified C trisomies

FEATURE	TRISOMY 8		UNIDENTIFIED C TRISOMIES	
	NUMBER	VALUE	NUMBER	VALUE
Fingertip patterns				
Arch	41	45.6%	22	18.3%
Radial loop	3	3.3%	7	5.8%
Ulnar loop	31	34.5%	56	46.7%
Whorl	13	14.4%	33	27.5%
Unknown	2	2.2%	2	1.7%
Distal axial triradius ^a	11	64.7%	12	50.0%
Palmar patterns ^a				
Thenar/I ₁	2	25.0%	10	62.5%
I ₂	2	33.3%	1	25.0%
I ₃	4	66.7%	12	85.7%
I ₄	8	100.0%	9	75.0%
Hypothenar	9	75.0%	11	50.0%
Simian crease ^a	6	37.5%	7	38.9%
Sole patterns:				
Great toe arch	6	100.0%	5	62.5%
Hallucal whorl	6	100.0%	7	70.0%
Mean TFRC	5	82.6	3	78.0
Mean <i>atd</i> angle	10	67.3°	0	—

^a Number of palms with the trait.

age (Bijlsma *et al.*, 1972), and consequently were not observed in the older patients. Conceivably, in some cases, comment on the furrows was omitted because their importance was not appreciated.

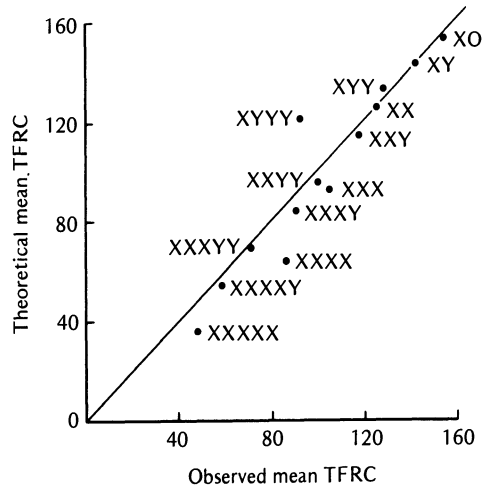
In spite of the small number of patients, the incompleteness of the dermatoglyphic data and lack of dermatoglyphic information concerning the patients' immediate families, certain combinations of unusual dermatoglyphic patterns seem to emerge as characteristic for trisomy 8 mosaicism syndrome as summarized in Figure 6.10 and Table 6.26. Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Atkins *et al.* (1974), Bargman *et al.* (1967), Caspersen *et al.* (1972), de Grouchy *et al.* (1971), Emberger *et al.* (1970), Gustavson *et al.* (1967), Higurashi *et al.* (1969), Jalbert *et al.* (1966), Laurent *et al.* (1971), Monnet *et al.* (1967), Oikawa *et al.* (1969), Pfeiffer *et al.* (1962), Reinwein *et al.* (1966), Riccardi *et al.* (1970), Smith (1964), Stalder *et al.* (1964), Tuncbilek *et al.* (1972), and Van Eys *et al.* (1970).

Aberrations of Sex Chromosomes

Abnormalities of the sex chromosomes do not have as much influence on ridge formation as do autosomal chromosomal aberrations. Nevertheless, there are some noteworthy dermatoglyphic features associated with sex chromosome defects.

In a personal communication to Holt in 1957, Penrose mentioned his observation of an increased distance between triradii *a* and *b* in Turner syndrome. Later, he elaborated on this relationship (Penrose, 1963). Alter (1965) demonstrated the semiquantitative inverse relationship between the number and type of sex chromosomes and the total finger ridge count. His observation was confirmed by Penrose (1967), who postulated that the sex chromosomes influence the size of a cell by controlling its fluid content; the more sex chromosomes present in a cell, the lower the fluid content. In XO individuals, the cells, and hence the finger fat pads, would be larger than those in XY individuals. The latter, in turn, would have larger cells than those of XX individuals, and so forth, for multiple sex chromosome disorders. Larger fat pads yield larger surface area, requiring more ridges to cover the pattern area during embryogenesis. Hence, the largest ridge count would be found in XO individuals, whereas low ridge counts would occur in multiple sex chromosome states. Penrose's hypothesis is sup-

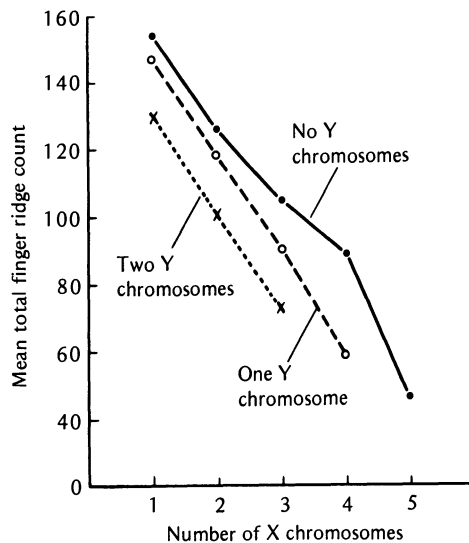
FIGURE 6.11 Relationship between sex chromosomes and the total finger ridge count (TFRC).



ported indirectly in humans by the observation that edema of the hands and feet is common at birth in Turner syndrome.

Penrose (1967) has noted that the presence of each additional X chromosome diminishes the TFRC nearly three times as much as does the presence of each additional Y chromosome. He expressed this relationship in an equation for the numerical value of an expected mean TFRC: $E = 187 - 30x - 12y$, where x is the number of X chromosomes and y is the number of Y chromosomes. Pooled data on mean TFRC, obtained from reports of patients with

FIGURE 6.12 Total finger ridge count related to number of X and Y chromosomes.



sex chromosome aneuploidies, correspond remarkably with the expected values derived from the equation, particularly in males (Figures 6.11 and 6.12). Females tend to have a slightly higher mean TFRC than predicted by the equation, suggesting the possibility that the effect of several X chromosomes acting together may be somewhat lower than the sum of their individual effects.

A general tendency for ridge breadth to increase with an increasing number of sex chromosomes has been shown but the Y chromosome was found to have more effect on ridge breadth than the X chromosome (Penrose and Loesch, 1969).

MONOSOMY OF THE X CHROMOSOME (TURNER SYNDROME)

Turner syndrome is caused by full or partial monosomy of an X chromosome, with or without mosaicism. There is a wide range of clinical findings that vary between severe expression of Turner syndrome to apparent normality. Characteristically, patients are phenotypic females of short stature. Their mean adult height is 140 cm and they usually have multiple anomalies. Ocular findings include ptosis, epicanthal folds, cataracts, and strabismus. The corners of the mouth are often depressed, the hard palate is narrow and high arched, and micrognathia and malocclusion are common. The ears are prominent and often low set. The back of the neck in newborns shows a redundancy of skin that in older children and adults becomes the characteristic webbing of the neck (pterygium colli). Usually, there is a low posterior hairline. The chest is shieldlike and congenital heart disease (usually coarctation of the aorta) may be present. Radiographs may reveal short metacarpal bones, particularly the fourth and fifth. Fingernails and toenails are usually narrow, deeply set, and transversely hyperconvex. Cubitus valgus and osteoporosis are common. Multiple pigmented nevi are often observed. There is a general lack of pubertal sexual development, breasts are undeveloped, and the nipples appear widely spaced. External genitalia are infantile with scanty pubic hair. These findings, as well as the primary amenorrhea, are caused by gonadal dysgenesis. Horseshoe kidney is frequent. Multiple intestinal telangiectases may be present. Intelligence is usually normal.

Palmar skin is often very thin and wrinkled, which produces the appearance of multiple secondary creases on dermatoglyphic prints. Palmar volar pads tend to be prominent.

In order to assure as homogeneous a group of patients with

Turner syndrome as possible, only individuals whose chromosomal constitution was cytologically proved to be 45,XO were used for dermatoglyphic analysis. Patients with mosaicism for the sex chromosomes or partial monosomy of an X chromosome were not included.

The TFRC was increased in patients with Turner syndrome, reflecting the inverse relationship between the number of sex chromosomes and TFRC. However, the frequency of finger pattern types (Table 6.27) was very similar to that of controls. The increased ridge count in Turner syndrome therefore could not be attributed to an increased frequency of high-intensity patterns. Instead, the usual patterns were increased in size.

The *a-b* ridge count was found to be somewhat increased in patients with Turner syndrome compared to controls. Frequency of patterns in both the third and fourth interdigital areas was elevated. A hypothenar pattern, often large, was found in about 50 percent of the palms. A distally displaced axial triradius, and hence an increased maximal *atd* angle, was usually associated with a hypothenar pattern. Both of these traits were present very frequently in patients with Turner syndrome. Either a full or a transitional single transverse crease was found in 28 percent of the palms investigated. A missing digital *c* triradius and a main-line A termination in the thenar area were other observed dermatoglyphic features. The hallual areas of the feet tended to have large whorls and large distal loops. Holt (1969) noted a marked decrease of *p* triradii and an excess of whorls in the first interdigital area of the soles. Penrose and Loesch (1970) found an average of 2.5 loops per sole in Turner syndrome, being a lower pattern intensity than in controls. They noted a deficit of distal loops, particularly in area III, and an increase of central loops III.

Dallapiccola *et al.* (1972) identified five dermatoglyphic characteristics (TFRC, *a-b* ridge count, maximal *atd* angle, T line terminating in the second interdigital area, and presence of hypothenar patterns on the palms) and four skeletal characteristics (carpal sign, metacarpal sign, phalangeal sign, and abnormally shaped distal phalanges), which show significant differences in frequency between patients with Turner syndrome and normal females. He used these nine characteristics to develop a scoring method to diagnose Turner syndrome. When both the dermatoglyphic and skeletal characteristics were considered together, there was little overlap between those with Turner syndrome and normal females. Only 4 percent of the controls reached a numerical score shown by 80 percent of Turner patients.

TABLE 6.27. Dermatoglyphics in Turner syndrome and in polysomies of the X chromosome

FEATURE	45,XO		47,XXX		48,XXXX		49,XXXXX	
	NUMBER	VALUE	NUMBER	VALUE	NUMBER	VALUE	NUMBER	VALUE
Fingertip patterns								
Arch	40	2.9%	15	25.0%	8	7.3%	13	43.3%
Radial loop	48	3.5%	3	5.0%	2	1.8%	1	3.3%
Ulnar loop	895	65.8%	25	41.7%	63	57.3%	5	16.7%
Whorl	366	26.9%	17	28.3%	36	32.7%	11	36.7%
Unknown	11	0.8%	0	0.0%	1	0.9%	0	0.0%
Palmar patterns^a								
Thenar/I ₁	26	13.1%	0	0.0%	1	6.2%	—	—
I ₂	5	3.4%	0	0.0%	3	18.8%	—	—
I ₃	84	57.5%	1	12.5%	6	33.3%	—	—
I ₄	86	58.9%	6	75.0%	11	55.0%	—	—
Hypothenar	158	51.1%	3	30.0%	10	45.4%	—	—
Distal axial triradius ^a	66	55.9%	4	33.3%	8	50.0%	0	0.0%
Single transverse palmar crease ^a	51	27.7%	0	0.0%	2	20.0%	2	50.0%
Mean TFRC	114	154.4	27	105.7	14	88.5	3	47.7
Mean summed <i>a-b</i> ridge count	108	89.5	—	—	3	79.7	—	—
Mean summed <i>atd</i> angle ^a	121	100.5°	6	90.5°	8	95.8°	—	—

^a Number of palms with the trait.

Using dermatoglyphic traits alone, 10 percent of the controls had a score shown by 70 percent of the Turner patients.

Some patients with Turner syndrome had a chromosome complement other than XO, such as an isochromosome, deletion, or mosaicism. Lindsten *et al.* (1963) and Engel and Forbes (1965) observed no difference in the dermatoglyphics between XO Turner syndrome and Turner syndrome associated with other chromosomal states, including XO/XXi, XO/XX_a, XX/XXd, and Xy. However, in one series of mosaic XO/XX patients (Pfeiffer and Kiera, 1968), a lower TFRC and a lower *a-b* ridge count were found than in XO Turner syndrome.

Dermatoglyphic data on Turner syndrome are summarized in Table 6.27. Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Almeida de Cabral *et al.* (1967), Ciovîrnache *et al.* (1968), Curcio (1967), Fischer and Haslund (1968), Forbes (1964), Franceschini *et al.* (1965), Holt and Lindsten (1964), Mutalik *et al.* (1968), Nance and Uchida (1964), Russell *et al.* (1966), Saksena and Kumar (1968), Shine and Corney (1966), Turner and Zanartu (1962), Yarema and Borgaonkar (1970), and our own unpublished data.

POLYSOMIES OF THE X AND Y CHROMOSOMES (KLINEFELTER PHENOTYPE)

Individuals with Klinefelter syndrome are sex-chromatin positive males with a normal penis but small testes. They appear grossly normal, although multiple minor anomalies are common, particularly in individuals with more complex sex chromosome aneuploidy, such as 48,XXXY; 49,XXXXY; or 49,XXYY.

47,XXY Klinefelter syndrome

The clinical features of the XXY phenotype become apparent only after puberty and include frequent gynecomastia, sparse facial hair, female pubic escutcheon, and disturbances of sexual function. Most individuals have normal intelligence, although reduced intelligence is not infrequent.

Dermatoglyphic characteristics of individuals with a 47,XXY chromosomal constitution are not very remarkable. The most distinctive feature in the pooled data (Table 6.28) was a lower TFRC. Penrose (1963) suggested a general reduction in the pattern size as a reason for the lower TFRC in XXY individuals. In support of this suggestion, the ulnar loop ridge count for separate fingers was

TABLE 6.28. Dermatoglyphics in Klinefelter phenotypes with various polysomies of X and Y chromosomes

FEATURE	46,XX MALES		47,XXY		48,XXYY		48,XXXXY		49,XXXXY		49,XXXXY	
	NUM- BER	VALUE	NUM- BER	VALUE	NUM- BER	VALUE	NUM- BER	VALUE	NUM- BER	VALUE	NUM- BER	VALUE
Fingertip patterns												
Arch	12	12.0%	116	9.5%	38	11.2%	31	38.8%	3	15.0%	38	23.8%
Radial loop	6	6.0%	56	4.6%	15	4.4%	2	2.5%	1	5.0%	6	3.8%
Ulnar loop	54	54.0%	595	48.8%	175	51.5%	27	33.8%	9	45.0%	73	45.6%
Whorl	28	28.0%	444	36.4%	112	32.9%	19	23.7%	7	35.0%	41	25.6%
Unknown	0	0.0%	9	0.7%	0	0.0%	1	1.2%	0		2	1.2%
Palmar patterns ^a												
Thenar/I ₁	2	50.0%	7	5.5%	0	0.0%	0	0.0%			0	0.0%
I ₂	1	25.0%	3	16.7%	1	3.6%	0	0.0%			0	0.0%
I ₃	8	50.0%	48	46.2%	21	45.6%	2	33.3%			7	70.0%
I ₄	4	40.0%	50	48.1%	18	37.5%	4	66.7%			4	40.0%
Hypothenar	5	41.7%	85	40.7%	31	45.6%					8	50.0%
Distal axial triradius ^a	13	86.7%	47	47.0%	27	67.5%	5	62.5%	0	0.0%	11	61.1%
Single transverse palmar crease ^a	2	12.5%	12	11.8%	11	22.0%	0	0.0%				
Mean TFRC	9	132.6	193	118.5	39	101.2	14	91.7	1	73.0	19	64.6
Mean summed <i>a-b</i> ridge count			102	79.9			2	64.0				
Mean summed <i>atd</i> angle	4	86.2°	120	86.3°	27	89.7°	3	83.3°			5	91.1°

^a Number of palms with the trait.

found to be reduced (Penrose and Loesch, 1969). A tendency to increased width of the ridges has also been reported (Cushman and Soltan, 1969; Penrose and Loesch, 1969; Wisniewski *et al.*, 1969) so that fewer ridges suffice to cover the pattern area.

Alternatively, the low TFRC may be the result of an increased frequency of arch patterns on fingertips (Alter, 1965). A somewhat increased frequency of fingertip arches in XXY patients and the fact that the mean TFRC of individuals without arches on any of the fingertips was 139.0, compared to only 63.7 in patients with at least one arch pattern, support this view. However, it may be that both mechanisms are operating in this chromosomal disorder to reduce the TFRC.

A reduced *a-b* ridge count has also been reported. In Hunter's (1968) series the mean *a-b* ridge count was 85.1, which was significantly lower than in his controls. In the Cushman and Soltan (1969) series, however, the mean *a-b* ridge count of 82.2 was not significantly different from controls. The overall mean *a-b* ridge count was somewhat lower than is usual in normal populations. The position of the axial triradius was reported as not being strikingly altered in XXY patients compared to the normal population (Penrose, 1963; Forbes, 1964; Hunter, 1968; Cushman and Soltan, 1969). In the pooled data, almost half of all investigated palms showed a distal axial triradius, mostly into the *t'* position. In spite of this, the *atd* angle was not found to be significantly increased. An ulnar triradius in association with a loop carpal, loop radial, or arch radial hypothenar pattern was found with increased frequency in XXY individuals when compared with normal males but not with normal females (Cushman and Soltan, 1969). An ulnar triradius was found in 13 percent of cases of XXY patients by Uchida *et al.* (1964) and in 35 percent by Cushman and Soltan (1969).

Other palmar dermatoglyphics, such as the thenar, hypothenar, and interdigital patterns, did not provide much information of diagnostic value. Their incidence generally corresponded to that of controls. A single palmar crease was observed in about 12 percent of the palms, which is a somewhat higher frequency than is usually reported in normal individuals but is far from being sufficiently increased to serve as a useful trait in diagnosing XXY individuals.

On the soles of XXY patients, Cushman and Soltan (1969) reported no significant differences in hallual or interdigital areas compared to controls. However, Penrose and Loesch (1970) found a significant reduction of pattern intensity on the soles. In particular, there was a deficit of proximal loops $\hat{\text{II}}$ and $\hat{\text{III}}$ and some decrease

of distal loops II, III, and IV, with a corresponding reduction in the frequency of the *p* triradius.

48,XXYY Klinefelter syndrome

Individuals with the XXYY chromosomal aneuploidy phenotypically resemble the XXY patients. However, they tend to be somewhat taller and more aggressive and are more frequently mentally retarded.

Pooled dermatoglyphic data of the XXYY individuals also resemble those found in the XXY Klinefelter syndrome (Table 6.28). The TFRC in patients with XXYY chromosome complement was found to be reduced even more than in XXY individuals. The low mean TFRC can probably be partially accounted for by the increased frequency of fingertip arches and partially by the general smallness of other patterns.

Frequencies of the palmar patterns did not differ appreciably from the control populations. However, the hypothenar area has been pointed out as being of particular interest in the XXYY syndrome by Uchida *et al.* (1964). The authors considered the presence of three hypothenar patterns—loop carpal, loop radial, and arch radial—associated with an ulnar triradius to be characteristic of the XXYY variant of Klinefelter syndrome but not of the XXY syndrome. Our pooled data support this observation, although the differences of combined frequencies of loop carpal, loop radial, and arch radial patterns between XXYY and XXY or XY individuals are not as large as in the smaller sample of Uchida *et al.* (1964). These patterns were found in 43.8 percent of all palms of the XXYY patients but in only 21.4 percent of the palms in the XXY Klinefelter syndrome and in 25.8 percent of control males.

Although the axial triradius was found to be distally displaced in about 68 percent of all palms, the mean maximal *atd* angle was found to be comparable with that of normal males.

Generally, the differences in dermatoglyphics between the XXYY and XXY syndromes, although present, are of a statistical nature and are so small that their application in differential diagnosis is rather limited.

48,XXXY; 49,XXXXY; 49,XXXXY Klinefelter syndrome

An increasing number of sex chromosomes is associated with poor development of the external genitalia and more severe mental retardation.

The dermatoglyphic features (Table 6.28) also reflect the greater number of X and Y chromosomes. In the pooled data, there was an

increase in arches on the fingertips and the mean TFRC was reduced progressively with the addition of each sex chromosome (Figures 6.11 and 6.12).

An ulnar triradius on the palm, associated with a radial loop in the hypothenar area, has been reported in two cases of XXXXY syndrome (Penrose, 1963; Joseph *et al.*, 1964). Abortive C main lines have also been reported in some XXXXY individuals (Schade *et al.*, 1963; Joseph *et al.*, 1964).

A single transverse palmar crease was a frequent finding in various variants of the Klinefelter syndrome.

46,XX Klinefelter syndrome

XX males have small testes and often a small penis and scrotum. Frequently, they have gynecomastia and lack of facial hair. Intelligence is normal.

Pooled dermatoglyphic data (Table 6.28) did not reveal any strikingly unusual features. The fingertip arches were somewhat increased in frequency and the mean TFRC was slightly decreased. Although most palms showed distally displaced triradii (usually into the t' position), the mean *atd* angle was not increased. Boczkowski *et al.* (1969) reported the ridge breadth on the fingertips to be $480\text{ }\mu\text{m}$, which is closer to the mean value for normal females ($509.0 \pm 46.0\text{ }\mu\text{m}$) than for normal males ($552.6 \pm 41.0\text{ }\mu\text{m}$).

Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Alter *et al.* (1966), Atkins *et al.* (1963), Bartsch-Sandhoff *et al.* (1974), Bitan *et al.* (1969), Blatsch (1964), Borgaonkar and Mules (1970), Borgaonkar *et al.* (1970), Christensen and Therkelsen (1970), de Grouchy *et al.* (1967), Ellis *et al.* (1961), Farquhar and Walker (1964), Ferguson-Smith *et al.* (1960), Ferrier and Ferrier (1968), Fraccaro *et al.* (1962), Garcia *et al.* (1967), Greenstein *et al.* (1970), Hayek *et al.* (1971), Herbeuval *et al.* (1965), Jancar (1968), Knorr *et al.* (1966), Lecluse-Van der Bilt *et al.* (1974), Lindsten *et al.* (1966), Lisker *et al.* (1970), Luciani *et al.* (1969), Morić-Petrović *et al.* (1973), Muller *et al.* (1967), Nicolis *et al.* (1972), Palutke *et al.* (1973), Parker *et al.* (1970), Penrose (1967), Pfeiffer (1962), Prader *et al.* (1964), Robinson *et al.* (1964), Sachsse *et al.* (1967), Scherz and Roedel (1963), Schlegel *et al.* (1965), Sebaoun *et al.* (1969), Shapiro *et al.* (1970), Shiono (1969), Simpson *et al.* (1974), Sperling *et al.* (1973), Tipton *et al.* (1966), Townes *et al.* (1965), Tumba (1972), Vormittag and Weninger (1972), Walker and Borgaonkar (1969), Walter *et al.* (1971), Zaleski *et al.* (1966), and Zellweger and Abbo (1966).

POLYSOMIES OF THE Y CHROMOSOME

47,XYY syndrome

The majority of the XYY patients are excessively tall and display a muscle weakness and poor coordination. Mild mental retardation is not uncommon and sexual development is usually normal.

The pooled dermatoglyphic data of the XYY individuals (Table 6.29) were unremarkable. The mean TFRC was somewhat lower than in normal males. The mean *atd* angle was not increased in spite of a frequent distal displacement of the axial triradius. The frequency of the palmar patterns was somewhat decreased.

48,XYYY

There is a lack of dermatoglyphic data among the very few reported cases of XYYY individuals (Table 6.29). Apart from the low TFRC, no unusual dermatoglyphic features have been noted.

TABLE 6.29. Dermatoglyphics in polysomies of the Y chromosome

FEATURE	47,XYY		48,XYYY	
	NUMBER	VALUE	NUMBER	VALUE
Fingertip patterns				
Arch	139	10.1%	1	3.3%
Radial loop	64	4.7%	1	3.3%
Ulnar loop	704	51.4%	24	80.0%
Whorl	450	32.9%	4	13.4%
Unknown	13	0.9%	0	0.0%
Palmar patterns^a				
Thenar/I ₁	1	1.7%	—	—
I ₂	5	7.8%	—	—
I ₃	38	33.9%	—	—
I ₄	39	34.8%	—	—
Hypothenar	36	28.6%	—	—
Distal axial triradius ^a	61	46.9%	2	33.3%
Single transverse palmar crease ^a	8	7.7%	2	100.0%
Mean TFRC	152	129.8	3	93.7
Mean summed <i>a-b</i> ridge count	82	78.0	1	75.0
Mean summed <i>atd</i> angle	94	90.7°	2	88.0°

^a Number of palms with the trait.

Dermatoglyphic data were pooled from the following sources: Abrams and Pergament (1971), Alam *et al.* (1972), Bartlett *et al.* (1968), Boczkowski (1969), Borgaonkar and Mules (1970), Cleveland *et al.* (1969), Daly *et al.* (1969), Dignon *et al.* (1972), Emberger *et al.* (1970), Emerit *et al.* (1968), Franks *et al.* (1967), Hubbell *et al.* (1973), Hunter and Quaife (1973), Keutel and Dauner (1970), Marinello *et al.* (1969), Mavalwala *et al.* (1969, 1970), Noel *et al.* (1969), Parker *et al.* (1969, 1970), Penrose *et al.* (1967), Penrose and Loesch (1967), Persson (1967), Ridler *et al.* (1973), Saldaña-Garcia (1973), Schoepflin and Centerwall (1972), Spencer *et al.* (1969), Townes *et al.* (1965), Tsuboi and Nielsen (1969), Uchida *et al.* (1964), and Valentine *et al.* (1971).

POLYSOMIES OF THE X CHROMOSOME

Although various congenital anomalies have been observed among 47,XXX females, there is no distinctive phenotype. Sexual disturbances (menstrual disorders, amenorrhea, sterility) and mental deficiency seem to be frequent. Females with tetrasomy or pentasomy X tend to be more severely affected. They tend to be mentally retarded and frequently have a number of congenital anomalies.

The pooled dermatoglyphic data were unremarkable (Table 6.27) except for the progressively lower mean TFRC correlated with an increasing number of X chromosomes and abnormal frequencies of the fingertip patterns. Both the TFRC and the fingertip pattern frequency of individual patients varied widely.

Dermatoglyphic data were pooled from the following sources: Berkley and Faed (1970), Blackston and Chen (1972), Borgaonkar and Leger (1969), de Grouchy *et al.* (1968), Di Cagno and Franceschini (1968), Duncan *et al.* (1970), Gardner *et al.* (1973), Kesaree and Woolley (1963), Kohn *et al.* (1968), Larget-Piet *et al.* (1972), Lejeune and Abonyi (1968), Peña *et al.* (1974), Penrose (1967), Rerrick (1972), Sergovich and Pozonyi (1971), Sokolowski *et al.* (1969), Telfer *et al.* (1970), and Zajackowska *et al.* (1970).

TRIPLOIDY

Common features of individuals with chromosomal triploidy are premature birth with very low birth weight, developmental retardation, and extremely low viability. Most of the patients either are

spontaneously aborted or stillborn or die very shortly after birth. Cutaneous syndactyly of the third and fourth fingers, coloboma of the iris, hypospadias, and testicular germinal cell hypoplasia are frequent findings. Otherwise, the clinical picture is variable, displaying a wide spectrum of major or minor abnormalities.

Triploidy seems to be frequent in abortuses and the number of liveborn individuals with triploidy surviving long enough to allow dermatoglyphic analysis is very low. In the pooled data of available cases with reported dermatoglyphics, the majority of patients (89.3 percent) showed a single transverse palmar crease. On the fingertips, whorls were found with markedly increased frequency, forming 66.6 percent of all patterns in pure triploidy and 42.5 percent in diploid/triploid mosaicism. Ulnar loops occurred considerably less frequently (16.7 percent and 42.5 percent, respectively) than in normal populations. In addition to a somewhat increased frequency, radial loops tended to be shifted from their usual site on the second digit to the third, fourth, and fifth digit. Missing or fused digital triradii in triploidy can be accounted for by syndactyly, which is a common finding in this chromosomal anomaly. Bilateral absence of distal flexion creases on the second, third, and fourth digits and two unusually close proximal creases on the fifth digits were observed in a case of 69,XXX triploidy (Butler *et al.*, 1969). A single interphalangeal crease has been observed on the left fifth finger of a female diploid/triploid patient (Jenkins *et al.*, 1971).

Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Bernard *et al.* (1967), Böök (1970), Edwards *et al.* (1967), Finley *et al.* (1972), Keutel *et al.* (1970), Lejeune *et al.* (1967), Niebuhr *et al.* (1972), Penrose (1968), Schindler and Mikamo (1970), Schmickel *et al.* (1971), Schmid and Vischer (1967), Simpson *et al.* (1972), Walker *et al.* (1973), and Zergollern *et al.* (1972).

Structural Chromosomal Aberrations

DELETION OF THE SHORT ARM OF CHROMOSOME 5 (CRI-DU-CHAT SYNDROME)

The most characteristic feature of this syndrome is a peculiar, high-pitched cry of infants which sometimes persists for months and even years. The cry resembles the mewing of a kitten, hence the name "cri-du-chat" syndrome. Low birth weight, failure to thrive, respiratory stridor, and laryngomalacia are frequent findings in the

cri-du-chat syndrome. Typically, the patients show profound mental retardation, microcephaly, hypertelorism, strabismus, broad based nose, oblique palpebral fissures, epicanthal folds, low-set ears, and micrognathia. Infants have a round face, whereas a thin face is usually observed in older patients. Premature greying of the hair, various skeletal anomalies, and muscular hypotonia are additional features of the syndrome.

The pooled dermatoglyphic data (Table 6.30) are based only on patients with deletion of part of the short arm of chromosome 5, which was cytologically proved and found to be the only chromosomal abnormality visible. Cases of cri-du-chat syndrome with a ring chromosome, translocation, mosaicism, or additional chromosomal aberrations were omitted.

Apart from somewhat increased frequency of whorls and a decrease in ulnar loops, which were more marked in affected females than affected males, no striking pattern abnormalities were found on the fingertips (Figure 6.13). An excess of whorls on the finger-

TABLE 6.30. Comparison of dermatoglyphic features in 4p— and 5p— syndromes

FEATURE	4p—	5p—
Fingertip patterns		
Arch	32.7%	10.0%
Radial loop	2.0%	4.0%
Ulnar loop	43.3%	50.3%
Whorl	10.0%	34.1%
Unknown	12.0%	1.6%
Mean TFRC	48.4	102.8
Mean <i>atd</i> angle	53.5°	50.8°
Distal axial <i>r</i> ^a	38.2%	82.3%
Palmar patterns ^a		
Thenar/I ₁	18.2%	24.5%
I ₂	0.0%	1.1%
I ₃	20.0%	36.0%
I ₄	70.0%	71.9%
Hypothenar	25.0%	26.8%
Single transverse crease ^a	52.3%	82.2%
Ridge dissociation	81.0%	0.0%

^a Percent of palms with the trait.

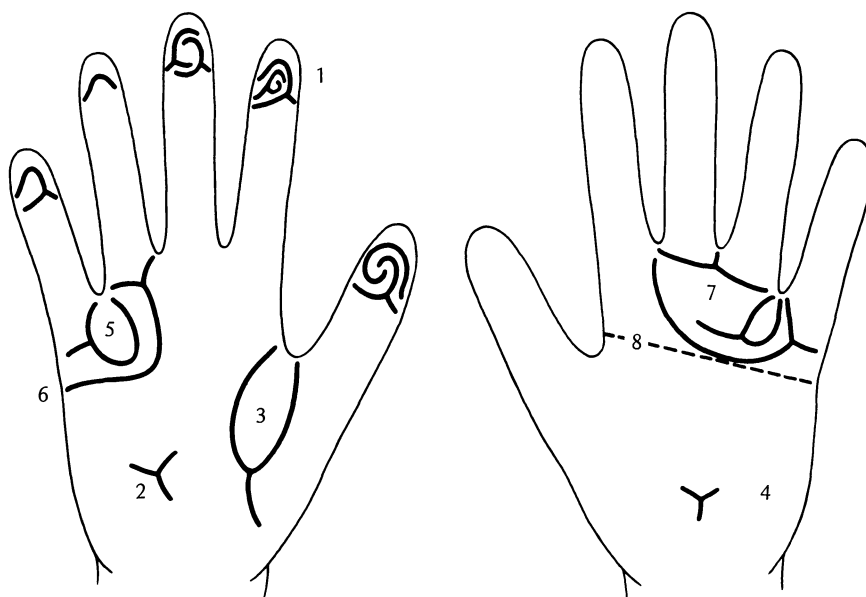


FIGURE 6.13 Dermatoglyphic features of deletion of the 5p— syndrome. (1) Whorls somewhat increased in frequency. (2) Distal axial triradius (r'). (3) Thenar patterns somewhat increased in frequency. (4) Hypothenar patterns decreased in frequency. (5) I_1 patterns increased in frequency; patterns result mostly from the D line. (6) Main-line C usually exits on the ulnar border of the palm. (7) Interdigital triradius bc (with an accessory triradius as part of the pattern in I_1 area). (8) Single transverse flexion crease increased in frequency.

tips may not really be characteristic of this syndrome because the apparent increase is caused by a small number of individuals (20 percent) with more than six whorls, whereas the majority of patients (60 percent) had three whorls or less, including 20 percent of the patients with no fingertip whorls. The whorls were predominantly of a simple type.

The mean TFRC, decreased in male patients, was increased among the females. However, because of a wide range of TFRC values (0–199) among individual patients, this feature cannot be considered diagnostically useful in the cri-du-chat syndrome.

The axial triradius was distally displaced in the majority of the palms, in 75 percent to the r' and in 10 percent to the r'' position. The elevation of the axial triradius was associated with an increase of the maximal mean atd angle to 50.8° per palm. The elevated axial triradius, in most cases, was not accompanied by a hypothenar

pattern. Hypothenar patterns were actually decreased in frequency in patients with the cri-du-chat syndrome and the frequency of thenar patterns, both loops and whorls, was increased. Among the interdigital patterns, there was an increase of pattern frequency in the I_4 area and a slight decrease in the I_3 area.

Warburton and Miller (1967) pointed out that 72.4 percent of the fourth interdigital loops, where main lines could be traced, resulted from the D main line. Additional data pooled from the literature were in agreement with this finding, bringing the total frequency of this type of fourth interdigital loop to 75.3 percent. This frequency is considerably higher than the frequencies of such loops found in a normal series of German males (Cummins and Midlo, 1961, p. 111) or in a series of phenotypically normal North American individuals, where 33 and 39 percent, respectively, of the I_4 loops resulted from D-line termination.

Unlike controls, where the most frequent terminations of the C line are in the third or fourth interdigital areas of the palms, the usual termination of the C line in patients with the 5p- syndrome was the ulnar border of the palm (Warburton and Miller, 1967). In pooled data, an ulnar exit of the C line was found in 60 percent of the palms in which the C line was present but only in 0.25 percent of the palms in the control series.

Partial syndactyly of the fingers or toes is relatively frequent among patients with the cri-du-chat syndrome (Hustinx and Wijffels, 1965; Rohde and Tompkins, 1965; Warburton and Miller, 1967; Grotzky *et al.*, 1971; Niebuhr, 1971). In some of these patients, as well as in several others without any visible syndactyly, fusion of the *b* and *c* triradii was observed. As has been pointed out by Warburton and Miller (1967), the fusion observed in the latter group of patients may nevertheless indicate a tendency toward syndactyly that has not been realized embryologically but that has affected the dermatoglyphic patterns. It has also been observed (Warburton and Miller, 1967) that in each case with fusion of *b* and *c* triradii, accessory triradii are present as part of whorls or loops in the fourth interdigital area.

A single transverse flexion palmar crease was found in 82.2 percent of all the palms analyzed. Seventy-four percent of all the simian creases were listed as true simian lines and the rest were transitional simian lines.

Analysis of the sole patterns did not reveal any striking abnormalities. In the hallucal area, an increased frequency of distally oriented loops (69.2 percent) was observed, whereas whorls, tibial

loops, and arches were somewhat decreased in frequency (21.1 percent, 3.9 percent, and 5.8 percent, respectively). Penrose and Loesch (1970) found normal pattern intensity in the analyzed soles of patients with the cri-du-chat syndrome. Using their topological approach, they found an excess of loops III and IV, which was balanced by a deficit of I and III loops.

Dermatoglyphic features characteristic for the cri-du-chat syndrome are summarized in Figure 6.13 and Table 6.30. Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Antich *et al.* (1968), Berg *et al.* (1965), Breg *et al.* (1970), Catti and Schmid (1971), De Capoa *et al.* (1967), de Grouchy *et al.* (1964, 1965), Dumars *et al.* (1964), Dyggve and Mikkelsen (1965), Esposito *et al.* (1970), Genest *et al.* (1965), Giorgi *et al.* (1970), Hijmans and Shearin (1965), Jackson and Barr (1970), Kajil *et al.* (1966), Laurent and Robert (1966), Lejeune *et al.* (1963, 1964, 1964, 1965), Macintyre *et al.* (1964), McCracken and Gordon (1965), McGavin *et al.* (1967), Mennicken *et al.* (1968), Miller *et al.* (1966), Milunsky and Chit-ham (1966), Niebuhr (1972), Philip *et al.* (1970), Punnett *et al.* (1964), Reinwein and Wolf (1965), Ricci *et al.* (1965), Schlegel *et al.* (1967), Schmid and Vischer (1967), Sedano *et al.* (1971), Singh *et al.* (1973), Steele *et al.* (1966), Taillemite *et al.* (1973), Taylor (1967), Turner *et al.* (1966), Vassella *et al.* (1967), Vis-sian *et al.* (1971), Wolf and Reinwein (1967), and Wolf *et al.* (1966).

DELETION OF THE SHORT ARM OF CHROMOSOME 4 (WOLF-HIRSCHHORN SYNDROME)

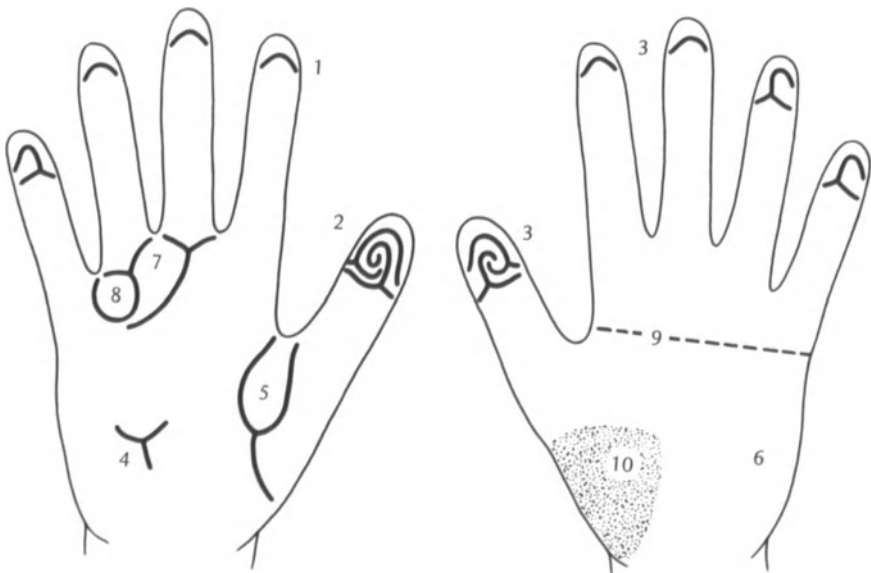
Patients with this syndrome share many clinical features in common with patients having the cri-du-chat syndrome. Among these are low birth weight, mental deficiency, growth retardation, microcephaly, hypertelorism, strabismus, epicanthal folds, a broad based nose, micrognathia, and muscular hypotonia. Mental and growth retardation tend to be more profound in patients with a deletion of the short arm of chromosome 4 than in those with chromosome 5 deletion. In addition, there are other signs that help differentiate the two chromosomal deletions: 4p- syndrome is associated with carplike mouth, cleft or high-arched palate, beaky nose, preauricular dimple, hypospadias, and delay in ossification of pelvic and carpal bones. Frequently, the patients have seizures, deformed ears, sacral dimple, foot deformities, prominent glabella, and

cryptorchidism. In some cases, hemangioma of the forehead, mid-line scalp defect, and coloboma of the iris or retina were reported. Moreover, the catlike cry of infants, characteristically associated with the 5p- syndrome, is not present in 4p- syndrome.

Dermatoglyphic data were pooled of only those cases where the deletion of part of the short arm of chromosome 4 was cytologically proved and was found to be the only chromosomal aberration visible.

Dermatoglyphic analysis revealed several unusual traits (Figure 6.14, Table 6.30). On the fingertips, there was a significant increase of arches, more marked in males (42.9 percent) than in females (23.8 percent). The majority of patients with a low number of arches rather than a small number of cases with high proportions of fingertip arches contributed to this increase. Seventy-five

FIGURE 6.14 Dermatoglyphic features of the 4p- syndrome. (1) Arches significantly increased in frequency; low TFRC. (2) Whorls significantly decreased in frequency. (3) Association of W^{d1} on thumb and A on the second and third digits. (4) Axial triradius somewhat distal (t'). (5) Thenar patterns somewhat increased in frequency. (6) Hypothenar patterns decreased in frequency. (7) I_s patterns decreased in frequency. (8) I_t patterns increased in frequency. (9) Single transverse flexion crease increased in frequency. (10) Ridge dissociation.



percent of patients had three or fewer arches on their fingertips. Whorls were very much diminished in frequency. The frequency of ulnar loops was also decreased in patients of both sexes. Warburton (1969) pointed out an association of a double loop on the thumb and arches on the second and third fingers, which was found in many cases of chromosome 4 deletion but not in patients with chromosome 5 deletions. This combination of patterns is relatively rare in normal individuals and Warburton thought it to be a discriminative feature in 4p- and 5p- syndromes.

The TFRC, although reported only in a small number of cases, appeared to be consistently low. The mean TFRC was found to be 45.0 in male and 57.0 in female patients. Decreased TFRC was also reported by Wolf *et al.* (1965) and in three patients of Miller *et al.* (1970).

The axial triradius was distally displaced in about 38 percent of palms, in most cases to t' rather than t'' position. Correspondingly, the mean summed *atd* angle was increased in males to 126.0°. In the single female patient whose *atd* angle was reported, the summed *atd* angle was 50°, which is less than is found in controls.

The thenar pattern frequency was increased, whereas hypothenar patterns were found with decreased frequency on the palms. In the interdigital areas, a decreased frequency of patterns was observed in the I_3 area, but an increase of pattern frequency was found in the I_4 area. Unlike patients with the cri-du-chat syndrome, those with the 4p- syndrome did not show any tendency toward an unusual source of the patterns in the fourth interdigital palmar area. Only about 37 percent of the loops in the I_4 area were derived from main-line D, which is comparable with our observations in healthy North American controls and considerably less than the 72.4 percent observed in the 5p- syndrome by Warburton and Miller (1967). In contrast to the cri-du-chat syndrome patients, the majority of patients with the Wolf-Hirschhorn syndrome showed an exit of main-line C in the I_3 or I_4 areas rather than on the ulnar border of the palm.

A single transverse palmar flexion crease was found on more than half of all palms inspected.

Perhaps the most striking dermatoglyphic trait of the 4p- patients is ridge dissociation. Although rare in controls and in most medical disorders, it was found in 81 percent of the reported cases of Wolf-Hirschhorn syndrome. The extent of involved epidermis varied between small areas (Wolf *et al.*, 1965) to the ridge dissociation involving the whole surface of the volar aspects of the hands and soles (Passarge *et al.*, 1970; Taylor *et al.*, 1970; Mikelsaar *et*

al., 1973). Ridge dissociation was responsible for the incompleteness of dermatoglyphic data in many reports of patients with 4p—deletions.

The dermatoglyphics in Wolf–Hirschhorn syndrome showed some similarities as well as important differences when compared to the cri-du-chat syndrome (Table 6.30). In both syndromes, there was an increase of pattern frequency in the thenar and I₄ areas, a decrease of patterns in the hypothenar area, elevated axial triradii, and a high proportion of palms with a simian crease. However, the frequency of both single transverse palmar creases and distally displaced axial triradii was much higher in the cri-du-chat syndrome than in the Wolf–Hirschhorn syndrome. Dermatoglyphic traits that differentiate 4p— from 5p— include an increased frequency of the fingertip arches, low TFRC, and ridge hypoplasia, whereas a high frequency of the fourth interdigital loops resulting from main-line D, and a high frequency of ulnar exits of main-line C are seen in 5p— and not in 4p— syndrome.

Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Arias *et al.* (1970), Carneiro *et al.* (1967), Carter *et al.* (1969), Citoler *et al.* (1971), Fryns *et al.* (1973), Golbus *et al.* (1973), Guthrie *et al.* (1971), Hijmans and Shearin (1965), Hirschhorn *et al.* (1965), Miller *et al.* (1966), Pfeiffer (1968), Šubrt and Blehová (1972), Šubrt *et al.* (1969), and Wilson *et al.* (1970).

DELETIONS OF CHROMOSOME 18

Deletion of the short arm of chromosome 18 (18p—)

Clinically, the 18p— syndrome shows a variable phenotype. The patients generally have low birth weight and somatic growth retardation. Mental retardation of markedly variable degree is a constant feature of the syndrome, ranging from borderline to very profound. Although there is no characteristic facial dysmorphism, such anomalies as hypertelorism, strabismus, epicanthal folds, and ptosis of the eyelids are common. The nasal bridge is often flattened or broad. The ears are usually large, floppy, poorly formed, and sometimes low set. The mandible is generally small and severe dental caries is frequently observed. Webbing of the neck, lymphedema, shield chest, and widely spaced nipples are noted. The hands are usually stubby with short fingers. Sometimes, there is a partial syndactyly of the toes.

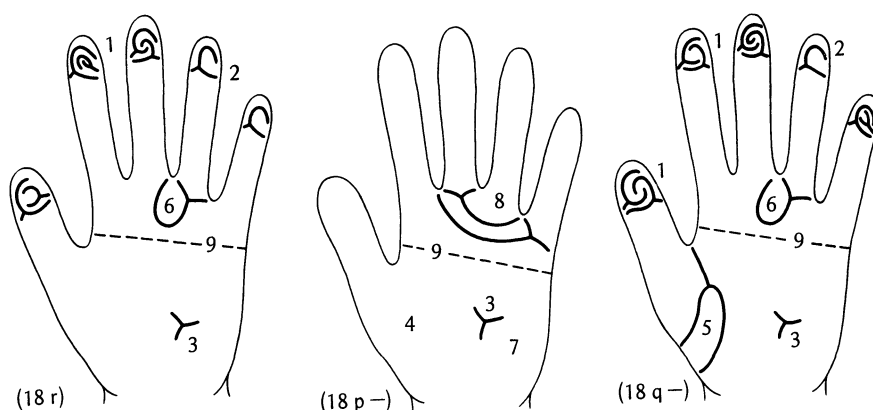


FIGURE 6.15 Dermatoglyphic features of the different deletions of chromosome 18. (1) Whorls increased in frequency. (2) Ulnar loops decreased in frequency. (3) Distal axial triradius. (4) Thenar patterns increased in frequency. (5) Thenar patterns decreased in frequency. (6) I_3 patterns increased in frequency. (7) Hypothenar patterns decreased in frequency. (8) Missing c triradius. (9) Single transverse palmar crease increased in frequency.

Dermatoglyphic findings in the 18p- syndrome are not striking (Figure 6.15). The pooled data indicated a slight decrease of arches and radial loops and marginal increase of whorls on the fingertips but there were only a few patients with 18p- whose dermatoglyphic data were reported (Table 6.31). The TFRC was somewhat increased in both sexes. An unusual finding was a very high occurrence of displaced axial triradii. Whereas 7.5 percent of the palms showed an ulnar triradius, another 72.5 percent had an elevated axial triradius. The distal displacement, however, was usually only into the t' (60.0 percent) rather than t'' (12.5 percent) position. The frequency of both the thenar and hypothenar patterns was diminished. The incidence of patterns in the third interdigital area was somewhat increased and a corresponding decrease of I_4 patterns was noted compared to the control values. The c triradius was missing bilaterally in three patients (Reinwein *et al.*, 1968; Ayraud *et al.*, 1969; Malpuech *et al.*, 1971) and a unilateral abortive c triradius occurred in one additional patient (Jacobsen and Mikkelsen, 1968) among 15 cases in which interdigital area patterns were reported.

Single transverse palmar creases were found on 14 percent of the palms. Marked palmar and plantar creases were noted in two patients (Laurent *et al.*, 1970; Sabater *et al.*, 1972).

TABLE 6.31. Comparison of dermatoglyphic features in deletions of chromosome 18

FEATURE	18p—	18q—	18r
Fingertip patterns			
Arch	2.8%	0.8%	3.0%
Radial loop	2.2%	2.5%	4.6%
Ulnar loop	61.1%	43.6%	46.2%
Whorl	33.3%	52.6%	46.2%
Unknown	0.6%	0.5%	0.0%
Number of fingertip whorls per individual			
0	16.7%	10.3%	19.2%
1-3	38.9%	20.5%	23.1%
4-5	22.2%	23.1%	15.4%
6-10	22.2%	46.1%	42.3%
Mean TFRC			
Males	179.7	150.0	100.0
Females	142.0	137.3	130.3
Total	158.1	142.0	126.5
Mean <i>a-b</i> ridge count	—	34.3	39.5
Mean <i>atd</i> angle	—	51.0°	44.5°
Distal axial triradius^a	—	53.1%	56.0%
Single transverse crease^a	14.3%	44.8%	13.0%
Palmar patterns^a			
Thenar/I ₁	0.0%	29.2%	14.7%
I ₃	53.3%	75.0%	63.9%
I ₄	38.5%	36.4%	52.9%
Hypothenar	15.8%	46.2%	33.3%

^a Percent of palms with the trait.

There is a lack of data concerning the plantar dermatoglyphics in the 18p— syndrome. Bilateral whorls were found in the hallual areas of all three patients in which plantar dermatoglyphics were reported (Schwanitz *et al.*, 1969; Fischer *et al.*, 1970; Laurent *et al.*, 1970) and bilateral fibular loops on the great toes of both patients in whom these areas were described (Schwanitz *et al.*, 1969; Fischer *et al.*, 1970).

Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: de Grouchy *et al.* (1963, 1966, 1967), Feingold *et al.* (1969), Finley *et al.* (1972), Gilgen-

krantz *et al.* (1968), Giraud *et al.* (1971), Lejeune *et al.* (1966), Migeon (1966), Pfeiffer (1966), Šubrt and Beránková (1970), Summitt (1964), Thieffry *et al.* (1963), Uchida *et al.* (1965), and Vaillaud *et al.* (1970).

*Deletion of the long arm of chromosome
18 (18q—)*

18q— syndrome is associated with characteristic clinical findings, such as low birth weight, mild microcephaly, profound mental retardation, retarded somatic growth, short stature, frequent hypotonia, and seizures. There is a characteristic midfacial dysplasia. Eyes are deeply set, with frequent ophthalmologic defects, such as glaucoma, strabismus, nystagmus, tapetoretinal degeneration, and optic atrophy. The nose is short and the mouth carp shaped in the majority of the patients. The pinnae are somewhat unusual, showing a prominent antitragus and antihelix. Atresia of ear canals is common. Conspicuous subacromial dimples and dimples on the epitrochlea, on the sides of the patellae, and on the back of the hands are frequently observed. Long, tapering fingers and clubfoot are common. About two-thirds of the patients have a congenital heart defect. Hypoplastic genitalia are found in patients of both sexes.

Dermatoglyphics of 18q— syndrome are clearly abnormal. Perhaps the most conspicuous finding has been the high frequency of whorls on the fingertips, whereas the frequencies of arches, ulnar loops, and radial loops have been decreased. Both sexes contributed similarly to the unusual fingertip pattern frequencies. Although about 10 percent of the patients did not have any fingertip whorls, 18 percent showed whorl patterns on all ten fingertips. Over 46 percent of the patients had six or more whorls on their fingertips. The whorls were often of composite character rather than simple, according to Parker *et al.* (1972). They included central loop, lateral loop, and double loop patterns as composites. It was not possible to confirm the conclusion of Parker *et al.* (1972) from reviewing the literature because many investigators did not specify the types of whorls in their reports.

Mean values of the TFRC for both males and females with the 18q— syndrome were slightly elevated compared to Caucasian controls, but less so than in the 18p— syndrome. The mean summed *a-b* ridge count was lower than that of controls. The mean *atd* angle was increased. The widened *atd* angle is explained by the frequent distal displacement of axial triradii, usually into the *t'* position.

A higher than normal pattern intensity was found on the palms,

with the thenar, hypothenar and, especially, the third interdigital area pattern occurrence being increased. I_4 pattern frequency was normal.

Single transverse palmar creases were observed in about 45 percent of all palms. Absent interphalangeal flexion creases were found on the fifth digit (de Grouchy *et al.*, 1964; Lejeune *et al.*, 1966), but information concerning these creases was frequently omitted in the clinical descriptions.

No unusual pattern frequencies were reported on the soles of 18q— patients.

Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Borkowf *et al.* (1969), Cenani *et al.* (1969), Curran *et al.* (1970), De Chieri *et al.* (1971), Destin   *et al.* (1967), Faed *et al.* (1972), Feingold *et al.* (1969), Fraccaro *et al.* (1971), Gouw *et al.* (1973), Insley (1967), Jacobsen *et al.* (1971), Juretic *et al.* (1970), Kushnick and Matsushita (1968), Law and Masterson (1969), Lejeune *et al.* (1967), Le Marec *et al.* (1971), Mavalwala *et al.* (1970), Plato *et al.* (1971), Reinwein *et al.* (1967), Schinzel *et al.* (1975), Stewart *et al.* (1970), Šubrt and Pokorn  y (1970), and Summitt (1969).

Ring chromosome 18 (18r)

Patients with 18r syndrome exhibit clinical features of both 18p— and 18q— syndrome, such as low birth weight, short stature, microcephaly, motor and mental retardation, hypotonia, congenital heart defects, and midfacial dysplasia. Ophthalmologic anomalies include nystagmus, strabismus, hypertelorism, epicanthus, and ptosis. Carp-shaped mouth, micrognathia, anomalies of pinnae, ear canal stenosis, short neck, and skeletal anomalies are frequent findings.

Dermatoglyphic features of 18r syndrome show an overlap between 18p— and 18q— syndromes. In the pooled data, there was a marked increase of whorls on the fingertips in both sexes. The whorl frequency was only slightly lower than in 18q— syndrome. Over 42 percent of the patients showed six or more fingertip whorls, compared to 22 percent of 18p— and 46 percent of 18q— patients.

The mean TFRC of the affected females was not abnormal. There was only one male patient whose TFRC (100) was reported. Both the mean summed *a-b* ridge count and *atd* angle were similar to the control data. The percentage of distally displaced axial tri-radii was closer to that of the 18q— syndrome than to that of the 18p— syndrome.

Pattern frequency in the thenar/first interdigital area was some-

what increased and that of the third interdigital area markedly increased. Frequencies of the fourth interdigital area and hypothernar area were normal.

A single transverse palmar crease was found in 13 percent of the palms, i.e., similar in frequency to that of 18p- patients but much lower than in those with 18q-.

No unusual pattern frequencies were observed on the soles of the 18r patients. In the hallucal area, the loop distal pattern was predominant, accounting for 63 percent of all patterns. The loop fibular pattern was found on 54 percent of the great toes.

Dermatoglyphic data were pooled from the following sources: Bernard *et al.* (1966), Borgaonkar and Scott (1969), Cenani *et al.* (1969), Christensen *et al.* (1970), de Grouchy *et al.* (1964, 1968), Deminatti *et al.* (1969, 1970), Dumars *et al.* (1970), Fujita *et al.* (1968), Gropp *et al.* (1964), Grosse *et al.* (1972), Henocq *et al.* (1973), Hooft *et al.* (1968), Kunze *et al.* (1972), Leisti *et al.* (1968), Lejeune *et al.* (1966), Lucas *et al.* (1963), Palmer *et al.* (1967), Petit and Poncelet (1967), Ricci *et al.* (1970), Richards *et al.* (1970), Stewart *et al.* (1970), Wald *et al.* (1969), and Winter *et al.* (1972).

Single-Gene Disorders and Disorders with Uncertain Genetic Transmission

DE LANGE SYNDROME

Individuals with the de Lange syndrome show mental and growth retardation, with low birth weight, generalized hirsutism, synophrys, long eyelashes, anteverted nostrils, and microcephaly, usually with brachycephaly. A long or protruding philtrum and thin lips also contribute to the characteristic appearance of the patients. The upper limbs are often severely malformed and many patients have oligodactyly or peromelia. Limitation in extension of the elbows, proximally placed thumbs, clinodactyly of fifth fingers, webbing of second and third toes, cardiac defects, undescended testes, and delayed eruption and wide spacing of teeth are also commonly found.

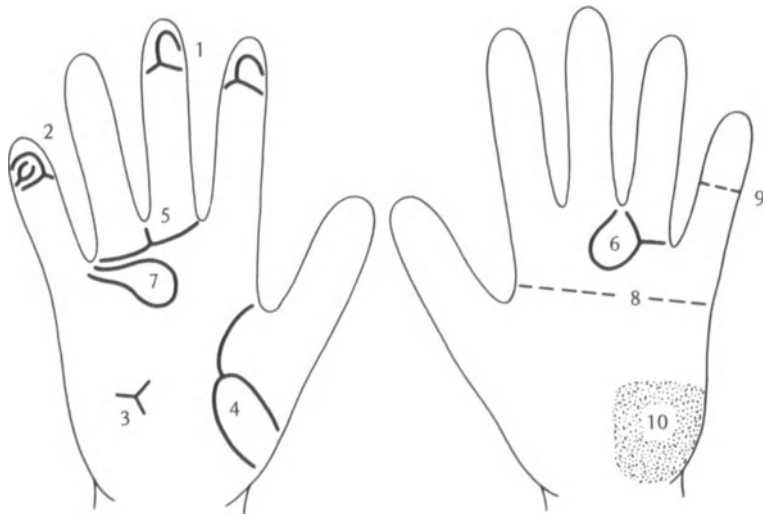
In view of the marked deformities of the hands and feet often present in the de Lange syndrome, associated abnormalities in dermatoglyphics are not surprising (Figure 6.16). However, not all dermatoglyphic peculiarities observed in the syndrome can be accounted for simply on this basis. One of these peculiarities is an unusual distribution of fingertip pattern types. In pooled material,

radial loops were markedly increased in frequency. Although the majority of them occur on the second digit, 44 percent of radial loops were found on other digits, particularly on the third digit. Whorl frequency was considerably decreased. About 76 percent of the reported patients did not have any fingertip whorls at all. The TFRC was low.

The axial triradius was often distally displaced, which resulted in a wider *atd* angle.

Striking changes affecting the distal palmar area proximal to the three ulnar digits were often observed on the palms. More than 41 percent of individuals with de Lange syndrome showed at least unilateral fusion of the *b* and *c* triradii, producing an interdigital or “zygodactylous” triradius in the third interdigital area (Figure 6.16). This configuration occurred even when fingers were well separated. The fused triradius was also observed on the feet (Broholm *et al.*, 1968).

FIGURE 6.16 Dermatoglyphic features of the de Lange syndrome. (1) Radial loops increased in frequency, often found on digits other than the second (particularly on the third). (2) Whorls markedly decreased in frequency. (3) Distal axial triradius. (4) Thenar patterns increased in frequency. (5) Interdigital triradius *bc*. (6) I_s loops increased in frequency. (7) Transverse or oblique loop in the I_s area. (8) True or transitional single transverse palmar crease increased considerably in frequency. (9) Single flexion crease occasionally present on the fifth digit. (10) Ridge dissociation common.



Another unusual dermatoglyphic feature of the de Lange syndrome was a transverse or oblique loop in the I_4 area (Pfeiffer and Kumbnani, 1967; illustrations of Smith, 1966; Grosse and Opitz, 1971). Grosse and Opitz (1971) found them in ten of 17 patients; in four of them they were bilateral. In the pooled data, there was a significantly increased frequency of I_3 loops, which were observed in the majority of the patients bilaterally.

Thenar patterns were considerably increased in frequency in the de Lange syndrome, whereas hypothenar patterns were somewhat less frequent than in controls.

TABLE 6.32. Dermatoglyphic features of the de Lange syndrome

FEATURE	NUMBER	VALUE
Fingertip patterns		
Arch	56	9.0%
Radial loop	110	17.8%
Ulnar loop	390	62.9%
Whorl	53	8.5%
Unknown	11	1.8%
Radial loops on individual digits		
First	0	0.0%
Second	62	56.4%
Third	37	33.6%
Fourth	9	8.2%
Fifth	2	1.8%
Mean TFRC		
Males	13	97.3
Females	8	63.1
Total	21	84.3
Mean summed <i>atd</i> angle		
Males	23	106.0°
Females	14	111.2°
Total	37	107.9°
Distal axial triradius ^a	56	41.5%
Palmar patterns ^a		
Thenar/ I_1	19	25.3%
I_3	57	59.4%
Hypothenar	40	33.3%
Interdigital triradius	18	28.6%
Single transverse palmar crease ^a	122	65.2%

^a Number of palms with the trait.

Ridge dissociation involving small or large areas of the digits, palms, and soles was relatively common. It has been reported in at least 30 patients, which represents 27 percent of cases with de Lange syndrome in which the abnormality was specifically sought.

Either a true or transitional single transverse palmar crease was found in over three-fourths of all individuals with the de Lange syndrome. In 66 percent of the patients, a simian crease was present bilaterally. A single flexion crease was occasionally observed on the fifth finger.

Dermatoglyphic features characteristics of the de Lange syndrome are summarized in Figure 6.16 and Table 6.32. Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Aberfeld and Pourfar (1965), Abraham and Russell (1968), Beck (1974), Beratis *et al.* (1971), Berg *et al.* (1970), Bryson *et al.* (1971), Cherington *et al.* (1969), Choo and Bianchi (1965), Dodge (1965), Falek *et al.* (1966), Gans and Thurston (1965), Hooft *et al.*, (1965), Jervis and Stimson (1963), Kurlander and DeMyer (1967), McArthur and Edwards (1967), Milot and Demay (1972), Noé (1964), Pashayan *et al.* (1969, 1970), Passarge *et al.* (1971), Payne and Maeda (1965), Pearce *et al.* (1967), Podhradská *et al.* (1968), Ptacek *et al.* (1963), Richter (1961), Salazar (1966), Schlesinger *et al.* (1963), Schuster and Johnson (1966), Shear *et al.* (1971), Silver (1964), Smithells (1965), Verma (1970), Vischer (1965), Walbaum *et al.* (1969), and our own unpublished data.

RUBINSTEIN-TAYBI SYNDROME

The Rubinstein-Taybi syndrome includes developmental retardation, broad terminal phalanges of thumbs and halluces, and an abnormal facial appearance. The forehead tends to be prominent. Peculiarities of the eyes include an antimongoloid slant of the palpebral fissures, strabismus, epicanthal folds, heavy or high-arched eyebrows, long eyelashes, and lid ptosis. The ears are abnormal in position, size, or shape. The nose has a beaked or straight appearance with a broad nasal bridge and the nasal septum extends below the alae. The mouth often appears small and a high-arched palate and mild retrognathia are common. Grimacing or an unusual smile is noted in the majority of patients.

The head circumference, stature, and bone age of almost all the reported patients was below the fiftieth percentile. Mental, motor, language, or social retardation is a constant feature of the syndrome.

The terminal phalanges of the thumbs and halluces are broad

TABLE 6.33. Dermatoglyphic features of the Rubinstein–Taybi syndrome

FEATURE	JAPANESE PATIENTS EXCLUDED		JAPANESE PATIENTS ONLY ^a		TOTAL	
	<i>n</i>	VALUE	<i>n</i>	VALUE	<i>n</i>	VALUE
Fingertip patterns						
Arch	82	15.8%	1	1.0 ^b %	83	13.4%
Radial loop	33	6.4%	5	5.0 ^b %	38	6.1%
Ulnar loop	259	49.8%	65	65.0 ^b %	324	52.3%
Whorl	138	26.5%	29	29.0 ^b %	167	26.9%
Unknown	8	1.5%	0	0.0 %	8	1.3%
Radial loops on individual digits						
First	4	12.1%				
Second	16	48.5%				
Third	10	30.3%				
Fourth	2	6.1%				
Fifth	1	3.0%				
Mean TFRC						
Males	8	149.4				
Females	17	94.9				
Total	25	112.3				
Mean <i>atd</i> angle						
Males	30	55.5°				
Females	50	48.0°				
Total	88	51.8°				

^a Shiono *et al.* (1971).
^b Japanese controls: arch = 1.7%, radial loop = 3.3%, ulnar loop = 47.5%, whorl = 47.5%.

and often the other fingers are similarly involved. Clinodactyly of the fifth fingers and overlapping toes are common. Angulation deformity of the thumbs and halluces with abnormal shape of the proximal phalanx is also frequently observed. A duplicated distal or proximal phalanx of the hallux was reported in several patients. Dermatoglyphics of patients with Rubinstein–Taybi syndrome show numerous unusual features (Table 6.33). From pooled data in the literature, there appeared to be a decreased frequency of ulnar loops and a higher frequency of arches on the fingertips. This tendency, however, was not observed in a series of ten Japanese patients (Shiono *et al.*, 1971), who had an increased frequency of ulnar loops and a decreased frequency of whorls in comparison to Japanese controls. No increase of arches was observed.

Although the frequency of radial loops in the pooled data was

FEATURE	JAPANESE PATIENTS EXCLUDED		JAPANESE PATIENTS ONLY ^a		TOTAL	
	<i>n</i>	VALUE	<i>n</i>	VALUE	<i>n</i>	VALUE
Distal axial triradius ^c	47	57.3%	15	75.0%	62	60.8%
Palmar patterns ^c						
Thenar/I ₁	60	55.6%	15	75.0%	75	58.6%
I ₂	19	27.1%	0	0.0%	19	21.1%
I ₃	47	52.8%	2	10.0%	49	45.0%
I ₄	21	50.0%	11	55.0%	32	51.6%
Hypothenar	44	42.7%	3	15.0%	47	38.2%
Single transverse palmar crease ^c	24	38.7%	8	40.0%	32	39.0%
Plantar patterns						
Great toe						
Arch	6	25.0%				
Loop fibular	12	50.0%				
Whorl	6	25.0%				
Hallucal area						
Arch	5	5.8%				
Loop distal	56	65.1%				
Loop tibial	11	12.8%				
Loop fibular	4	4.7%				
Whorl	8	9.3%				
Unknown	2	2.3%				

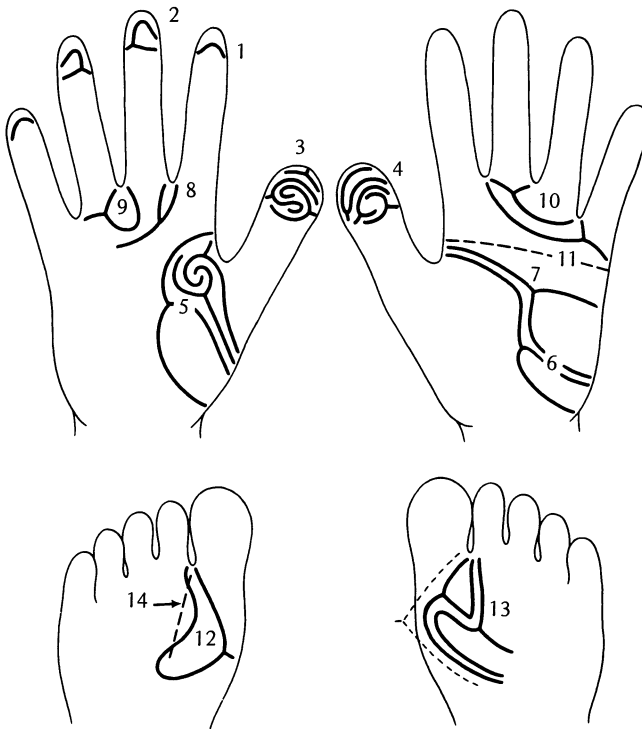
^a Number of palms with the trait.

normal, more than half of all radial loops were found on fingertips other than the second, usually on the third finger. Perhaps the most unusual finding on the fingertips was an additional triradius on the apex of the thumb (Figure 6.17) or big toe in some of the patients with Rubinstein–Taybi syndrome (Berg *et al.*, 1966; Robinson *et al.*, 1966; Salmon, 1968; Bejar and Smith, 1969; Smith *et al.*, 1970). One of the patients (Padfield *et al.*, 1968) possessed this highly unusual dermatoglyphic configuration on both fifth fingers. Unless looked for specifically, the additional apical triradius may be missed. Its actual frequency may be even higher than that suggested from the published reports, therefore, particularly as five of the six patients of Berg *et al.* (1966) showed this trait, four having it bilaterally. Apical triradii were observed in association with all pattern types. Rare double patterns (ulnar loop/radial loop, whorl/loop) were observed

on the thumbs of several patients (Robinson *et al.*, 1966; Giroux and Miller, 1967; Rubinstein, 1969).

The TFRC in Rubinstein–Taybi syndrome was somewhat decreased. However, this apparent decrease was caused by the low value of the mean TFRC among females, whereas males showed a

FIGURE 6.17 Dermatoglyphic features of the Rubinstein–Taybi syndrome. (1) Arches increased in frequency. (2) Radial loops shifted to fingers other than the second. (3) Additional apical triradius on the thumb or great toe. (4) Double patterns on the thumbs. (5) Thenar/I₁ patterns increased in frequency, size, and complexity. (6) Ulnar loop is a predominant pattern in the hypothenar area, associated with (7). (7) Distal axial triradius (and therefore an increased maximal *atd* angle). (8) I₂ patterns increased in frequency. (9) I₃ patterns increased in frequency. (10) Missing *c* triradius. (11) Single transverse palmar crease increased in frequency. (12) Distorted, unusually long distal loop in hallucal area; distal loops increased in frequency. (13) Combination of two loops (L^a/L⁵) in the hallucal area. (14) Deep plantar crease in the first interdigital plantar area.



normal TFRC. Also, the range in the TFRC among reported cases is very wide (9–216); therefore, the TFRC cannot be considered as diagnostically useful in this syndrome.

Although thenar patterns are relatively rare among phenotypically normal individuals, over half of all palms of patients with Rubinstein–Taybi syndrome were found to have large and complex patterns in the thenar/first interdigital area. The patterns were observed usually bilaterally in the same patient and very frequently in both the thenar and the I_1 area simultaneously. In the hypothenar area, the pattern frequency was normal among patients with the Rubinstein–Taybi syndrome, excluding the Japanese series (Shiono *et al.*, 1971). However, ulnar rather than radial loops were predominant, accounting for almost 60 percent of true hypothenar patterns. Ulnar loops, associated with a distal axial triradius, contributed significantly to an increase in the values of the *atd* angle.

An increased frequency of patterns was observed in the second interdigital area on the palms of patients with Rubinstein–Taybi syndrome with the exception of the Japanese patients, none of whom had an I_2 pattern. Pattern frequency in the I_3 area was somewhat increased, again excluding the Japanese series. In the I_4 area, the frequency of patterns was comparable to that of controls in both the Caucasian and the Japanese patients. Distal loops in the interdigital areas were often accompanied by an accessory triradius. A missing *c* triradius has been reported by several investigators (Robinson *et al.*, 1966; Davison *et al.*, 1967; Filippi, 1972). Filippi (1972) found this trait, unilaterally or bilaterally, in five of seven apparently unrelated patients. This is a considerably higher frequency than that encountered in controls. More data concerning the missing *c* triradius in patients with Rubinstein–Taybi syndrome are needed to establish the value of this trait.

A single transverse palmar crease, including its transitional forms, was a common finding.

Distorted and unusually long distal loops were found in the hallucal area in 11 of 16 patients by Padfield *et al.* (1968) and in several patients reported by Giroux and Miller (1967). Berg *et al.* (1966) observed a hallucal pattern containing two loops (L^d/L^f) in three of their six patients. They considered this pattern to be the most useful one for diagnostic purposes. Distal loops in the hallucal area were often described as having a laterally displaced *f* triradius, with or without an associated *e'* triradius, or a distal loop opening to the I_1 space associated with a more proximal fibular loop (Rubinstein, 1969). In the pooled data, loop distal patterns were increased

and whorls decreased in frequency compared to the general population.

A deep crease in the first interdigital plantar area was another unusual trait among patients with the Rubinstein–Taybi syndrome. It was observed in ten of 19 patients summarized by Rubinstein (1969) and in five of seven reported by Filippi (1972). The crease occurred bilaterally in four patients in the latter series.

The pooled information on the frequency of great toe patterns was based on numbers too small to be considered representative for the Rubinstein–Taybi syndrome. An unusual pattern consisting of two fibular loops was observed on one great toe in the series of seven patients reported by Filippi (1972).

Dermatoglyphic features characteristic of the Rubinstein–Taybi syndrome are summarized in Figure 6.17 and Table 6.33. Apart from the references cited in the text, dermatoglyphic data were pooled from the following sources: Buchinger and Ströder (1973), Coffin (1964), Filippi (1969a, b), Galluzzi *et al.* (1974), Herrmann and Opitz (1969), Jancar (1965), Jéliu and Saint-Rome (1967), Johnson (1966), Roy *et al.* (1968), and Simpson and Brissenden (1973).

SMITH–LEMLI–OPITZ SYNDROME

The characteristic features of the Smith–Lemli–Opitz syndrome include moderate to severe mental retardation, growth retardation of prenatal onset, microcephaly, ptosis of eyelids, inner epicanthal folds, internal strabismus, short nose with broad nasal tip and anteverted nostrils, increased nasolabial distance, broad maxillary alveolar ridges, mild micrognathia, low-set ears, short neck, short stature, narrow shoulders, cutaneous syndactyly of the second and third toes, and genital anomalies in the male, including cryptorchidism and hypospadias. Vomiting in early infancy, changes in muscle tone, and a tendency toward a shrill cry are also frequently encountered. Occasionally, the patients have a cleft palate or bifid uvula, metatarsus adductus, deep sacral dimple, pyloric stenosis, and cardiac anomalies.

There were some unusual features in the pooled data on dermatoglyphics of patients with Smith–Lemli–Opitz syndrome. On the fingertips, the frequency of ulnar loops was considerably lower than in controls, whereas arches and whorls were elevated in frequency. In spite of the abnormally high number of whorl patterns in the pooled series of patients, more than half of all patients with Smith–Lemli–Opitz syndrome had only three or fewer whorls on their fin-

gertips, and about 35 percent had only one or none. Approximately a third of the patients had seven or more whorls on their fingertips. Several of those were known to have come from families where at least one of the parents had an excessive number of whorls (Smith *et al.*, 1964; Lowry *et al.*, 1968; Dallaire, 1969; Hoefnagel *et al.*, 1969). However, in the majority of patients, the familial dermatoglyphic data were lacking so that it is still uncertain whether an elevated whorl frequency is a characteristic dermatoglyphic feature of the syndrome.

The mean value of the TFRC was decreased. Very low TFRC's were reported in patients whose predominant fingertip patterns were arches (Schumacher, 1969; Sinclair *et al.*, 1969; Chakanovskis and Sutherland, 1971) as well as in those with a high number of whorls (Lendvai *et al.*, 1969; Nevo *et al.*, 1972). Nevo *et al.* (1972) reported that a low TFRC was observed not only in the patients but also in both the father, who had four fingertip whorls, and the

TABLE 6.34. Dermatoglyphics in the Smith-Lemli-Opitz syndrome

TRAIT	NUMBER	VALUE
Fingertip patterns		
Arch	46	14.2%
Radial loop	20	6.2%
Ulnar loop	102	31.5%
Whorl	156	48.1%
Number of whorls per individual		
0	6	17.1%
1-3	12	34.3%
4-6	4	11.4%
7-10	13	37.1%
Mean TFRC	6	96.3
Distal axial triradius^a	32	50.8%
Palmar patterns^a		
Thenar/I ₁	5	22.7%
I ₃	8	36.4%
I ₄	4	25.0%
Hypothenar	4	18.2%
Single transverse palmar crease^a	58	78.4%

^a Number of palms with the trait.

mother, who had one whorl. A high TFRC was observed in a patient reported by Hoefnagel *et al.* (1969). Both parents and two sisters of this patient also showed an elevated TFRC, which, however, was not as high as that of the patient.

On the palms, there was a paucity of patterns in the hypothenar, third, and fourth interdigital areas. The frequency of thenar patterns seemed to be elevated, but the small number of patients providing information on thenar patterns made this finding inconclusive.

Approximately half of the palms of the patients showed a distally displaced axial triradius, into either the t' or the t'' position.

A majority of the patients had a true or transitional simian crease on the palm and approximately three-quarters of the simian creases were bilateral.

Other peculiarities reported in individual patients with the syndrome include hypoplasia of dermal ridges (Opitz *et al.*, 1969) and a missing triradius c bilaterally (Chakanovskis and Sutherland, 1971). In one patient having bilateral absence of triradius c (Nevo *et al.*, 1972), both parents had a unilaterally missing c triradius. A patient with exit of main-line A in the thenar areas bilaterally was reported by Fried and Fraser (1972) and a single flexion crease on the fifth digit with hypoplasia of distal flexion creases on the third and fourth digits was noted by Opitz *et al.* (1969).

No unusual dermatoglyphic features were reported on the soles.

Dermatoglyphic features characteristic of the Smith-Lemli-Opitz syndrome are summarized in Table 6.34.

CLEFT LIP AND PALATE

Several studies have been carried out in order to determine whether a correlation exists between cleft lip and/or palate and unusual dermatoglyphics. Silver (1966) investigated the fingertips, third interdigital area, and hallual patterns of 71 patients, 60 having cleft lip and palate (CLP), three having cleft lip (CL), and eight having cleft palate (CP) only. He found no significant difference in any dermatoglyphic configurations between patients and controls. Adams and Niswander (1967) studied only the atd angle in patients with cleft lip, with or without cleft palate ($CL \pm P$). After excluding patients with an asymmetry in the hypothenar pattern on the two hands and those who lacked an axial triradius in either hand, they observed an increased asymmetry of atd angles in the group of patients with familial $CL \pm P$. However, no difference in asymmetry was found between either the sporadic cases of $CL \pm P$ or the CP and controls. Some unusual dermatoglyphic features were reported in a

group of 15 patients with several types of CL/P (Wittwer, 1967). Among these were several atypically located triradii and duplications of triradii and a tendency of the palmar main lines toward a vertical alignment. However, because the group of patients investigated was very small and heterogeneous as to the type of cleft, no generalizations could be drawn about dermatoglyphic anomalies in relation to facial clefts.

In a larger study of 152 CLP patients (Dziuba, 1972), the frequencies of thenar and first interdigital area patterns were increased on the left palms in patients when both sexes were combined. In male patients, the TFRC was found to be lower than in controls and the fingertip pattern intensity index was decreased by an increased frequency of ulnar loops and arches and a consequent decrease of whorls. Female patients had a lower frequency of the patterns in the fourth interdigital area of the left hand than did controls.

Piatkowska and Sokolowski (1973) compared dermatoglyphic features of 42 patients with partial or complete clefts of the primary palate (CP I) with those of 91 patients with clefts of the secondary palate (CP II). Among 50 traits that were evaluated, only two significant differences were observed: females with CP I showed a significant decrease of patterns in the fourth interdigital area, particularly of peripheral loops. In a group of males and a combined group of males and females with CP II, a significant decrease of tented loops (III^T) in the third interdigital area was found. A similar but statistically insignificant tendency was observed in female patients with CP II. The authors pointed out the high probability of obtaining these two differences in a study of a large number of dermatoglyphic parameters by chance alone and concluded that their results indicated no significant differences in dermatoglyphics on the palms of patients with CP I and CP II.

Study of the frequency of the simian line in a group of 132 children with cleft palate (Jaworska, 1969) revealed this line to be present in 5.3 percent compared to 3.2 percent of controls. The difference was not significant.

de Bie and Matton (1973) found no statistical differences in the dermatoglyphics of patients with oral clefts and controls in either sex. The CL(P) patients did not differ significantly from those with CP, nor were there any differences between unilateral and bilateral clefts. Similarly, Gall *et al.* (1973) reported no significant differences between dermatoglyphic features of patients with isolated oral clefts and those of controls. However, they found an unusual frequency of some dermatoglyphic patterns among patients whose oral cleft was accompanied by other minor dysmorphogenic

features. It is possible that such patients are responsible for some unusual dermatoglyphics observed in previously cited studies. Silver's (1966, p. 375) conclusion, that "cleft lip/palate is a congenital anomaly whose developmental basis seems to be independent of the production of aberrant dermatoglyphic patterns," seems appropriate on the basis of information now available.

CEREBRAL GIGANTISM

Cerebral gigantism is characterized by excessive length at birth, rapid linear growth in the first years of life, advanced bone age, and acromegalic features, including large head, frontal bossing, prognathism, large hands and feet, poor coordination, clumsiness, and mild mental retardation. Cerebral ventricles may be dilated without increased intracranial pressure. Laboratory studies, including extensive endocrinologic evaluations, have shown no consistent abnormalities and the etiology and pathophysiology of the syndrome remain unclear.

Although unusual dermatoglyphics have been repeatedly cited in early reports of cerebral gigantism, the references have almost invariably been based on individual case reports and have lacked familial control data. Among the reported dermatoglyphic anomalies were increased TFRC (Abraham and Snodgrass, 1969; Ott and Robinson, 1969; Bejar *et al.*, 1970), both high (Milunsky *et al.*, 1967; Abraham and Snodgrass, 1969) and normal (Bejar *et al.*, 1970) *a-b* ridge count, decreased distance between palmar triradii *b* and *c* (Hooft *et al.*, 1968), considerable increase in the mean ridge breadth (Abraham and Snodgrass, 1969), tendency toward vertical (Milunsky *et al.*, 1967; Ott and Robinson, 1969; Bejar *et al.*, 1970) or horizontal (Hooft *et al.*, 1968) alignment of palmar ridges, and unusual exit of main-line A in the thenar area (Milunsky *et al.*, 1967; Ott and Robinson, 1969; Bejar *et al.*, 1970; Fauchier *et al.*, 1970; Schlack and Pfeiffer, 1970). The latter trait was noticed to be associated with a wide separation of triradii *a* and *b* (Milunsky *et al.*, 1967). Bilateral simian creases were reported in one patient of Schlack and Pfeiffer (1970). Unremarkable dermatoglyphics were found in the patients reported by Mace and Gotlin (1970) and by Schneider and Vassella (1971). In a study of six patients with cerebral gigantism and their families (Schaumann and Alter, 1973), the affected individuals showed a significantly increased TFRC and a decreased main-line index compared to their unaffected relatives. A thenar exit of main-line A was not observed in any of the patients, nor were there any other dermatoglyphic

TABLE 6.35. Dermatoglyphic features of cerebral gigantism

	MALES		FEMALES	
	NUMBER	VALUE	NUMBER	VALUE
Fingertip patterns				
Arch	1	1.4%	2	3.3%
Radial loop	4	5.7%	3	5.0%
Ulnar loop	37	52.9%	44	73.4%
Whorl	28	40.0%	11	18.3%
Mean TFRC	6	190.8	5	171.0
Mean <i>a-b</i> ridge count	12	43.9	14	42.7
Mean <i>atd</i> angle	10	39.9°	4	63.3°
Mean main line index	7	16.6	10	14.4

changes. Pooled dermatoglyphic data (Schaumann and Alter, 1973) revealed a significant increase of mean TFRC and the frequency of the thenar exit of main-line A among the patients with cerebral gigantism.

It is tempting to attribute some of the unusual dermatoglyphic features simply to the large size of the hands in cerebral gigantism. For example, a larger number of ridges would be required to cover the volar surface, resulting in an increased TFRC and *a-b* ridge count. Vertical alignment of main lines, reflected by a tendency of the A line to exit in the thenar area and a lower main-line index, could be attributed to the elongation of the hands that is also present in this syndrome. Hence, it remains uncertain whether the unusual dermatoglyphics reflect an underlying direct genetic influence or are merely the result of mechanical modification associated with the large limbs.

Dermatoglyphic features of cerebral gigantism are summarized in Table 6.35.

Nongenetic and Exogenous Factors

RUBELLA EMBRYOPATHY

The marked teratogenicity of the rubella virus during early gestation of the fetus was recognized by Gregg (1941). Affected individuals showed a variety of severe congenital anomalies, including microcephaly, cataracts, heart defects, and mental retardation.

Abnormal dermatoglyphics have also been reported in children

with proved rubella embryopathy, and even in apparently normal children whose mothers were exposed to rubella early during gestation. This suggests that dermatoglyphics may be a sensitive indicator of even subtle intrauterine rubella damage. Achs *et al.* (1966) found an increased frequency of simian lines in both rubella-damaged and rubella-exposed groups of children (10.5 and 10.0 percent, respectively, compared to 1.6 percent in controls). Similarly, distally displaced axial triradii were found in 26.3 and 20.0 percent of the rubella groups, respectively, whereas only 7.6 percent of controls showed this trait. Radial loops on other than second digits were observed more often in both rubella-affected and rubella-exposed groups than in controls. Alter and Schulenberg (1966) noticed an increased frequency (29 percent) of transitional and full simian palmar creases among their patients with proved rubella embryopathy. They also observed several additional unusual dermatoglyphic traits, such as an increased frequency of whorls on the fingertips, a decreased *a-b* ridge count, an increased *atd* angle, and a tendency toward more patterns in the interdigital areas, thenar/first interdigital area, and hypothenar area. Interestingly, the increase in fingertip whorls was markedly higher in male than in female patients (61 and 38 percent, respectively). This observation has since been confirmed in several other studies (Purvis-Smith and Menser, 1968, 1973). In yet another study (Purvis-Smith *et al.*, 1969) of 100 children with congenital rubella, the fingertip whorls were also found to be significantly increased in children of both sexes. However, female patients had somewhat more whorls than male patients. An increased frequency of I_3 patterns and a decreased frequency of I_4 patterns were reported (Purvis-Smith *et al.*, 1969). Both simian and Sydney creases occurred significantly more often among the rubella group than in controls. Only Sydney creases were elevated among 50 adult patients with rubella embryopathy, whereas the frequency of simian creases was found to be normal (Purvis-Smith and Menser, 1968).

von Rott and Jolk (1971) described an infant with congenital rubella who showed four fingertip whorls, distally displaced axial triradii bilaterally, and bilateral I_3 patterns and, in addition, had a ridge dysplasia, which was not observed in the patient's parents.

In an investigation undertaken to determine whether any teratogenic effect of rubella would be demonstrable in altered dermatoglyphic symmetry (Hook *et al.*, 1971), dermatoglyphic prints of 20 infants born to mothers who contracted rubella in early pregnancy were analyzed and compared to 20 controls. Contrary to ex-

pectation, symmetry was increased in the rubella infants. It therefore seems that the teratogenic effects of the rubella virus may override otherwise normal lateral differences of dermatoglyphic traits during development and influence the patterns toward greater symmetry.

In a more recent rubella study, Purvis-Smith and Menser (1973) attempted to ascertain the relative contributions of genetic and viral influences on high whorl frequency, which seems to be the most consistent dermatoglyphic finding in rubella embryopathy. They studied the fingertip frequencies of 120 patients with congenital rubella (both children and adults), their mothers, and 79 of their fathers. An excess of whorls was found in patients of both sexes compared to controls but was expressed more frequently in male patients. The latter possessed significantly more whorls than female patients, 51.5 and 38 percent, respectively. Whereas the mothers of the patients had a whorl frequency similar to that of female controls, the fathers showed a significantly raised whorl frequency compared to the male controls. Because the patients had a markedly higher whorl frequency than either their mothers or fathers, Purvis-Smith and Menser attributed high whorl frequency to direct teratogenesis. However, an underlying inherited component in digital whorl determination was also suggested. Parental pairs were divided into those with high whorl frequency ("high-whorl parents," or HWP) and those with low whorl frequency (LWP). The rubella children of HWP had significantly more whorls than the children of LWP. Nonetheless, an environmental influence on whorl development was apparent even in this comparison. The children with rubella born to both the HWP and LWP had a significantly increased frequency of the fingertip whorls compared to their parents and even the children of LWP had more whorls than the controls. The authors concluded that although both genetic factors and the viral influence may have accounted for the very high whorl frequency in rubella-affected individuals, the teratogenic effect of the virus appeared to predominate.

LEUKEMIA

A number of studies of dermatoglyphics in patients with leukemia have been carried out since Aleksandrowicz *et al.* (1966) reported an increased frequency of fingertip radial loops in males and of "radial whorls" in females. However, there is as yet little consistency in findings among the various investigators and even some

question of whether a deviation from normal really exists. Many of the difficulties in interpreting the results of different studies stem from the fact that different types of leukemia have often been lumped together and, in some studies, the types have not been specified. Moreover, dermatoglyphics have not always been analyzed for each sex separately and the sample size has often been small. Inadequate controls and bias in the statistical analyses characterize other studies. These difficulties have been discussed by Rosner (1970) and by Berka *et al.* (1971).

Acute lymphocytic leukemia

There appear to be no consistent differences in fingertip pattern frequency in patients with acute lymphocytic leukemia compared to the general population (Wertelecki *et al.*, 1969; Berka *et al.*, 1971). Colombo *et al.* (1973) found whorls to be decreased and ulnar loops to be increased in females, whereas Purvis-Smith and Menser (1973) found a higher frequency of whorls in patients of both sexes. The latter investigators noted that fathers of the male patients also had a higher frequency of whorls than the general population and postulated a paternal effect on risk of leukemia. However, in the Wertelecki *et al.* (1973) series, the fathers and the brothers both had a lower frequency of whorls than the male patients. They reported that the frequency of whorls on the fingertips was 33, 27.8, and 26.7 percent in male patients, their fathers, and their brothers, respectively.

Reports concerning the frequency of unusual palmar creases in acute lymphocytic leukemia vary. An increase of Sydney lines without an increase of single transverse creases was observed in several investigated groups of patients (Wertelecki *et al.*, 1969, 1973; Colombo *et al.*, 1973). In the Colombo *et al.* (1973) study, the increase in frequency of the Sydney line was limited to male patients. Purvis-Smith and Menser (1973) found an increase of single transverse creases among the female patients and an increase of single transverse creases together with Sydney lines in female patients and their mothers. In contrast to these reports, no abnormal frequencies of the single transverse palmar crease and Sydney line were observed by other investigators (Rosner, 1969; Berka *et al.*, 1971).

Acute myelogenous leukemia

In four patients with acute myelogenous leukemia studied by Berka *et al.* (1971), the fingertip patterns were not unusual. Rosner

(1969) reported increased radial loops on the right hand of males and, in females, there was an increased *atd* angle and a higher frequency of hypothenar patterns of the left palms.

Acute leukemia (not otherwise designated)

No significant fingertip pattern deviations from normal were observed by Zahálková and Běluša (1970) or by Berka *et al.* (1971) in patients with acute leukemia not otherwise designated. However, Verbov (1970) reported an increase of whorls and a decrease in ulnar loops in the male patients in his series. Menser and Purvis-Smith (1972) found a decreased frequency of ulnar loops and an increased frequency of arches on the fingertips of male children with acute leukemia. Female children had an increase of fingertip whorls and a decrease of arches. Single transverse creases were increased among the latter group of patients. Nora *et al.* (1969) did not find any significant increase of either single transverse palmar creases or Sydney lines among patients with acute leukemia.

Acute blast cell leukemia

Menser and Purvis-Smith (1969) reported an increase of arches and a decrease of ulnar loops in the fingertips of a group of patients with an acute blast cell leukemia. The patients had a significantly increased frequency of Sydney lines and an insignificant increase of single transverse palmar creases.

Chronic lymphocytic leukemia

Rosner (1969) did not find any abnormal dermatoglyphic features among male patients with chronic lymphocytic leukemia. However, female patients had increased frequencies of fingertip arches and hypothenar patterns on the right palms, wider *atd* angles, and a decreased frequency of I_4 patterns. Aleksandrowicz *et al.* (1969) found an increase of the fingertip radial loops in males and of the "radial whorls" in females. Verbov (1969) observed an increased frequency of radial loops and a decrease of fingertip arches in males whereas females had an increased frequency of fingertip arches.

Chronic myelogenous leukemia

An increased frequency of whorls and a decreased frequency of ulnar loops on the fingertips of the patients was reported by Rosner (1969). In addition, male patients in this study showed an increase in patterns in the third interdigital area. Loesch and Mdzewski

(1971) did not find any differences in frequency of the dermatoglyphic pattern types on fingers, palms, or soles between the patients and healthy controls, nor were there any differences in mean *atd* angle, TFRC, or ridge breadth.

CYTOMEGALIC INCLUSION DISEASE

Because the cytomegalovirus, like rubella virus, may have a teratogenic effect on the fetus during the first trimester of gestation, a question has been raised as to whether it influences the formation of epidermal ridge patterns. Wright *et al.* (1972) analyzed the dermatoglyphics in 15 infants with cytomegalic inclusion disease (CID), in eight of their mothers, and in five of their fathers. They found the frequency of whorl patterns on the fingertips increased to 43 percent. The mothers' whorl frequency was comparable to controls. However, the fathers whose prints were available showed a fingertip whorl frequency of 66 percent, significantly higher than in controls. The TFRC varied greatly among the patients between 0 and 208, with an average of 155 in males and 80 in females; data on only four females could be analyzed. On the palms, a classical simian line was observed unilaterally in one patient and transitional transverse creases were found bilaterally in eight other patients, i.e., in 53 percent of the palms.

In another study of eleven children (six males and five females) with CID, Purvis-Smith *et al.* (1972) also observed an increase in fingertip whorls in patients (31 percent) and an even higher increase in their fathers (39 percent), whereas the mothers and siblings showed only 21 and 23 percent, respectively, compared to 26 percent whorls in controls. No patient had a simian line, but four individuals showed a Sydney line, two of them bilaterally. Otherwise, no significant abnormalities were found among dermatoglyphic characteristics of patients or their parents. As Purvis-Smith *et al.* (1972) suggested, a certain bias might have been introduced in both CID studies, because the time of maternal infection during pregnancy in the majority of cases was either not known or not stated. It is therefore possible that in some of the patients the infection may have been acquired during later stages of pregnancy, after differentiation of the epidermal ridges has been completed. Purvis-Smith *et al.* (1972) also pointed out parallels in the results of dermatoglyphic studies in patients with congenital rubella and congenital CID in which an increased digital whorl frequency was found. The fathers of these individuals consistently had substan-

tially higher whorl frequencies than male controls. The authors concluded that genetic factors controlling immune responses may be inherited from the father and predispose a particular fetus to damage from certain viral infections. The dermatoglyphics may constitute a sensitive indicator of this damage.

CELIAC DISEASE

David *et al.* (1970) reported fingerprint changes in patients with celiac disease, a disorder characterized by malabsorption, abnormal small bowel structure, and intolerance to gluten. Dermatoglyphic changes varied between moderate epidermal ridge atrophy associated with the appearance of white lines and actual loss of visible ridges and disappearance of white lines. These abnormalities were described in adults with celiac sprue, 95 percent of whom showed some ridge atrophy. Whether children also showed the same changes was unclear in this study as the number of children with untreated celiac disease was too small to draw conclusions.

Epidermal ridge atrophy was found to improve on treatment with a gluten-free diet. Because the ridges also deteriorated in patients with clinical deterioration, the authors suggested that dermatoglyphics could be employed as an indicator of the patients' response to the diet therapy.

McCrae *et al.* (1971) analyzed the fingerprints of 23 children suffering from celiac disease, six of them untreated, five partially treated, and 12 treated and judged completely well. Epidermal ridge atrophy was not found in any of the patients and white lines did not occur more frequently than in controls. The authors did not consider abnormal dermatoglyphics as a diagnostic feature of celiac sprue. In another study, Mylotte *et al.* (1972) reported that the degree of abnormality was not different from controls. David *et al.* (1973) reexamined the prints of Mylotte *et al.* (1972) and criticized their conclusions on the grounds that only the fingertips were studied, patients were printed only once, and the quality of the prints was poor. David *et al.* (1973) agreed that none of the 31 adults in the Mylotte *et al.* (1972) study had the severe epidermal ridge atrophy that had been found in nine of 105 adults with celiac disease in their own study. However, ridge atrophy of lesser degree was present. David *et al.* repeated their study in 1973 and, among 48 untreated adult patients, 43 had epidermal ridge atrophy in fingerprints and palm prints, usually worse on the palms than on the fingers. Fifty-two of 57 patients already on the diet also had ridge

atrophy. Although the ridge atrophy improved with a gluten-free diet, it did not completely return to normal until several years had elapsed. Among 22 untreated children, only one had abnormal ridges. Even this patient, a 3-month-old infant, subsequently developed normal ridges without treatment. All 34 children already on the diet had normal prints. The actual severity of the ridge atrophy in adults was found to be poorly correlated with the severity of the malabsorption. Although the authors suggested that ridge atrophy in an adult should alert the physician to the possibility of celiac sprue, they did not find dermatoglyphic analysis to be a reliable method of monitoring the response of the celiac patient to the gluten-restricted diet.

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CONGENITAL MALFORMATIONS OF HANDS AND FEET

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AUTOSOMAL TRISOMIES

Trisomy 21 (Down syndrome)

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CHAPTER 6: MEDICAL DISORDERS

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STRUCTURAL CHROMOSOMAL ABERRATIONS

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SINGLE-GENE DISORDERS AND DISORDERS WITH UNCERTAIN GENETIC TRANSMISSION

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