DERMATOGLYPHICAL FINDINGS IN PRIMARY AMENORRHOEA@

by

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Introduction

Dermatoglyphics or the study of dermal patterns on the finger prints has become a science for the medical profession in the last few decades. The term dermatologlyphics, first coined by Cummins and Midlo in 1926, is a Greek word, derma meaning 'skin' and glyphic meaning 'to carve'. It consists of the study of epidermal ridges on the fingers, palms and soles. However, its usefulness as a science in medical biology was demonstrated by Galton in 1892 who first systematically described the various configurations.

These epidermal ridges are formed in the third month of intrauterine life. No change occurs after that, either in the detailed structure or in the arrangement of the ridges. Even after birth, no developmental changes occur in the ridges.

The ridges are made up of the spores of the sweat glands. These are present on the palms, palmar surface of the fingers, on the soles and the plantar surface of the toes. The ridges are not continuous but consist of segments of various lengths. Variability is so much that the details of small areas are never repeated either in the same individual or in a different individual. Even identical twins differ in minor ridge characteristics. That is how these palm prints today are extensively used for the identification of the persons.

Penrose, Cummins and others described the various techniques to obtain prints of palms and soles, the quantitative estimation of ridge counts, the knowledge of influence during foetal development and of heredity on the configurations of pattern and its association with various congenital and chromosomal aberrations and various disorders.

At the junction of three ridge systems, a radiate structure is formed which is called a 'Triradius'. In the palm, at the base of each finger is a triradius called 'a, b, c and d'. There is also an axial triradius called 't'. By marking the ridges from triradius features of ridge arrangement are obtained.

The palms were divided into thenar and hypothenar patterns and interdigitate patterns by the ridge

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systems. Joining the axial triradius 't' to the triradii 'a' and 'd', the angle 'atd' is formed. In the normal persons, the angle 'atd', is about 48°. The position of axial triradius 't' is determined by heredity. The angle will be more in cases of Turner's syndrome, Mongolism and other conditions.

In the fingers, ridges form three main types—an arch, a loop and a whorl. The arch has no triradius, the loop has one triradius and the whorl has two triradii. Ridge-count is done from the triradius to the core of loop or whorl. In the arch, the ridge count is zero as there is no triradius, the loop has usually 12 ridges and the whorl about 19 ridges. The loops are named ulnar and radial depending upon which side they open. In the general population, loops form 69%, whorls form 26% and arches 5%. Men have more whorls and so have a higher ridge count. The average ridge count in women is 127.

Though the sole is also divided into different patterns, only the hallucal area under the big toe is important. It has a triradius 'f' on the tibial side, 'p' on the proximal side.

This paper deals with the applications of dermatoglyphics in patients with sex chromatin abnormalities and genital tract anomalies, studied at the genetic clinic of the Department of Medicine, B. J. Medical College and Sassoon General Hospitals, Poona.

Material and Methods

Ten female patients were referred from the Department of Gynaecology to the genetic clinic for the study of dermatoglyphics, buccal smears for sex chromatin and chromosome studies. Seven of the patients, who had primary amenorrhoea, showed hypoplastic or absent genital tract. The remaining three cases were diagnosed as Turner's syndrome.

The prints of palms and soles were obtained on drawing paper with a special ink on a foam leather pad. These were also visually studied and recorded on a proforma.

The patterns were classified as described by Penrose (1963). The fingers were studied for the presence of loops, radial or ulnar, arches or whorls. Their ridge count was calculated, the calculation being done from the tri-radius of a pattern to the centre of that pattern (ridge count for arch is zero as it has no tri-radius). The palms were studied for digital and axial tri-radius (Fig. 1), thenar

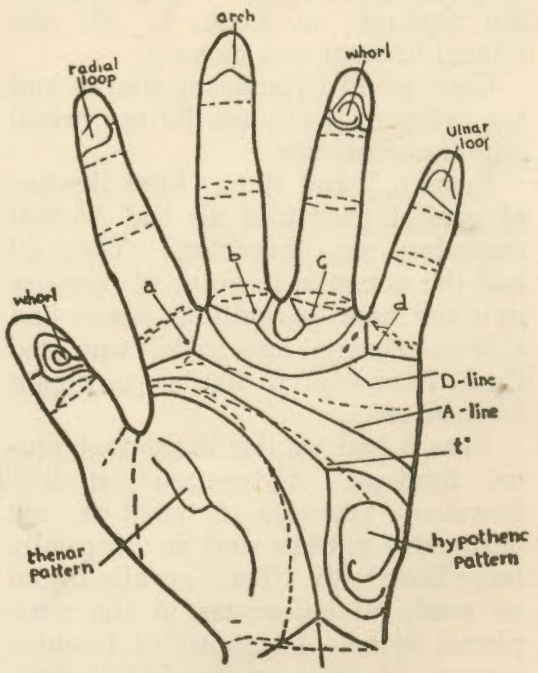

Fig. 1
Dermatoghyphical features illustrating various patterns.

and hypothenar patterns, transverse palmar creases, the interdigital patterns, and the 'atd' angle. The sole prints were specially studied for patterns at the hallucal areas.

Observation

The age, gynaecological findings and the main dermatoglyphical findings are outlined in Table 1.

Ten cases of primary amenorrhoea were studied. They could be classified as follows:

(1) Turner's syndrome—3.
(2) Isochromosome X (Xx)—1.
(3) Testicular feminisation—1.
(4) Abnormal developmental anomalies of genital tract—5.

The dermatoglyphics of Turner's syndrome as found in this series are summarised in Table 1. Fig. 2 shows the findings in Case 1. All the typical findings can be seen.

Case 5 had hypoplastic uterus and the cytogenetic studies did not reveal any abnormalities.

Cases 6, 7 and 10 had total absence of genital tract and all had normal secondary sex characters. They all had the common findings of increase in ulnar loops. In addition, case 6 had also a horizontal loop on left sole, and Case 7 had arch fibular on right hallucal area.

Case 9 had similar dermatoglyphical findings. Cytogenetic studies, however, revealed a positive sex chromatin pattern and an unusually large Barr body. This was also found on study of leucocytes in the peripheral blood. Diagnosis of Isochromosome X (Xx) was made but chromosome culture could not be done in this case.

Case 8, a phenotypic female, aged 17 years, on physical examination revealed absent uterus, swelling of labia and a blind vagina. The swellings of the labia on exploration, revealed gonads, which on histology showed testicular tissue. The buccal smear showed negative sex chromatin pattern and the karyotype showed normal male pattern (46-xy). Dermatoglyphical findings were mainly confined to the finding of horizontal loops on both soles, in hallucal area —a pattern which the authors have not come across before. Fig. 3.

Comments

Dermatoglyphics is a new diagnostic tool and an impressive literature has already been amassed on abnormalities in dermal patterns found in various congenital and heritable diseases. Thus, specific abnormalities have been reported in Down's syndrome (Fang 1950, Ford Walker 1950, Penrose 1963), in other autosomal trisomies, such as D,-trisomy (Uchida 1963), E-trisomy, (Ford Walker 1965, Holt 1964) and in sex chromosome anomalies. (Holt 1964, Lindstein 1963, Penrose 1963, Uchida 1963 and Forbes 1964).

Although the present series is admittedly small, containing only 10 cases, in many of the groups studied dermatoglyphical abnormalities were specific and were pointers to effective investigatory measures and subsequent diagnosis.

Case 1., N. S., for instance, was referred to the clinic for stunted growth. She was a twelve year old girl and on physical examination did not show any stigmata of Turner's syndrome. However, she had typi-

TABLE 1

Showing Age, Sex, Gynaecological and the main Dermatoglyphical findings in the Series

No.	Patient	Age	Gynaecological findings	Diagnosis	Main dermatoglyphical findings
1	N. S.	12 yrs.	Stunted growth ; primary amenorrhoea.	Turner Mosiac (xo/xx)	Excess of worls on fingers, increased 'atd' angle, increased ridge count, ulnar deviations of 'b' tri-radius.
2	P. S.	17½ ,,	Short stature ; primary amenorrhoea.	Turner's syndrome	do.
3	S. S.	15 ,,	Short stature ; primary amenorhoea.	Turner's syndrome	do.
4	D. V.	18 ,,	Primary amenorrhoea. Reduced rugosity of vagina, small rudimentary uterus. Secondary sex normal.	Hypoplastic uterus	Increase in ulnar loops.
5	S. K.	16 ,,	Primary amenorrhoea. Small hypoplastic uterus.	Hypoplastic uterus	Increase in ulnar loops.
6	R. S.	25 ,,	Primary amenorrhoea. Complete absence of genital tract.	Absent genital tract	(a) increased ulnar loops. (b) horizontal loops on the left sole.
7	S. P.	22 ,,	Primary amenorrhoea. Complete absence of uterus, cervix and vagina.	Absent genital tract	Increase in ulnar loops with very low ridge count, and arch fibular on rt. sole.
8	S. P.	17 ,,	Primary amenorrhoea. Absent uterus ; gonads in labia, identified as testes.	Testicular feminisation syndrome. (xy) chromatin negative.	horizontal loops on both the soles. soles.
9	T. M.	18 ,,	Primary amenorrhoea. Hypoplastic vagina, absent uterus.	Malformed genital tract (Xx Isochrome)—	(a) Increase in ulnar loops. (b) horizontal loop on right sole.
10	R. S.	18 ,,	Primary amenorrhoea. Complete absence of genital tract Normal secondary sex.	Absent genital tract.	Increase in ulnar loops.

cal dermatoglyphical abnormalities of Turner's syndrome i.e. increase in whorls, ulnar deviation of 'b' triradius and a high ridge count. These findings strongly indicated further cytogenetic studies and she was found to be a Turner-Mosaic on chromosomal studies (xo/xx).

Similar indications were provided by cases 2, 3, 8 and 9. Case 8 presented peculiar horizontal loops on both the soles and although this abnormality has not been described in any specific anomaly, it led us to cytogenetic studies which showed the patient to be chromatin negative and with a karyotype of a typical male (46 chromosomes with xy constitution).

Case 9 also is worthy of comment. An 18 year old girl was admitted for primary amenorrhoea and on gynaecological examination had hypoplastic vagina and absent uterus. Dermatoglyphical abnormalities showed increase in ulnar loops and again we encountered the horizontal loop that was described in case 8. The sex chromatin studies were revealing. Although she was chromatin positive, the Barr bodies were abnormally large, and this characteristic was also shared by the drumsticks found in 3% of the polymorphonuclear leucocytes. In view of these findings, as well as the genital tract abnormalities, a diagnosis of Isochrome X (Xx) was made. Chromosome studies, however, could not be carried out in this case.

The dermatoglyphical abnormalities in these cases were broadly of three type. Cases 1, 2 and 3, were more or less of the classical types.

There were, as described before, 3 cases of cytogenetically proved Turner's syndrome who had the following dermatoglyphical abnormalities.

(1) Increase in 'atd' angle.
(2) Increase in ridge count.
(3) Ulnar deviations of 'b' triradius.
(4) Excess of whorls.

These abnormalities have been described by a number of authors. (Holt 1963, Penrose 1963).

The second group of abnormalities belonged to the 6 cases (Cases, 4, 5, 6, 7, 9 and 10) where maldeveloped or under-developed genital tract was the presenting feature. The dermatoglyphical abnormalities in these cases were remarkably similar i.e. increase in ulnar loops, and in some case (Cases 6, 8 and 9) horizontal loops in hallucal areas.

The third group of abnormalities is found in the lone case of testicular feminisation syndrome of horizontal loops on soles. The authors have not come across references to these findings in literature.

Summary

Dermatoglyphics is a growing science of great practical value in diagnosis. These studies, apart from being additional clinical signs, can aid the cytogeneticist to look for specific abnormalities and identification of the abnormal chromosomes.

Ten cases of primary amenorrhoea were studied for dermatoglyphical abnormalities. These have been classified and discussed with reference to similar findings reported in the literature.

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References

1. Fang, T. C.: J. Ment. Sc. **96:** 780, 1950.
2. Forbes, A. P.: New Eng. J. Med. **270:** 1268, 1964.
3. Ford Walker, N.: J. Paed. **50:** 19, 1957.
4. Ford Walker, N.: The Current Status of Research on Dermatoglyphics in Medical constitution. In Genetics today. (Geerts, S. J. ed) Pergamon Press, Oxford, (1965), 981.
5. Holt, S. B.: Role of Dermatoglyphics in Medical Biology, Medical World, Aug. 1964.
6. Holt, S. B. and Lindsten, J.: Ann. Hum. Genet. **28:** 87, 1964.
7. Lindstein, J., Fraccaro, M., Ikkos, D., Kaijser, K., Klinger, H. P. and Luft, R.: Ann. Hum. Genet. **26:** 383, 1963.
8. Penrose, L. S.: Nature. **197:** 933, 1963.
9. Uchida, I. A. and Soltan, H. C.: Paed. Clin. of N. Am. **10:** 409, 1963.

Figs. on Art Paper X