Dermatoglyphics and Cleft Lip and Palate

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Dermatoglyphics comprise the varied and intricate patterns present on the surface of the skin in man and other mammals. These configurations are unmistakable and are readily classifiable marks which can be used to distinguish one individual from another. Most importantly, they remain constant from before birth until death, unaffected by any constitutional or environmental disturbances during the remaining gestative period and beyond.

These dermal configurations make their appearance early. Appendage buds are observed at about the fourth to fifth week of fetal life (7). During the sixth week the terminal portion of the bud, which is destined to become the hand or foot, becomes expanded and flattened with a marginal flange along its free border. Four radial grooves in this marginal flange suggest the formation of the digits. The digits develop during the third month and then mounds are formed on the tips of the digits. These mounds are the precursors of the dermatoglyphic areas of the palm and sole, the thenar and hypothenar areas of the palm, and the calcar and hallucal areas of the sole (Figure 1).

The plane of union between the epithelium and the dermal connective tissue on these mounds, as elsewhere, is smooth when first formed. During the third fetal month, the mounds begin to recede. As the mounds recede, the epithelium thickens and its lower border becomes irregular, exhibiting ridges and hollows into which connective tissue grows. This epithelium forms the dermal ridges. Once these configurations have been established during the third to fourth fetal month, they never change, except in size, throughout life.

These dermal patterns provide significant information as stigmata of various constitutional disorders of man (1, 2, 16). Aberrant pattern formation can be important as a chronological landmark in the occurrence of congenital anomalies or as a clue to specific chromosomal abnormalities (14). Hunt (8) states that any peculiarities of these print patterns in individuals with dentofacial deformity, that is, malocclusion, would be highly suggestive of a significant prevalence of generalized disturb-

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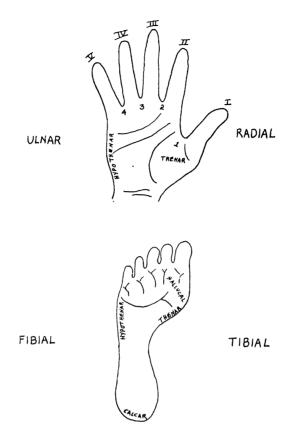


FIGURE 1. Dermatoglyphic areas of the palm and sole.

ances of development in the early embryonic and fetal months. Uchida and Soltan (15) point out that similarities of dermal patterns among relatives, differences between unrelated individuals, and still greater variation among members of different races indicate that these patterns are genetically determined. Patients with chromosomal abberations, however, have dermal configurations that are more similar to those of other patients with the same syndrome, even though of different races, than to those of their own relatives.

Experiments by Ingalls (θ) suggest that certain anomalies of the teeth, skull, and face of the type associated with Down's syndrome and cleft lip/palate anomalies have much in common in regard to time of onset and perhaps also in regard to causation.

The investigations of Walker (18) have demonstrated that finger, palm, and sole patterns differ significantly in frequency among mongoloids as compared with a normal control series. Seventy per cent of mongoloids possess combinations of fingerprint patterns which are not repeated in normals. There are also significant differences in print pat-

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terns between patients with congenital and those with acquired heart disease, as shown by Hale (6).

Recent investigations of chromosomal abnormalities (14) have led to the better description of syndromes where dermatoglyphic determinations have been related. In the Trisomy 18 syndrome, there are characteristic arches present on most of the digits. All D₁ Trisomys studied demonstrate distal axial triradii on the palms. Other unusual dermal ridge patterns have been observed in individuals where reciprocal translocations are probably present (4). Interestingly enough, the patterns found on the palms of D Trisomics (15), who also show cleft lip/palate (13), resemble those of Down's syndrome (Trisomy 21).

The causative factors of fetal malformations are multiple. In spite of the sheltered conditions of intrauterine life, the human embryo is not entirely insulated from the external environment. In addition to the genetic causes which are apparently implicated in some malformations, Fraser (5) lists a number of teratogenic agents such as radiation, virus, toxoplasma organism, acute folic acid deficiency, progestins, and anoxia.

Whether etiologic factors are local, such as embryonic infarction of blood supply to primitive facial processes (11), systemic, such as toxic substances (5), genetic, or whether they are a combination of these, as in certain abnormalities of protein metabolism, makes little difference. The ontogenetic timing of the insult is the critical and determining factor as to the affected part.

Since dermal ridges are present in all individuals and have been demonstrated to be stigmata in other congenital anomalies, it is important to determine whether deformities of the face may also be related to the same ontogenetic horizon as the formation of dermal ridges. This would provide a clue to the establishment of the occurrence of the event, or even to the ascertainment of unaffected carriers of a genotype which might result in predisposition toward particular cleft lip or palatal anomalies. To devise techniques for detecting carriers of recessive or relatively recessive traits is one application of dermatoglyphic investigation. Genetic counseling regarding future progeny, as well as etiology regarding other teratogenic factors, could equally apply such methods.

There are a multitude of deformities in the dentofacial area. Malocclusion is by far the most common deformity, and probably involves the greatest variation and number of etiologic factors. Since the etiology of malocclusion can involve local, systemic, or environmental sources both before and after birth, it would be difficult to separate them. For purposes of this research, one channel was followed: cleft lip and/or palate, a specific trait without any postnatal etiology. The incidence of this condition in white populations has been found to be 1 out of 929 births (3).

In this investigation, a total of 39 white boys and 32 white girls with cleft lip/palate were examined. All individuals were residents of the Bos-

	Boys	Girls	Total
Cleft lip Cleft lip/palate Cleft palate	3 33 3	0 27 5	3 60 8
	39	32	71

TABLE 1. Cleft type, by sex.

ton area and were selected as they presented for treatment or consultation at the Children's Hospital Cleft Palate Orthodontic Clinic and at the Tufts University Cleft Palate Institute. A control group of normals was selected from the elementary school population in the same geographical area. The normal children were chosen at random as they presented for routine oral examination and prophylaxis at the Forsyth Dental Infirmary. This group consisted of 43 white boys and 49 white girls.

Method and Material

Each subject had a complete set of dermatoglyphic prints taken by the investigator employing the Faurot inkless method (17). The set consisted of right and left full palm prints, individually rolled prints of each finger, and right and left sole prints. In addition, a complete history was taken on each patient and recorded on specially printed McBee Keysort cards.

Distribution of cleft type according to sex is presented in Table 1. Cleft lip and palate together is the most prevalent condition. Cleft lip or cleft palate alone comprises a sample which is too small to provide significance in this series.

Evidence of history of miscarriage among the mothers of both boys and girls in the affected group was only 11% as compared with an incidence of 19% as observed by Mall (10) in a general American population (Table 2). However, it is interesting to note that the occurrence of clefts in pregnancies among the affected group is 1 out of 4. This com-

	Cleft group		Total	Mall	
-	Boys	Girls	10101	111 000	
No. of pregnancies No. of miscarriages Live births No. of clefts	$162 \\ 17 \\ 145 \\ 41$	$ 123 \\ 13 \\ 110 \\ 32 $	285 30 255 73	100,000 19,000 80,572 not reported	

TABLE 2. Incidence of history of miscarriage among the families of children with clefts, by sex, and as reported by Mall (10).

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	Younger than	Matern	ial ages	Older than	Younger than	Patern	al ages	Older than
	25 yrs.	25 to 29	30 to 35	25	25 yrs.	25 to 29	30 to 35	35 yrs.
Boys	9	16	5	4	5	13	7	10
Girls	10	12	4	4	4	9	9	9
Total	19	28	9	8	9	22	16	19

TABLE 3. Distribution of parental ages at birth of children with a cleft.

TABLE 4. Birth orders for cleft and normal groups, by sex.

Birth order	Bo	ys	Ga	irls	Total
Dirin order	Normal	Cleft	Normal	Cleft	10101
$\overline{\text{First}}$	13	12	17	9	51
Second	16	11	19	10	56
Third	7	8	7	5	27
Fourth or below	6	8	6	7	27

pares with a rate of approximately 1 out of 925 (3) in the general population. This ratio of one to four has genetic implications of a Mendelian recessive trait that deserve further investigation.

The effects of parental age and birth order have been the subjects of investigation in many congenital anomalies. The condition of the reproductive system in the male or female has been suspected of contributing to the production of affected offspring. The age of the mother and the father at the time of birth of the affected child (Table 3), as well as the birth order of the child (Table 4) were recorded in this investigation.

The distribution of parental ages at the birth of the child and the birth order in the present series did not differ significantly between the normal controls and the affected children.

In the investigation of the dermatoglyphics, there were three important areas: the third interdigital area of the palm, the individual fingerprint patterns, and the hallucal area of the sole. Of all areas, these can be 'read' most reliably.

The third interdigital area of the palm can print two types of patterns, a 'true' pattern or a 'not true' pattern. Analysis of these print patterns in the third interdigital area of the palm indicates that there was no significant difference between normal and cleft groups or between sexes (Table 5).

The hallucal pattern on the ball of the foot may show six different

patterns. Chi Square tests for significance were done combining arches, open field and whorl as one classification against all loops. In this study, there was no significant difference in the hallucal pattern formation between boys and girls or between clefts and normals (Table 6).

In the evaluation of fingerprints, easily discernible patterns are important. The obtaining of fully rolled prints is also critical since only a part of a pattern may be misleading. Four fundamental patterns were considered in this study: arch, radial loop (opening toward the radial margin of the digit), ulnar loop (opening toward the ulnar margin of the digit), and whorl (Figure 2). These patterns have been classified according to the standards of the Federal Bureau of Investigation Handbook. The occurrence of all four patterns in this study is listed in Table 7. The whorl and ulnar loop patterns are the most frequent while radial loops and arches are the least frequent in all fingers in both sexes.

In the statistical analysis, whorls and arches were combined and radial and ulnar loops were combined. This was done because of the small cell size in radial loops and in arches and because of similarity in pattern formation. All patterns tend to occur in all fingers with some frequency. Statistical analysis reveals no significant difference in any pattern for-

Pattern	Boys		Girls		Total	
1 40011	Normal	Cleft	Normal	Cleft	10100	
"True"		33 32	39 53	30 27	$140 \\ 156$	

TABLE 5. Distribution of types of patterns in the third interdigital area of the palm for cleft and normal groups, by sex.

TABLE 6. Distribution of patterns in hallucal area of the sole for cleft and normal groups, by sex.

Pattern	Boys		Gir	Total	
r uuern	Normal	Cleft	Normal	Cleft	10141
Loop distal	38	32	54	25	149
Loop tibial	8	2	11	8	29
Loop fibial	0	0	0	1	1
Whorl	31	32	28	20	111
Open field	3	4	1	4	12
Arch	5	2	4	2	13

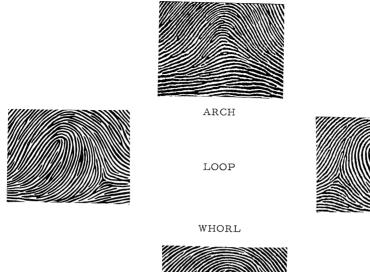




FIGURE 2. Fingerprint patterns.

TABLE 7. Distribution of types of	fingerprint patterns	(right and left combined)
for cleft and normal groups, by sex.	•	

Pattern	Bo	ys	Girls	
r uuern –	Normal	Cleft	Normal	Cleft
Ulnar loops	249	204	259	184
Radial loops	23	10	22	9
Arches	11	47	48	38
Whorls	147	119	161	89

mation between cleft and normal groups in any individual finger or in either hand.

Summary

Review of the literature reveals cleft lip/palate to be a relatively common (1:929) congenital anomaly among the white population. This investigation set out to compare a cleft group of 71 boys and girls with a normal group of 92 boys and girls in the Boston area. The purpose was to obtain a full history on all patients with particular emphasis on dermatoglyphic patterns. In this series, the cleft lip and palate together was the most prevalent condition. Difference in parental age and birth order were not significant. There was, however, a 1:4 occurrence of clefts within cleft siblings which may imply a Mendelian recessive trait which deserves further investigation. There was no significant difference between any of the dermatoglyphic configurations of the cleft group and the normal group. Cleft lip/palate is a congenital anomaly whose developmental basis seems to be independent of the production of aberrant dermatoglyphic patterns.

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References

- 1. CUMMINS, H., Dermatoglyphic stigmata in mongolian idiocy. Anat. Record, 64, 11, 1936 and 73, 407, 1939.
- 2. CUMMINS, H., and MIDLO, C., Fingerprints, Palms and Soles, An Introduction to Dermatoglyphics. New York: The Blakiston Co., 1943.
- 3. Donahue, R. F., Birth variables and the incidence of cleft palate. Part I. Cleft Palate J., 2, 282-290, 1965.
- 4. EDWARDS, J. H., FRACCARO, M., DAVIES, P., and YOUNG, R. B., Structural heterozygosis in man: Analysis of two families. Annals hum. Genet., 26, 163-178, 1962.
- 5. FRASER, F. C., Causes of congenital maliformations in human beings. Conference on congenital malformations. J. chronic Dis., 10, 102–112, 1959.
- 6. HALE, A. R., PHILLIPS, J. H., and BURCH, G. E., Features of Palmar dermatoglyphics in congenital heart disease. J. Amer. Med. Assoc., 176, 41-45, 1961.
- 7. PATTEN, B. M., Human Embryology. New York: The Blakiston Co., 1960.
- 8. HUNT, E. E., JR., Malocclusion and civilization. Amer. J. Orthod., 47, 406-425, 1961.
- 9. INGALLS, T. H., Intra-uterine causes of cleft palate, mongolism and associated defects of the skull. Cleft Palate Bull., 2, 7-8, 1952.
- 10. MALL, F. P., On the frequency of localized anomalies in human embryos and infants at birth. Amer. J. Anat., 22, 49-72, 1917.
- 11. McCov, F. J., Congenital malformations of the face. Cleft Palate Bull., 6, 8-10, 1956.
- 12. PENROSE, L. S., Outline of Human Genetics. New York: W. Heinemann Ltd., 1959.
- 13. SMITH, DAVID W., The No. 18 trisomy and D₁ trisomy syndromes. *Pediat. Clin.* North Amer., 10, 389-408, 1963.
- 14. UCHIDA, I. A., PATAU, K., and SMITH, D. W., Dermal patterns of 18 and D₁ trisomics. Amer. J. hum. Genet., 14, 345-352, 1962.
- UCHIDA, I. A., and SOLTAN, H. C., Evaluation of dermatoglyphics in medical genetics. *Pediat. Clin. North Amer.*, 10, 409-422, 1963.
- 16. Von HIRSCH, W., and GEIPEL, G., Das Papillarleisten system der hand und seine beziehung zu cerebralen storungen. Acta Genet., 10, 103–182, 1960.
- 17. WALKER, N. F., Inkless methods of finger, palm and sole printing. J. Pediat., 50, 27-29, 1957.
- WALKER, N. F., The use of dermal configurations in the diagnosis of mongolism. J. Pediat., 50, 19-26, 1957.