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Chromosomal and Dermatoglyphic Changes in Twins

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SUMMARY

A review of 15 chromosomally abnormal twin pairs indicates that finger and hallucal patterns of the affected twin are more similar to other patients than to the normal cotwin.

Correlation coefficients of TFRC and *atd* angle differences tend to remain similar in spite of the presence of chromosome abnormality in the cotwin. This supports the use of dermatoglyphic analysis as a means for diagnosing twin zygosity. Correlation coefficients of intrapair TFRC and *atd* angle differences in 25 control MZ twin pairs shows good agreement in zygosity determinations, when compared to results obtained by blood group and clinical observation studies.

In a series of 155 female and 142 male MZ twin pairs, TFRC and *atd* angle differences plotted on a graph against the frequency of occurrence illustrate that the range of variation is greater among females. Further study of a large series of twin pairs, however, is necessary to determine whether this is an effect of lyonization or the alteration of the polygenic complex of dermal configuration by the X chromosome.

Introduction

Twin studies have been used to observe concordance in hereditary traits, even though differences due to intrauterine influences have been noted (Osborne and De George, 1959; Gedda, 1961). In a review of 23 pairs of MZ twins reported in the literature, Nielsen (1967) pointed out the discordance of the genotype including chromosomal differences, when only one is afflicted with an inherited abnormality. Random inactivation of the paternal or maternal X chromosome in the female, as postulated by the Lyon hypothesis (Lyon, 1961*a*), might lead to intrapair differences in female MZ twins (Vanderberg et al, 1962), although this has not been established (McKusick, 1969). Evidence of intrapair variation in dermatoglyphic configuration in male and female twin pairs are findings of this report.

Dermatoglyphic analyses have been useful in zygosity determination (Smith and Penrose, 1955; Lamy et al, 1956). However, in cases where one MZ twin is chromosomally abnormal, the abnormal twins' dermatoglyphics are so affected that the twin is more similar to other patients affected with this disease, than to his chromosomally normal cotwin. These studies thus indicate and support other independently derived conclusions that dermatoglyphics are characteristic of the abnormal karyotype (Uchida and Soltan, 1963). Studies of dermal patterns in some chromosomally abnormal twin pairs are presented in this paper.

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Material and Methods

This study comprises original observations on 12 female and 13 male MZ twin pairs. The zygosity was established by means of clinical observations and blood group studies (Smith, 1965). Later, dermatoglyphic analyses were performed and correlation coefficients for intrapair *atd* angles and total finger ridge count were calculated. Intrapair ridge count differences are presented with their frequency of occurrence in Fig. 1. Also, similar calculations were performed on 143 female and 129 male MZ twin pairs reported by Lamy (1956) and are included in the figure.

Dermatoglyphics on 11 chromosomally abnormal MZ twin pairs obtained from the literature and/or correspondence are presented. In addition, data on 4 chromosomally abnormal twin pairs are presented. These are 1 pair of MZ twins (Tab. 1, case 7) both

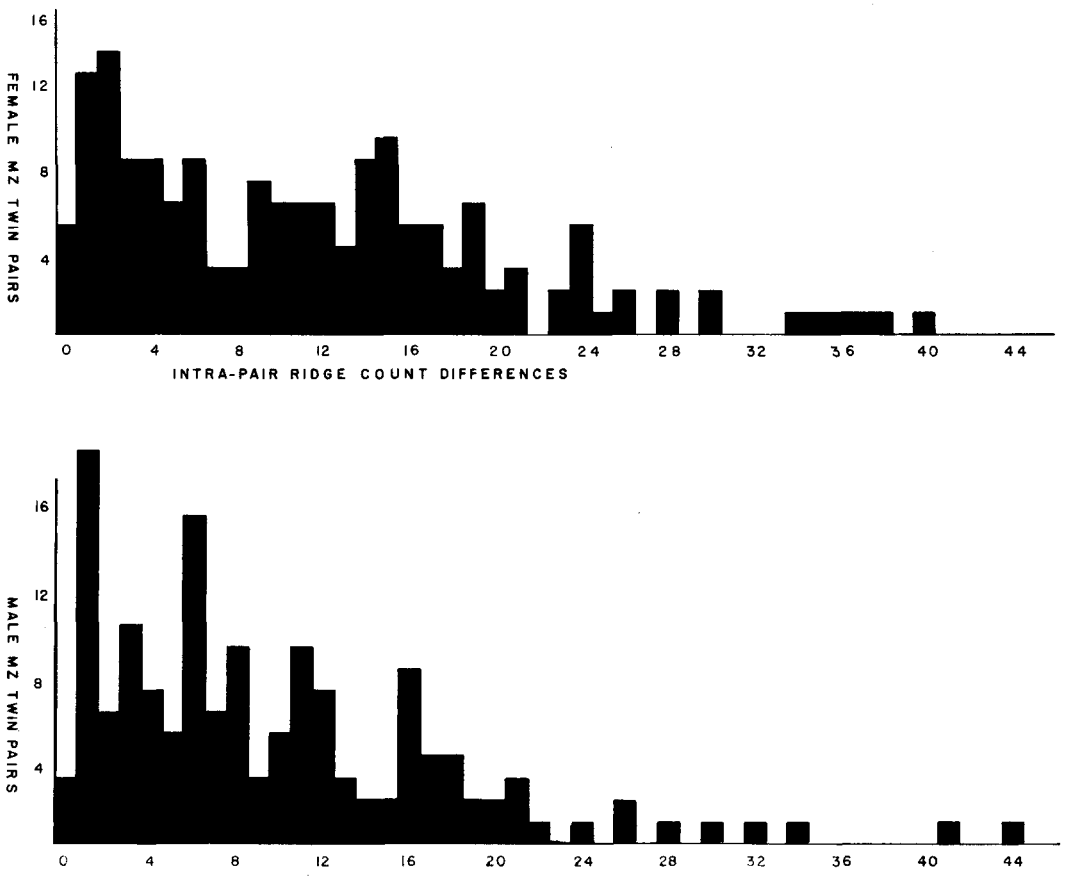


Fig. 1. Histogram showing intrapair total finger ridge count differences in MZ twins

affected with Turner's syndrome, and 3 DZ twin pairs where only one twin is affected with Down's syndrome (cases 5 and 6), or Turner's syndrome (case 11).

Dermatoglyphic prints were obtained using the Faurot inkless method (Walker, 1958) and/or the Hollister method.

Chromosome studies were performed by the standard micromethod of leucocyte culture, and sex chromatin studies were performed from buccal smears.

Results and Discussion

Lyonization effect

Calculations of intrapair ridge count differences from 12 male and 13 female MZ twin pairs, combined with 129 male and 143 female MZ twin pairs reported by Lamy et al (1952) have been plotted against their frequency of occurrence (Fig. 1). Intrapair ridge count differences in the 155 female and 142 male MZ twin pairs indicate a greater spread of variation among the female twin pairs. A χ^2 analysis of these data shows that this variation is statistically significant ($P = 0.03$). The source of this observed degree of variation in the intrapair total ridge count differences may be explained as due to random inactivation of maternal or paternal X chromosome, as postulated originally by Lyon (1961*b*). In contrast, male MZ twin pairs, with only one X chromosome, have a lesser degree of variation. Also, in some female MZ twin pairs, a lesser degree of variation may be due to inactivation of the maternal or paternal X chromosome exclusively. The assumption that the cause and effect of random inactivation of the X chromosome in females is the source of genetic variation, as reflected in the dermal pattern analysis, leads us to believe that the X chromosome in some way alters the polygenic complex of dermal pattern configuration. Although this seems to clearly support the effect of lyonization, further studies on a large series of twin pairs may demonstrate the alteration of the polygenic complex of dermal configuration by the X chromosome.

Chromosomally abnormal DZ twins

Intrapair differences of total ridge counts and *atd* angles for twins A and B in three DZ chromosomally abnormal twin pairs (5, 6 and 11) are summarized in Tab. I. As expected, intrapair ridge count and *atd* angle differences in chromosomally abnormal DZ twin pairs are greater. In each of the twin pairs represented in Tables I and II, within-pair correlation coefficients have been determined for total ridge count and the *atd* angle (Smith and Penrose, 1955), or both. Zygosity and chromosome constitutions have been previously established, each independently of the other. Twin pairs 5, 6 and 11 represent three DZ chromosomally abnormal sets. Dermatoglyphic patterns are illustrated in Fig. 2 and detailed findings are presented in Tab. III. The most remarkable dissimilarity is seen in the position of the palmar triradius, with normal sibs of each pair presenting a proximally positioned palmar triradius. Lesser dissimilarities occur in the hypothenar and hallucal areas.

Tab. I. Summary of twin data in ridge count and *atd* angle differences

Case N.	Twin	Phenotype	Karyotype	Zygosity	Ridge count		Intrapair difference	*atd* angle		Intrapair difference	Reference
					Left	Right		Left	Right		
1	A	Normal	46, XX	MZ	26	26	0	53	49	8	Dekaban, 1965
	B	Down's	47, XX, Tri 21		32	20		46	48		
2	A	Normal	46, XY	MZ	—	—	—	42	40.5	9	de Wolff et al, 1962
	B	Down's	47, XY, Tri 21		—	—		37.5	36		
3	A	Down's	47, XX, Tri 21	MZ	49	58	1	51	81	0	Nance and
	B	Down's	47, XX, Tri 21		53	55		61	71	0	Uchida, 1964*
4	A	Down's	47, XX, Tri 21	MZ	102	76	1	—	—	1	Ford and Fumkin,
	B	Down's	47, XX, Tri 21		90	87		—	—		1942
5	A	Normal	46, XX	DZ	27	22	90	47	47	1	This report
	B	Down's	47, XX, Tri 21		73	66		48	45		
6	A	Normal	46, XX	DZ	17	4	104	40	40	1	This report
	B	Down's	47, XX, Tri 21		56	69		60	71		
7	A	Turner's	45, XO	MZ	76	95	15	42	40.5	0.5	This report
	B	Turner's	45, XO		81	79		43	40		
8	A	Turner's	45, XO	MZ	—	—	—	49	45	3	Turner and Zanartu*
	B	Turner's	45, XO		—	—		47	50		1962
9	A	Turner's	45, XO	MZ	56	71	8	47	45	1.5	Nance and
	B	Turner's	45, XO		64	71		46	54.5		Uchida 1964*
10	A	Normal	46, XX	MZ	173**		10	—	—	—	Shine and Corney,
	B	Turner's	45, XO		183			—	—	—	1966
11	A	Normal	46, XX	DZ	80	65	43	48	44	34	This report
	B	Turner's	45, XO		88	100		70	58		
12	A	Turner's	45, XO	MZ	55	60	17	40	36	3	Edwards et al,
	B	Turner's	46/46, XO/XY		65	67		40	39		1966
13	A	Turner's	45/46, XO/XX	MZ	21	13	13	127**		9	Edwards et al,
	B	Turner's	45/46, XO/XX		13	8		136			1966
14	A	Klinefelter	47, XXY	MZ	54	64	8	56	55	17	Bertrand et al,
	B	Klinefelter	47, XXY		62	50		42	42		1966
15	A	Klinefelter	47, XXY	MZ	123**		20	—	—	—	Nowakowski et al,
	B	Klinefelter	47, XXY		103						1963

* Information included in this table was obtained by personal communication.
** Total ridge counts available only.
— Data unavailable.

Tab. II. Correlation coefficients of total ridge count and *atd* angle differences in chromosomally abnormal twin pairs

Case N.	Zygosity	Correlation coefficients	
		Total ridge count	*atd* angle
1	MZ	1.0000	0.9215
2	MZ	—	0.8909
3	MZ	0.9907	1.0000
4	MZ	0.9943	0.9493
5	DZ	0.3525	0.9893
6	DZ	0.1680	0.8631
7	MZ	0.9356	1.0000
8	MZ	—	0.9690
9	MZ	0.9407	0.9200
10	MZ	0.9453	—
11	DZ	0.7712	0.7187
12	MZ	0.8712	0.9620
13	MZ	0.6176	0.9338
14	MZ	0.9322	0.7567
15	MZ	0.8373	—

Tab. III. Dermatoglyphic patterns of hands and feet of four chromosomally abnormal twin pairs

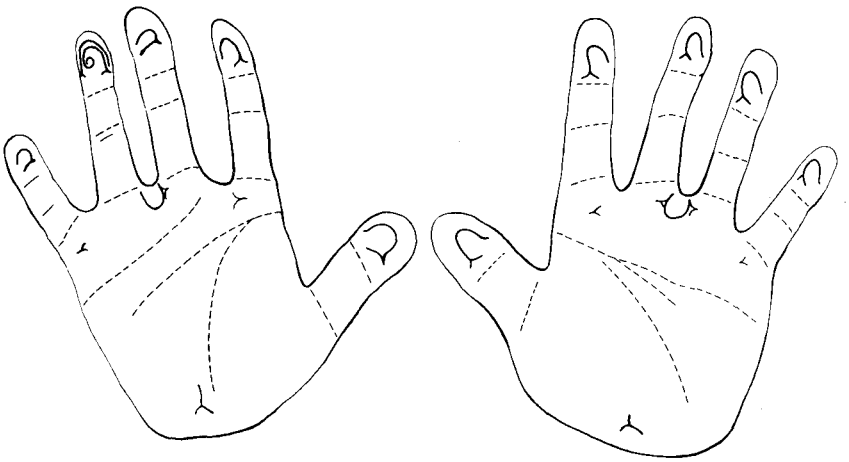
Case N.	Twin	Finger patterns		Palmar triradius		Hypothenar		Interdigital		Hallucal	
		Left 1 2 3 4 5	Right 1 2 3 4 5	Left	Right	Left	Right	Left 1 2 3 4	Right 1 2 4 3	Left	Right
5	A	UUUWU	UUUUU	t	t′	–	–	– – L –	– – L –	Ld	Ld
	B	UAUUU	UAUUU	t′	t′	–	–	– – L –	– – L –	Ld	Lp
6	A	AAUUU	AAUUA	t	t	L	–	– – L –	– – L –	L	A
	B	AWUWU	UUUWU	t″	t″	L	W	– – L –	– L L –	L	L
7	A	UUUWU	WWUWU	t′	t′	– –	–	– – L –	– – L –	A	A
	B	UWUWU	UWUWU	t′	t′	–	–	– – – –	– – L –	A	A
11	A	WWUUU	WRUUU	t	t	U	L	– – – L	– – – L	O	O
	B	WRUUW	UUUUU	t″	t″	L	L	– – – L	– – – L	O	O

U = ulnar loop; W = whorl; A = arch; R = radial loop; L = loop; O = unavailable; – = absent; d = distal; p = proximal.

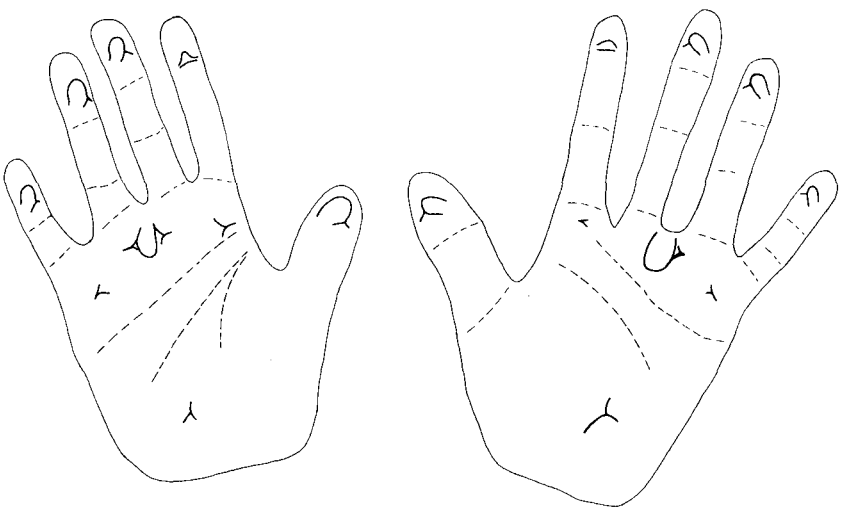
Chromosomally abnormal MZ twins

In three female MZ twin pairs (7, 8, 9), all concordant for Turner's syndrome (XO), total ridge count and *atd* angle correlation coefficients are greater than 0.90 with a small range of variation (Tab. II).

In two twin pairs concordant for Down's syndrome (cases 3 and 4), and two pairs in which only one twin is afflicted (cases 1 and 2), correlation coefficients of intra-

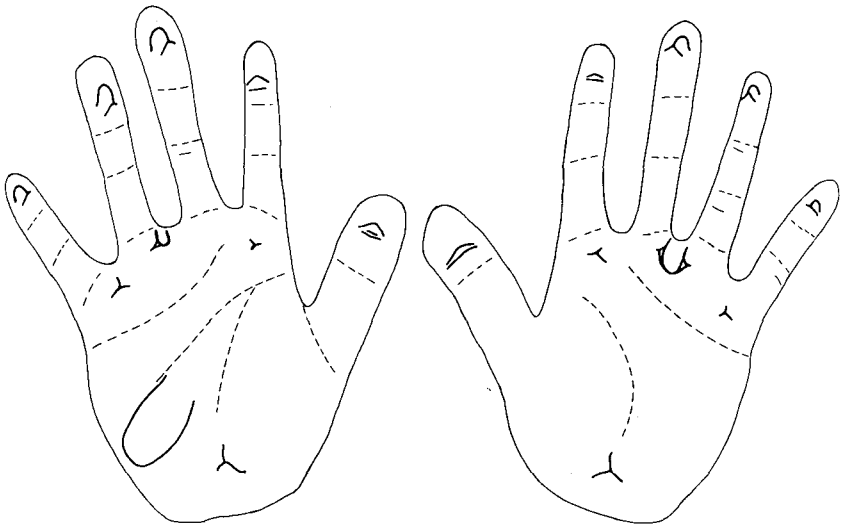


A) Normal female

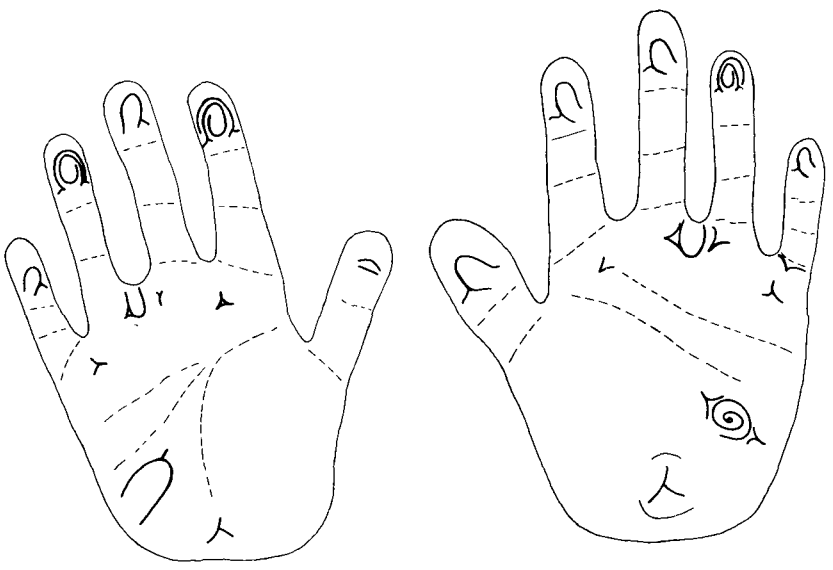


B) Mongoloid female

Fig. 2*a*. Case 5: DZ. Diagrammatic illustrations of palm patterns in a chromosomally abnormal, discordant twin pair



A) Normal female



B) Mongoloid female

Fig. 2*b*. Case 6: DZ. Diagrammatic illustrations of palm patterns in a chromosomally abnormal, discordant twin pair

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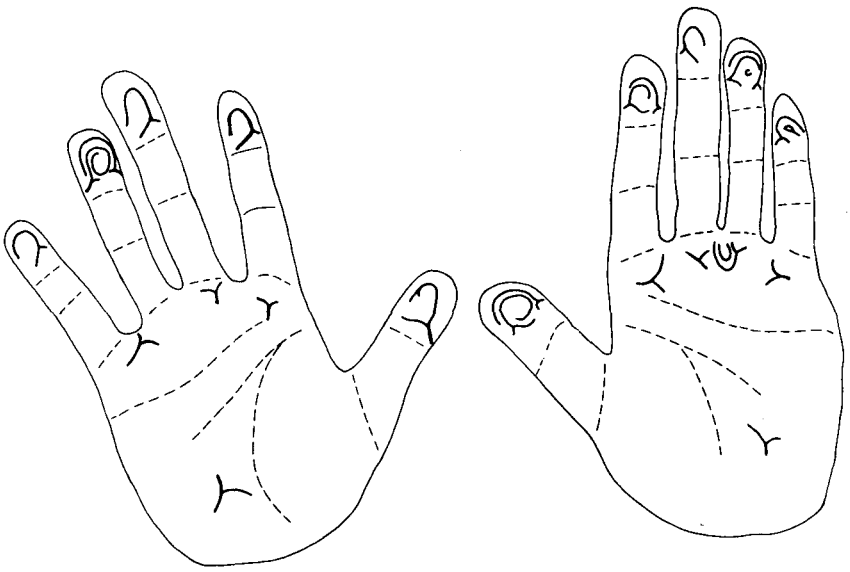
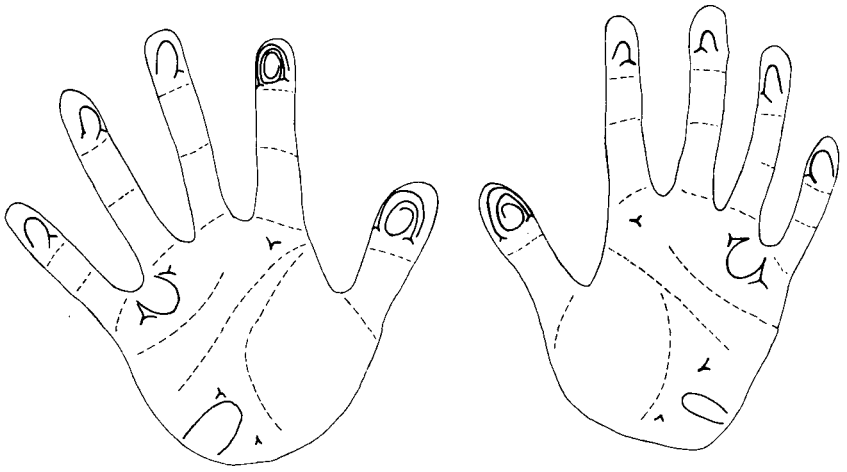


A) Turner's syndrome



B) Turner's syndrome

Fig. 2c. Case 7: MZ. Diagrammatic illustrations of palm patterns in a chromosomally abnormal, concordant twin pair

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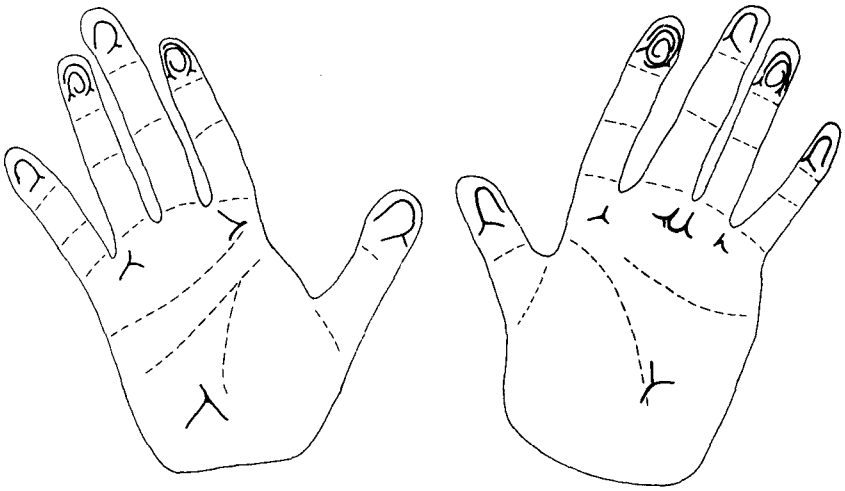
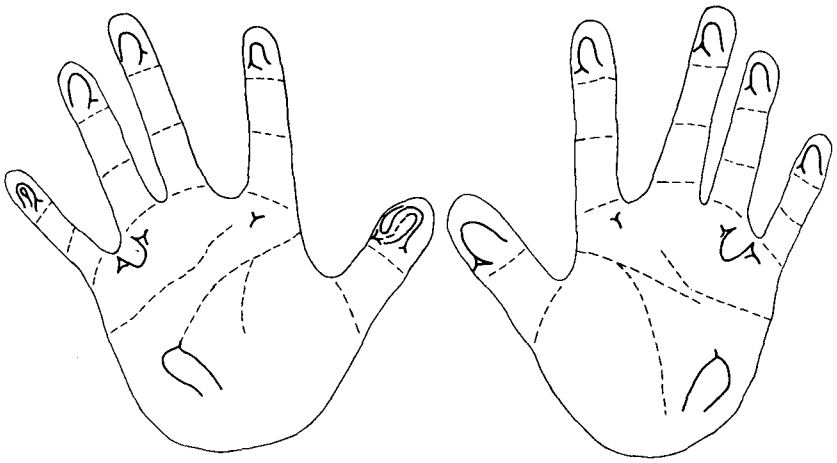


A) Normal female



B) Turner's syndrome

Fig. 2*d*. Case 11: DZ. Diagrammatic illustrations of palm patterns in a chromosomally abnormal, discordant twin pair

pair ridge count indicate little or no variation spread. The intrapair variation of *atd* angle is high. Twins A and B (cases 1 and 2) show differences in the *atd* angle, probably because this character is known to be affected by the trisomy 21 condition, and one of the twins has trisomy 21; on the other hand, no ridge count difference is evident as a result of this discordance, and in fact this trait is not known to be influenced by trisomy 21. Support for this is obtained from discriminant analysis derived from dermatoglyphic features of mongoloid patients (Penrose and Smith, 1966). Furthermore, in case 10, where one twin is a Turner syndrome patient and the other is a normal female, the total ridge count is affected, since this character is known to be a consistently uniform feature of this pathologic condition (Holt and Lindsten, 1964).

The MZ twins of case 7, both affected with Turner's syndrome, exhibit remarkable similarity in their dermatoglyphic patterns, especially with reference to the *atd* angle.

In studies of MZ twin pairs where one twin is chromosomally abnormal, dermatoglyphic characteristics of finger and hallucal patterns are unlike those of the normal cotwin, but similar to those of patients with the same disease (Down's syndrome, Turner's syndrome, and Klinefelter's syndrome). Correlation coefficients of ridge counts and *atd* angles as seen in cases, 1, 2, and 10, however, do not suggest such definite characteristics. This reflects the value of these parameters for diagnosing twin zygosity. This is again demonstrated in DZ twin pairs 5, 6, and 11, where correlation coefficients vary over a wide range, but to approximately the same extent as found in the control population of DZ twin pairs. Blind studies of intrapair correlation coefficients of ridge counts and *atd* angles of 25 control MZ and DZ twin pairs show good agreement with blood group data and clinical observations.

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Riassunto

Un'indagine dermatoglifica condotta su 15 coppie di gemelli con anomalie cromosomiche ha evidenziato che i gemelli affetti sono più simili agli altri pazienti con la stessa sindrome piuttosto che ai loro co-gemelli. I coefficienti di correlazione delle differenze del TFRC e dell'angolo *atd* tendono a rimanere elevati, nonostante l'aberrazione cromosomica discordante, il che depone a favore dell'uso dei dermatoglifi per la diagnosi di zigotismo. L'uso dei coefficienti di correlazione delle differenze intracoppia del TFRC e dell'angolo *atd* in 25 coppie MZ di controllo concorda sufficientemente nelle determinazioni di zigotismo, con i risultati dell'analisi dei gruppi sanguigni e dell'osservazione clinica.

In un'ulteriore serie di coppie MZ (155 ♀ e 142 ♂), l'insieme delle differenze del TFRC e dell'angolo *atd*, valutate in base alle rispettive frequenze, indicano una maggiore variabilità nelle femmine. Saranno necessarie ulteriori indagini gemellari per stabilire se ciò sia dovuto ad un effetto di lionizzazione oppure all'alterazione del complesso poligenico delle configurazioni dermiche da parte del cromosoma X.

Résumé

L'analyse des dermatoglyphes chez 15 couples de jumeaux avec des anomalies chromosomiques a démontré que les jumeaux atteints sont plus proches aux autres patients avec le même syndrome qu'à leurs co-jumeaux.

Les coefficients de corrélation entre jumeaux pour le compte total des crêtes (TFRC) et l'angle *atd* demeurent toutefois élevés, malgré la discordance chromosomique, ce qui confirme la possibilité de l'usage des dermatoglyphes dans le diagnostic de zygotie. Les diagnostics de zygotie d'après les corrélations de TFRC et de l'angle *atd* sont en accord, chez 25 couples de contrôle, avec les résultats de l'analyse des groupes sanguins et l'observation clinique.

L'analyse des différences du TFRC et de l'angle *atd* chez un autre échantillon de couples MZ (155 ♀ et

142 ♂) indique une variabilité plus élevée chez les femmes. De nouvelles recherches gémellaires seront nécessaires pour établir si cela est dû à un effet de lyonisation, ou bien à l'altération du complexe polygénique des configurations dermiques de la part du chromosome X.

ZUSAMMENFASSUNG

Aus einer Untersuchung der Hautleisten bei 15 Zwillingspaaren mit Chromosomenanomalien ergab sich, dass die Hautleisten der kranken Zwillinge mehr den Mitkranken als denen ihrer Zwillingspartner ähneln.

Die Korrelationskoeffizienten der Unterschiede von TFRC und *atd*-Winkel neigen dazu, trotz Diskordanz der Hautleisten ähnlich zu bleiben, was die Anwendung der Hautleistenkontrolle für die Eiigkeitsdiagnose befürwortet. Die Anwendung der Korrelationskoeffizienten der Unterschiede von TFRC und *atd*-Winkel zur Eiigkeitsbestimmung stimmt bei 25 Kontroll EZ-Paaren mit den glänzenden Ergebnissen der Blutgruppenanalyse und mit der klinischen Beobachtung überein.

In einer weiteren Reihe (155 ♀ und 143 ♂) zeigt das auf Grund der jeweiligen Frequenz bewertete Gesamtergebnis der Unterschiede von TFRC und *atd*-Winkel beim weiblichen Geschlecht eine grössere Variabilität. Es werden weitere Forschungen an Zwillingspaaren notwendig sein, um festzustellen, ob diese auf Lionisierung oder auf Alteration des polygenen Komplexes der Hautleistenzeichnung durch das X-Chromosom zurückzuführen ist.

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