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DERMATOGLYPHIC AND SKELETAL HAND ABNORMALITIES IN TURNER'S SYNDROME

A Tentative Scoring Method[1]

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SUMMARY

A scoring method for the diagnosis of Turner's syndrome (T.S.), based on five dermatoglyphic and four skeletal characteristics, showing significant frequency variations between patients and normal females, is presented. The dermatoglyphic characteristics include: total finger ridge count, *ab* ridge count, maximal *atd* angle, *T* line terminating in the second interdigital interval, and presence of hypothenar patterns. The skeletal characteristics include: carpal sign, metacarpal sign, phalangeal sign, and abnormally shaped distal phalanges. Arbitrary score numbers have been assigned to each characteristic, depending on the magnitude of the difference between patients and controls; the partial scores were added up to form the final T.S. score. The diagnostic value of T.S. score has been proved, since it is well separating the patients from normal females. The probability that an individual with a given T.S. score has the Turner's syndrome has been estimated.

The finger and palm pattern characteristics in Turner's syndrome (T.S.) have been described as being abnormal, particularly with respect to the finger pattern intensity, distance between the palmar triradii *a* and *b*, and position of the axial triradius *t*; other dermatoglyphic abnormal traits include a marked tendency for *T* line to terminate in the second interdigital interval, with the A line ending near the base of the thumb, and the presence of large hypothenar patterns. Even if these abnormalities are not specific of T.S. patients, their diagnostical importance has been proved and confirmed by different investigators (Uchida and Soltan 1963, Holt and Lindsten 1964, Forbes 1964, Dallapiccola 1966 and 1968, Walbaum et al. 1967, Pfeiffer and Kiera 1968).

However, despite the theoretical and practical interest of the dermal configurations in T.S., as far as we know, no attempt has hitherto been done to devise a dermatoglyphic scoring method that permits the discrimination of T.S. patients from normal females, with a fairly high degree of probability. In this paper we present an attempt to construct a scoring method that would be of practical importance in the diagnosis of T.S.

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MATERIALS AND METHODS

The material used in this investigation consists of data on 50 T.S. patients and 50 normal females as controls. The karyotypes of the patients included in this study are reported in Table I.

In all subjects, prints were obtained by standard techniques, using printer's ink and good-quality bond paper. In addition, standard radiographs of the hands were obtained in all patients and controls. In 10 patients and 8 controls X-ray studies of dermatoglyphics

TABLE I
KARYOTYPES OF THE T.S. PATIENTS

	N
45,X	39
45,X/46,XX	1
45,X/46,XXr	1
45,X/47,XXX	1
45,X/46,XXqi	3
46,XXqi	5
	50

were done. The skin of these subjects was smeared with lanolin; an X-ray opaque medium, bismuth carbonate, was then applied and distributed by gentle massage. The excess was wiped off and a shadowgraph was taken by the usual X-ray procedure. In this way, a detailed picture of the skin surface, in addition to bones and soft parts, was obtained.

Five different dermatoglyphic traits (1-5) and four skeletal characteristics (6-9), showing significant frequency variations between patients and controls, were selected. According to the scoring method employed by Beckman et al. (1965) for the diagnosis of Down's syndrome, we decided to use a simple scale, assigning to each dermatoglyphic characteristic the arbitrary score numbers 1 or 2, and to each skeletal characteristic the score numbers 5, 12, 25, or 30, depending on the magnitude of the difference between patients and controls. The nine different score items are shown in Fig. 1. The percentage frequencies of the dermatoglyphic and skeletal characteristics used in the diagnosis of T.S., the ratio between the frequencies in the two groups of subjects, and the scores assigned to each characteristic are listed in Table II.

The three metrical dermatoglyphic characteristics, i.e., (1) total ridge count, (2) *ab* ridge count, and (3) maximal *atd* angle, were considered abnormal when their values were higher than the respective means in controls. In normal females the mean total ridge count was 128, the mean *ab* ridge count 84 (sum of both hands) and the mean maximal *atd* angle 86° (sum of both hands). The diagnosis of the other dermatoglyphic traits (4-5) and of the skeletal characteristics (6-9) meets no difficulties in palmprints and in radiographs of good quality. The carpal sign (6: Kosowicz 1962) was considered positive when the lines delineating the scaphoid, lunate, and triquetral bones formed an angle less than 130°, i.e., less than the mean value for the controls. The metacarpal sign (7: Archibald et al. 1959) is positive when the tangent touching the outlines of the 5th and 4th metacarpal heads runs through

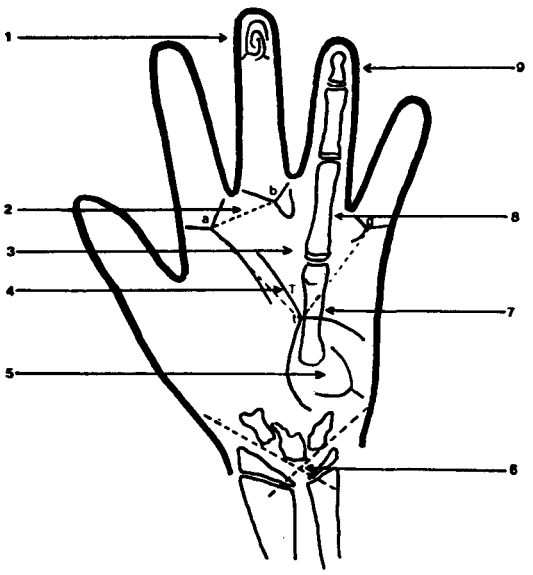

FIG. 1. *Schematic picture showing the dermatoglyphic and skeletal characteristics used in the diagnosis of Turner's syndrome:* (1) *total finger ridge count,* (2) ab *ridge count,* (3) *maximal* atd *angle,* (4) T *line terminating in the second interdigital interval,* (5) *hypothenar pattern,* (6) *carpal sign,* (7) *metacarpal sign,* (8) *phalangeal sign,* (9) *abnormally shaped distal phalange.*

TABLE II
THE SCORING METHOD

Characteristic	Percentage frequencies		Ratio between frequencies	Partial scores
	Patients	Controls		
1	72	44	1.63	1
2	90	40	2.25	2
3	62	38	1.78	1
4	52	18	2.89	2
5	63	50	1.26	1
6	62	5	22.4	12
7	50	2	25.0	25
8	58	2	29.0	30
9	25	5	5.0	5

the head of the 3rd metacarpal. The phalangeal sign (8: Leszcynski and Kosowicz 1965) is positive when the sum of the length of the distal and proximal phalanges is larger than the length of the metacarpal bones.

The partial scores were added up to form the final score, which we call the T.S. score.

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RESULTS

A comparison between the percentage distributions of the different characteristics in patients and controls, according to Student's *t* method, is shown in Table III. The same comparison between the percentage distributions of the nine characteristics in the two groups, but taking into account the score numbers, is reported in Table IV.

The mean T.S. score numbers, standard deviations, and *t* values for patients and

TABLE III
PERCENTAGE DISTRIBUTIONS OF THE DIFFERENT CHARACTERISTICS IN PATIENTS AND CONTROLS

		M	*SD*	*t*	*DF*
Only dermatoglyphic characteristics	Controls	38	22.08	3.76	8
	Patients	69	13.99		
Only skeletal characteristics	Controls	35	1.73	5.42	6
	Patients	48.75	16.60		
Dermatoglyphic and skeletal characteristics	Controls	22	20.12	4.18	16
	Patients	60	17.73		

TABLE IV
SCORE NUMBERS' PERCENTAGE DISTRIBUTIONS OF THE DIFFERENT CHARACTERISTICS IN PATIENTS AND CONTROLS

		M	*SD*	*t*	*DF*
Only dermatoglyphic characteristics	Controls	49.6	17.05	2.05	8
	Patients	97.4	48.89		
Only skeletal characteristics	Controls	40.75	16.52	2.65	6
	Patients	946.75	691.93		
Dermatoglyphic and skeletal characteristics	Controls	49.22	16.18	2.08	16
	Patients	482.29	62.26		

controls are given in Table V. In T.S. patients the mean score number is 4.88 when only the dermatoglyphic characteristics are considerd, 38.20 when only the skeletal characteristics are considered, and 43.14 when all the nine characteristics are considered together. The group of patients shows a highly significant T.S. score difference

from normal females, both when dermatoglyphic characteristics and bone abnormalities are examined separately or together ($P>0.001$).

The fitting of the normal distributions of T.S. scores, for dermatoglyphic traits, skeletal abnormalities, and combined dermatoglyphic and skeletal characteristics, in patients and controls, are given in Figs. 2, 3, and 4, respectively. When the derma-

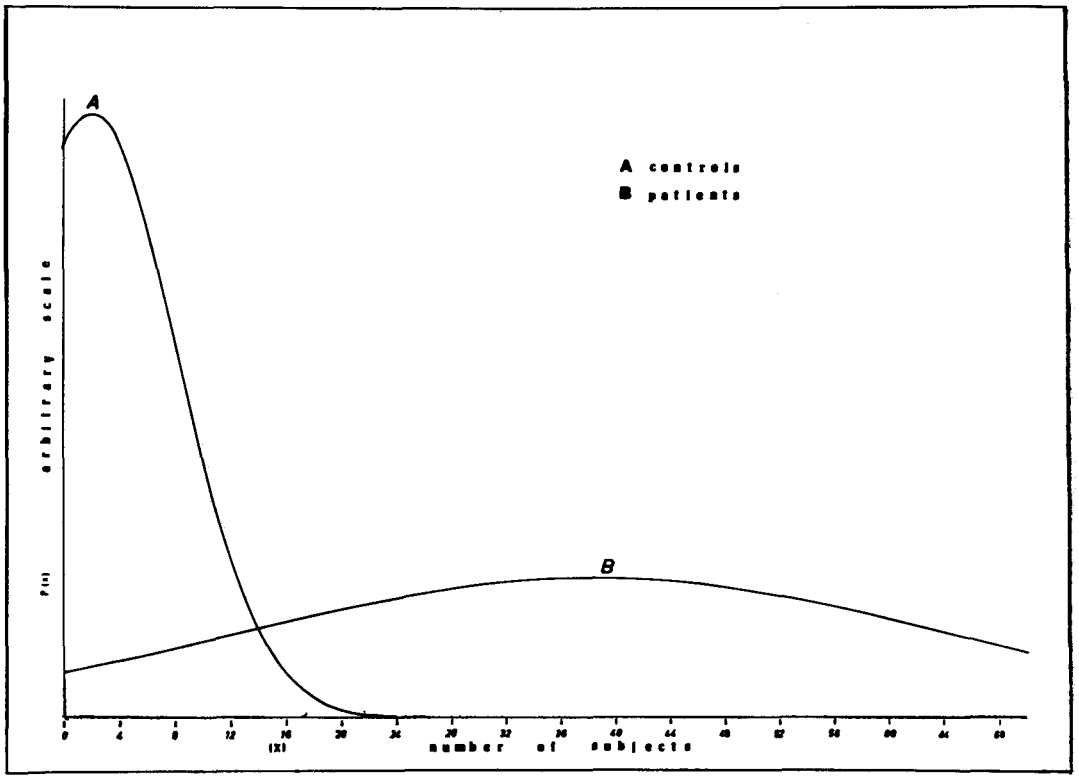

FIG. 2. *Fitting of the normal distributions of T.S. scores for dermatoglyphic characteristics in normal females and in patients*

toglyphic characteristics are considered, there is a satisfactory separation of patients from controls: about 10% of the controls had score numbers 5 and 6, which were present in 70% of the patients, and are considered to give a good degree of separation between the two groups of subjects. However, when the dermatoglyphic and skeletal characteristics are considered together, only 4% of the controls, compared with 80% of the patients, had score numbers higher than 20, which is considered to give a good degree of separation between the two groups of subjects.

On the basis of the figures obtained we made an attempt at a calculation of the probability that an individual with a given score number would be affected by the

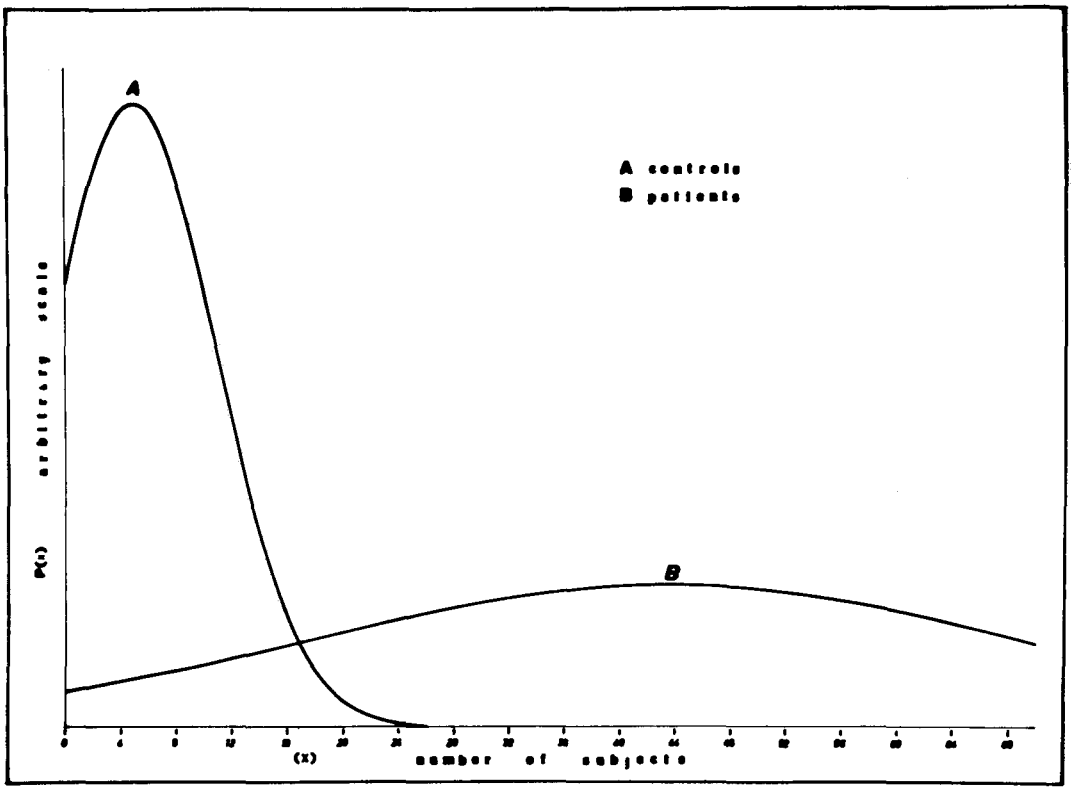

FIG. 3. *Fitting of the normal distributions of T.S. scores for skeletal characteristics in normal females and in patients.*

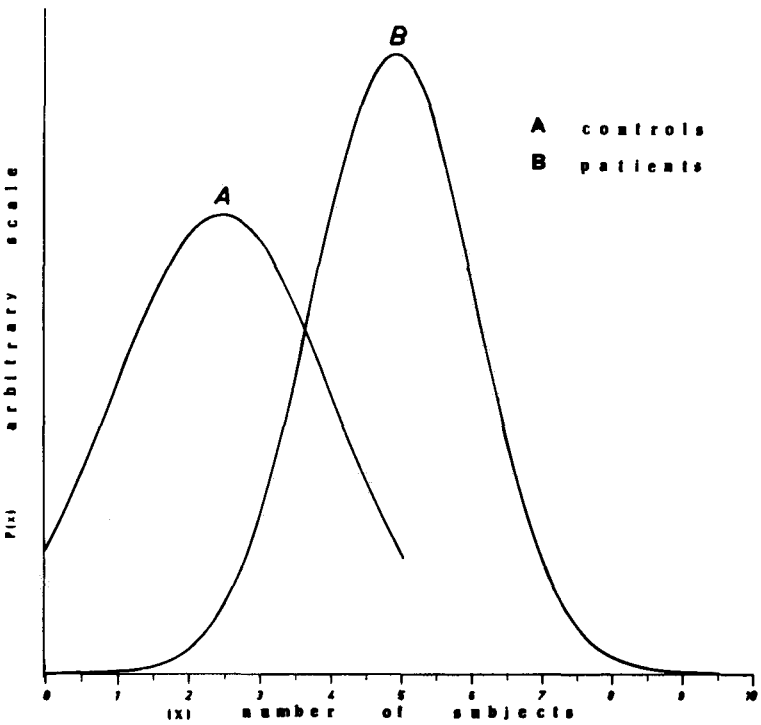

FIG. 4. *Fitting of the normal distributions of T.S. scores for dermatoglyphic and skeletal characteristics in normal females and in patients.*

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syndrome. Assuming normal distributions of the scores, we calculated for each score number the probability that an individual with that number would be a T.S. patient. The *M* and *SD* figures reported in Table V were used for normals and T.S. patients. The differences between the score number and the respective means were

TABLE V
MEAN SCORE NUMBERS IN PATIENTS AND CONTROLS

		M	*SD*	*t*
Only dermatoglyphic characteristics	Controls	2.48	1.53	8.91
	Patients	4.88	1.14	
Only skeletal characteristics	Controls	2.02	6.07	9.65
	Patients	38.20	25.79	
Dermatoglyphic and skeletal characteristics	Controls	4.80	5.97	10.15
	Patients	43.14	25.78	

expressed in *SD* units and the corresponding probability values (P^N = normal and P^T = T.S. patients) were extracted by fitting in the Gaussian distribution. The final probability for being a T.S. patient was calculated according to the formula suggested by Beckman et al. (1965):

$$\frac{P^T}{P^T + P^N} \%$$

The probability figures obtained for different score numbers and for different groups of characteristics considered are given in Table VI. It appears that, when only the dermatoglyphic characteristics are analyzed, the score numbers 2-4 will not be informative; when the dermatoglyphic and skeletal characteristics are considered together the score numbers 1-5 will not be informative. However, score numbers 5-7 and 20-35, respectively, suggest that an individual is a T.S. patient.

DISCUSSION

The present investigation was an attempt at a scoring method that would be of practical importance in the diagnosis of T.S. The information extractable from the five dermatoglyphic characteristics and from the four skeletal abnormalities selected have been measured, assigning to each characteristic partial scores, which were added up to a final T.S. score. The validity of this T.S. score has been proved, since it is well separating the patients from normal females.

TABLE VI
T.S. SCORES AND PROBABILITY OF BEING AFFECTED

T.S. score	Dermatoglyphic abnormalities	Skeletal abnormalities	Dermatoglyphic and skeletal abnormalities
0	0.05	7.7	7.3
2	5.5	8.0	
4	27.0		
5	83.0	10.4	
7	94.9	13.7	8.5
9		19.4	
10		23.5	12.9
12		35.2	
14		51.5	
15		60.7	35.5
20		93.7	78.8
25		99.6	98.2
30		99.98	99.9
35		99.99	99.999

In our series of patients a highly significant T.S. score difference from controls was found, both when only the dermatoglyphic characteristics were analyzed, or when they were analyzed together with the bone abnormalities. In this way it has been shown that the dermatoglyphic patterns are *per se* typical enough to separate the T.S. patients from normal females. However, it is interesting to note that, when the dermal configurations are considered in individual cases, 70% of patients have a T.S. score higher than 4, which is considered the score that permits to discriminate the patients from normals with a fairly high degree of probability. Furthermore, in about 80% of patients, a score number higher than 20 is obtained from the combined analysis of dermatoglyphic and skeletal characteristics. It means that the latter analysis is more informative in the diagnosis of T.S. than the single study of dermatoglyphics. Since the combined study of dermal configurations and of bone abnormalities is easily available by the X-ray technique we have employed in some of our cases, the present scoring method may be regarded as a useful tool in the diagnosis of T.S.

Other more simple techniques may have a more practical diagnostic interest especially in newborn babies, in which the T.S. scoring method cannot be employed for the immaturity of the skin ridges and the skeletal system. However, the present investigation has shown that a high proportion of cases which were diagnosed cytogenetically could have been safely diagnosed by means of the T.S. score. This finding proves that a correct diagnosis may be done by means of this scoring method and that cytogenetic analyses can be restricted to subjects where the T.S. score has not provided conclusive evidence.

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Riassunto

Viene descritto un metodo per la diagnosi della sindrome di Turner. Tale metodo è basato sullo studio di cinque segni dermatoglifici e quattro segni radiologici, i quali mostrano una significativa diversità di incidenza nelle pazienti, rispetto ai soggetti di sesso femminile di controllo. I cinque segni dermatoglifici comprendono: il conteggio totale delle creste sui polpastrelli, la distanza *ab*, l'angolo *atd*, l'emergenza della linea *T* nel secondo spazio interdigitale, la presenza di configurazioni sull'ipotenar. I segni radiologici comprendono: il segno dell'angolo carpale, il segno del metacarpo breve, il segno falangeale, la presenza di falangi distali dismorfiche. A ciascun segno sono stati assegnati valori arbitrari, calcolati sulla base della loro diversa incidenza nelle pazienti rispetto ai soggetti di controllo. La somma dei nove valori costituisce un utile indice per la diagnosi della sindrome di Turner, in quanto separa le pazienti dai soggetti di controllo. È stato infine calcolato per ogni punteggio finale la corrispondente probabilità di essere affetto da sindrome di Turner.

Résumé

Une méthode est décrite pour le diagnostic du syndrome de Turner sur la base de cinq traits dermatoglyphiques et quatre radiologiques dont la fréquence se présente significativement différente chez les patientes par rapport aux sujets féminins de contrôle. Les traits dermatoglyphiques sont les suivants: nombre total des crêtes digitales, distance *ab*, angle *atd*, terminaison de la ligne *T* dans la deuxième région interdigitale, présence de configurations sur l'hypothénar. Les traits radiologiques sont les suivants: signe de l'angle carpal, signe du métacarpe bref, signe phalangéal, présence de phalanges distales dysmorphiques. Chaque signe a été assigné une valeur arbitraire calculée sur la base de leur différence de fréquence chez les patientes par rapport aux contrôles. Finalement, pour toute valeur totale, la probabilité correspondante d'être atteint par le syndrome de Turner a été calculée.

Zusammenfassung

Beschreibung einer Methode zur Diagnose des Turnersyndroms. Diese Methode stützt sich auf die Untersuchung von fünf Hautleisten- und vier Röntgenmerkmalen, welche gegenüber den weiblichen Kontrollpersonen wesentlich in der Häufigkeit wechseln. Die fünf Hautleistenmerkmale sind: Gesamtzahl der Fingers-

pitzenlinien, der Abstand *ab*, der Winkel *atd*, das Hervortreten der *T*-Linie im zweiten Fingerzwischenraum, das Vorhandensein von Zeichnungen am kleinen Handballen. Die Röntgenmerkmale sind: das Handknochenwinkel, das kurze Mittelhandknochen, das Fingergliedmerkmal und das Vorhandensein von dysmorphen distalen Fingergliedern. Jedem Merkmal wurden willkürliche Werte beigemessen, je nach ihrem verschiedenen Vorkommen gegenüber den Kontrollpersonen. Die Summe der neun Werte stellt einen nützlichen Index für das Turnersyndrom dar, da sie eine deutliche Unterscheidung zwischen Patienten und Kontrollpersonen ermöglicht. Zum Schluss wurde noch für jede einzelne Schlussbewertung die entsprechende Wahrscheinlichkeit eines Turnersyndroms berechnet.

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