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Dermatoglyphs of Chinese Children with Down's Syndrome

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The characteristic stigmata of Down's syndrome (mongolism, trisomy 21) are rarely obscured by racial features (Emanuel, Huang, and Yeh, 1968). However, occasional patients, particularly infants, from any ethnic group may be difficult to diagnose clinically. In these equivocal situations and whenever confirmatory tests are indicated, a dermatoglyphic analysis of finger, hand, and foot prints has been shown to be a valuable objective tool (Cummins and Platou, 1946; Walker, 1958; Beckman, Gustavson, and Norring, 1965). The ideal of karyotyping all Down's syndrome patients usually is impractical, and karyotyping, even where facilities are available, commonly is restricted to patients of young mothers, those with a familial history of Down's syndrome, or those in whom the clinical picture is atypical.

Dermatoglyphic studies from several large European series showed good discrimination between Down's syndrome and control groups (Snedeker, 1948; Penrose, 1949, 1954; Turpin and Lejeune, 1953; Walker, 1957), but only one ethnic investigation on a non-European patient and control series, a Japanese study by Matsui, Nakagome, and Higurashi (1966), appears to have been done.‡ Such studies are required since various ethnic groups have different frequency ratios of dermatoglyphic features (Cummins and Midlo, 1943; Tiwari and Chattopadhyay, 1967). The present communication gives a dermatoglyphic analysis of 314 Chinese normal subjects and 53 Chinese patients with the clinical and cytogenetic diagnosis of Down's syndrome. Physical features and the cytogenetic findings of the patient group have been described previously (Emanuel *et al.*, 1968; Huang *et al.*, 1967).

Material and Methods

The patient sample, part of a comprehensive study of Chinese children with mongolism, consisted of 32 males and 21 females, none related. All of these were born in Taipei City and County in Taiwan. Most of these were referred from medical and educational centres in Taipei. Of these, 50 had simple trisomy 21 with 47 chromosomes (47,XX,21+ or 47,XY,21+)*; two had 46 chromosomes, a boy with a D/G translocation (46,XY,D−, t(DqGq)+) and a girl with a G/G translocation (46,XX, G−,t(GqGq)+); and one girl had a trisomy 21/normal mosaicism (47,XX,21+/46,XX). Previous studies of the dermatoglyphs of translocation and mosaic patients have shown them to be comparable with trisomic patients though the frequencies of some typical patterns are slightly reduced (Soltan and Clearwater, 1965; Rosner and Ong, 1967; Penrose 1965). The dermatoglyphs of these three patients are consistent with the findings of the remaining 50 trisomic patients and have been included in the following results. Their separate analyses have been filed with the National Auxiliary Publications Service (N.A.P.S.).

The control sample consisted of 163 male and 151 female unrelated Chinese children living in orphanages. Data broken down by sex, ethnic origin, and age have been filed with the N.A.P.S.

Finger, hand, and foot prints were taken using either a roller with printer's ink or Hollister† pad techniques. Conventional methods were used in dermatoglyphic interpretation (Cummins and Midlo, 1943; Penrose, 1968).

Results

The frequencies of fingerprint patterns as seen in our group of Chinese normal subjects are in general accord with previous population studies which have

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‡ An additional Japanese study has been recently published: Shiono, Kadowaki, and Kasahara (1969).

* Nomenclature as standardized by Chicago Conference (1966).
† Hollister, Inc., 833 North Orleans Street, Chicago, Illinois.

TABLE I

FINGERPRINT PATTERN PERCENTAGES IN DOWN'S SYNDROME (53) AND CONTROLS (314)

Pattern	Group	Left V	Left IV	Left III	Left II	Left I	Right I	Right II	Right III	Right IV	Right V
Ulnar loop	Down's	67·9	67·9***	88·7***	86·8***	66·0***	62·3***	98·1***	92·5***	66·0***	73·6
	Control	70·7	34·4	56·1	34·7	40·1	33·8	33·8	58·0	30·6	60·5
Whorl	Down's	20·8	22·6***	11·3***	9·4***	30·2***	34·0***	1·9***	7·6***	24·5***	20·8*
	Control	29·0	64·7	40·5	53·2	57·8	63·7	50·7	40·0	69·1	38·5
Radial loop	Down's	9·4***	9·4***	0·0	1·9	0·0	1·9	0·0*	0·0	7·6***	5·7**
	Control	0·3	0·3	1·0	7·3	0·6	0·3	11·2	0·6	0·3	0·6
Arch	Down's	1·9*	0·0	0·0*	1·9	3·8	1·9	0·0	0·0	1·9*	0·0
	Control	0·0	0·6	2·6	4·8	4·5	2·2	4·5	2·0	0·0	0·3

* $p < 0·05$.
** $p < 0·01$.
*** $p < 0·001$.

shown high intensity fingerprint patterns (whorls) in the Orient and also in the Near East (Cummins and Midlo, 1943; Matsui *et al.*, 1966).

Our patients had an increased frequency of ulnar loops on digits i to iv bilaterally, largely at the expense of whorls and to a lesser degree at the expense of radial loops on the second digit. There was an increase in radial loops on the fourth and fifth digits (Table I).

TABLE II

AXIAL TRIRADIAL HEIGHT 40% OR HIGHER* (PERCENTAGES)

	Controls (313) Males (163)	Females (150)	Total	Down's Syndrome (51) Males (31)	Females (20)	Total
Trait present:						
Bilateral	0·0	0·0	0·0	35·5	35·0	35·3
Unilateral left	0·0	0·7	0·3	9·7	25·0	15·7
Unilateral right	1·2	1·3	1·3	6·5	10·0	7·8
Total	1·2	2·0	1·6	51·6	70·0	58·8***
Trait absent:	98·8	98·0	98·4	48·4	30·0	41·2

* Percentage distance of most distal axial triradius measured on perpendicular line drawn from distal wrist crease to proximal metacarpophalangeal crease of third digit.
*** $p < 0·001$.

TABLE III

PALMAR THIRD INTERDIGITAL PATTERNS AND REDUCED* C MAINLINE PERCENTAGES

	Controls (314)	Down's Syndrome (51)
Left pattern	6·1	19·6***
Left reduced C	15·0	54·9***
Total	21·0	74·5
Normal C and no pattern—left	79·0	25·5
Right pattern	29·3	49·0**
Right reduced C	14·3	17·7
Total	43·6	66·7
Normal C and no pattern—right	56·4	33·3

* Mainline abortive or absent (X, x, or O).
** $p < 0·01$.
*** $p < 0·001$.

Palms. On the palm the dermatoglyphic feature of trisomy 21 primarily was the occurrence of distal axial triradii (t'') and a consequent increase in hypothenar patterns with some shifting of patterns

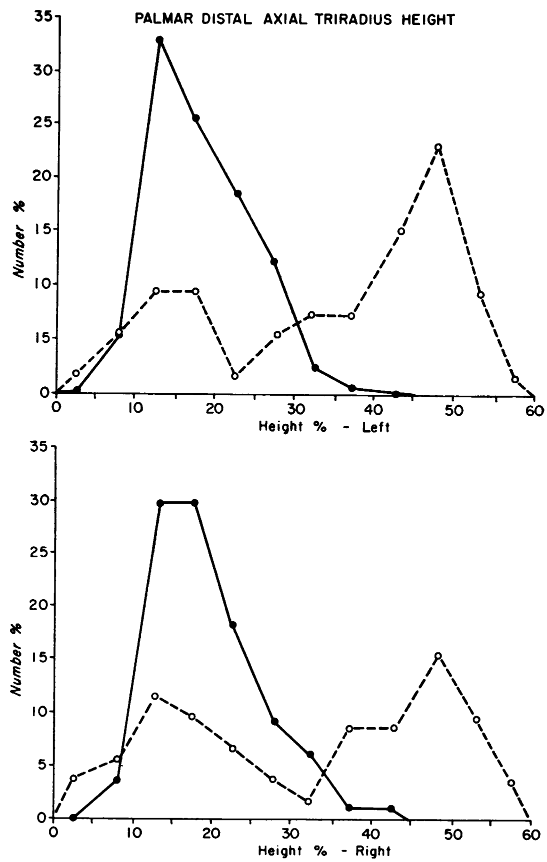

FIG. 1. The palmar distal axial triradius height of 53 Chinese Down's syndrome patients (broken line) is bimodal in contrast to 314 Chinese controls (solid line).

from the fourth interdigital to the third interdigital areas (Tables II and III). The distal axial triradius was expressed in percentage distance from the distal wrist crease to the proximal metacarpophalangeal crease of the third digit. Over 98% of the control hands had triradial heights lower than 40% in contrast to 50% left and 57% right of the patients. Curves of the control and patient triradial heights show the patient curve to be bimodal in contrast to the controls (Fig. 1).

The C mainline is the ridge that begins as the proximal radiant from triradius *c*, which is situated at the base of the ring finger. The line usually extends to an exit at the distal or ulnar edge of the palm but sometimes terminates abruptly or even is absent. There was a striking increase in frequency of reduced C mainlines on the left palm of the patient group. Abortive and absent C mainlines were found on 55% of the patient left palms as compared with 15% in the controls (Table III).

Feet. Over 90% of the plantar hallucal patterns of the patients showed an arch tibial in contrast to 7% of left and 5% right in the controls (Table IV).

TABLE IV

PLANTAR HALLUCAL ARCH TIBIAL PATTERN PERCENTAGES

	Controls (313)			Down's Syndrome (53)		
	Males (162)	Females (151)	Total	Males (32)	Females (21)	Total
Arch tibial present:						
Bilateral	3·1	2·7	2·8	90·6	85·7	88·7
Unilateral left	4·3	4·0	4·2	3·1	0·0	1·9
Unilateral right	2·5	2·0	2·2	3·1	4·8	3·8
Total	9·9	8·6	9·3	96·9	90·5	94·3***
Other patterns	90·1	91·4	90·7	3·1	9·5	5·7

*** $p < 0·001$.

More than 50% of controls had a loop distal and over 25% had a whorl pattern.

True patterns in the plantar second interdigital area were absent in the patient group and present in over 7% left and 9% right of the control group. No significant difference was seen in the third interdigital area. There was only a 5% left and 9% right increase in patterns of the fourth interdigital area of the patient group when data from males and females were pooled. However, when males and females were analysed separately, 20% both left and right of the females had a pattern here as opposed to only 2% left and 4% right of the female controls. While 6% left and 19% right of male patients had a pattern, 10% left and 16% right male controls likewise had fourth interdigital plantar patterns (Table V).

TABLE V

TRUE PATTERNS IN PLANTAR FOURTH INTERDIGITAL AREA PERCENTAGES

	Control Females (151)	Down's Syndrome Females (20)	Control Males (162)	Down's Syndrome Males (31)
Left pattern	2·0	20·0***	9·9	6·5
No pattern left	98·0	80·0	90·1	93·5
Right pattern	4·0	20·0***	16·1	19·4
No pattern right	96·0	80·0	83·9	80·6

*** $p < 0·001$.

Great toe patterns showed a slight increase in whorls and tibial loops at the expense of arches in the patient group.

Creases. Though creases are not components of dermatoglyphs in the strict sense, they are treated here because certain departures in mongolism are well known. The presence or absence of a single crease on the fifth digit replacing the normal two interphalangeal creases and the presence or absence of a single transverse palmar crease were scored.

A transverse palmar crease (simian crease, four-finger fold) was defined strictly as a single, prominent, horizontal crease, approximately straight, and extending from border to border. Transitional forms, while observed, were not scored as positive traits. 50% of the male patients and over 28% female patients had a transverse palmar crease as opposed to less than 10% on control palms (Table VI). There was an increased frequency in males in both groups and additionally on the left side in the patient group. No controls were seen with a single crease on the fifth finger while over 43% of the males and 28% of the females with mongolism had this trait.

Discussion

One purpose of this survey was to investigate how closely the dermatoglyphic frequencies of a group of Chinese patients with chromosomally proved Down's syndrome resembled European patients, particularly in the light of the differences between the two ethnic control groups. The results of this series of patients are much closer to those of the Japanese study by Matsui *et al.* (1966) than to previously reported European series (Table VII).

TABLE VI—CREASES PERCENTAGES

A—TRANSVERSE PALMAR CREASE

	Controls (314)			Down's Syndrome (53)		
	Males (163)	Females (151)	Total	Males (32)	Females (21)	Total
Trait present						
Bilateral	4·9	0·7	2·9	15·6	14·3	15·1
Unilateral left	3·7	2·7	3·2	25·0	9·5	18·9
Unilateral right	3·7	4·0	3·8	9·4	4·8	7·6
Total	12·3	7·3	9·9	50·0	28·6	41·5***
Trait absent	87·7	92·7	90·1	50·0	71·4	58·5

B—SINGLE CREASE 5th DIGIT (DOWN'S SYNDROME—53)* ***

	Males (32)	Females (21)	Total
Trait present			
Bilateral	21·9	23·8	22·6
Unilateral left	3·1	4·8	3·8
Unilateral right	18·8	0·0	11·3
Total	43·8	28·6	37·7
Trait absent	56·2	71·4	62·3

* No controls were seen with this trait.
*** $p < 0·001$.

Fingerprints. Fig. 2 shows the fingerprint frequencies of this group of Chinese controls and patients, and the European controls and patients, largely from North European stock, as reported by Walker in her North American survey (1957).

The most consistent difference between the control groups was the greater incidence of whorls on all digits in the Chinese group. The increased whorl frequency in our Chinese group corresponded mainly to a proportional increase of ulnar loops on digits i, iii, iv, and v; and radial loops on digits ii as well as a smaller increase in arches on digits ii and iii in the European group. The differences between these two control groups, with few exceptions, are greater than, and in the same direction away from, the differences of the patient groups.

The effect on fingertip patterns of trisomy 21 is remarkably similar regardless of whether the genome is of Chinese or Northern European extraction. However, this Chinese patient group had a frequency of radial loops on the left fourth and fifth digits twice the European group in addition to a reduction of whorls on the second digit right side only. This latter observation may have been a fortuitous finding in this particular sample. Additional data are needed to clarify the point. It should be noted from Fig. 2 that the majority of the slight pattern frequency differences of the Chinese patients resulted in a greater distinction from the Chinese controls than that found in European populations. Contributing to this effect was the increase of ulnar loops and decrease of whorls on the second through fourth digits bilaterally in the Chinese patients as compared to the European patients. This was an unexpected finding because a study by Walker and Johnson (1964) showed that Italian patients with Down's syndrome reflected their Mediterranean ancestry by more whorls than the patients from Walker's earlier study (1958).

Palms. On the other hand, the palmar dermatoglyphs of the Chinese patients do reflect the lowered incidence of a distal axial triradius (40% or higher) and third interdigital pattern of the Chinese controls as compared both to Walker's series (1957) and to the British series by Penrose (1954) and Fang (1950) (Table VII).

TABLE VII

ETHNIC COMPARISONS (PERCENTAGES)

	Down's Syndrome				Control			
	Chinese*	Japanese†	N. American‡	British§	Chinese*	Japanese†	N. American‡	British§
Radial loop 4th digit	8·5	6·2	5·4	4·8	0·3	0·3	0·6	0·7
High axial triradius	46·6	46·3	85·1	83·2	0·8	4·3	11·8	8·5
Palmar 3rd interdigital pattern—left	19·6	24·0	54·0	52·2	6·0	3·0	31·3	25·7
—right	49·0	43·8	85·4	83·9	29·3	13·9	55·5	48·2
Hallucal arch tibial	91·5	87·7	47·0	50·4	6·1	4·6	0·3	0·2
Total whorls	18·3	20·8	20·1	12·7	46·6	48·3	28·4	26·1
Total ulnar loops	77·0	76·3	75·1	82·8	49·1	46·7	61·2	63·5
Total radial loops	3·6	2·1	2·5	1·8	2·3	2·7	4·9	5·4
Total arches	1·1	0·8	2·3	2·7	2·1	2·3	5·6	5·0

* Present study.
† Matsui *et al.* (1966).
‡ Walker (1957).
§ Fingerprint data from Holt (1964); axial triradii from Penrose (1954); interdigital patterns from Fang (1950); hallucal patterns from Penrose and Smith (1966).

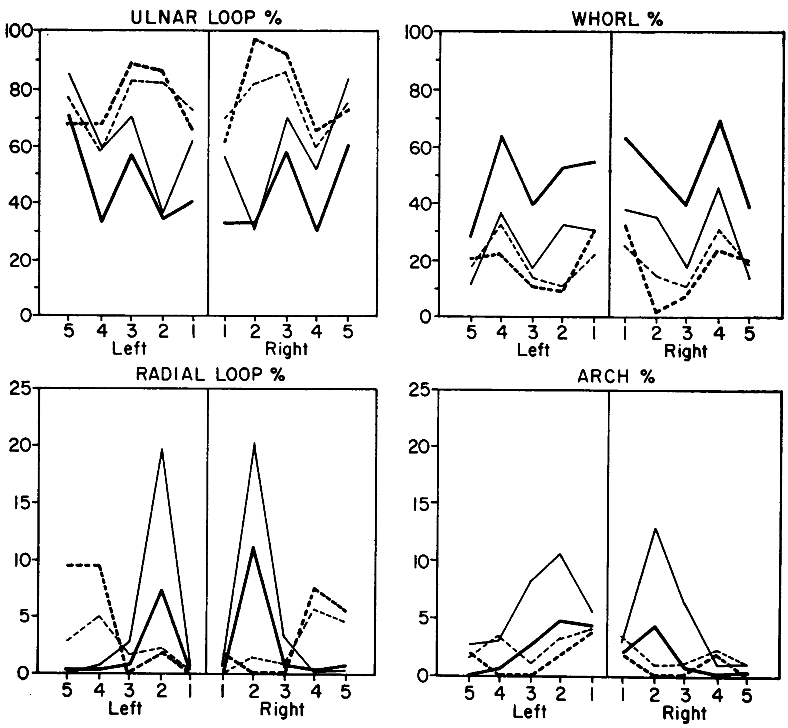

FIG. 2. A comparison of percentage frequencies of fingerprint patterns in Down's syndrome patients and controls in Caucasoid and Chinese populations. The broken lines indicate patient groups and the solid lines indicate controls. The thick lines represent the Chinese figures from this study and the thin lines indicate Caucasoid figures from Walker's series (1957).

All the groups examined have shown a larger frequency of third interdigital patterns on the right hand, much accentuated in the Down's syndrome groups. This is in contrast to the general observation that the dermatoglyphs of patients with Down's syndrome are more symmetrical than control groups (Cummins, 1939). However, if an absent or abortive C mainline is viewed as a variation of this dermatoglyphic disorder (Cummins and Midlo, 1943) and these frequencies are combined (Table II), the unilateral phenomenon seen in the controls disappears in our patient group, and instead a slightly greater frequency of abnormalities is seen in the distal portion of the left palm than in the right, which perhaps may be correlated with the slightly greater frequency of a raised axial triradius in the left hand. The transverse palmar crease also has a slightly higher frequency on the left hand of our patient group.

As noted above, Fig. 1 shows that the curve of the Chinese patient axial triradial height is bimodal in contrast to the control group. A delay in the regression of the fetal hypothenar pad at the time the dermal ridges are forming could produce this effect by causing the triradius to form in a more distal site than normal, or sometimes in a proximal position, but rarely in the intermediate elevated pad area (Cummins, 1926; Cummins and Midlo, 1943). Frequently the hands of patients who lacked a well-defined distal triradius showed a disordered arrangement of ridges that suggested a vestigial triradius in the distal area of the palm.

It may be that all of the observed palmar stigmata of Down's syndrome—increased height of axial triradii, increased hypothenar patterns, increased transverse palmar creases, increased third interdigital patterns, and reduced C mainlines, basically are secondary dermatoglyphic reflections of an abnormally persistent prenatal hypothenar pad.

Feet. The high concordance of arch tibial patterns on the plantar hallucal area with Down's syndrome in the Chinese was most striking. Only 3 patients in this series lacked an arch tibial on both feet, and only an additional 3 patients did not have bilateral arch tibials. 3% of the controls did have arch tibials on both feet, and an additional 6% had a single arch tibial. This was only slightly more than the Japanese survey (Matsui *et al.*, 1966) but in marked contrast to the North American (Walker, 1957) and British (Penrose and Smith, 1966) series (Table VII).

While our Chinese male controls had a higher frequency of fourth interdigital patterns than our female controls and our female patients had a significant increase as compared with female controls in agreement with a European study by Smith (1964), it was surprising to find that our male patients did not have the increased pattern frequency as observed by Smith.

Creases. While no normal controls have shown the occurrence of a single crease on fifth digits, our Chinese patients did have a higher frequency than reported in other series (Penrose, 1931; Uchida and Soltan, 1963; Hall, 1964; Soltan and Clearwater, 1965).

The strict definition of a transverse palmar crease used in this work makes comparison with most other series difficult, but our Chinese controls had a higher frequency than found in European populations, though the patient groups were similar (Beckman *et al.*, 1962; Davies and Smallpeice, 1963; Beckman *et al.*, 1965). This observation is in agreement with an extensive comparative study by Van Der Wiel (1953).

Comment

In this Chinese study, the hallucal arch tibial pattern had strong diagnostic value in differentiating Down's syndrome patients from normal controls. In each of the few exceptional cases, analysis of the palmar axial triradii provided further discrimination required to place the individual in the normal or abnormal group consistent with his cytogenetic findings.

The characteristic dermatoglyphic profile in Down's syndrome as described originally in European populations has been verified in this group of Chinese patients. The frequency expression of some of these features was different in the patient groups, usually, though not consistently, in direct relation to the pattern differences between the ethnic groups. Therefore we conclude that knowledge of ethnic pattern frequencies is valuable when using dermatoglyphic criteria as a diagnostic aid. An important precaution and extension of this observation is that a particular familial inheritance of dermatoglyphic patterns, in any ethnic group, also may have the effect of 'masking' or perhaps enhancing the characteristic patterns of a specific disorder such as Down's syndrome. Dermatoglyphic examinations whenever possible should include parents and normal sibs, and interpretations be made in light of expected familial inheritance as well as abnormal dermatoglyphs.

Summary*

A dermatoglyphic study of a group of 314 Chinese controls and 53 Chinese patients with the clinical and cytogenetic diagnosis of Down's syndrome has been presented, and results compared with other studies of European groups. The Chinese patient group had findings similar, but not identical, to European patient studies. Some, but not all, of the differences were in direct relation to the differences observed between the control groups. The hallucal arch tibial pattern had strong diagnostic value in the Chinese group. Other valuable traits included the palmar axial triradii height, transverse palmar creases, and single flexion creases on the fifth digit. It was suggested that the bimodal curve of the palmar axial triradii height of the patient group provides evidence for the developmental mechanism leading to palmar abnormalities seen in Down's syndrome.

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* Documentary tables regarding ethnic origin, sex, age, and all dermatoglyphic data referred to have been filed with the National Auxiliary Publications Service of the American Society for Information Science, and may be retrieved by writing: c/o CCM Information Sciences, Inc., 22 West 34th Street, New York, New York 10001, U.S.A.

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