Palmar Dermatoglyphs in Tuberous Sclerosis

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Dermatoglyphs are well established as useful physical signs in patients with chromosome abnormalities, as well as being of interest in patients with certain congenital malformations. What is still unclear, however, is whether single gene disorders can directly affect dermatoglyphs. The dermatoglyphic changes found in the Holt-Oram syndrome (Rosner and Aberfeld, 1970) are often claimed to be an example of a single gene disorder affecting the dermatoglyphs, although the dermatoglyphic abnormalities in this condition are quite clearly due to the hand deformity and not the gene *per se*. In the same way, onychia affects fingerprint patterns (Penrose, 1968), but this is only a secondary effect of the nail abnormality. The claim that Wilson's disease is associated with increased whorls (Hodges and Simon, 1962) has been refuted (David, 1971a; David and Ajdukiewicz, 1972) and normal dermatoglyphs have been reported in phenylketonuria (Alter, 1967). The only remaining claims, that patients with thalassaemia (Rosner and Spriggs, 1969) and patients with Huntington's chorea (Barbeau, Trudeau, and Coiteux, 1965) have minor dermatoglyphic differences from control groups, both require confirmation. Recently, females with congenital virilizing adrenal hyperplasia have been reported to have significantly larger pattern size, but it was suggested that this was the result of increased growth from the 3rd month of intrauterine life, and therefore this is not a direct genetic effect (Qazi and Thompson, 1971).

Dermatoglyphs have been studied in patients with tuberous sclerosis for 2 reasons. Firstly, it is known that this disease is inherited as an autosomal dominant condition (Nevin and Pearce, 1968)—with a substantial proportion of cases representing new mutations (Bundey and Evans, 1969)—and hence this is an appropriate condition to look for evidence of a single gene disorder having characteristic dermatoglyphs. Secondly, striking dermatoglyphic abnormalities, if found, would be a useful aid to diagnosis.

Patients and Methods

Everyone studied lived in the south and west of England. The controls, 500 males and 500 females, consisted of unrelated individuals representing as far as possible a cross section of social classes.

The patients were either studied in mental subnormality institutions or they were studied at home after prior consent had been obtained from the paediatrician and the relatives. The diagnostic criteria for every case were either (1) severe mental subnormality with grand mal epilepsy and adenoma sebaceum or (2) grand mal epilepsy, adenoma sebaceum, and an air encephalogram which supported the diagnosis. Two patients who did not meet these criteria and were living at home were not approached. Three patients had to be excluded because severe spasticity precluded palm printing, and a 4th patient in an institution refused to cooperate, leaving a total of 54 patients who were fully studied.

The prints were recorded on specially prepared fingerprint forms, using black fingerprint ink. Rolled and plain impressions were obtained from all fingers, and 3 sets of palm prints were recorded, one of which was taken with the fingers fully adducted (for *atd* angle measurement).

Features studied were fingerprint patterns; total finger ridge count; summed *a-b* ridge count; patterns in the thenar, hypothenar, and interdigital areas; summed corrected *atd* angles, mean ridge breadth, and palmar flexion creases. The left and right *atd* angles were corrected for lateral deviation of the *t* triradius (David, 1971b) before they were added together to give the summed corrected *atd* angle. The mean ridge breadth was calculated by dividing the summed *a-b* distance in millimetres by the summed *a-b* ridge count + 2.

Results

Fingerprint Patterns. Tables I and II show the frequencies and actual numbers of fingerprint pattern types. These were compared with the controls either by the chi-square test, or if the numbers were small, by Fisher's exact significance taking individual patterns on single fingers (eg, there were 66 whorls on the left thumb in female controls and 3 in the female patients). Out of 100 such comparisons, 2 results were significant at $p < 0.025$. This incidence was to be expected by chance alone.

TABLE I
FREQUENCIES OF FINGERPRINT PATTERNS IN 500 MALE CONTROLS AND 29 MALE PATIENTS

	Left Hand*					Right Hand*				
	5	4	3	2	1	1	2	3	4	5
<i>Arches + tented arches</i>										
Controls	3 (0.6%)	5 (1.0%)	46 (9.2%)	55 (11.0%)	16 (3.2%)	7 (1.4%)	68 (13.6%)	54 (10.8%)	2 (0.4%)	4 (0.8%)
Patients	0	0	3 (10.3%)	1 (3.4%)	0	0	2 (6.9%)	1 (3.4%)	0	0
<i>Ulnar loops</i>										
Controls	426 (85.2%)	314 (62.8%)	346 (69.2%)	182 (36.4%)	324 (64.8%)	279 (55.8%)	139 (27.8%)	334 (67.0%)	247 (49.4%)	394 (78.8%)
Patients	27 (93.1%)	21 (72.4%)	22 (75.9%)	15 (51.7%)	20 (69.0%)	17 (58.6%)	11 (37.9%)	23 (79.3%)	15 (51.8%)	23 (79.3%)
<i>Radial loops</i>										
Controls	0	1 (0.2%)	9 (1.8%)	110 (22.0%)	0	1 (0.2%)	119 (23.8%)	15 (3.0%)	10 (2.0%)	3 (0.6%)
Patients	0	0	0	4 (13.9%)	0	0	7 (24.1%)	0	1 (3.4%)	0
<i>Whorls</i>										
Controls	60 (12.0%)	170 (34.0%)	85 (17.0%)	130 (26.0%)	72 (14.4%)	144 (28.8%)	149 (29.8%)	100 (18.0%)	237 (47.4%)	90 (18.0%)
Patients	2 (6.9%)	6 (20.7%)	3 (10.4%)	8 (27.6%)	2 (6.9%)	6 (20.7%)	6 (20.7%)	5 (17.3%)	13 (44.8%)	6 (20.7%)
<i>Twinned loops</i>										
Controls	11 (2.2%)	10 (2.0%)	14 (2.8%)	23 (4.6%)	88 (17.6%)	69 (13.8%)	25 (5.0%)	6 (1.2%)	4 (0.8%)	9 (1.8%)
Patients	0	0 (6.9%)	0 (3.4%)	0 (3.4%)	0 (24.1%)	6 (20.7%)	3 (10.4%)	0	0	0

* Fingers are numbered 1 (thumb) to 5 (little finger).

TABLE II
FREQUENCIES OF FINGERPRINT PATTERNS IN 500 FEMALE CONTROLS AND 25 FEMALE PATIENTS

	Left Hand*					Right Hand*				
	5	4	3	2	1	1	2	3	4	5
<i>Arches + tented arches</i>										
Controls	11 (2.2%)	15 (3.0%)	67 (13.4%)	69 (13.8%)	30 (6.0%)	15 (3.0%)	76 (15.2%)	44 (8.8%)	10 (2.0%)	7 (1.4%)
Patients	3† (12.0%)	2 (8.0%)	3 (12.0%)	3 (12.0%)	3 (12.0%)	3 (12.0%)	3 (12.0%)	4 (16.0%)	2 (8.0%)	2 (8.0%)
<i>Ulnar loops</i>										
Controls	432 (86.4%)	332 (66.4%)	359 (71.8%)	198 (39.6%)	329 (65.8%)	316 (63.2%)	203 (40.6%)	392 (78.4%)	286 (57.2%)	436 (87.2%)
Patients	20 (80.0%)	12 (48.0%)	18 (72.0%)	8 (32.0%)	17 (68.0%)	15 (60.0%)	12 (48.0%)	20 (80.0%)	12 (48.0%)	20 (80.0%)
<i>Radial loops</i>										
Controls	2 (0.4%)	6 (1.2%)	9 (1.8%)	99 (19.8%)	2 (0.4%)	1 (0.2%)	72 (14.4%)	9 (1.8%)	4 (0.8%)	1 (0.2%)
Patients	0	1 (4.0%)	0	3 (12.0%)	0	0	0†	0	0	0
<i>Whorls</i>										
Controls	50 (10.0%)	140 (28.0%)	52 (10.4%)	109 (21.8%)	66 (13.2%)	101 (20.2%)	125 (25.0%)	48 (9.6%)	195 (39.0%)	55 (11.0%)
Patients	2 (8.0%)	10 (40.0%)	3 (12.0%)	9 (36.0%)	3 (12.0%)	4 (16.0%)	7 (28.0%)	1 (4.0%)	10 (40.0%)	3 (12.0%)
<i>Twinned loops</i>										
Controls	5 (1.0%)	7 (1.4%)	13 (2.6%)	25 (5.0%)	73 (14.6%)	67 (13.4%)	24 (4.8%)	7 (1.4%)	5 (1.0%)	1 (0.2%)
Patients	0	0	1 (4.0%)	2 (8.0%)	2 (8.0%)	3 (12.0%)	3 (12.0%)	0	1 (4.0%)	0

* Fingers are numbered from 1 (thumb) to 5 (little finger).

† Significant at $p < 0.025$.

One was an increase in arches plus tented arches on the left little finger of female patients, and the other was a decrease in the radial loops on the right index finger of female patients. The same digits in male patients showed the opposite trends, and therefore no importance is attached to these 2 results.

Total Finger Ridge Count (TRC). As previous investigators have found, the distribution of the TRC is not normal (Holt, 1955/1956), and this was so in both the patients and controls (sexes studied separately), so that it is invalid to compare the means (Table III) by Student's *t* test. Comparison was made by the Wilcoxon-White test, which showed that there was no significant difference between the patients and controls in either sex, *p* being 0.805 in males and 0.172 in females.

TABLE III
TOTAL FINGER RIDGE COUNT (TRC)

	Mean TRC and Standard Deviation	Median	Inter- quartile Range	Total Range
<i>Males</i>				
Controls (500)	141.2 ± 48.7	144	105-177	12-273
Patients (29)	143.7 ± 45.5	146	105-166	38-235
<i>Females</i>				
Controls (500)	126.4 ± 48.1	130	92-161	0-284
Patients (25)	112.1 ± 54.5	112	77-163	1-193

Summed (right + left) *a-b* Ridge Count. The distribution was normal in patients and controls, and the means are shown in Table IV. The *a-b* count was lower in the patients than the controls for

TABLE IV
SUMMED *a-b* RIDGE COUNT

	Mean and Standard Deviation	<i>p</i>
<i>Males</i>		
Controls (500)	86.1 ± 11.3	< 0.05
Patients (29)	81 ± 12.4	
<i>Females</i>		
Controls (500)	84.5 ± 10.2	Not significant
Patients (25)	81.4 ± 9.1	

both sexes, but the difference was only significant in the males (*p* < 0.05).

Palmar Flexion Creases. None of the patients had any abnormalities of the palmar creases.

Palmar Interdigital Patterns. The patterns in the IIInd, IIIrd, and IVth spaces were studied separately. Only true patterns were recognized (loops and whorls) so that open fields and absent triradii were not considered to be patterns. The results are shown in Table V. There were no significant differences between the patients and controls (by the chi-square test).

Thenar Patterns. These are nearly always vestigial patterns rather than true loops or whorls. The results are shown in Table VI. There were no significant differences between the patients and controls (by the chi-square test).

Hypothenar Patterns. These were classified into the following groups: (a) no pattern, (b) distal

TABLE V
PALMAR INTERDIGITAL PATTERNS

	IIInd Space			IIIrd Space			IVth Space		
	None	Loop	Whorl	None	Loop	Whorl	None	Loop	Whorl
<i>Right palm</i>									
Male controls (500)	472 (94.4%)	28 (5.6%)	0	239 (47.8%)	259 (51.8%)	2 (0.4%)	275 (55.0%)	225 (45.0%)	0
Male patients (29)	28 (96.6%)	1 (3.4%)	0	14 (48.3%)	15 (51.7%)	0	17 (58.6%)	12 (41.4%)	0
Female controls (500)	481 (96.2%)	19 (3.8%)	0	239 (47.8%)	258 (51.6%)	3 (0.6%)	261 (52.2%)	236 (47.2%)	3 (0.6%)
Female patients (25)	24 (96.0%)	1 (4.0%)	0	11 (44.0%)	14 (56.0%)	0	15 (60.0%)	10 (40.0%)	0
<i>Left palm</i>									
Male controls (500)	484 (96.8%)	16 (3.2%)	0	370 (74.0%)	130 (26.0%)	0	192 (38.4%)	299 (59.8%)	9 (1.8%)
Male patients (29)	28 (96.6%)	1 (3.4%)	0	23 (79.3%)	6 (20.7%)	0	9 (31.0%)	20 (69.0%)	0
Female controls (500)	495 (99.0%)	5 (1.0%)	0	355 (71.0%)	145 (29.0%)	0	187 (37.4%)	307 (61.4%)	6 (1.2%)
Female patients (25)	25 (100.0%)	0	0	16 (64.0%)	9 (36.0%)	0	13 (52.0%)	12 (48.0%)	0

TABLE VI
THENAR PATTERNS

	None	Bilateral	Right	Left
Males				
Controls (500)	410 (82.0%)	37 (7.4%)	9 (1.8%)	44 (8.8%)
Patients (29)	27 (93.2%)	1 (3.4%)	0	1 (3.4%)
Females				
Controls (500)	419 (83.8%)	32 (6.4%)	9 (1.8%)	40 (8.0%)
Patients (25)	23 (92.0%)	1 (4.0%)	1 (4.0%)	0

loop, (c) outward (ulnar) loop, (d) whorl, (e) twinned loop, and (f) other patterns (arch, tented arch, proximal loop, lateral pocket loop, nutant loop, composite, double proximal axial triradius). The results are shown in Table VII. There were no significant differences between the patients and controls, using the chi-square and Fisher's exact significance tests.

Summed (right + left) Corrected *atd* Angles.

Since the *atd* angle varies with several factors, attempts were made to exclude these factors by: (1) dividing patients and controls into different age groups (because the *atd* increases with age during growth); (2) taking the palm print for *atd* angle measurement with the fingers maximally adducted (because abduction of the fingers can greatly increase the *atd* angle); (3) correcting each *atd* angle by a special formula (David, 1971b)—to exclude the effect of lateral deviation of the *t* triradius; and (4) measuring the *atd* angle from the most proximal *t* triradius where a hypothenar pattern was present (otherwise the genuine distal displacement of the *t*

triradius cannot be distinguished from the presence of a hypothenar pattern).

The summed corrected *atd* angles were found to be normally distributed in all groups of patients and controls where there were sufficient numbers, and statistical comparisons were made on 3 age groups—4–7 years, 8–11 years, and 20 years and over. The results are shown in Table VIII. There were no significant differences between the patients and controls by Student's *t* test.

TABLE VIII
SUMMED CORRECTED *atd* ANGLES BY AGE WITH
SEXES COMBINED

	Mean and Standard Deviation	p
4–7 years		
Controls (33)	94.3 ± 23.1	Not significant
Patients (8)	92.6 ± 11.5	
8–11 years		
Controls (66)	85.1 ± 10.2	Not significant
Patients (5)	91.4 ± 6.5	
20 years and over		
Controls (640)	85.3 ± 13.2	Not significant
Patients (30)	82.3 ± 10.1	

Mean Ridge Breadth (MRB). This is estimated by using the *a–b* distance and ridge count. It is influenced both by age and sex, so that the patients and controls were studied in the same age groups as for the *atd* angle, but with the sexes separate. Only in those patients over 20 years of age group were the numbers sufficient to establish that the distributions were normal and to enable statistical comparison. No significant differences were found comparing patients and controls. The means are shown in Table IX.

TABLE VII
HYPOTHENAR PATTERNS

	None	Distal Loop	Outward Loop	Whorl	Twinned Loop	Others
Right palm						
Male controls (500)	329 (65.8%)	99 (19.8%)	27 (5.4%)	13 (2.6%)	8 (1.6%)	24 (4.8%)
Male patients (29)	16 (55.2%)	9 (31.0%)	2 (6.9%)	0	2 (6.9%)	0
Female controls (500)	301 (60.2%)	110 (22.0%)	25 (5.0%)	11 (2.2%)	12 (2.4%)	41 (8.2%)
Female patients (25)	12 (48.0%)	9 (36.0%)	2 (8.0%)	0	0	2 (8.0%)
Left palm						
Male Controls (500)	351 (70.2%)	94 (18.8%)	35 (7.0%)	2 (0.4%)	2 (0.4%)	16 (3.2%)
Male patients (29)	21 (72.4%)	6 (20.7%)	2 (6.9%)	0	0	0
Female controls (500)	331 (66.2%)	103 (20.6%)	30 (6.0%)	4 (0.8%)	11 (2.2%)	21 (4.2%)
Female patients (25)	15 (60.0%)	7 (28.0%)	2 (8.0%)	0	0	1 (4.0%)

TABLE IX

MEAN RIDGE BREADTH (MRB) WITH SEXES
SEPARATE (AGE GROUP—20 YEARS OR OVER)

	Mean and Standard Deviation	P
<i>Males</i>		
Controls (259)	541.3 ± 48.2	Not significant
Patients (15)	532.7 ± 43.9	
<i>Females</i>		
Controls (381)	492.5 ± 42.8	Not significant
Patients (15)	501.7 ± 63.6	

Discussion

The palmar dermatoglyphics of 54 patients with tuberous sclerosis have been compared with 1000 controls. Out of 165 statistical comparisons there were 3 significant results, 2 at $p < 0.025$ and 1 at $p < 0.05$. Two concerned fingerprint pattern trends in female patients, but the male patients showed opposite trends. The other positive result was a small decrease in the summed $a-b$ ridge count, significant at $p < 0.05$ in the male patients. Taking $p < 0.05$ to be significant one would expect more than 3 significant results, and unless these 3 trends can be confirmed in other patients with tuberous sclerosis, it must be concluded that this disease does not appear to be associated with any specific palmar dermatoglyphic features. Indeed, it is hard to envisage how a single gene disorder such as this could be associated with any special dermatoglyphic features, and the suggestion that: 'It may be rewarding to examine the dermatoglyphics in such genetically determined conditions as tuberous sclerosis, incontinentia pigmenti, the many forms of ichthyosis, Ehlers-Danlos syndrome. . . .' (*Lancet*, 1972) seems difficult to justify.

Summary

A palmar dermatoglyphic study of 54 patients with tuberous sclerosis, inherited as an autosomal dominant condition, has failed to reveal statistically significant differences when compared with 1000 controls. It is suggested that single gene disorders do

not directly affect dermatoglyphs unless the hands or feet are prenatally affected by the disorder.

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