

Dermatoglyphics in Patients with a Syndrome of Brachydactyly, Short Stature and Hypertension*

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A hitherto undescribed, dominantly inherited syndrome of brachydactyly associated with short stature and hypertension has been reported.¹ Dermatoglyphic studies were carried out on 19 affected individuals from this inbred family living in the eastern Black Sea coast region of Turkey. Studies were also made of 13 unaffected family members as well as of 33 control subjects who were born in the same geographic area. Finger-tip pattern frequencies, total finger ridge counts, sums of the a-b ridge counts on the left and the right hands, and angle values on both hands, frequencies of thenar and hypothenar patterns of brachydactylous subjects were analyzed and compared with those of normal relatives and control individuals. Dermatoglyphics of all these 65 individuals were obtained by an inkless method using sensitized paper and a special pad developed by the Faurot Company.

Samples of typical patterns of one of the brachydactylous and normal subjects are shown in Fig. 1.

Results

Finger-tip patterns did not differ in the brachydactylous subjects when compared with the unaffected family members and the controls (Tables 1a and b).

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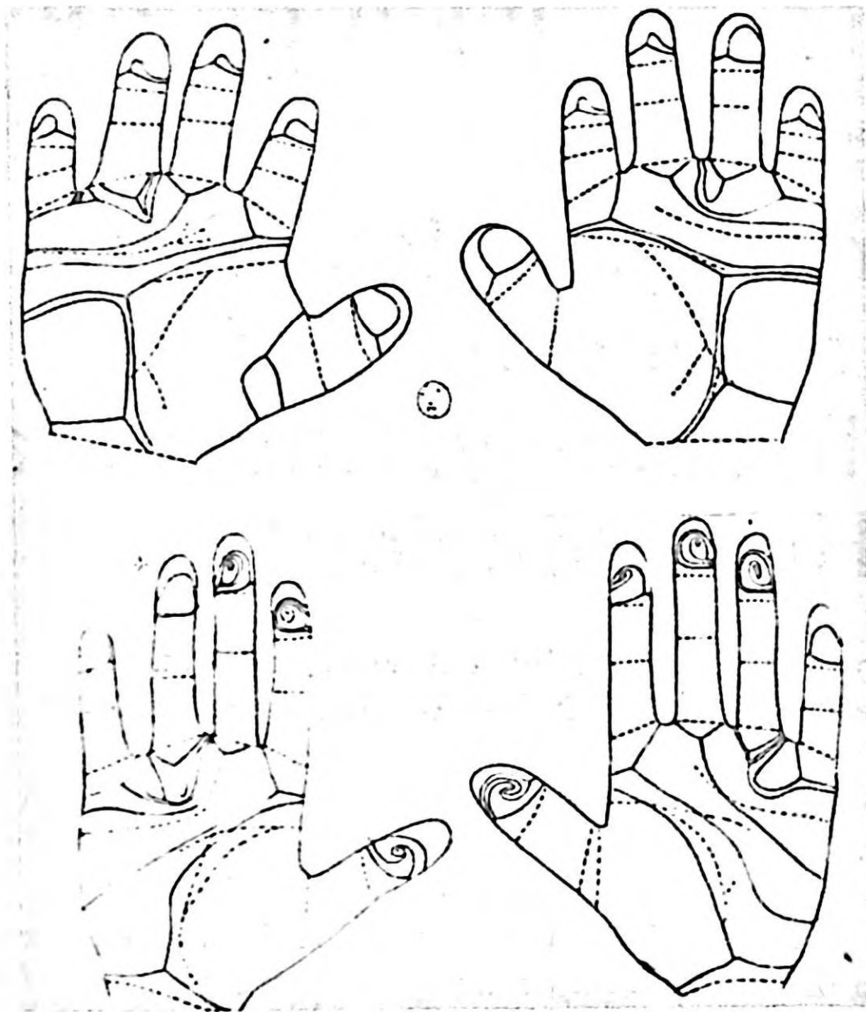


Figure 1

Drawings of main lines on prints of hands. Brachydactylous (top) and normal (bottom) subjects.

Total finger ridge count (TRC) analyses were done separately in both sexes because of a well-established quantitative difference between them. Male and female brachydactylous subjects alone did not differ in their mean values of TRC counts from the male and female controls, respectively. Unaffected male family members had a lower mean value of TRC counts of borderline significance than the brachydactylous and the control subjects, but in this respect, unaffected female family members did not show any deviation from the normal (Table 2).

The mean values of the sum of a-b ridge counts on both palms of brachydactylous subjects were compared with controls without separating the males and the females. Statistically, a significant difference was found between them ($P < 0.01$) as well as between the brachydactylous and the unaffected family members ($P < 0.01$) (Table 3a). When the mean values of the male and the female subjects were anal-

TABLE 1 a
STATISTICS CONNECTED WITH FINGER-TIP PATTERNS OF BRACHYDACTYLOUS AND CONTROL SUBJECTS

	Left Fingers										Right Fingers									
	V		IV		III		II		I		I		II		III		IV		V	
B., n = 19	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.
C., n = 33	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.	B.	C.
Whorl (W)	31.6	18.2	57.9	33.3	26.3	15.1	42.1	36.4	68.4	48.5	63.2	51.5	47.4	36.3	26.3	9.1	57.9	51.5	31.6	18.2
Ulnar Loop (U)	63.1	78.8	26.3	63.7	57.9	78.8	31.6	36.4	31.6	48.5	36.8	45.5	15.8	27.3	63.2	87.9	36.8	45.5	63.2	78.8
Radial Loop (R)	-	-	-	-	5.3	-	15.8	18.2	-	-	-	-	21.1	30.3	5.3	-	-	-	-	-
Simple Arch (A)	5.3	3.0	15.8	3.0	10.5	6.1	10.5	9.0	-	3.0	-	3.0	15.8	6.1	5.2	3.0	5.2	3.0	5.2	3.0
X ²	0.661		2.124		0.341		0.188		1.218		0.271		0.004		1.633		0.030		0.564	
p	> 0.05		> 0.05		> 0.05		> 0.05		> 0.05		> 0.05		> 0.05		> 0.05		> 0.05		> 0.05	

B. : Brachydactylous subjects

C. : Controls

TABLE 1 b

STATISTICS CONNECTED WITH FINGER-TIP PATTERNS OF BRACHYDACTYLOUS AND UNAFFECTED FAMILY SUBJECTS

	Left Fingers										Right Fingers									
	V		IV		III		II		I		I		II		III		IV		V	
Brac., n = 19	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.
U.F.M., n = 13	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.
Whorl (W)	31.6	43.8	57.9	46.1	26.3	23.1	42.1	30.8	68.4	38.5	63.2	53.9	47.4	38.5	26.3	15.4	57.9	69.2	31.6	23.1
Ulnar Loop (U)	63.2	40.8	26.3	38.5	57.9	53.8	31.6	30.8	31.6	46.1	36.8	46.1	15.8	23.1	63.2	76.9	36.9	23.1	63.2	69.2
Radial Loop (R)	-	-	-	-	5.3	7.7	15.8	23.0	-	-	-	-	21.0	15.4	5.8	-	-	-	-	-
Simple Arch (A)	5.2	15.4	15.8	15.4	10.5	15.4	10.5	15.4	-	15.4	-	-	15.8	23.0	5.2	7.7	5.2	7.7	5.2	7.7
X ²	0.019		0.084		0.089		0.090		1.704		0.020		0.021		0.089		0.086		0.018	
p	> 0.05		> 0.05		> 0.05		> 0.05		> 0.05		> 0.05		> 0.05		> 0.05		> 0.05		> 0.05	

B. : Brachydactylous subjects

U.F.M. : Unaffected family members

TABLE 2

STATISTICS CONNECTED WITH TOTAL FINGER RIDGE COUNTS AT FINGER TIPS OF BRACHYDACTYLOUS SUBJECTS, CONTROLS AND UNAFFECTED FAMILY MEMBERS

	Male		Female		Male		Female		Male		Female	
	B.	C.	B.	C.	B.	U.F.M.	B.	U.F.M.	U.F.M.	C.	U.F.M.	C.
n	9	25	9	8	9	7	9	6	7	25	6	8
$\bar{x}$	151,11	139,32	114,77	89,12	151,11	97,57	114,77	129,66	97,571	139,32	129,667	89,125
S_x	± 47	$\pm 32,9$	$\pm 59,9$	$\pm 48,1$	± 47	$\pm 43,4$	$\pm 59,9$	$\pm 65,8$	$\pm 43,400$	$\pm 32,90$	$\pm 65,800$	$\pm 48,100$
D	11,791		25,653		53,540		14,889		41,749		40,542	
S _D	$\pm 14,348$		$\pm 26,568$		$\pm 22,929$		$\pm 32,784$		$\pm 15,068$		$\pm 30,311$	
t	$\pm 0,821$		$\pm 0,965$		$\pm 2,335$		$\pm 0,454$		$\pm 2,771$		$\pm 1,337$	
P	> 0,05		> 0,05		< 0,05		> 0,05		< 0,01		> 0,05	

B. : Brachydactylous subjects,

C. : Controls,

U.F.M. : Unaffected family members

TABLE 3 a

STATISTICS CONNECTED WITH a-b RIDGE COUNTS ON THE PALMS OF BRACHYDACTYLOUS SUBJECTS, CONTROLS AND UNAFFECTED FAMILY MEMBERS (MALES AND FEMALES COMBINED)

	B.	C.	B.	U.F.M.
n	19	33	19	13
$\bar{x}$	84.842	73.454	84.842	73
$S_{\bar{x}}$	± 2.904	± 2.308	± 2.904	± 3.607
D	11.388		11.842	
S_D	± 3.759		± 4.272	
t	± 3.03		± 2.779	
P	< 0.01		< 0.01	

B. = Brachydactylous subjects,

U.F.M. = Unaffected family members,

C. = Controls.

alyzed separately however, only the a-b mean value of male brachydactylous subjects was markedly higher than those of both unaffected male family members and male controls. Females showed no difference in this respect (Table 3b).

First the mean values of the sum of maximal atd angles on both hands of both sexes of brachydactylous and control subjects were compared (Table 4a). The brachydactylous subjects showed markedly higher mean atd angles than either the controls ($P < 0.001$) or the unaffected family members ($P < 0.02$). By contrast, when the analysis was done in the males and the females separately, only the male brachydactylous subjects showed a difference (see in Table 4b).

There were no significant differences between the means of axial "t" distance of the affected subjects and the controls, or between the means of axial "t" distance of the affected subjects and the normal family members in either sex (Tables 5a and b).

Thenar pattern frequencies on the palms of brachydactylous subjects were higher than the controls, but there was no difference between the affected and the unaffected family members. Therefore, this increased frequency may merely represent a familial trait (Table 6).

TABLE 3 b

STATISTICS CONNECTED WITH a-b RIDGE COUNTS ON THE PALMS OF BRACHYDACTYLOUS SUBJECTS, CONTROLS AND UNAFFECTED FAMILY MEMBERS (MALES AND FEMALES SEPARATELY)

	Male		Female		Male		Female	
	B.	C.	B.	C.	B.	U.F.M.	B.	U.F.M.
n	9	25	10	8	9	7	10	6
$\bar{x}$	87	72,52	82,9	76,375	87	68,478	82,9	78,333
S_x^-	$\pm 12,247$	$\pm 12,124$	$\pm 13,3$	$\pm 16,941$	$\pm 12,247$	$\pm 9,219$	$\pm 13,3$	$\pm 10,049$
D	14,48		6,525		15,522		4,567	
S_D	$\pm 4,729$		$\pm 7,128$		$\pm 5,582$		$\pm 6,342$	
t	$\pm 3,061$		$\pm 0,915$		$\pm 2,781$		$\pm 0,720$	
P	< 0,01		> 0,05		< 0,05		> 0,05	

B. = Brachydactylous subjects,

C. = Controls,

U.F.M. = Unaffected family members

TABLE 4 a

STATISTICS CONNECTED WITH MAXIMAL and ANGLES ON THE PALMS OF BRACHYDACTYLOUS SUBJECTS, CONTROLS AND UNAFFECTED FAMILY MEMBERS (MALES AND FEMALES)

	B.	C.	B.	U.F.M.
n	19	25	19	13
$\bar{x}$	132.263	95.394	132.263	101.846
$S_{\bar{x}}$	± 8.451	± 5.531	± 8.451	± 5.524
D	36.869		30.417	
S_D	± 8.447		± 11.226	
t	± 4.365		± 2.709	
P	< 0.001		< 0.02	

B. = Brachydactylous subjects

U.F.M. = Unaffected family members

There was no difference between the frequencies of hypothenar patterns on the palms of the brachydactylous subjects, and those of the controls or the healthy relatives ($P > 0.05$) (see Table 7).

Discussion

Brachydactyly constitutes one of the major findings in the various clinical entities. McKusick, in his catalogue of human hereditary disorders, describes more than a dozen, apparently different conditions associated with brachydactyly.² In only two of these are the results of dermatoglyphic studies available. In the first family, obviously suffering from an entity different from that of our patients, Pensore and Holt³ found that the brachydactylous members had a high frequency of simple arch patterns on the finger tips and consequently had low total ridge counts. In our family members either affected or unaffected, total finger ridge counts were normal and there was no increase of any particular digital pattern. Likewise, the increased frequency of radial loop patterns on the thumbs and the great toes reported in the first brachydactylous family was not observed in our study. On the other hand, our male brachydactylous subjects had higher palmar a-b counts than the controls and the unaffected family members while these values were normal in the family studied by Penrose and Holt.

TABLE 4 b

STATISTICS CONNECTED WITH MAXIMAL α AND ANGLES ON THE PALMS OF BRACHYDACTYLOUS SUBJECTS, CONTROLS AND UNAFFECTED FAMILY MEMBERS (MALES AND FEMALES, SEPARATELY)

	Male		Female		Male		Female	
	B.	C.	B.	C.	B.	U.F.M.	B.	U.F.M.
n	9	25	10	8	9	7	10	6
$\bar{x}$	138.778	93	126.4	102.875	138.778	95.857	126.4	108.833
S_x	± 43.9	± 24.553	± 36.7	± 27.946	± 43.9	± 13.711	± 36.7	± 24.062
D	45.778		23.575		42.921		17.567	
S_D	± 10.673		± 15.727		± 15.294		± 16.916	
t	± 4.289		± 1.499		± 2.806		± 1.039	
P	< 0.001		> 0.05		< 0.02		> 0.05	

B. = Brachydactylous subjects,

C. = Controls

TABLE 4 c

STATISTICS CONNECTED WITH a-b RIDGE COUNTS AND MAXIMAL atd ANGLES ON THE PALMS OF CONTROLS AND OF THE NORMAL UNAFFECTED FAMILY MEMBERS IN DIFFERENT SEXES

	a-b Ridge Count				atd Angle Values			
	Controls		U.F.M.		Controls		U.F.M.	
	Male	Female	Male	Female	Male	Female	Male	Female
n=	25	8	7	6	25	8	7	6
$\bar{x}$ =	72.52	76.375	68.478	78.333	93	102.875	95.857	108.833
S_x^- =	± 12.124	± 16.941	± 9.219	± 10.049	± 24.533	± 27.946	± 13.711	± 24.062
D	3.855		9.855		9.875		12.976	
S	± 5.430		± 5.35		± 9.791		± 10.9	
t	± 0.709		± 1.840		± 1.008		± 1.190	
P	> 0.05		> 0.05		> 0.05		> 0.05	

U.F.M. = Unaffected family members

TABLE 5 a

STATISTICS CONNECTED WITH AXIAL t DISTANCE OF BRACHYDACTYLOUS AND CONTROL SUBJECTS (MALES AND FEMALES, SEPARATELY)

	Male				Female			
	Left		Right		Left		Right	
	B.	C.	B.	C.	B.	C.	B.	C.
n	9	25	9	25	10	8	10	8
$\bar{x}$	37.889	22.48	30.556	23.24	25.3	27	24.4	28.625
S	± 14.46	± 12.09	± 18.25	± 13.04	± 17.4	± 12.28	± 14.4	± 16.92
$S_{\bar{x}}$	± 4.82	± 2.418	± 6.083	± 2.608	± 6.152	± 4.341	± 5.092	± 5.981
D	15.409		7.316		1.7		4.225	
S_D	± 8.219		± 6.17		± 7.293		± 7.42	
t	± 1.874		± 1.185		± 0.233		± 0.569	
P	> 0.05		> 0.05		> 0.05		> 0.05	

B. = Brachydactylous subjects,

C. = Controls

TABLE 5 b
STATISTICS CONNECTED WITH AXIAL TRIRADIUS t DISTANCE OF THE BRACHYDACTYLOUS AND THE UNAFFECTED FAMILY MEMBERS

	Male				Female			
	Left		Right		Left		Right	
	B.	U.F.M	B.	U.F.M.	B.	U.F.M.	B.	U.F.M.
n	9	7	9	7	10	6	10	6
$\bar{x}$	37.889	23.571	30.556	30.857	25.3	27.667	24.4	28.883
S	± 14.46	± 14.67	± 18.25	± 17.54	± 17.4	± 18.5	± 14.4	± 21.49
$S_{\bar{x}}$	± 4.82	± 5.544	± 6.083	± 5.544	± 6.152	± 7.554	± 5.092	± 8.775
D	14.318		0.301		2.367		4.433	
S_D	± 7.336		± 9.002		± 9.195		± 8.961	
t	± 1.951		± 0.033		± 0.257		± 0.494	
P	> 0.05		> 0.05		> 0.05		> 0.05	

B. = Brachydactylous subjects,

U.F.M. = Unaffected family members

TABLE 6

TRUE THENAR PATTERN PERCENTAGES OF BRACHYDACTYLOUS SUBJECTS, CONTROLS AND UNAFFECTED FAMILY MEMBERS

Subject	Left	Right	Subject	Left	Right
B.	53	42	B.	53	42
C.	12	9	U.F.M.	31	23
P*	0.0043	0.0047		0.169	0.171

B. = Brachydactylous subjects,

C. = Controls

U.F.M. = Unaffected family members

P* = Exact X^2 test was applied

One finding common to both Penrose and Holt's and our study was the greater width of the atd angle. It is of interest that the affected male subjects had markedly wide atd angles while these angles were normal in affected females. Since the axial t distances were normal (Table 5), and both palmar a-b ridge counts and atd angles were high only in males in our study, the wider atd angles apparently resulted from increased a-b ridge counts which were presumably due to the wide palms of the male affected individuals rather than to central displacement of the axial triradii. However, in this respect, it should be remembered that normally palmar a-b ridge counts and atd angles show slight and insignificant differences between males and females (Table 4c), while total finger ridge count (TRC) values are markedly higher in normal males compared with normal females.⁴

In still another type of brachydactyly associated with a hip joint disorder, Robinson et al.⁵ reported the occurrence of a high frequency of digital radial loops and a high frequency of true patterns on the thenar areas. This latter finding was found to be a common feature in the families studied by Penrose and Holt and Robinson et al., and in our subjects.

It is also worth while to note that the abnormal dermatoglyphic findings such as increased palmar a-b ridge counts and wider atd angles were observed only in the male brachydactylous individuals in our family. In order to make such a statement we must be sure that there is no statistically significant difference between the sexes in controls

TABLE 7

HYPOTHENAR PATTERN PERCENTAGES AND RELATED STATISTICS OF BRACHYDACTYLOUS SUBJECTS, CONTROLS AND UNAFFECTED FAMILY MEMBERS

		A ^c	L ^u	W	W-S	L ^r	A ^r	L ^c	X ²	P
Left	B.	47.3	42.1	—	5.3	5.3	—	—	1.114	> 0.05
	C.	66.6	12.2	6.1	—	12.2	6.1	—		
Right	B.	57.9	36.8	—	—	5.3	—	—	0.014	> 0.05
	C.	63.7	12.2	3.0	3.0	12.2	—	3.0		
Left	B.	47.3	42.1	—	5.3	5.3	—	—	0.0	> 0.05
	U.F.M.	53.9	38.4	—	—	7.7	—	—		
Right	B.	57.9	36.8	—	—	5.3	—	—	0.019	> 0.05
	U.F.M.	53.9	38.4	—	—	7.7	—	—		

B. = Brachydactylous subjects, C. = Controls, U.F.M. = Unaffected family members.
 A^c = Carpal arch, L^u = Ulnar loop, W = Whorl, W-S = S-shaped whorl,
 L^r = Radial loop, A^r = Radial arch, L^c = Carpal loop.

and normal family members with regard to palmar a-b counts and atd angles. In fact, when palmar a-b counts and atd angles were compared in the male and female controls and in the male and female family members, these differences were found to be statistically insignificant ($P > 0.05$ in Table 4c). Both Penrose and Robinson noticed the same kind of diversity in different sexes. In their families only the affected male individuals had a high frequency of radial loops. Since the severity of the other clinical features of this syndrome does not differ between the sexes, it is difficult to explain this interesting finding on the basis of a difference in the expressivity of the disease between the sexes.

Summary

The results of dermatoglyphic studies in a family with hereditary brachydactyly, short stature and high blood pressure are presented. Affected male individuals showed higher a-b ridge counts and wider atd angles than either controls or unaffected family members.

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