

Dermatoglyphics Associated with the XXYY Chromosome Complement

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PATIENTS WITH AUTOSOMAL TRISOMY have characteristic dermal configurations (Uchida, Patau, and Smith, 1962). Individuals with sex chromosome aneuploidy, however, show less striking deviations from normal pattern frequencies. In patients with Turner's syndrome, the axial triradius of the palm is displaced somewhat distally (Uchida and Soltan, 1963; Penrose, 1963) and higher frequencies of simian and partial simian creases have been observed. On the fingers of XXY Klinefelter's syndrome, Penrose describes a tendency towards low ridge counts and more arches.

In the present communication, observations are presented which suggest that individuals with an XXYY sex chromosome complement have characteristic dermal patterns.

MATERIAL

Individuals with the XXYY variant of Klinefelter's syndrome have been described (Ellis *et al.*, 1961; Carr, Barr, and Plunkett, 1961; Vague *et al.*, 1961; Muldal *et al.*, 1962; Laurence, Ishmael, and Davies, 1963). In the course of a study of two patients with this chromosomal complement (Robinson *et al.*, 1964), one of us (J. R. M.) was impressed by the striking similarity between the configurations present on the palms of both patients and those reported by Ellis and his colleagues. The same configurations have since been observed in an XXYY patient recently identified in Winnipeg, Manitoba (Fig. 1).

The dermatoglyphic analyses presented below are based on a study of seven cases: the above three patients; three others from London, Ontario (Carr *et al.*, 1961; Barr *et al.*, 1963); and the published case of Ellis *et al.* (1961). Included for comparison are two cases with an XYY complement (Hauschka *et al.*, 1962; Sandberg *et al.*, 1963), one with an XXXYY complement (Bray and Sr. Ann Josephine, 1963), and 23 patients with the usual XXY Klinefelter's

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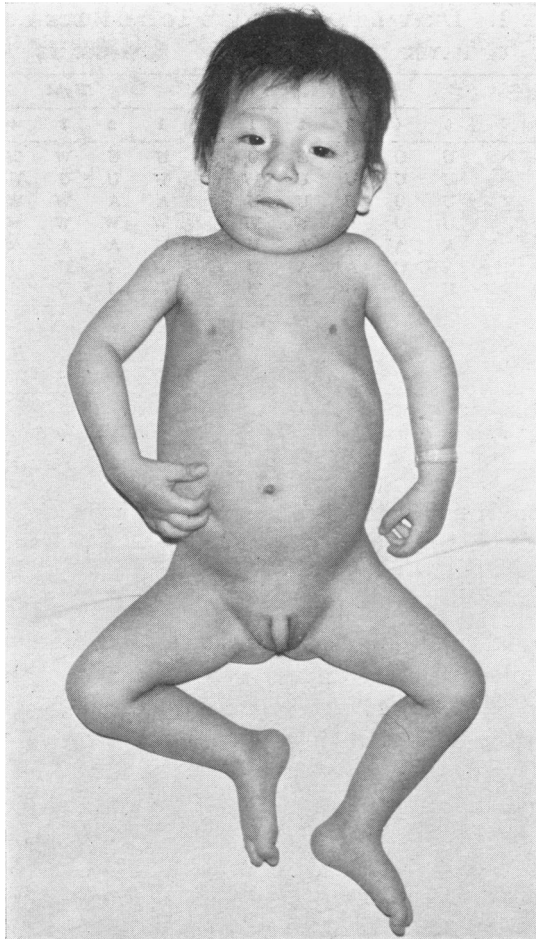


FIG. 1. Patient with XXYY sex chromosome complement, age 19 months, of Cree Indian origin.

syndrome. Patients with known or suspected mosaicism have not been included. The controls consist of 353 male and 342 female elementary school children. They are all of Caucasian origin, unrelated, and without obvious physical defects. These controls are part of a larger sample of control dermatoglyphics which are presently being analyzed.

RESULTS

Digital patterns. The patterns and total ridge counts found on the digits of the patients with two Y chromosomes are shown in Table 1. Half of these patients have at least one simple arch pattern (Fig. 2). In the control sample, only 13% of the males and 23% of the females had an arch pattern. The frequencies of arches among the aneuploids and the controls are shown in Table 2. It is obvious, without applying any statistical test, that arches are found with a significantly higher frequency among patients with two Y

TABLE 1. DIGITAL PATTERNS AND TOTAL RIDGE COUNTS OF PATIENTS WITH TWO Y CHROMOSOMES

Cases	Left					Right					Total ridge count
	5	4	3	2	1	1	2	3	4	5	
XXYY	V ₁	U	U	U	U	U	U	W	W	U	160
	V ₂	U	U	U	U	W	W	U	U	U	82
	Wg	U	U	U	U	U	A	A	W	U	69
	L ₁	U	U	W	W	W	W	W	W	W	188
	L ₂	A	A	A	A	U	U	A	A	A	11
	L ₂	A	U	A	A	U	W	A	U	U	43
	E*	U	U	U	U	U	U	U	U	U	68
XYY	B ₁	U	U	U	W	U	R	U	U	U	156
	B ₂	U	U	U	A	A	U	A	U	U	49
XXXYY	SL	U	U	A	A	W	W	A	U	U	73

A = simple arch, U = ulnar loop, R = radial loop, W = whorl.

Cases: V = Vancouver, Wg = Winnipeg, L = London, B = Buffalo, SL = Salt Lake City.

*Ellis *et al.* (1961).

chromosomes than in any other category. A significant difference also exists between XXY individuals and male controls ($\chi^2 = 9.7$, $P = 0.002$) while no difference was found between XXY and female controls.

The ridge count of a loop or whorl is determined by the number of ridges between the core and the periphery, marked by the triradius (Fig. 2). The count is therefore indicative of the size of the pattern. Arches have a count of zero. For the present analysis we have followed convention by using only the larger of the two counts in whorls. The mean total ridge counts are shown in Table 3. Mean differences significant at the 1% level or lower exist only between the double Y patients and male controls and between male and female controls.

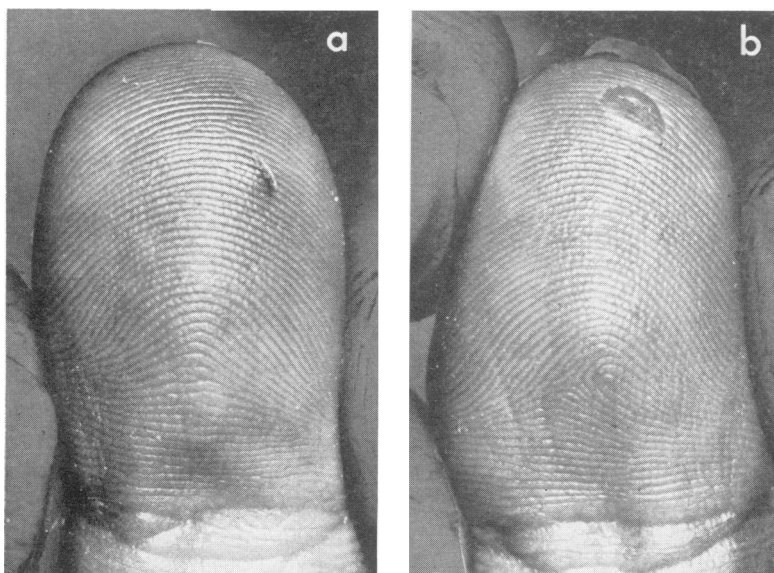


FIG. 2. Thumbs of patient shown in Fig. 1. a. Simple arch pattern. b. Small loop ulnar with ridge count of 2.

TABLE 2. FREQUENCIES OF SIMPLE ARCH PATTERNS ON THE DIGITS OF SEX ANEUPLOIDS AND CONTROLS

No. of arches per person	XXYY	XXY	XXXYY	Total -YY	XXY	Controls	
						Males	Females
0	4	1	-	5	16	306	265
1	-	-	-	-	4	21	31
2	1	-	-	1	-	12	15
3	-	-	1	1	2	8	13
4	1	1	-	2	-	2	4
5	-	-	-	-	1	2	2
6	-	-	-	-	-	1	6
7	-	-	-	-	-	-	-
8	1	-	-	1	-	-	2
9	-	-	-	-	-	1	1
10	-	-	-	-	-	-	3
Total no. of arches	14 (20%)	4	3	21 (21%)	15 (6.5%)	102 (2.9%)	204 (6.0%)

TABLE 3. MEAN TOTAL RIDGE COUNTS OF SEX ANEUPLOIDS AND CONTROLS

	XXYY	All -YY	XXY	Controls	
				Male	Female
Number	7	10	23	152	170
Mean	88.71	89.90	122.17	139.25	121.96
Standard error	23.87	18.34	11.02	3.60	3.75
XXYY vs. male controls: $P < 0.01$		YY vs. male controls: $P < 0.01$			
XXYY vs. female controls: $P \sim 0.08$		YY vs. female controls: $P \sim 0.05$			
XXYY vs. XXY: $P < 0.20$		YY vs. XXY: $P > 0.10$			
XXY vs. male controls: $P > 0.08$		Male controls vs. female controls: $P < 0.001$			

Palmar patterns. The important dermatoglyphic area of the palm of XXYY patients appears to be the hypothenar. The patterns observed and their frequencies are recorded in Table 4. The characteristic configurations present in the XXYY patients—loop carpal (Fig. 3) and loop radial and arch radial (Fig. 4)—have in common the presence of a palmar triradius on the ulnar border which we propose to call the "ulnar triradius." With both types of loops the axial and ulnar triradii are present while only the ulnar triradius is present with the arch radial. These patterns were found bilaterally in five of the seven XXYY cases, in 13% of XXY patients, 13% of male controls, and 16% of female controls. The differences between XXYY and the other categories are all highly significant (see Table 4). These characteristic patterns were not observed in the three other cases with two Y chromosomes.

Plantar patterns. The configurations found in the hallucal area of the soles of XXYY patients are variable and show no striking trend in the present sample.

DISCUSSION

Patients who are not mongoloid but possess an extra small acrocentric chromosome have been observed (Hayward and Bower, 1960; Turner and

TABLE 4. HYPOTHENAR PATTERNS OF SEX ANEUPLOIDS AND CONTROLS

*Pattern combination	XXYY	XYY	XXXYY	XXY	Controls	
					Male	Female
L ^r × A ^r	3	—	—	—	4	2
L ^r × L ^c	1	—	—	—	—	1
L ^r × L ^r	1	—	—	2	45	40
L ^c × L ^c	—	—	—	—	—	—
L ^c × L ^r	—	—	—	1	—	3
A ^r × A ^r	—	—	—	—	—	1
Subtotal (T ₁)	5	—	—	3	49	48
A ^u × A ^u	2	2	1	14	178	164
Others	—	—	—	6	126	130
Subtotal (T ₂)	2	2	1	20	304	294
Total individuals	7	2	1	23	353	342

*One pattern on each hand. L^r = loop radial, A^r = arch radial, L^c = loop carpal, A^u = arch ulnar. Fisher's test of significance between T₁ and T₂—XXYY compared with male controls: $P = 0.001$; with female controls: $P = 0.001$; with XXY: $P = 0.006$.

Jennings, 1961; Dunn *et al.*, 1961; Biesele, Schmid, and Lawlis, 1962; Fitzgerald, 1962). Two of these reported cases have been reinvestigated and they are now known to be instances of XYY. The case reported by Hayward and Bower was re-examined by Dent, Edwards, and Delhanty (1963) who were able to demonstrate that the extra chromosome was not an autosome but a Y chromosome giving a sex chromosome complement of XYY. We (J.R.M.) have reinvestigated our case, reported by Dunn *et al.*, and have come to the same conclusion.

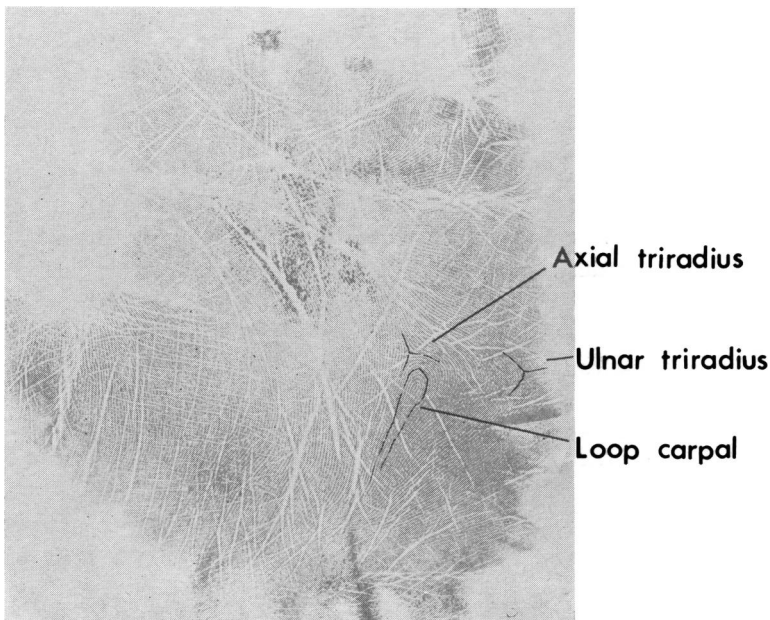


FIG. 3. Right palm of XXYY patient (Case V₁) showing loop carpal in hypthenar area.

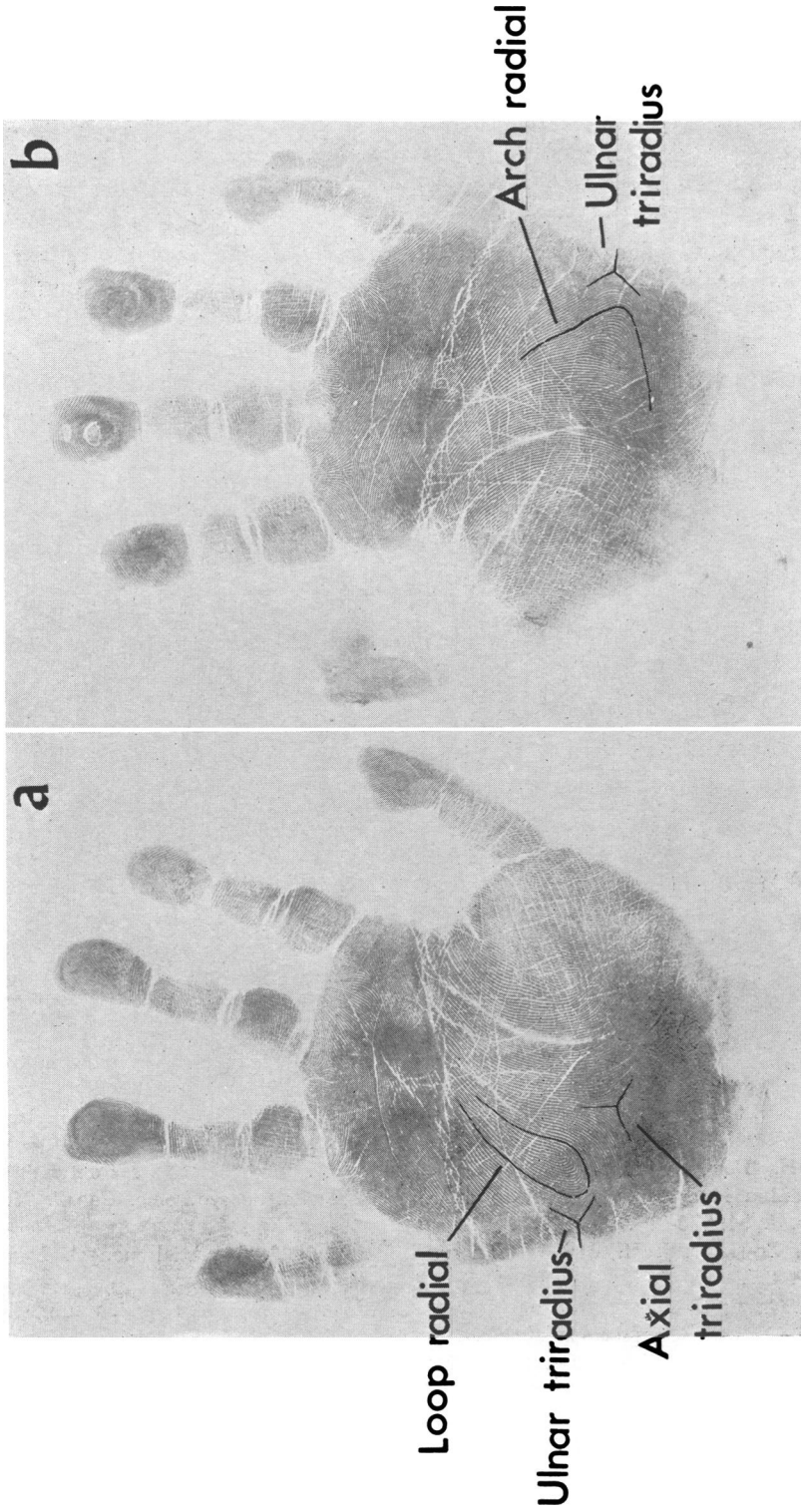


FIG. 4. Palms of patient shown in Fig. 1. a. Left palm with loop radial in hypothenar area and simian crease. b. Right palm with arch radial in hypothenar area.

A screening test for the presence of an extra Y chromosome, analogous to the sex chromatin body for the X chromosome, would be useful in cases where there might be difficulty in differentiating the Y chromosome from the other small acrocentrics. Such a test could lie in the demonstration of a dermal configuration characteristic of the presence of an extra Y chromosome.

Since the hypothenar patterns found in XXYY cases are not characteristic of the XXY complement, it was thought that they might be associated with the presence of two Y chromosomes. However, examination of the prints of the three cases with two Y chromosomes in combination with a single or triple X did not reveal patterns with an ulnar triradius. These patterns therefore appear to be associated with the specific combination XXYY and are useful as a criterion to rule out a diagnosis of the double aneuploid, Klinefelter-mongolism.

SUMMARY

Dermatoglyphic analyses of seven patients with an XXYY chromosomal complement have been made. It appears that these patients have characteristic configurations of the hypothenar area involving the presence of an ulnar triradius with a loop carpal, loop radial, or arch radial pattern. In addition, arches and small patterns are frequently found on the fingers. The hypothenar patterns appear to be specific for this chromosome complement and not for other double Y combinations or XXY.

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