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Chromosomal aberration dermatoglyphics
fingerprints palmar patterns 18q-syndrome

Dermatoglyphics in the 18q-Syndrome

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Extract

The dermatoglyphics of 6 Caucasian patients with partial deletion of the long arm of chromosome 18 (18q-syndrome) were studied together with those of their parents and normal sibs. A second control sample, consisting of 19 randomly selected Caucasian families, was used for comparison.

The patients showed significantly higher pattern intensity index, mainly higher frequency of whorls on their fingers. The direction of the main palmar lines was also significantly more transverse. The distribution of pattern intensity index, however, and the main line terminations of the parents and sibs of the patients were similar to those of the control families.

Speculation

The present study indicates the relevance of dermatoglyphics as an aid in the diagnosis of the 18q-syndrome, and reveals the usefulness of intrafamily comparisons in establishing dermatoglyphic peculiarities, especially when dealing with small samples.

Introduction

The first patient with partial deletion of the long arm of a 17–18 chromosome was described by de Grouchy *et al.* [6]. Shortly thereafter, this cytogenetic anomaly served as the basis for the recognition of a new clinical syndrome by Lejeune *et al.* [9] and Wertelecki *et al.* [15], herein referred to as the 18q-syndrome. The clinical diagnostic criteria have changed little since our first report and are summarized in Table I.

These studies suggest that the 18q-patients have an excess of whorls on their fingers. It is the objective of the present study to evaluate in detail the significance of the fingerprint patterns and other dermatoglyphic features in six patients with 18q-syndrome and to ascertain whether these features are also encountered in other family members.

Materials and Methods

All six cases were first diagnosed at the Clinical Genetics Division of Boston Children's Hospital Medical Center. Clinical and cytogenetic details are described by Wertelecki and Gerald [14]. The nature and amount of genetic material deleted are probably not the same in each of the six unrelated patients.

Palm prints and fingerprints were collected from patients and their first degree relatives by the standard ink method. Nineteen control families (with 55 normal children) were selected at random from a control sample previously studied by Niswander and Adams [11]. The patients and controls were all Caucasian.

All dermatoglyphics were evaluated for the following features: (*1*) fingerprint patterns and ridge count using single count in whorls as described by Holt [7]; (*2*) pattern intensity index, which is the sum of the coded values of the total finger patterns and serves as a measure of fingerprint pattern complexity. The values of 0, 1, and 2 were assigned to arches, loops, and whorls, respectively; (*3*) main line terminations, with special emphasis on the modal types of the D line (Fig. 1) as described by Cummins and Midlo [2, 3]. The modal types of the D line serve as indicators of the

Table 1. Main clinical diagnostic signs of the 18q-syndrome

	Cases					
	I	II	III	IV	V	VI
Mental retardation (IQ)	+ (52)	+ (45)	+ (42)	+ (85)	+	+ (46)
Short stature	+	+	+	−	+	+
Characteristic facies	+	+	+	+	+	+
Prominent antihelix and/or antitragus	+	+	+	+	+	+
Narrow or atretic external ear canal	+	+	+	−	−	+
Hearing loss (usually conductive)	+	+	+	+	−	+
Horizontal nystagmus	+	+	−	+	−	+
Generalized hypotonia	+	+	+	+	+	+
Incoordination	+	+	+	+	+	+
Club feet	−	+	−	+	+	+
Abnormal toe implantation	−	+	+	+	−	+
Proximally implanted or short thumb	+	+	+	+	+	+
Ocular anomalies (fundoscopic)	+	+	+	−	−	+
Genital anomalies	−	+	+	−	−	+
Congenital heart disease	+	+	+	−	+	−

transversality of the palm ridges. The higher the modal type of the D line the greater the transversality of the palmar ridges. The main line index was used as another measure of the transversality of the palmar ridges. It incorporates the terminations of both the A and the D lines, [2, 3]; (*4*) presence of patterns in the palmar configurational areas (hypothenar, thenar/I, II, III, and IV); (5) presence of accessory triradii; (*6*) width of the atd angle; and (*7*) presence of simian creases. In addition, the general morphology of the palms was scrutinized.

Statistical comparisons were made between the patients and their parents and sibs, as well as between the patients and control families whenever possible.

Results

The pertinent dermatoglyphic features of all members of the patients' families are shown in Table II. Significant differences were observed in fingerprint patterns and main line terminations.

Fingerprints

The patients have an excess of whorls even when they are compared with their own siblings and parents. Figure 2 presents the same data in terms of pattern intensity index and shows clearly that with exception of *family VI* the patients have the highest intensity index within their family groups. The mean pattern intensity indices were: 17.1 ± 0.64 for patients, 11.6 ± 0.36 for parents, and 10.5 ± 0.90 for sibs. The difference between patients and parents or sibs was significant ($P < 0.01$) as determined by the t test.

The average pattern intensity index of two parents (the mid-parent value) was calculated for each family in the study. The intensity index of each child was compared with that of the corresponding mid-parent (Fig. 3). If the child's index was higher than that of his mid-parent, he was recorded on the left side of Figure 3, if lower on the right. The differences between mid-parent and offspring were coded for convenience so that 0 signifies a difference of less than 1, 1 = 1 to 2.9, 2 = 3 to 4.9, and so on. The distributions of the mid-parent *vs.* offspring differences in the patients', siblings, and controls (Fig. 3, *b* and *c*) are very similar to each other and are suggestive of a normal curve. All of the patients' indices, however, are clearly higher than those of their mid-parent (Fig. 3*a*). The similarity of Figures 3 *b* and 3 *c* suggests that the higher pattern intensity score of the patients is due to their inherent differences which are not shared by their normal sibs.

Total ridge counts of the patients and their relatives are shown in Table II. The patients have a significantly higher ($P < 0.05$) ridge count than their par-

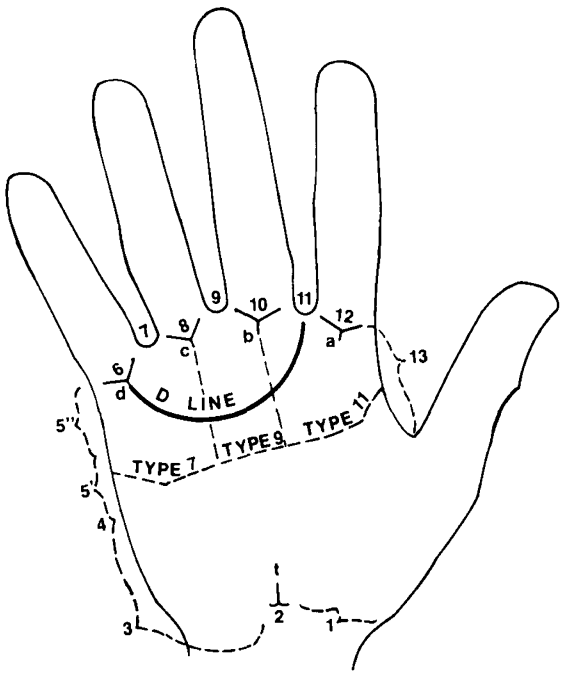

Fig. 1. The areas of the modal types of the D line of the palm. *Modal type 7* consists of terminations in *areas* 5, 7, and 8; *type 9* for termination in *9* and *10*, and *type 11* for terminations in *areas 11, 12*, and *13*.

Table II. Principal dermatoglyphic observations on six 18q-patients and their relatives.

	Fingerprint patterns[1]										Line D termination		Hypothenar patterns[2]		Simian crease		Atd angle, degree		Total ridge count
	Left					Right													
	I	II	III	IV	V	I	II	III	IV	V	Left	Right	Left	Right	Left	Right	Left	Right	
Family I																			
Propositus female	U	W	R	W	U	U	W	R	W	W	10	11	L^r	L^r	+	−	57	51	129
Mother	U	W	U	U	U	W	R	U	W	U	7	9	O	O	−	−	0	0	85
Father	U	W	U	U	U	U	U	U	U	U	9	11	O	O	+	−	38	41	64
Male sib	U	A	U	U	U	W	A	A	U	U	10	9	O	O	−	−	41	42	76
Female sib	A	A	A	U	U	A	A	A	A	U	7	9	L^r	L^r	−	−	42	44	16
Male sib	U	A	U	U	U	U	A	U	U	U	9	10	O	O	−	+	46	44	86
Female sib	U	A	U	U	U	U	R	U	W	U	9 7	9/7	L^r	L^r	+	−	51	48	77
Family II																			
Propositus female	W	W	W	W	W	W	W	W	W	U	11	11	O	L^u	−	−	43	46	117
Mother	A	A	U	U	U	U	U	U	U	U	10	9	L^r	L^r	−	+	37	39	84
Father	U	R	U	U	U	W	R	U	W	U	9	11	L^r	O	−	−	44	40	149
Female sib	U	R	U	U	U	U	R	R	W	U	10	10	L^r/L^u	L^u	+	−	50/58	45 51	117
Male sib	U	A	U	U	U	U	R	U	U	U	9	10	L^r	O	+	−	40	41	97
Female sib	U	W	U	W	U	U	W	U	W	U	9	10	L^u	L^u	−	−	45/55	42/52	129
Male sib	W	W	U	U	U	W	R	W	W	U	9	9	L^r	L^r	−	−	39	41	201
Family III																			
Propositus male	W	W	W	U	U	W	W	W	W	U	11	11	O	O	−	−	36	33	165
Mother	U	U	U	U	U	U	U	U	W	U	9	11	L^r	W	−	−	40	38/48	103
Father	U	U	W	W	U	U	W	W	W	U	9	10	O	O	−	−	37	34	130
Male sib	U	U	W	W	U	U	U	W	W	U	11	10	O	O	−	−	47	42	—
Female sib	A	U	U	U	U	U	A	U	W	U	11	11	L^r/L^u	L^r	−	−	35 41	33	93
Female sib	U	U	U	U	U	U	U	U	U	U	9	11	O	O	−	−	37	39	131
Male sib	U	U	U	U	U	U	W	W	W	U	11-5	11-11	O	V	−	+	41	33/40	126
Female sib	U	W	U	U	U	U	U	U	U	U	10	11	L^r	L^r	−	−	36	44	148
Family IV																			
Propositus male	U	W	U	W	W	U	U	U	W	W	10	11	L^u	L^u	−	−	39/77	40 77	135
Mother	U	R	U	U	U	U	A	U	U	U	9	11	O	O	−	−	37	38	67
Father	U	W	U	U	U	U	R	U	U	U	8	9	L^u	W	−	−	37 53	38/63	158
Family V																			
Propositus male	U	W	W	W	W	U	W	W	W	W	11	11	O	O	−	+	43	45	143
Mother	U	U	U	U	U	U	W	A	U	U	11-7	11-11	O	O	−	−	43	41	132
Father	U	R	R	U	U	U	R	U	U	U	9	9	L^u	V	−	−	36/49	36	125
Family VI																			
Propositus male	W	U	W	W	W	W	U	U	W	W	11	11	O	O	−	−	37	38	152
Mother	W	W	W	W	U	W	U	W	W	W	10/7	10-10	O	O	−	−	39	40	127
Father	U	U	U	U	U	U	U	U	U	U	11	10	O	O	−	−	42	44	71

[1] U = ulnar loop; R = radial loop; W = whorl; A = arch.
[2] O = open field; L^r = radial loop; L^u = ulnar loop; W = whorl; V = vestigial pattern.

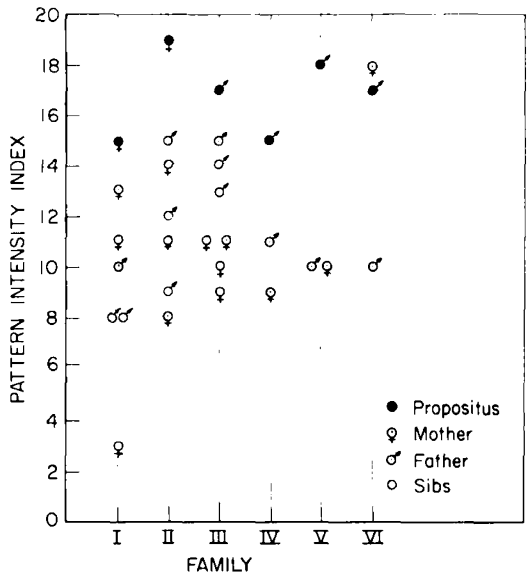

Fig. 2. The pattern intensity indices of patients with the syndrome 18q- and their first degree relatives.

ents' or mid-parent value. On the average the patients also have a higher ridge count than their sibs; however, this difference is not statistically significant.

Main Line Terminations

The percentage distribution of the modal types of line D is shown in Figure 4, from which the following conclusions could be drawn: (*1*) the modal types of the D lines of the patients are much higher than those of their parents (Fig. 4*a*); (*2*) the distribution of modal types of the normal sibs of the patients and their parents are similar to each other (Fig. 4*b*), and both differ from that of the patients; (*3*) the modal types of the control parents and their children are also similar to each other (Fig. 4*c*); and (*4*) there is striking similarity in the correlation observed for the parents and their normal offspring of both the patient and the control families (Figs. 4, *b* and *c*).

The modal types of the patients, their parents, and their sibs were compared with each other by contingency chi-square tests. Significant differences ($P < 0.01$) were found in comparisons of patients with parents or sibs. Comparisons of parents and normal sibs were not significantly different.

The mean of the main line indices for the patients was 10.33 ± 0.60, for the parents, 7.83 ± 0.66, and for the patients' sibs, 8.50 ± 0.38. Again, significant differences were found when the patients were compared with their normal sibs ($P < 0.05$) or parents ($P < 0.01$).

There were no significant differences between patients and their parents or sibs in the following comparisons: patterns in the palmar configurational areas, number of accessory interdigital or axial triradii, width of the atd angles, and incidence of simian crease.

At first impression the palms strike the observer as being unduly long. The thumbs are proximally implanted and frequently appear short. The first metacarpal may also be short. The thumb is often rotated radially along its longitudinal axis, so that when at rest, its palmar surface faces almost the same plane as that of the other fingers. The end of the distal flexion crease frequently tends to point toward the radial palm border or the proximal flexion crease rather than the index area.

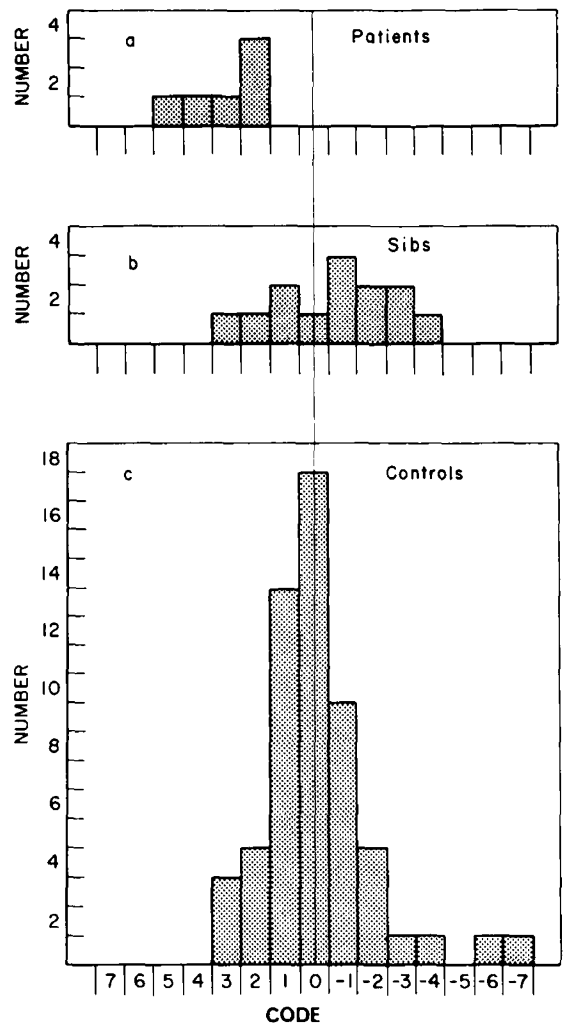

Fig. 3. Deviations of the pattern intensity indices of patients with 18q-syndrome, their normal sibs, and normal controls from that of their respective mid-parent intensity index. (See text for code explanation.)

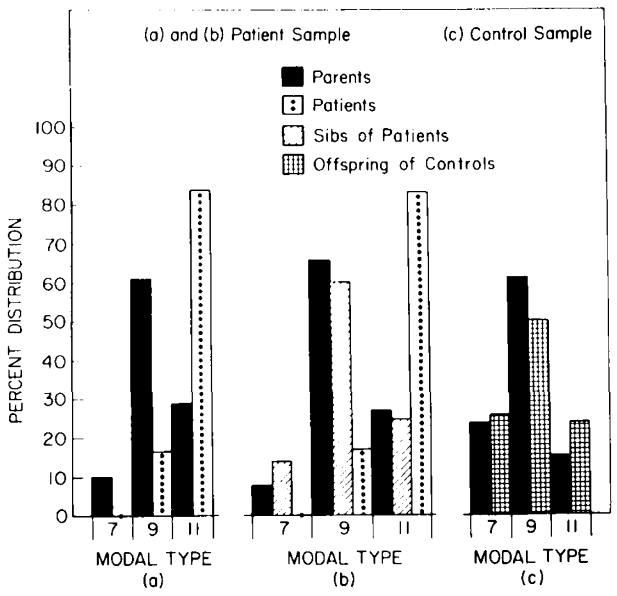

Fig. 1. The distribution of the modal type of the D line of the patients with 18q-syndrome and their parents (*a*); the 18q-, their parents, and their sibs (*b*); and the control subjects and their parents (*c*).

Discussion

The 18q-syndrome, although more recently identified, could be as frequent as the better known "*cri du chat*" disorder (5p-). The typical clinical signs frequently suggest the diagnosis prior to the confirming chromosome analysis. The dermatoglyphic alterations are also part of the overall dysmorphic complex of this syndrome. The present study confirms prior suggestions that the high frequency of whorls may be used as one of the diagnostic signs of this syndrome. The transversality of the D line may serve as another characteristic of diagnostic value, particularly since it is not a result of, or associated with, higher axial triradii or the presence of hypothenar patterns. This feature has not been stressed before, although it is seen in the palm prints shown in several previous reports [4, 9, 10, 12, 13].

We noted in published photographs that high pattern intensity index and high modal type (number) of the D line is also present in patients with ring chromosome 18 (18r) [1, 5, 6, 8]. This similarity is not surprising in view of the belief that the formation of a ring chromosome involves the loss of chromosomal material from both the long and short arms. For the stated reasons, a search for dermatoglyphic features found in 18q- could be applied to the 18r anomaly.

Most dermatoglyphic characteristics are believed to be inherited through polygenic factors. In our study this is supported by the very close similarity between mid-parent values and their normal offspring.

The pattern intensity index and the transversality of palmar ridges of the patients are distinctive not only in terms of absolute values but also in the lack of correlation to the values of the normal family members. Therefore, the use of such dermatoglyphic features for diagnostic purposes is enhanced when entire families are studied. Occasionally patients having the 18q-syndrome with low pattern intensity index will be found but still will be distinctive from the remaining family members.

Summary

Studies of dermatoglyphic patterns were carried out on 6 patients with 18q-syndrome, their parents, and normal sibs; 19 normal families were studied as controls.

Significant differences between the patients and normal subjects were observed in: (*1*) fingerprint pattern intensity index (excess of whorls); and (*2*) transversality of the palm ridges as shown by the modal type of the D line and the main line index.

No differences were found in the incidence of simian creases, width of the atd angle, number of accessory triradii, and palmar interdigital patterns.

The comparisons used in this study acquire greater significance when the dermatoglyphic features of the patients are considered not in terms of absolute values, but compared with those of first degree relatives.

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