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Dermatoglyphics in Saethre-Chotzen syndrome: a family study

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The dermatoglyphic findings in a Cuban family with the Saethre-Chotzen syndrome are reported. The family consisted of the parents who were first cousins and their three children. A new classification of zygodactylous patterns was used. Characteristic dermatoglyphic patterns which appeared in these cases were representative of the syndrome.

Dermatoglyphics also helped to discover minor expressions of syndactyly and showed that all the members of the family had zygodactylous patterns on palms and soles.

Craniosynostosis as part of genetically determined syndromes is often associated with limb deformities, especially with syndactylism. Perez Comas [8] differentiated the following varieties of acrocephalosyndactyly: Apert syndrome, Pfeiffer syndrome, Summitt syndrome, Saethre—Chotzen syndrome and two types with polysyndactyly: Carpenter syndrome and Sakati—Nyhan—Tisdale syndrome. Dermatoglyphics have a diagnostic value in such syndromes; when there is a malformation of the hand or foot, dermatoglyphics are abnormal even when the morphologic defect is not easily perceived.

In this paper, distinctive dermatoglyphic features found in a family of five members with the Saethre—Chotzen syndrome are reported. This entity was first described by Saethre [10] in a mother and her two daughters, and a year later by Chotzen [4] in a father and his two sons. The principal abnormalities of the entity are craniosynostosis, asymmetry of skull and facies, hypertelorism, maxillary hypoplasia, deviation of nasal septum, shallow orbits, ear deformities, clinodactyly, brachydactyly, single palmar crease and soft tissue syndactyly usually of second-third fingers and/or third-fourth toes. The diagnostic usefulness of a dermatoglyphic study in these disorders will be discussed.

MATERIALS AND METHODS

Dermatoglyphics of the hands and soles were taken by an ink method of five members of a Cuban family with the Saethre–Chotzen syndrome. The family consisted of the propositus, a two-year-old boy who had cranial deformity, his eight-year-old sister, his six-year-old brother, his 29-year-old father and the 30-year-old mother. The

parents were first cousins. In all the cases, a clinical and radiological study was made. The more important clinical findings were, craniosynostosis in the propositus and his sister (Cases 1 and 2), palpable cranial sutures in these children and the brother (Case 3); hypertelorism and malformed ears in all. Ear crus extended from the root of the helix across the concha in the children, but not in the parents. All except the father (Case 4) had a high forehead, deviation of the nasal septum and the mother (Case 5), an aquiline nose. Maxillary hypoplasia was seen in all except the father, prognathism in Cases 2 and 5, facial asymmetry in Cases 1, 2 and 5, shallow orbits in the mother and the children. The propositus and his brother had clinodactyly of the fifth finger on both sides. The sister had a short but not curved fifth finger on both hands with a single digital flexion crease. Brachydactyly of the fourth and fifth toes was present in Cases 2, 3 and 4. The toes were close together and a hallux valgus was seen in all the cases. The propositus and his sister had an equivalent of simian crease on one hand and the former had a Sydney line on the left hand and also cryptorchidism. Intelligence was normal in all.

Syndactyly was classified as slight when there was only an excess of membrane between the fingers or toes, mild when the web reached the 1st interphalangeal joint, moderate when it attained the 2nd interphalangeal joint, severe when it involved the tips of fingers or toes and total if the syndactyly was cutaneous and osseous. Based on this classification, the propositus had a moderate syndactyly of the right 2–3 fingers and a mild bilateral syndactyly of the 2–3 toes. The sister had a moderate bilateral syndactyly of fingers 4–5 and severe syndactyly of the right 2–3 toes. The brother had severe syndactyly of the right toes 2–3 and a moderate one of the same toes on the left side. He and the father had no syndactyly of the fingers, but a moderate bilateral one of the toes 2–3. The mother had no syndactyly.

For the classification of dermatoglyphic patterns on hands and feet, determination of the finger pattern intensity index, the main-line index, the terminations of the main-lines on palms and the modal types of main-line D, the method of Cummins and Midlo [5] was used. The modal types of main-line A were analysed following the criteria of these authors with a variant: apart from the modal types of main-line A 1, 3 and 5, a modal type 11 was included to describe the exit of this line in the 11, 12 and 13 positions. For the modal types of main-line C and for the position of the axial triradius, the methods of Plato [9] and Walker [11] were consulted.

Our classification [3] of syndactylous patterns, based on the study of dermatoglyphics of 42 patients with different varieties and grades of finger fusion was used. Nine distinctive pattern types had been established, but in this family only some of them were seen and only these will be commented. Type 1 is an interdigital triradius [5, 7] replacing the two normal adjacent subdigital triradii which had disappeared. Type 2 is a more proximally situated interdigital triradius which involves two or more digits and with the proximal radiant adopting a course similar to a main-line. Type 3 is only seen in severe and total forms of syndactyly. In the midpoint, where the tips of fingers and toes are fusioned, a triradius of the two closely joined finger or toe patterns (a loop and a loop, a whorl and a loop or two whorls) is missing, and only one remains with its proximal triradius extending proximally in a slightly diagonal line. Type 4 is characterized by the absence of triradii and only a band of transversal ridges cross the united digits and the web. Type 5 is a combination of type 1 and type 2; type 6 is the association of type 2 and 3 and type 7, the combination of type 2 and 4. Slight syndactyly is associated with types 1 and 5, mild syndactyly with types 1, 2 and 5; moderate forms are observed with types 1, 2, 4, 5 and 7, severe and total syndactyly

TABLE I

Dermatoglyphics of the hands of the five patients

Case No.	Digital patterns 1	2	3	4	5	PII	FRC	TFRC	Atd angle	position of t	Th/I	Hy	12	13	14
1 L	U	U	U	U	U	5	38	71	—	11% t	O	A^u	O	O	cd
R	U	U	U	U	R	5	33		—	12% t	O	A^u	ab a′	O	cd
2 L	W	R	U	U	R	6	56	100	—	19% t′	O	A^u/A^c	O	L	cd
R	W	U	U	U	R	6	44		—	20% t′ 29% t′ 38% t′	O	W/A^c	O	L	c′ cd c′
3 L	W	U	W	W	W	9	87	165	38°	16% t′	O	L^r/A^c	O	bc	O
R	W	W	U	W	W	9	78		34°	10% t	O	A^u	O	O	O
4 L	W	A	U	U	U	5	70	135	34° 63°	11% t 49% t″	O	W	O	O	O
R	U	U	U	U	U	5	65		40° 64°	13% t 47% t″	O	L^u	O	O	L
5 L	W	R	A	U	W	6	32	57	34°	19% t′	O	A^u/A^c	O	O	L
R	W	A	U	U	W	6	25		35°	16% t′	O	A^u/A^c	O	L	O

PII, pattern intensity index; FRC, finger ridge count; TFRC, total finger ridge count; Th/I, thenar area; HY, hypothenar area; 12, 13, 14 interdigital spaces Nos II, III, IV; U, ulnar loop; R, radial loop; A, arch; W, whorl; O, open field; A^u, ulnar arch; A^c, carpal arch; L^r, radial loop; L^u, ulnar loop; L, loop; ab, abc, cd, interdigital triradii; a, c, interdigital triradii situated more proximally; t, axial triradius

with types 2, 5, 6 and 3, 5, 6. The types 8 and 9 of our classification [3] are present in very special varieties of syndactyly and will not be discussed here.

The configurations of the toes were examined with the magnifying lens because it is difficult to evaluate them on prints.

Results

Distribution of patterns on all the fingers showed a preponderance of ulnar loops (54%) and whorls (30%) with a low proportion of arches (6%) and a higher number of radial loops (10%) than in our control series [2] (Table I). An important fact was that the radial loops were located in the 5th finger in two cases. This pattern is never observed in this finger in the normal population. The pattern intensity index was within normal limits. The finger ridge count for each hand and the total finger ridge count were low in the propositus and his mother as compared to normal values and it was due to the presence of small loops and whorls. Cases 2 and 4 had lower counts than the mean value of normal controls [2] but within the normal range. They also had small patterns and all the radial loops had low counts.

On the palms high values for the atd angle were found in the father. In Cases 1 and 2 it was not possible to measure it, because they had no sub-digital triradius d. There was a high distal position t' or t'' of the axial triradius in all the cases, except in the propositus. The hypothenar area displayed a seam whorl on the right palm of Case 2 and Case 4 had an ulnar loop on the right and a whorl on the left side. These figures have low frequencies in the general population. True patterns were not seen in the thenar/I and II interdigital areas in all the patients. An open loop at the third interdigital space was present on both sides of Case 2 and on the right palm of Case 5. An open loop was at the fourth interdigital space of the right palm of Case 4 and of the left palm of Case 5.

On the palms the a—b ridge count gave low values in all the cases except in the father (Table II). The b—c ridge count was low in two patients (Cases 2 and 4) and normal in the mother (Case 5). The b—c ridge count for normal controls is on the left hand 27.76 ± 5.92 and 28.60 ± 5.80; and on the right hand 28.51 ± 5.59 and 29.09 ± 5.97. The c—d ridge count was low in the mother.

The modal type 5 of main line A was seen in three patients. The modal type of main line C was absent in the right palm of Case 3 and proximal in the left palm of the father. The main line index showed transversality of the ridges in Cases 3 and 4. The propositus had in one hand an equivalent of simian crease (transitional type) and in the other hand a Sydney line. His sister had also an equivalent of simian crease in the left palm.

As shown in Table III, on the toes the fibular loop (66%) predominated; whorls (14%) were located principally in the first toe (8%). Arches (18%) were found in the fifth toe and

TABLE II

Dermatoglyphics on the hands of the five patients

Case No.		a–b ridge count	b–c ridge count	c–d ridge count	Modal types of the main lines			MLI	Commins formula	Simian crease	Sydney line
					A	C	D				
1	L	20	—	—	5	—	—	—	Oid0.5″5′	—	×
—	R	—	—	—	—	—	—	—	Oid0.5″	trans.	—
2	L	35	18	—	5	—	—	—	Oid0.5″.5″.	trans	—
—	R	28	20	—	5	—	—	—	Oid0.5″.5″.	—	—
3	L	—	—	—	11	—	11	13	11.Oid0.12.	—	—
—	R	37	—	—	3	absent	11	9	11.0.7.3.	—	—
4	L	49	21	34	5	proximal	11	11	11.X.7.5′.	—	—
—	R	47	20	37	5	ulnar	9	10	10.7.6.5′.	—	—
5	L	32	35	16	3	ulnar	7	6	8.7.3.3.	—	—
—	R	26	36	22	3	radial	9	7	9.9.5′.3.	—	—

MLI, main line index; Simian crease trans, simian crease transitional

TABLE III

Dermatoglyphics on the soles of the five patients

Case No.		Patterns on toes					Patterns on soles				Zygodactylous triradii	
		1	2	3	4	5	Th/I hallucal	I_2	I_3	I_4	I_2	I_4
1	L	L^f	L^f	L^f	L^f	A	T	O	O	O	a′	
	R	L^f	L^f	L^f	T	A	L^t	O	O	O	a′	
2	L	W	L^f	L^f	L^f	A	L^d	O	O	O	—	d′
	R	W	L^t	L^f	L^f	A	L^d	O	O	O	ab a′	d′
3	L	W	W	W	L^f	L^f	L^d	O	L^p	O	a′	d′
	R	W	W	L^f	L^f	L^f	L^d	O	L^p	O	ab a′	d′
4	L	L^f	L^f	L^f	L^f	A	L^d	O	V^p	O	ab a′	d′
	R	L^f	L^f	L^f	L^f	A	W	O	L^p	O	ab a′	d′
5	L	L^f	L^f	L^f	L^f	A	L^d	O	O	O	—	—
	R	L^f	L^f	L^f	L^f	A	L^d	O	O	O	—	—

L^f, Fibular loop; L^p, proximal loop, L^d, distal loop, L^t, tibial loop; T, tented arch; A, arch; W, whorl; O, open field; V^p, proximal curved vestige; ab, interdigital triradius; d, interdigital triradius; a, interdigital triradius situated more proximally

a tented arch was on the fourth right toe of the propositus. A tibial loop was observed on the second right toe of the sister. On the hallucal/I area, distal loops were present in 4 cases, in this area the propositus had a tented arch on the left and a tibial loop on the right. In this region the father had a distal loop on the left and a whorl on the right hand. There were few figures in the interdigital areas. Case 3 had a proximal loop in the third interdigital space, the father in the same area besides the same pattern on the right and on the left side a proximal vestigial loop. This paucity of the interdigital patterns was due to the presence of interdigital triradii

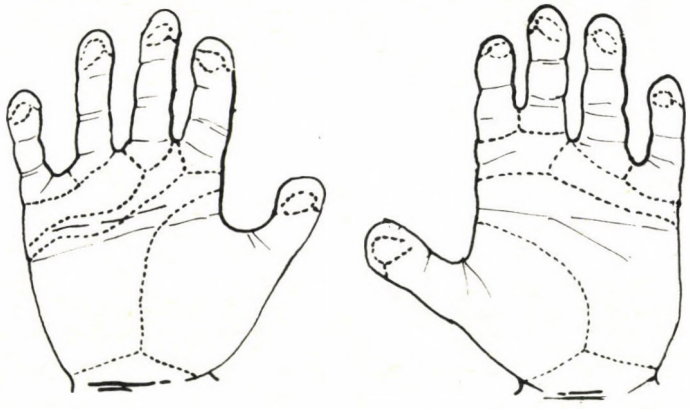

FIG. 1. Diagram of dermatoglyphics in Case 1, the propositus. Note the zygodactylous pattern types of our classification, bilateral pattern type 1 in the fourth interdigital space and pattern type 5 in the right second interdigital area

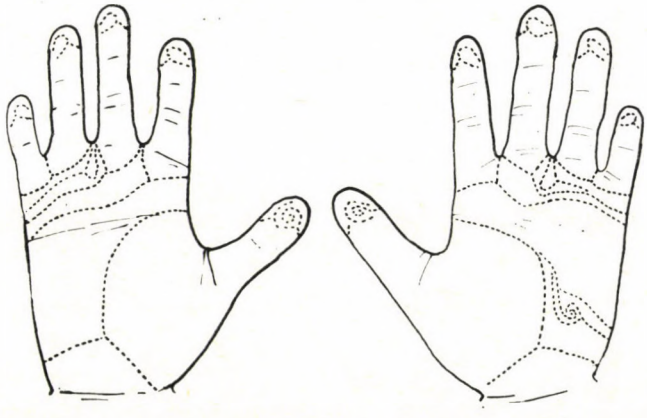

FIG. 2. Dermatoglyphics of the sister (Case 2). Zygodactylous pattern type 5 in the fourth interdigital space of both palms, with a whorl in the right hypothenar area

in 4/5 cases, seventeen in number. No triradius p was found in the family.

The propositus had on both hands a zygodactylous pattern type 1 in the fourth interdigital space, and pattern type 5 in the second right interdigital space (Fig 1). In both palms of his sister pattern type 5 was observed in the fourth interdigital area (Fig 2). The brother had a type 1 pattern in the left third interdigital space (Fig. 3) Figures 4 and 5 showed that the father and mother had no syndactylic triradii in the palms.

On the soles of both feet the propositus had a type 2 pattern in the second interdigital space (Fig 6). The sister showed a type 2 pattern in the fourth interdigital space of both

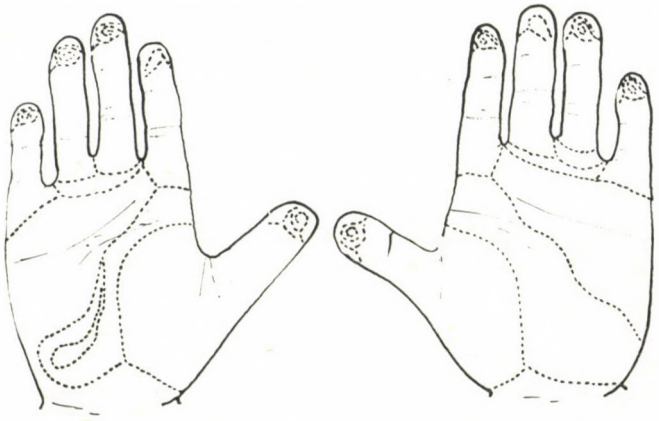

FIG. 3. Dermatoglyphics of the brother (Case 3). Syndactylous pattern type 1 in the third left interdigital space. Modal type absent of main line C on the right

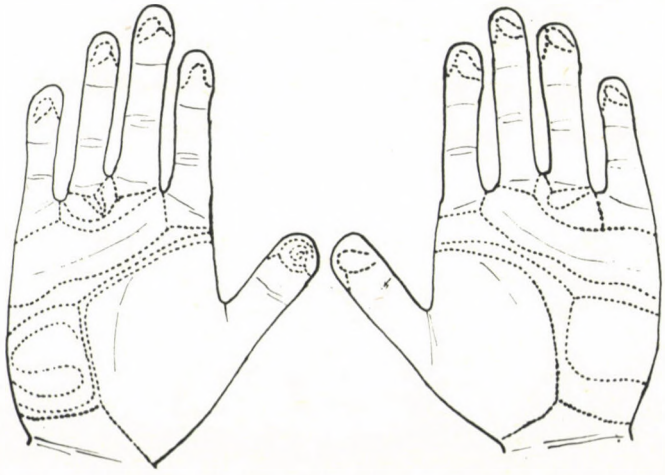

FIG. 4. Dermatoglyphics of the father (Case 4). High distal position of the axial triradius, hypothenar figures on both palms, modal type proximal of main line C on the left hand

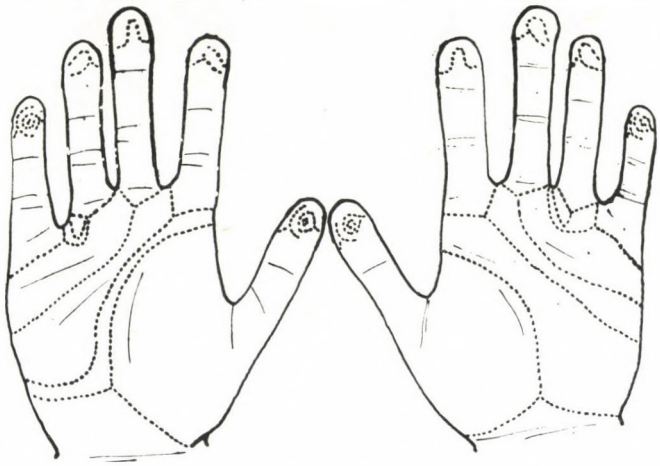

FIG. 5. Dermatoglyphics of the mother (Case 5). No zygodactylous patterns are present, but there is an a–b ridge count and also a c–d ridge count with low values

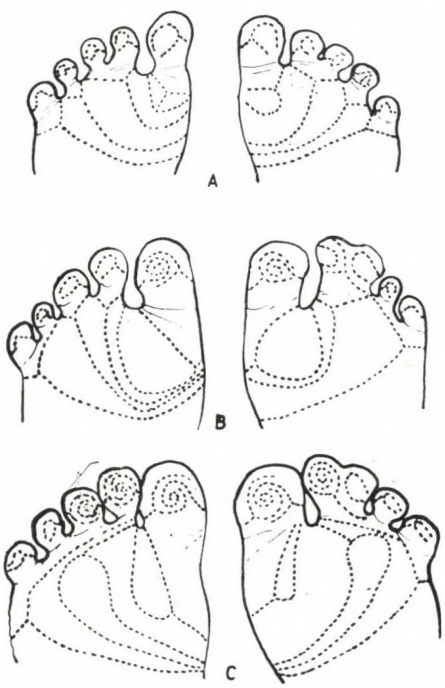

FIG. 6. Dermatoglyphics of the feet of the three children. *a* The propositus shows a bilateral zygodactylous pattern type 2 in the 2nd interdigital space. *b* The sister (Case 2) has syndactylous pattern types 2 in both fourth interdigital spaces and pattern type 6 in the right second interdigital area. *c* The brother (Case 3) has on his left sole a type 2 pattern in the 2nd interdigital space and type 2 pattern in the 4th interdigital space. On the right sole, pattern type 6 in the 2nd interdigital space and pattern type 2 in the 4th interdigital area

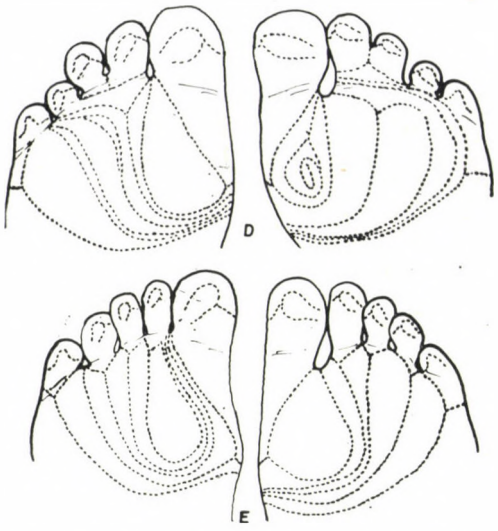

FIG. 7. *d* Dermatoglyphics of the father (Case 4). Syndactylous pattern type 5 in both second interdigital spaces. *e* Dermatoglyphics of the mother (Case 5), all the sub-digital triradii are present on the soles

soles and a type 6 pattern in the right second interdigital space (Fig 6). Type 2 patterns were present in the second and fourth interdigital spaces on the left sole of the brother (Fig 6) and he had on the right side a type 6 pattern in the second interdigital space and a pattern type 2 in the fourth.

The father had on both soles a type 5 pattern in the second interdigital space and a type 2 pattern in the fourth interdigital areas (Fig 7).

No zygodactylous configurations were seen on the soles of the mother (Fig 7).

DISCUSSION

Characteristic dermatoglyphic abnormalities were observed in this family with Saethre—Chotzen syndrome, *viz.*

1. Low finger ridge count due to reduction of finger pattern size
2. High frequency of radial loops on fingers and of fibular loops on toes, with a radial loop located on the fifth finger
3. Distal displacement of the axial triradius in either, the t' or the t'' position
4. Figures in the hypothenar area
5. Simian crease or equivalent and Sydney line
6. Transversality of the distal ridges of the palm
7. Absence of the p triradius on the soles
8. Typical zygodactylous patterns on the palm and sole.

Our classification of syndactylous patterns helps to determine and appreciate the syndactyly and the grades of severity of this defect. In this family, syndactyly was more

marked in the feet than in the hands, but the peculiar dermatoglyphic abnormalities permit the diagnosis of low grades of the malformation.

Clinically, there was only syndactyly of the right fingers 2–3 of the propositus and of the fingers 4–5 on both hands of his sister. Dermatoglyphic pattern type 1, in the fourth interdigital space in both hands of Case 1 and in the third interdigital space in the left hand of the brother allowed to diagnose syndactyly between fingers 4–5 and fingers 3–4.

The low values of the a–b ridge count in Cases 1, 2, 3 and 5 and the low b–c count in Cases 2 and 4 and the low values of the c–d ridge count in Case 5 could be interpreted as a minor expression of syndactyly [5]. It then became apparent that the whole family had syndactyly on the hands and that practically all the fingers were involved: fingers 2–3 in the propositus, his sister, brother and mother; fingers 3–4 of the father and the brother of the propositus; fingers 4–5 of the propositus, his sister and mother. It was the same on the feet where there was obvious syndactyly of toes 2–3 of the sister and the brother of the patient. The father and the propositus had only mild syndactyly of the same toes. Dermatoglyphic patterns of the soles revealed syndactyly of toes 4–5 in the father, the sister and the brother of the propositus. These same cases had also brachydactyly of toes 4–5, and it would be better to classify the defect as brachysyndactyly of the 4th and 5th toes. Only toes 3–4 were unaffected and the mother had no zygodactylous patterns on the feet.

The dermatoglyphic findings in these patients were in agreement with those reported in the literature [8]: low finger ridge count, high number of arches, high number of hypothenar patterns and simian crease. Our patients had few arches on the fingers and toes but they had small loops and whorls, and these small patterns should be interpreted as transitional forms which tend to be arches. It has to be stressed that the zygodaytylous pattern is indispensable in the diagnostics of the Saethre—Chotzen syndrome.

Aue—Hauser [1] published a classification of plantar zygodactylous triradii in a normal population of 500 males. Zygodactylous triradii were classified as strong, medium and weak expressions. In her classification, only our types 1 and 2 were considered but these forms were seen in a normal population whereas the patterns described by us were observed in abnormal cases.

The great variability of expression of the dominant gene in this family where the parents were first cousins and affected like their children, has been demonstrated by the great number and different varieties of the zygodactylous configurations.

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