Camptodactyly, with muscular hypoplasia, skeletal dysplasia, and abnormal palmar creases: Tel Hashomer camptodactyly syndrome*

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Summary. A syndrome characterized by camptodactyly, distinct facial features, multiple musculoskeletal defects, and unique dermatoglyphic changes is described in two sisters born of consanguineous parents. In 1972 this same constellation of findings was first reported in two sibs from a different ethnic origin. This heritable disorder of connective tissue termed the *Tel Hashomer camptodactyly syndrome* is thought to be transmitted as an autosomal recessive trait. The basic defect is unknown.

In 1972, we (Goodman, Bat-Miriam Katznelson, and Manor) described two new genetic disorders of connective tissue which presented with camptodactyly. Recently we had the opportunity of seeing for the second time one of these syndromes in two sibs from an Arab family. The purposes of this report will be to give a detailed account of the clinical features in this syndrome and to discuss its mode of inheritance.

Family studies

A 20-year-old unmarried Bedouin Arab woman with multiple malformations was referred to our diagnostic clinic for evaluation. On questioning the patient about her family, two important facts became known. She stated that she had an older sister with identical features and that her parents were first cousins.

The family pedigree is shown in Fig. 1. In addition to the two affected sibs and their mother, it was not possible to examine any of the other family members. The mother stated that her husband was in good health and did not have any physical defects. The mother did not know her true age but estimated that she was in her late forties. All of her children were born at home and she could not give a reliable history regarding all 13

pregnancies. Five of her children died during the first year of life and the cause of death in each is not known. She denied that any of her other children were abnormal or in any way resembled her two affected daughters.

Physical examination of the proband showed that she had a strikingly similar constellation of findings that we first previously observed in two other sibs some three years ago. For purposes of evaluation and possible treatment both she and her sister, who was identically affected, were admitted to hospital. The positive physical findings in these two sibs are presented in

FIG. 1. The family pedigree.

Received 21 March 1975.

* This study was supported in part by a grant from the Lester Aronberg Foundation.

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TABLE I
CLINICAL FINDINGS IN THE TEL HASHOMER CAMPTODACTYLY SYNDROME

Location and Defect	Previous Cases		New Cases	
	III.7 ♀ aged 17	III.3 ♂ aged 13	III.6 ♀ aged 20	III.7 ♂ aged 19
Stature				
Short	+	+	+	+
Skull				
Brachycephaly	+	++	+	+
Prominent forehead	+	+	+	+
Facies				
Asymmetry	++	+	±	+
Ocular hypertelorism	+	+	+	+
Small mouth	+	+	+	+
High arched palate	+	+	+	+
Increased philtrum length	+	+	+	+
Dental crowding	+	+	+	+
Chest				
Thoracic scoliosis	+	++	+	+
Winging of scapulae	+	++	+	+
Extremities				
Camptodactyly	+	++	+	+
Syndactyly	+	++	+	+
Clinodactyly	-	-	+	+
Brachydactyly thumbs	-	-	+	+
Spindle-shaped fingers	+	+	+	+
Abnormal hand prints	+	+	+	+
Dislocated radii	-	-	+	+
Clubbed feet	+	+	-	-
Pes planus	-	-	+	+
Malformed toes	+	+	+	+
Muscular system				
Hypoplasia of chest	+	++	+	+
Hypoplasia of pelvis	+	++	+	+
Hypoplasia of limb and hand muscles	+	++	+	+

- = absent, ± = mild, + = present, ++ = severe.

TABLE II
RADIOGRAPHIC FINDINGS IN TWO SIBS WITH THE TEL HASHOMER CAMPTODACTYLY SYNDROME

Site	III.6 ♀ aged 20	III.7 ♀ aged 19
Skull	Brachycephalic; prominent maxilla on A-P; broad mandible	Same as III.6
Spine		
Cervical	Spina bifida C ₁ ; enlarged vertebral foramina C ₃	Spina bifida C ₁ ; enlarged vertebral foramina C ₃ ; high vertebral bodies
Thoracic	Mild rotational deformities of dorsal vertebrae	Vertebral annular epiphyses present in all vertebrae; mild scoliosis centred at th. 9-10
Lumbar	Agenesis of left pedicle L ₄ with secondary scoliosis	Mild scoliosis of lumbar vertebrae to right
Pelvis	Normal	Normal
Thorax	Mild lateral retraction of chest wall	Same as III.6
Upper extremities		
Humerus	Hypoplasia of trochlea and capitellum	Same as III.6
Ulnar-radius	Posterior dislocation of radial head	Deformed and enlarged radial head
Hands	Clinodactyly; syndactyly skin only; brachydactyly 1st metacarpal right; narrow distal phalanges	Same as III.6
Lower extremities		
Femur	Normal	Normal
Tibia, Fibula	Elongated fibula; subluxation of tibiotalar joint	Normal
Feet	Vertical talus right with deformity of left talus; subluxation of subtalar joints; deformed navicular; deformed and flattened calcaneus; hallux valgus both big toes; subluxation of interphalangeal joints of great toes, bilateral; severe deformity of terminal digits of feet bilateral	Talus flat and deformed; subluxation of subtalar joint; deformed navicular; deformed and flattened calcaneus; brachydactyly of right metatarsals; hallux valgus big toe right; subluxation of interphalangeal joints of great toes, bilateral; severe deformity of terminal digits of feet bilateral

TABLE III

DERMATOGLYPHIC FINDINGS IN THE TEL HASHOMER CAMPTODACTYLY SYNDROME

Family Members	No. of Whorls	Total Ridge Count	a-b Ridge Count	Ridge Breadth (microns)	Maximal <i>atd</i> angle	Modification of Creases
Present family						
III.6 affected	9	177	77	493	75°	+
III.7 affected	7	209	76	697	92°	+
Mother	4	162	62	523	68.5°	-
Previous family						
III.1 affected	8	271	73	606	110.5°	+
III.3 affected	8	350	120	536	101°	+
III.4 normal sib	3	135	75	428	76°	-
III.6 normal sib	4	179	90	385	90.5°	-
Father	3	163	79	561	71°	-
Mother	1	181	82	533	82°	-

Table I and they are compared with our two previous cases. Fig. 2 and 3 illustrate some of their characteristic physical features.

All routine laboratory studies in each patient were within normal limits including such investigations as serum and urine amino acids, serum aldolase, creatine kinase, transaminase (SGOT and SGPT), phosphorus, calcium, and electrolytes. Electromyographic studies along with electrocardiographic examination were also

within a normal range. Serum gamma-globulin levels were slightly raised in each patient but serum protein immunoelectrophoretic studies did not show any significant alteration. Chromosome studies in the proband showed a normal female karyotype.

Muscle evaluation studies revealed that both patients had bilateral absence of the peronei muscles and extensor hallucis longus muscle of the lower extremities.

Multiple skeletal abnormalities were observed on

FIG. 2. The proband showing a prominent forehead, facial asymmetry, ocular hypertelorism, small mouth, increased philtrum length, poor muscle development involving the shoulders, lower extremities, and palmar muscles of the hand. One can also note the malformed toes, spindle-shaped fingers with camptodactyly. Syndactyly was also present but could only be detected when the fingers were spread.

various x-ray studies, and these are presented in Table II and some are illustrated in Fig. 4.

Dermatoglyphic studies were done in the two affected sibs and their mother. A summary of these findings along with our previous observations are presented in Table III. Fig. 5 shows the very abnormal dermatoglyphic pattern in each affected sib.

The proband complained of pain and difficulty in walking. Her heels were in a valgus position, with short and overlapping toes. Both great toes were also in a valgus position and the left great toe was flexed so that the toe-nail came into contact with the ground (Fig. 2).

To ease her condition a corrective osteotomy was performed at the base of the proximal phalanx and the interphalangeal joint of the left great toe was arthrodesed.

Discussion

There is little doubt that these two sisters share almost the identical pattern of physical findings with our previous two cases of a sister and brother. Their common facial appearance and musculo-skeletal deformities make this a distinct syndrome.

Of significant interest is the striking similarity in the dermatoglyphic configuration noted not only between the two affected sibs but also in the other two previous cases. Genetic disorders of chromosomal origin tend to have a higher degree of similarity (often diagnostic) in their dermatoglyphic patterns, in contrast, to autosomal or X-linked disorders. This heritable syndrome may well be one of the best exceptions to the above. The presence of digital whorls on seven or more digits which extend beyond the borders of the terminal phalanges, a low main line index caused by the very vertical orientation of the A-D radiants, coupled with numerous palmar creases obliterating the normal structure of the ridges and openings of the sweat pores, suggests a possible diagnosis through the use of dermatoglyphs, since, to the best of our knowledge, these findings are unique for this syndrome.

In our original report (Goodman *et al*, 1972), we postulated that this syndrome was transmitted as an

FIG. 3. The proband's 19-year-old sister showing a strong facial and body resemblance with a prominent forehead, facial asymmetry, ocular hypertelorism, small mouth, increased philtrum length, poor muscular development, as exhibited by a stooped posture, poor lower leg muscle development, and hypoplasia of the thenar and hypothenar eminences of the hands. Camptodactyly and spindle-shaped fingers can also be noted.

FIG. 4. X-rays from the two affected sisters showing the cervical spine with spina bifida of C-1 and an enlarged foramina of C-3; short metatarsals with thin shafts of metatarsals and phalanges, hallux valgus with subluxation of interphalangeal joints bilaterally, and overlapping of distal joints; a deformed calcaneus with a vertical talus and a deformed navicular; agenesis of the left pedicle of L-4 resulting in scoliosis.

autosomal recessive trait. Both parents were normal and no other family members were known to be affected. Though the parents denied consanguinity, they both came from small neighbouring communities in Morocco and the overall consanguinity rate for the Jewish Moroccan community is estimated to be 10.7% (Goodman, 1974). In the present family the parents are first cousins and though we did not examine the father, both parents are thought to be normal. Based on the above family information, it now seems that we have convincing evidence for an autosomal recessive mode of transmission in this syndrome.

To describe a syndrome and not to understand the basic defect is a frustration that most observers must live with. Essentially we know nothing of how the mutant gene in this disorder is producing the various malformations. However, it seems that this genetic disorder does share a common bond with all the other genetic syndromes in which camptodactyly is a key feature (Goodman *et al*, 1972). That common bond being that these disorders all involve various alterations in connective tissue.

Originally we termed this disorder, a *syndrome of camptodactyly with muscular hypoplasia, skeletal dysplasia, and abnormal palmar creases*. Since this is a very cumbersome name for a syndrome and since we now have recognized two separate families (one of Jewish Moroccan origin and the other of Arab Bedouin origin) with this condition, we would propose to call this the *Tel Hashomer camptodactyly syndrome*. Without going into the nosological reasons for this terminology, we hope that soon such terminology would be replaced by those words describing the basic defect in this disorder.

We would like to express our appreciation to Dr B. Goldman, E. Akstein, and R. Chaki for performing the cytogenetic studies and to Mrs R. Langridge and Mrs R. Grossman for their secretarial help.

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FIG. 5. The top dermatoglyphic prints are of the proband while the lower are of her affected sister. Four outstanding identical features are noted, the first three of which were also observed in our previous two cases. These features are (1) the numerous palmar creases obliterating the normal structure of the ridges and openings of the sweat pores; (2) the many digital whorls extending beyond the first interphalangeal creases; (3) the strikingly abnormal vertical orientation of the radiants; and (4) the presence of simian creases (the horizontal dashed lines).