Familial Streblodactyly

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A family is described in which female members of two generations were affected from birth from a flexion deformity of the fingers. The pedigree analysis suggests the possibility of a sex-limited autosomal dominant pattern of inheritance. This rare condition, which has been reported only once in the literature, is known by the term streblodactyly (streblos=Gr. twisted, crooked). *Key words: Streblodactyly; Camptodactyly; Sex-limited autosomal dominant inheritance; Hereditary disease.* (Received November 9, 1982.)

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In 1963 Parish et al. (4) first described a hand malformation characterized by a congenital flexion contracture of the fingers and aminoaciduria in 10 females of three generations in a family. The affection is known by the name of streblodactyly. Since all females in the direct line were affected, the authors postulated a sex-linked dominant inheritance. We recently had the opportunity to study two sisters in a family in which female members showed this disorder (Fig. 1).

CASE REPORTS

Case 1 (III, 3). The first propositus, a 27-year-old unmarried woman, otherwise healthy, was first seen in April 1982, for a hyperkeratosis of the soles. During clinical examination it was noted that she had unusual deformities of the fingers (Fig. 2), characterized by flexion contractures of the metacarpophalangeal and interphalangeal joints. The deformity had been present from birth.

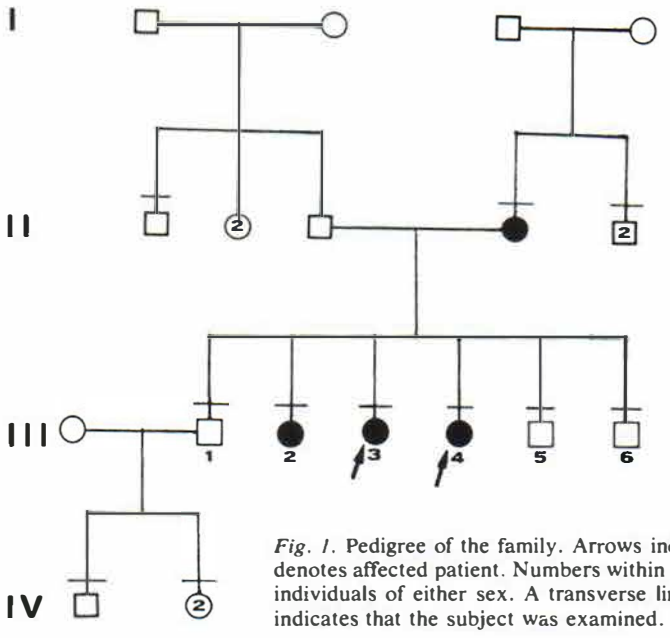
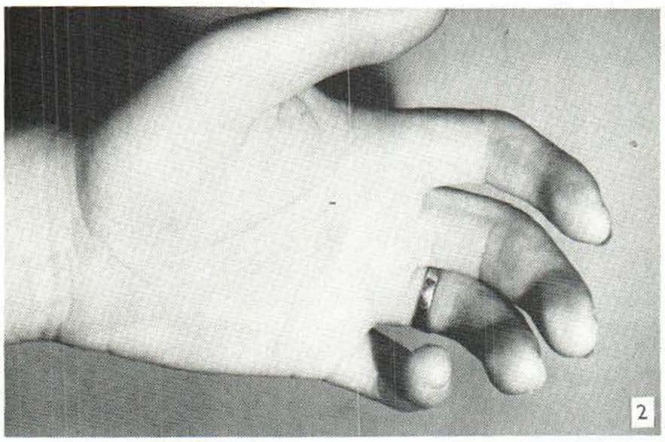
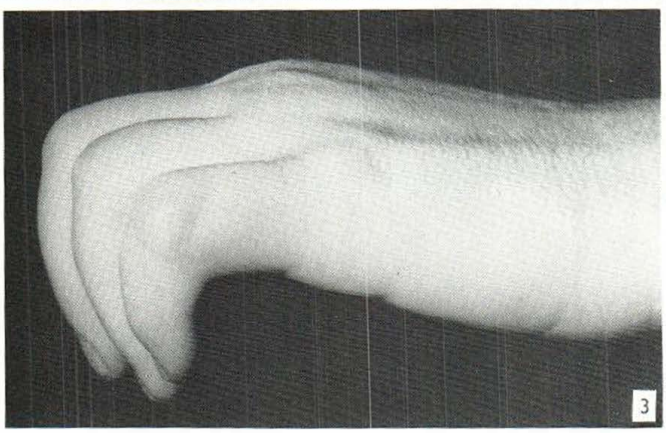


Fig. 1. Pedigree of the family. Arrows indicate propositi. Black symbol denotes affected patient. Numbers within symbols refer to the number of individuals of either sex. A transverse line immediately over a symbol indicates that the subject was examined.





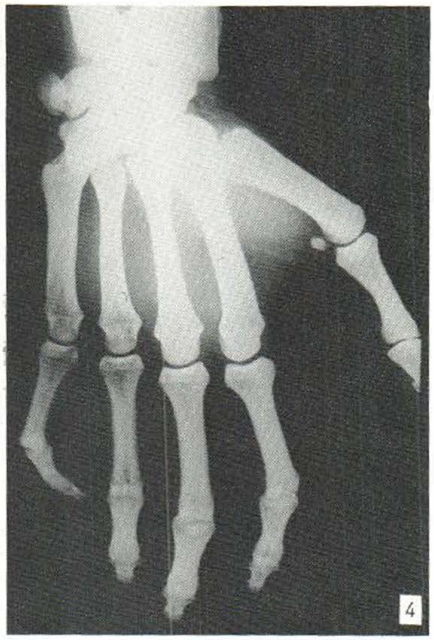


Fig. 2. Left hand of propositus (III, 3), palmar vies.

Fig. 3. Left hand of propositus (III, 4), lateral view.

Fig. 4. Hand X-ray film showing flexion at metacarpophalangeal and interphalangeal joints, bone structure and metacarpal index being normal.

Case 2 (III, 4). The second propositus, a 25-year-old unmarried woman, otherwise healthy, was seen at the same time as her sister (case 1). Her medical history was identical with case 1, as was the physical examination, noting flexion contractures of the fingers (Fig. 3).

In both patients, routine laboratory evaluations were within normal limits. A urinary amino acids pattern, as well as neurological and ophthalmological examinations, revealed no abnormalities. X-ray of the hands showed identical features in both cases (Fig. 4).

DISCUSSION

To our knowledge, only one case of familial streblodactyly (FS) has been reported hitherto in the literature (4). The congenital deformity is characterized by a permanent flexion contracture of all the fingers at the proximal interphalangeal joints. The defect differs from camptodactyly, also known as streblomicrodactyly (5), in which disease only the little finger may be affected and both sexes are equally involved; it is inherited as an autosomal dominant trait with variable penetrance (3). The only occurrence of FS in female members and the absence of miscarriages in the pedigree of this family suggest a possible sex-limited autosomal dominant inheritance (1). This pattern of inheritance is extremely rare and has been postulated in male-limited autosomal dominant precocious puberty and in X-linked testicular feminization syndrome (2).

We stress the interest of this disease for the rarity and the pattern of inheritance.

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