Familial Streblosactyly

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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 streblosactyly (streblos=Gr. twisted, crooked). Key words: Streblosactyly; 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 streblosactyly. 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 I (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.
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 strebiodactyly (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 strebomicrodactyly (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.
Elephantiasis of External Ears: A Rare Manifestation of Pediculosis capitis

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A case of elephantiasis of the external ears associated with pediculosis capitis is reported. Hypersensitivity to the louse bite and Melkersson-Rosenthal syndrome are discussed. (Received September 8, 1982.)

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Elephantiasis and elephantiasiform enlargement of the ears have rarely been described, following eczema, frostbite, erysipelas and piercing of the ears (3). Elephantiasis of the ears associated with pediculosis does not previously appear to have been reported.

CASE REPORT

A 33-year-old woman with enlarged and mildly pruritic ears of 3 years’ duration had been heavily infested with pediculus capitis for several years. Both ears were erythematous and markedly swollen; the right one (Fig. 1) was more severely affected. A few post-auricular subcutaneous nodules (5–10 mm) were present bilaterally. The skin over the nape of the neck and upper back was indurated and hyperpigmented. Right cervical lymph nodes were enlarged. She had a persistent right peripheral facial palsy, thought to be Bell’s palsy. The white cell count ranged 8.2–11.3 x 10^9/l, with 8–11% eosinophils. The erythrocyte sedimentation rate was 45 mm/hour (normal 20 mm/hour).

Other tests including serum protein electrophoresis, liver function tests and antinuclear antibodies were normal. Scrapings of ear lobes and nasal mucosa showed anaerobic fast bacilli. Chest X-ray, and bone survey were normal. Patch tests with the whole battery of European standard allergens proved negative. Cervical lymph node biopsy showed non-specific reactive changes. Skin biopsies of the right ear (Fig. 2) showed hyperkeratosis, follicular plugging, focal spongiosis, elongated rete ridges and numerous branching hair follicles. The dermis was infiltrated by chronic inflammatory cells composed predominantly of lymphocytes together with eosinophils, plasma cells and a few melanin-laden macrophages. The cellular infiltrate formed aggregates with no germinal centres around dilated capillary branches. Numerous dilated and conspicuous lymphatic spaces were present, especially in the superficial and mid-dermis. Mast cells were apparently increased in number. No acid-fast bacilli, epithelioid or foreign-body type granuloma were seen. There was no iron pigment or amyloid deposition. Biopsies of post-auricular nodules and the skin of the back showed similar changes. However, the lymphatics were not dilated or conspicuous in the biopsies of the back lesions.

Her pediculosis was treated with DDT. Intralesional injections of 10 mg/ml triamcinolone acetonide (Kenalog) resulted in a decrease in the size and induration of the ears, with almost complete resolution of subcutaneous nodules.

REFERENCES