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Case Report

FAMILIAL HYPEREXTENSIBLE PROXIMAL INTERPHALANGEAL JOINTS

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Summary Hyperextensibility of the proximal interphalangeal joints was noted in 3 males and 6 females in 3 generations of a Japanese family. The proposita, a 14-year-old girl, had hyperextensible proximal interphalangeal joints of the 2nd, 3rd, 4th, and 5th fingers giving "swan-neck" appearance during the extension. She was asymptomatic and had no other features of skin involvement. Her father, younger sister, grandmother, two aunts, and three cousins on the father's side had hyperextensible proximal interphalangeal joints. No instance of male to male transmission was present. The condition was thus inherited as an autosomal or X-linked dominant trait.

Key Words dominant trait, hyperextensible joints, proximal interphalangeal joints

INTRODUCTION

Familial articular hypermobility syndromes are a heterogeneous group of disorders characterized by generalized articular hypermobility with or without subluxation or dislocation (Beighton *et al.*, 1986, 1989). In addition to generalized type, localized familial joint laxity is also seen (Whitney, 1932; Sturkie, 1941; Glass and Kistler, 1953).

We report here a Japanese family in which 9 members in three generations are affected by the hypermobile proximal interphalangeal joints.

CASE REPORT

The proposita, a 14-year-old girl (1127694), was referred to us because of the evaluation of hyperextensible joints of the fingers. She was born to a 28-year-old

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gravida I, para 0 mother and a 31-year-old father, both healthy and nonconsanguineous.

Hyperextensible finger joints were first noted at the age of 8 years. Radiographs of the hand which were taken at the age of 9 years revealed no abnormalities.

When first seen by us at the age of 14 years, she weighed 66 kg and measured 154.2 cm. She had hyperextensible proximal interphalangeal joints from the 2nd to the 5th finger of both hands (Table 1). The voluntary extension of the proximal interphalangeal joints was associated with reciprocal flexion of the distal interphalangeal joints and gave "swan-neck" appearance (Fig. 1A). Metacarpophalangeal and distal interphalangeal joints of the fingers and interphalangeal joints of the toes were not hypermobile. She also had mild hyperextensible elbow joints, whereas the rest of the joints including shoulders, wrists, hips, knees, and ankles did not show any hyperextensibility. She was asymptomatic and did not have skin laxity, hyperextensibility, fragility, or scar formation after injury.

Family studies (Fig. 2). The proposita's father (II-2) had hypermobile joints (Fig. 1B). On the father's side, the proposita's younger sister (III-5), grandmother (I-2), aunts (II-1 and II-3) and three cousins (III-1, III-6, and III-7), all had hyperextensible proximal interphalangeal joints according to the father's description. Of these, the paternal aunt (II-3) and paternal cousin (III-1) were reported to be affected in the same way as the proposita without any symptoms. There was no family history of subluxation or dislocation of the joints, rheumatoid arthritis, skin fragility, or other skin disorders.

DISCUSSION

Familial articular hypermobility syndrome is divided into two types; generalized, uncomplicated type and complicated type (Beighton *et al.*, 1986). Ehlers-Danlos syndromes and skeletal dysplasias with hypermobility are excluded from the syndrome. In addition to the generalized hypermobility syndromes, localized

Table 1. Movement at proximal interphalangeal, elbow and wrist joints.

	Extension (degrees)	
	Right	Left
Proximal interphalangeals		
Index	20°	5°
Middle	42°	10°
Ring	50°	15°
Little	30°	0°
Elbow	20°	30°
Wrist	90°	90°

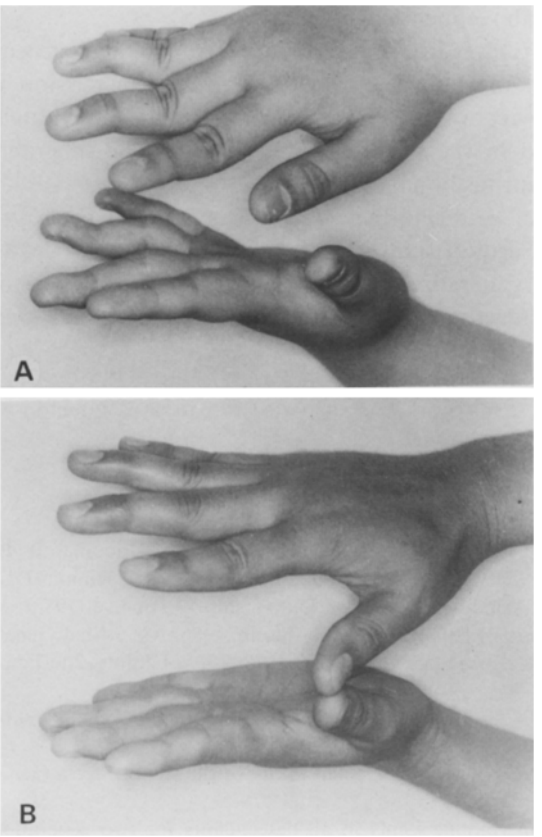

Fig. 1. Hand of the proposita (A) and her father (B) showing hyperextension at proximalinterphalangeal joints and flexion at distal interphalangeal joints.

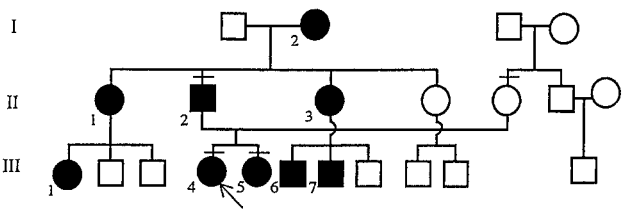

Fig. 2. Pedigree of the family.

familial hypermobility has been so far reported. These include hypermobility of the interphalangeal joints of the thumb (Whitney, 1932), hypermobility of the fingers and thumb (Sturkie, 1941), and distal hyperextensibility of the thumb (Glass and Kistler, 1953; MIM 274200). Proximal joint hyperextensibility and distal joint restriction of the fingers are also noted in Aarskog syndrome (Berman *et al.*, 1975). However, the condition described in our reported family is different from

those found in the above mentioned.

The facts that 1) vertical transmission through three generations were noted, 2) both sexes were affected, 3) no male to male transmission was observed, and 4) variable expressivity was noted among the affected individuals, lead us to think that the trait found in the present family seems to be inherited in an autosomal or X-linked dominant fashion.

Basic defects of articular hypermobility syndrome is as yet unknown at the molecular level. Further studies will clarify the defects leading to phenotypic variations between generalized and localized articular hypermobility as noted in other heritable connective tissue disorders such as Ehlers-Danlos syndrome or osteogenesis imperfecta.

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