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Symphalangism in an Indian Family

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Abstract

Symphalangism is an uncommon congenital abnormality characterized by fusion of interphalangeal joints of fingers and toes. It exhibits autosomal dominant inheritance pattern. Involvement of both proximal and distal interphalangeal joints can occur. But proximal interphalangeal joint involvement is more common. It may be associated with other skeletal or non-skeletal abnormalities. Compensatory hyperflexibility of the joints proximal and distal to the fused joints may be present. Here we are presenting two brothers with proximal symphalangism in bilateral hands and feet. Involvement of index finger and thumb was also present in these patients. They did not complain of any functional impairment in daily activities and hence did not seek any intervention to regain movement in the involved joints.

Introduction

Symphalangism is an uncommon syndrome characterized by ankylosis of interphalangeal joints of fingers and toes. It was first described by Harvey Cushing in the first issue of Genetics (1916).[1] He interviewed three affected members of a family and presented a family record of seven generations, where he found 84 affected members among 312 descendants of an affected person.

The fusion can occur in any finger. Little finger is the most frequently involved finger followed by ring, middle and index finger.[2,3] Index finger is generally involved only when other fingers are also involved. Involvement of thumb has also been reported.[5] The fusion can occur in proximal interphalangeal (PIP) or distal interphalangeal (DIP) joints; however, involvement of proximal interphalangeal joints is more common.[3,4,5] Accordingly it can be classified into proximal and distal symphalangism. Proximal as well as distal symphalangism have an autosomal dominant inheritance pattern.[1,2,3,4,5,7,8] It has been reported to be associated with additional skeletal and non-skeletal abnormalities [3,4,7,9], however, expression of these abnormalities is variable and depends on the type of symphalangism. Commonly seen skeletal abnormalities include brachydactyly, camptodactyly, clinodactyly, syndactyly, radio-humeral fusion, carpal and metacarpal anomalies, pes planus, bilateral hip dislocation, tarsal coalition, congenital fusion of cervical or thoracic spine, compensatory hyperflexibility of unaffected joints of same digit. Conductive hearing loss and absence of cutaneous creases over the affected joints are the associated non-skeletal abnormalities. Symphalangism may be found in association with some syndromes such as Poland syndrome, Apert syndrome, Herrmann's syndrome etc. According to Flatt and Wood symphalangism can be divided into three main groups- true symphalangism without additional skeletal abnormality, symphalangism associated with symbrachydactyly, and symphalangism with syndactyly.[3]

In India symphalangism have been reported earlier by Gemma Savarinathan and Willard R. Centerwall.[10] They reported a south Indian family of which twelve members spanning over four generations were affected. They found fibrous symphalangism of thumb associated with variable syndactyly and polydactyly in nine out of twelve affected members. Osteoarthritis of joints proximal and distal to the fused joints has been observed as a long term complication. This may be partly because of increased mechanical stress in these adjacent joints.[11]

Case History

Two cases of symphalangism in a family are presented. The affected patients were two brothers out of five siblings. The family pedigree is shown in the Figure 1. Both the brothers were interviewed and examined. They confirmed the involvement of four generations of their family tree of which they represent the third generation. They could not provide details of extent of involvement in the first and second generations except for their father.

Case 1: The younger brother aged 40 years presented in our department with absence of movements in some of the joints of the fingers and toes since birth. His physical examination revealed slightly short fingers and normal

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length toes except short great toes. No active or passive movements could be elicited in the PIP joints of all fingers and lateral four toes and in the interphalangeal joints of thumb and big toe of both sides. All the nails were normally developed. The DIP and metacarpophalangeal joints of the affected finger were having increased flexibility. Rest of the joints of the extremities including bilateral wrist, elbow, shoulder, ankle, knee and hip joints showed no abnormality in movement. There was absence of skin creases on the dorsal and volar aspects of the affected joints (Figure 2, 3); although single poorly developed skin crease was present in the volar aspect of PIP joints of bilateral little and middle fingers. Examination of the rest of the body did not reveal any other abnormality. Radiographic examination showed complete fusion of PIP joints of medial four digits of both the hands (Figure 4, 5). Fusion of IP joints of thumbs was incomplete. Radiographs of the feet showed complete fusion of the PIP joints of lateral four toes (Figure 6). There was incomplete fusion of IP joints of both the great toes. On the left side 1st metatarsal and proximal phalanx was markedly shorter in comparison to normal, whereas on the right side it was slightly shorter.

Case 2: The elder brother aged 48 years accompanied case 1. During history taking of case 1 he also exposed his deformities. His physical examination showed normal length fingers and toes. No active or passive movement was present in PIP joints of middle, ring and little fingers of both the hands. In the PIP joints of index fingers and IP joints of thumbs some flexion was possible. In PIP joints of lateral four toes and IP joints of great toe of both feet there was no passive or active movement. Joints just proximal and distal to the affected joints were having more flexibility in comparison to normal range. Rest of the joints in the extremities were found to have normal mobility. There was no skin crease on the dorsal aspect of the affected joints and on volar aspect of ring and little finger of both hands (Figure 7, 8). But poorly developed skin crease was present on the volar aspect of the little and index finger and thumb. No other abnormality was detected in the rest of the body.

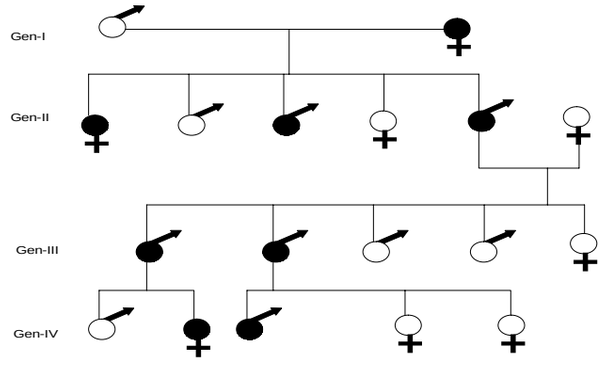

Fig. 1: *Pedigree pattern of symphalangism involving four generations*

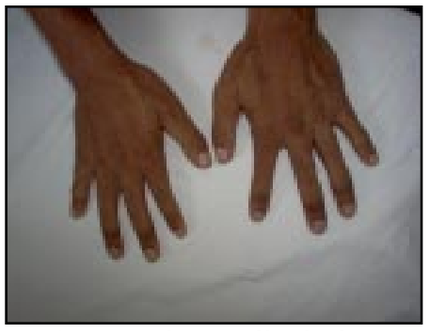

Fig. 2: *Dorsal aspect of hands of case 1*

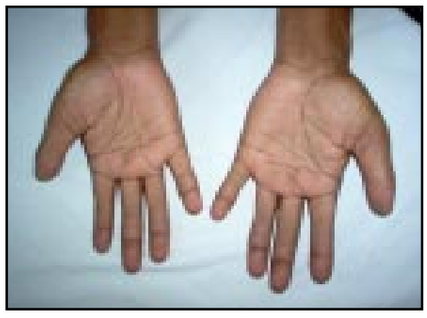

Fig. 3: *Volar aspect of hands of case 1*

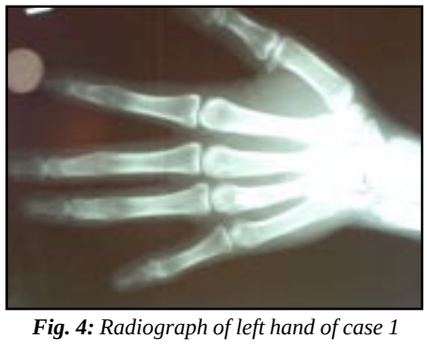

Fig. 4: *Radiograph of left hand of case 1*

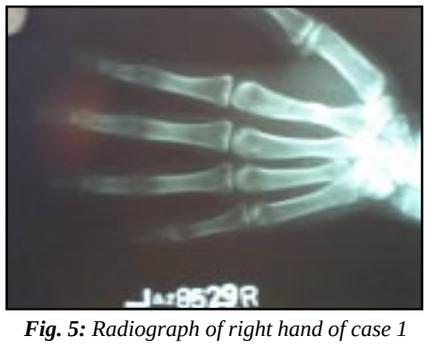

Fig. 5: *Radiograph of right hand of case 1*

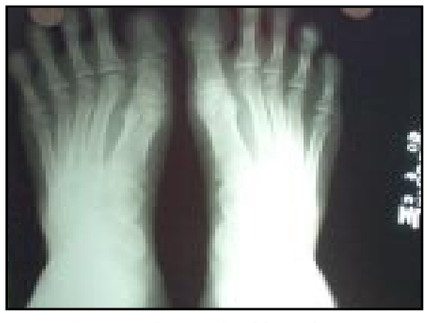

Fig. 6: *Radiograph of feet of case 1*

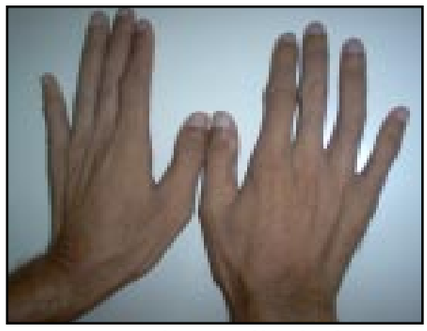

Fig. 7: *Dorsal aspect of hands of case 2*

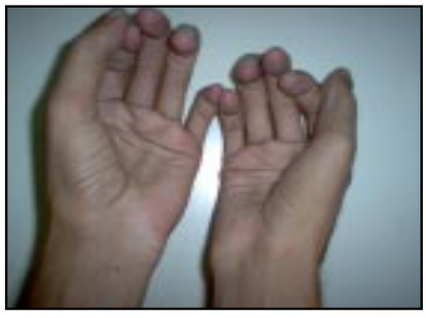

Fig. 8: *Volar aspect of hands of case 2*

Discussion

Symphalangism is an autosomal dominant disorder. [1,2,3,4,5,7,8] The pedigree pattern of our cases also reflects the same. As evident from the literature available, proximal symphalangism is more common than the distal variety. [3, 4, 5] In a review of literature Flatt and Wood documented only 34 DIP fusions out of 649 affected fingers. The two cases discussed here fall under the category of proximal symphalngism. Occurrence of the abnormality in the index finger and thumb is less common, but in our cases involvement of these digits were also noticed. In the Indian family reported by Gemma Savarinathan and Willard R. Centerwall affection of thumb was described and the ankylosis was fibrous whereas in our cases it was bony. [10] Their cases were associated with syndactyly and polydactyly but in our cases these findings were not present. It is known to be associated with additional skeletal abnormalities and in our case 1, short 5th metacarpal and short proximal phalanx of great toes of both sides were the only such findings. The only non-skeletal association found in our cases was the absence of skin creases over the affected joints. Occurrence of osteoarthritis in the joints proximal and distal to the fused joints has been reported.[11] Our patients were aged 40 years and above but they did not show signs of osteoarthritis as per ACR criteria. Probably because the condition manifests since birth in most of the instances, persons with symphalangism learn to perform tasks without movement of the affected joints and do not have functional impairment.[3, 6, 12] Both the cases we are presenting did not have any functional impairment and did not want any intervention for restoration of movement in the affected joints or to increase functional ability. These cases are unique in another respect that involvement of thumb and index finger being rare, was present in these cases.

References:

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