

# Distal symphalangism associated with camptodactyly

SHOZO OHDO, YOSHIKAZUMI YAMAUCHI, AND KUNIO HAYAKAWA

From the Department of Pediatrics, Miyazaki Medical College, 5200 Kihara, Kiyotake-cho, Miyazaki-gun, Miyazaki, Japan

**SUMMARY** A Japanese family in which four patients in three generations had distal symphalangism associated with camptodactyly is reported. All of these patients had extension limitation of the proximal interphalangeal joints of the toes of both feet. Radiographs of the hands and feet, undertaken in three cases, showed no bony fusion of the distal and proximal interphalangeal joints. This malformation is caused by an autosomal dominant gene. To our knowledge, no previous case of distal symphalangism with extension limitation of the proximal interphalangeal joints has been reported.

## Case reports

### CASE 1

The family pedigree is shown in fig 1.

The proband was born in October 1978. The mother had no complications during pregnancy. He was 3360 g in weight and 52.9 cm in height when delivered. Consanguinity was denied.

Although the proximal interphalangeal joints of fingers 2, 3, and 4 of both hands showed extension limitation (up to 150°), flexion of these fingers was normal. Slight limitation of movement was noted in the distal interphalangeal joints. Distal interphalangeal creases on both the dorsal and volar sides were completely absent on fingers 2, 3, 4, and 5 of both hands (fig 2).

The proximal interphalangeal joints of the second toes of both feet had extension limitation (up to 150°).

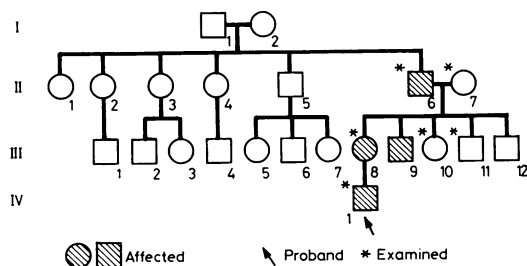


FIG 1 Family pedigree.

Received for publication 12 February 1981

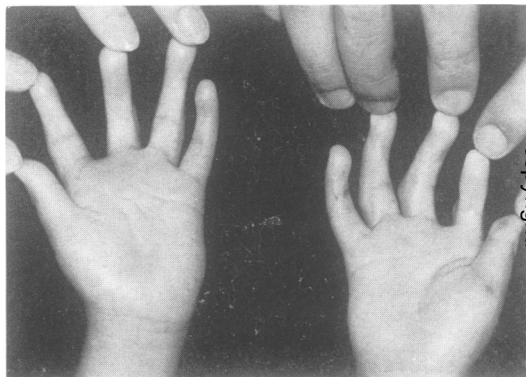


FIG 2 Hands of proband.

Growth rate was in the normal range and no chromosome abnormalities were noted. There were no abnormal findings in the clinical laboratory tests performed and x-ray studies showed no abnormality (fig 3).

### CASE 2

The mother of the proband, a housewife, was medically examined at 28 years of age. She had extension limitation (up to 110 to 140°) of the proximal interphalangeal joints of fingers 3, 4, and 5 of both hands. She could not flex the distal interphalangeal joints by herself, but they could be flexed by force up to 150°. Distal interphalangeal creases on both the dorsal and volar sides of fingers 2, 3, 4, and 5 of both hands were completely absent (fig 4). She was able to clench her fist in a normal

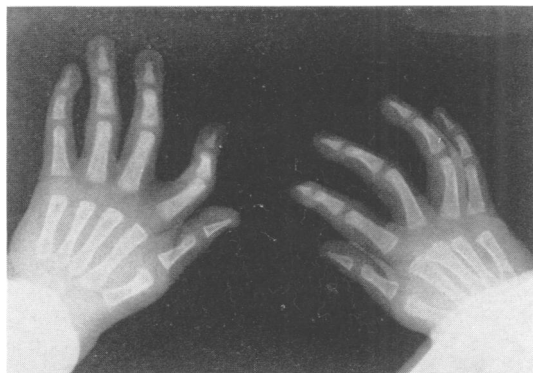


FIG 3 X-ray of hands of proband.

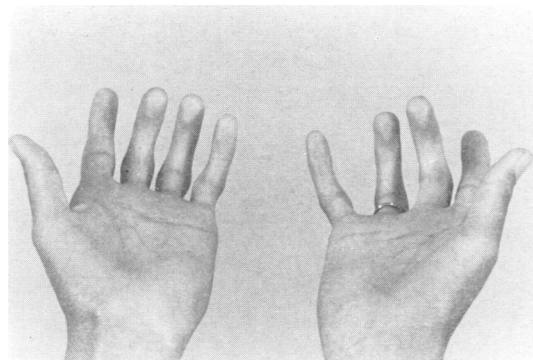


FIG 4 Hands of case 2.

manner. The metacarpophalangeal joints of both thumbs could be fully extended with difficulty. There was extension limitation (up to  $140^\circ$ ) of the proximal interphalangeal joints of the third toes of both feet.

#### CASE 3

The maternal grandfather of the proband, a farmer, was 62 years of age when examined. He had extension limitation of the proximal interphalangeal joints of fingers 2, 3, 4, and 5 of both hands and limitation was most pronounced in finger 3.

Fingers 2, 3, and 4 of both hands showed limitation of movement at their distal interphalangeal joints. Slight flexion, however, was possible when forced. On the distal interphalangeal joints, creases on the dorsal side of fingers 2, 3, 4, and 5 of both hands were absent. Creases on the volar side were also absent on right fingers 2, 3, and 4 and on left fingers 3, 4, and 5 and incomplete on the right finger 5 and on the left finger 2.

The thumbs could be fully extended with difficulty

at the metacarpophalangeal joints. The fourth toes of both feet showed extension limitation of the proximal interphalangeal joints. Simian creases were present on both palms.

#### CASE 4

The maternal uncle of the proband was 26 years of age at the time of this study. Although he was known to have similar deformities to those described above, we could not examine him because he lived too far away.

#### Discussion

Symphalangism, as an independent deformity, has been classified into at least two types: distal symphalangism and proximal symphalangism.<sup>1</sup> Steinberg and Reynolds<sup>2</sup> suggested that the locus for distal symphalangism is different from that for proximal symphalangism. This suggestion was based on the finding that there had not been a case in which patients with distal symphalangism and those with proximal symphalangism were present in the same family.

As regards proximal symphalangism, a number of clinical reports have been presented since the first report by Cushing.<sup>3</sup> Five families with this disease have been reported in Japan.

Distal symphalangism, on the other hand, has been reported only by Inman,<sup>4</sup> Daniel,<sup>5</sup> Fried and Mundel,<sup>6</sup> and Lambert *et al.*<sup>7</sup> Both Fried and Mundel<sup>6</sup> and Lambert *et al.*<sup>7</sup> stated that their cases were different from the cases reported by Inman<sup>4</sup> and Daniel<sup>5</sup> because of the lack of bony fusion between the distal and middle phalanges in their cases. They have thus suggested possible heterogeneity in distal symphalangism, that is, those with and without bony fusion. The presence or absence of bony fusion has also been reported in cases of proximal symphalangism.

The outstanding clinical feature of the cases reported here is that distal symphalangism in the family was associated with camptodactyly of the proximal interphalangeal joints.

Symphalangism of both distal and proximal types is known to be the result of autosomal dominant heredity and camptodactyly is also inherited as an autosomal dominant trait. Therefore, we inferred that in this family the original mutation might have occurred in the germ cells of the father, the mother, or the maternal grandfather of the proband.

#### References

- 1 Temtamy S, McKusick V. *The genetics of hand malformations*. New York: Liss, 1978.

- <sup>2</sup> Steinberg AG, Reynolds EL. Further data on symphalangism. *J Hered* 1948;**39**:23-7.
- <sup>3</sup> Cushing H. Hereditary ankylosis of proximal phalangeal joints (sympalangism). *Genetics* 1916;**1**:90-106.
- <sup>4</sup> Inman OL. Four generations of symphalangism, *J Hered* 1924;**15**:329-34.
- <sup>5</sup> Daniel GH. A case of hereditary anarthrosis of the index finger, with associated abnormalities in the proportions of the fingers. *Ann Eugen* 1936;**7**:281-97.
- <sup>6</sup> Fried K, Mundel G. Absence of distal interphalangeal creases of fingers with flexion limitation. *J Med Genet* 1976;**13**:127-30.
- <sup>7</sup> Lambert D, Nivelon-Chevallier A, Chapuis JL. Absence of distal interphalangeal fold causing difficulty in extending fingers. *J Med Genet* 1977;**14**:466-7.

Requests for reprints to Dr S Ohdo, Department of Pediatrics, Miyazaki Medical College, 5200 Kihara, Kiyotake-cho, Miyazaki-gun, Miyazaki, Japan.