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Congenital Contractural Arachnodactyle*

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Hecht and Beals recently delineated a new syndrome which mimics Marfan's syndrome.[1, 2] They named this new etity congenital contractural arachnodactyly (CCA). The purpose of this communication is to present another such case and to propose that metatarsus varus with hallu valgus secondary to it, is probably a common feature of this syndrome.

Case Report

A. U., a 13/1/2-year-old girl, was the sixth child of a 37-year-old mother and a 40-year-old father. She was the product of a full-term pregnancy and an uneventful labor. The parents were not relatives and there was no history ot exposure to X-ray, infections or drugs.

The patient was referred to this hospital with complaints of excessive height and inability to gain weight. Her mother's height was 168 cm. and the father's height was 178 cm. She had three sisters, 17, 28 and 32 years of age; the tallest being 168 cm. Her two brothers, 23 and 26 years of age, had a height of 168 and 172 cm., respectively. Physical examination of the family members revealed no abnormalities.

The patient was 166 cm. in height (90 percentile) and weighed 37.6 kg. (10 percentile). No abnormalities with regard to facial features existed but she could open her mouth only 2.5 cm. Her arms and legs were thin and long with decreased muscle mass; her arm span was 174 cm (Figure 1.) She had flexion contractures involving wrists and proximal interphalangeal joints; the fingers were thin and very long. (Figure 2.) She was unable to fully extend her lower extremities which were located past the knee joint. She also had pes planus, metatarsus varus and thin,

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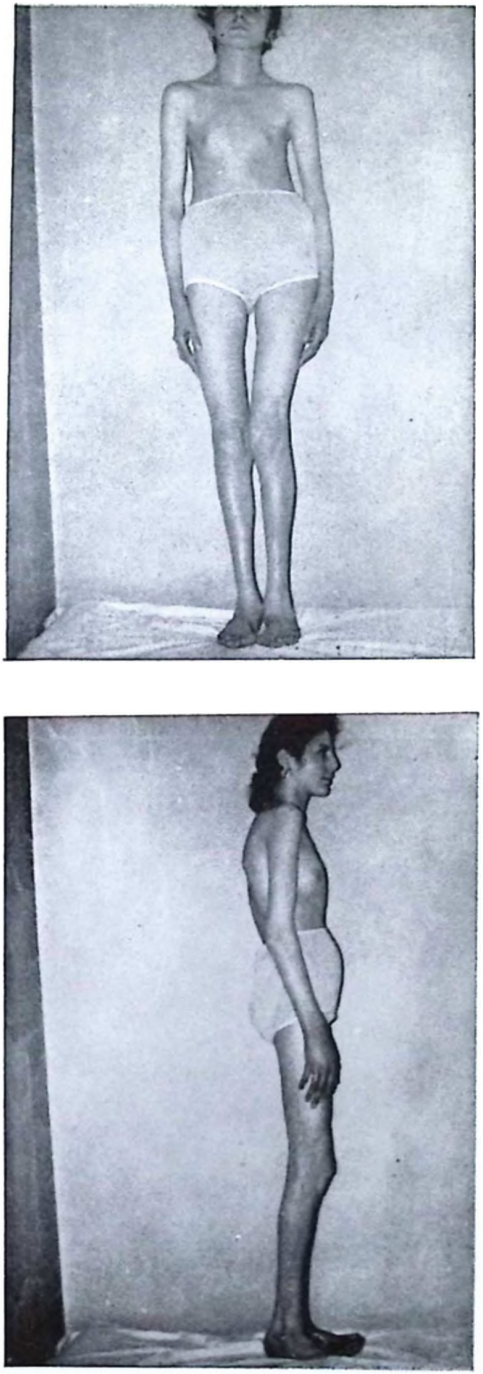

Figure 1 a and 1 b

Picture of the patient showing long and thin extremities with decreased muscle mass.

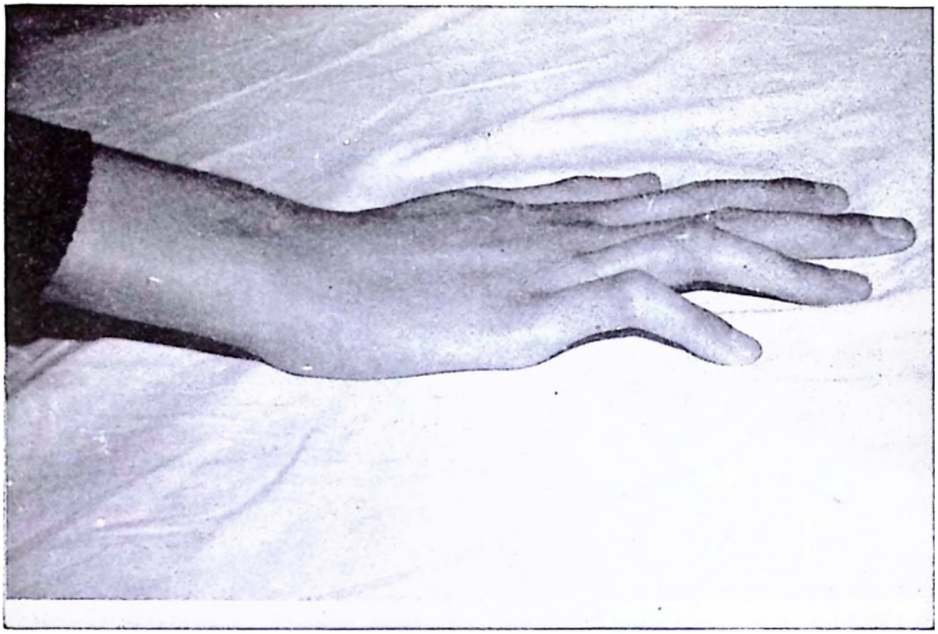

Figure 2
Long and thin fingers with contractures at the proximal interphalangeal joints. Limitation of extension is more marked on the fourth and fifth fingers.

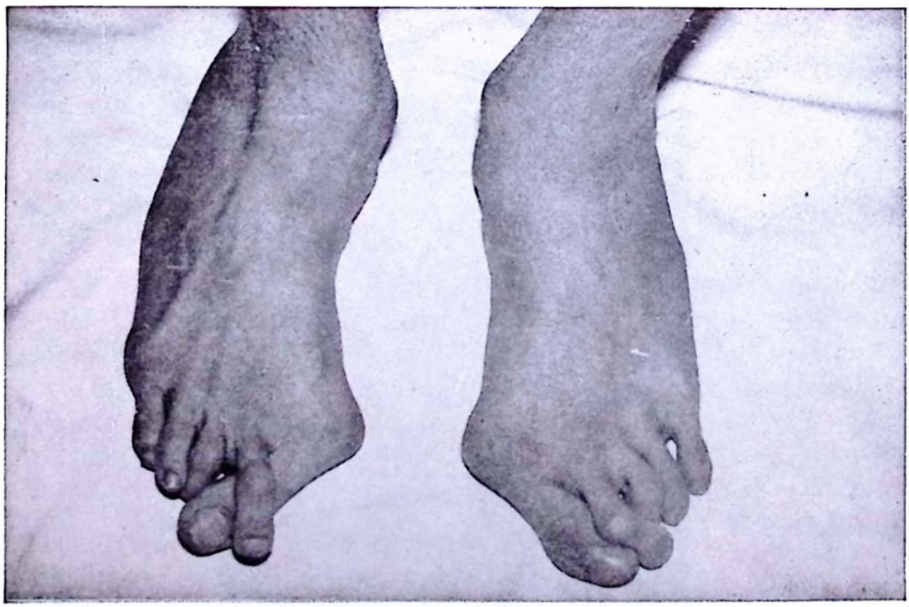

Figure 3
Bilateral hallux valgus, metatarsus varus and incurving of the toes. Second toe overrides the first on the right.

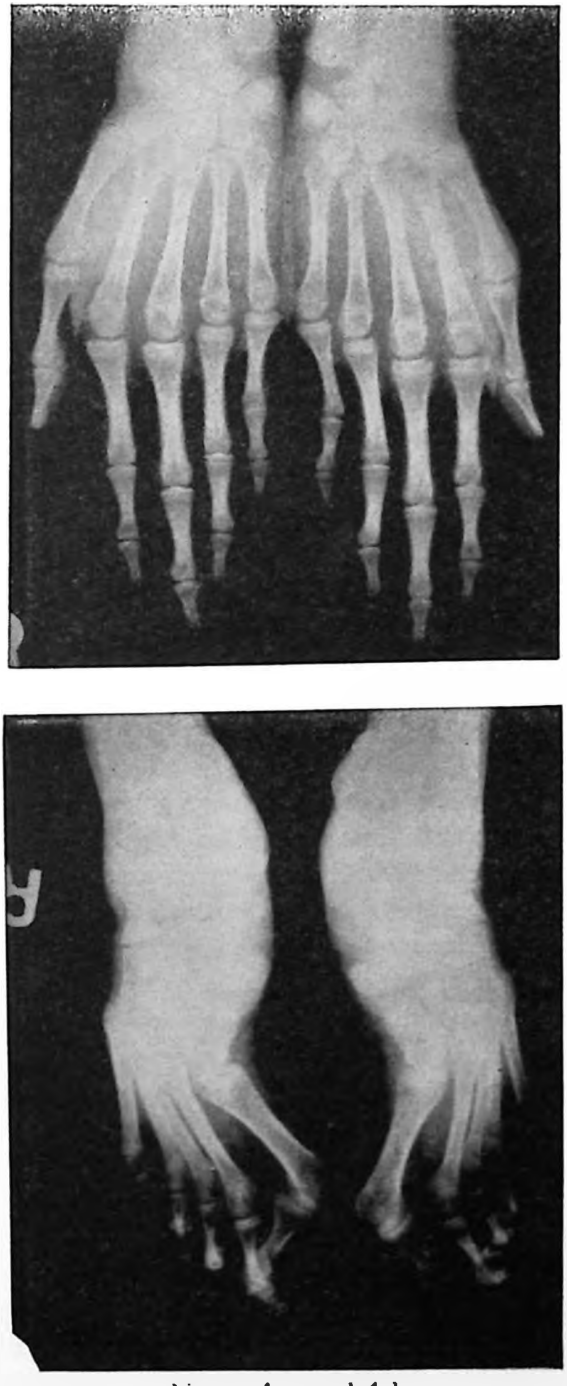

Figure 4 a and 4 b

X-ray of the hands and feet showing bony deformities which are explained in the text.

long toes which showed interphalangeal flexion contractures, especially, involving the proximal interphalangeal joints. There was a 60° bilateral hallux valgus. (Figure 3.) Other findings included a moderate pectus excavatus, slight scoliosis at the thoracal region and thoracolumbar kyphosis. The remainder of the examination revealed no other abnormalities. The lenses were not dislocated.

X-ray examination of hands and feet showed markedly elongated phalanges, metatarsal and metacarpal bones. The metacarpal index was definitely in favor of Marfan's syndrome.10 Bilateral metatarsus varus and hallux valgus were present. (Figure 4 a and b). X-rays of the spine showed flattening of the thoracal vertebrae, scoliosis at the thoracal region and thoracolumbal kyphosis. In this latter area the ventral height of the corpus vertebrae was less than the dorsal height, producing a beak-like anterior vertebral margin. No secondary ossification centers could be visualized. The heart appeared normal in size and configuration.

Routine blood chemistry, including calcium, phosphorus and alkaline phosphatase, showed no abnormality. Urine amino acid chromatography was also normal. Chromosome analysis of peripheral leucocytes revealed a 46, XX pattern.

Discussion

Congenital contractural arachnodactyly is a recently described entity.[1:2] Its characteristic features include multiple congenital joint contractures, arachnodactyly and crumpled earsand kyphoscoliosis. There is usually no ocular or cardiovascular involvement. The main difference between it and Marfan's syndrome is the fact that in CCA, the patients have joint contractures, rather than loose ligaments and joint capsules. No dislocation of lens and cardiovascular pathology is found in CCA, which also helps differentiate these two entities. Other conditions that should be considered in differential diagnosis include homocystinuria, Achard syndrome and the condition reported as syndrome of "pigmentary retinal degeneration, cataract, microcephaly and severe mental retardation.[3]" Homocystinuria, of course, could be definitely diagnosed by a proper labortaroy examination of urine by amino acid chromatography. It should also be remembered that the patient with homocystinuria may show dislocation of lens and cardiovascular lesinos, while congenital contractures are usually not encountered.[4] The patient described by Achard had cylindiric and elongated fingers, but she lacked contractures and long, norrow extremities.[5]

We were impressed by the severity of the bilateral hallux valgus deformity due to metatarsus varus in our patient. After examining the drawing of the foot of Marfan's original patient,[6] we wondered if this finding could be another one of the characteristic features of CCA. A brief survey of the available literature[1, 2, 7, 8, 9] further strengthened this opinion, since one of the patients of Ghosh7 (case 1) had a similar malformation. Interestingly enough, both families reported by Beals et, al.[2] also had metatarsus varus deformity. In the first case (kindred-1, DH), malformation was mild, while in the second it was severe enough to require the application of plaster casts.

In the case of A. U. ,as well as in those of Ghosh and Beals, severe hallux valgus did not seem to be a pirimar., but secondary deformity to that of metatarsus varus. We hope further publication of cases with congenital contractural arachnodactyly will throw more light on this matter.

Summary

A case of congenital contracutral arachnodactyly is presented. It is proposed that metatarsus varus is probably a common feature of this syndrome.

Acknowledgement

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