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CONGENITAL CONTRACTURAL ARACHNODACTYLY

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Five cases of congenital contractural arachnodactyly (CCA) are reported. Three belong to the one family. CCA has often been mistaken for Marfan's disease and arthrogryposis multiplex. Because CCA has a more favourable prognosis, it is very important to be able to recognize this syndrome.

Key words: congenital contractural arachnodactyly; Marfan's syndrome; arthrogryposis multiplex congenita; connective tissue disorder; joint contractures

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Congenital contractural arachnodactyly (CCA) is a dominantly inherited disorder of the connective tissue. A few years ago the syndrome was separated from Marfan's disease and delineated as a pathological entity (Epstein et al. 1968, Beals & Hecht 1971). The most characteristic features are arachnodactyly, multiple congenital contractures, dolichostenomelia (long, thin limbs), deformed external ears and kyphoscoliosis. In CCA there is no lens displacement, and usually no cardiac involvement. Because the prognosis is generally favourable, it is important to distinguish this syndrome from both Marfan's disease and arthrogryposis multiplex congenita.

The purpose of the present paper is to call attention to the syndrome by reporting some cases treated at our hospital.

CASE REPORTS

Case 1. T.S. (born 21 January, 1962). He was the product of a normal pregnancy and delivery and was referred to our hospital at the age of 4 weeks because of congenital contractures of the knees, shoulders and elbows. He had noticeably long, thin fingers and toes and a talipes calcaneovalgus deformity of the feet. The condition was thought to be arthrogryposis multiplex. The foot deformity was treated by plaster casts and braces. No treatment was given for the joint contractures. At the age of 21 months the contractures of the elbows, shoulders and knees had lessened, but he still walked with a flexion deformity of 15° in both knees, and a slight kyphoscoliosis was noticed. At the age of 7 years he had marked arachnodactyly, and his feet were long and slender. He had only slight contractures of the knees and elbows, but the kyphoscoliosis had increased. The condition was now diagnosed as Marfan's syndrome, although there was no sign of cardiovascular or ocular abnormities.

When re-examined at the age of 13 years, he was a tall boy, height 189 cm, with long extremities. His head had an oval shape. A mild retrognathia was present, and the antihelices of both ears were somewhat crumpled. The range of movement in the shoulder was normal, but there was a 10° contracture of both elbows. Pronation/supination of the forearm was normal. He had arachnodactyly, but no finger contractures. His feet were abnormally long, with long slender toes and a slight talipes calcaneovalgus was present. A marked kyphoscoliosis had developed in the thoracic region.

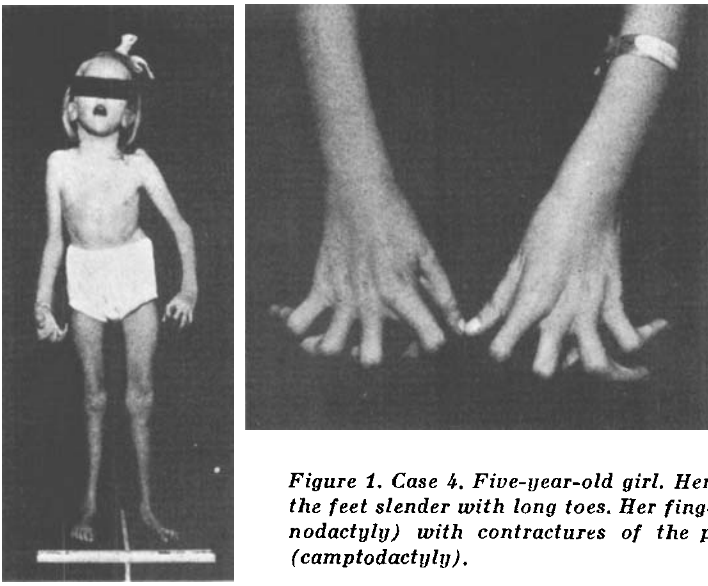

Figure 1. Case 4. Five-year-old girl. Her extremities are long and thin, the feet slender with long toes. Her fingers are long and thin (arachnodactyly) with contractures of the proximal interphalangeal joints (camptodactyly).

No ocular or cardiovascular abnormalities were found. Investigation of his family revealed that the patient's two brothers were normal, but his mother and uncle were both affected and are described below. His grandfather had finger contractures and a moderate deformity of the spine.

Case 2. R.S. (born 6 October, 1939), the mother of T.S., was seen at the hospital at the age of 35 years. In her childhood she had contractures of both elbows. She now had long extremities and long, slender fingers and toes. Slight contractures of the proximal interphalangeal (PIP) joints were present on both hands, but otherwise joint movement was normal. She had a moderate scoliosis of the thoracic spine, and very distinctive antihelices of her ears.

Case 3. K.G. (born 31 July, 1938), R.S.'s brother had been examined at the hospital at the age of 18 years because of low back pain. A follow-up examination was carried out at the age of 37 years. His extremities were long and thin, with long fingers and toes. A flexion contracture of 15° of the PIP joints of the fingers was detected. Moreover, there was a flexion contracture of 10° in the left elbow and 15° in the right. Examination of the spine revealed a slight left-sided scoliosis in the lumbar region. The patient stated that the contractures of the elbows and fingers had been more marked when he was young.

Case 4. W.B. (born 27 February, 1970) was seen in the pediatric department when 5 days old. She was treated in an incubator because of breathing difficulties. Her weight at birth was 3000 grams. She had extraordinarily long fingers and toes and contractures of the knees (40°), elbows and fingers. Both thumbs were lying across the palm of the hand. The feet were deformed, the right in the equinovarus and the left in the calcaneovalgus position. Examination of the hips showed reduced abduction, but no contractures. The condition was diagnosed as arthrogryposis. The foot deformities were treated with repeated plaster casts and the contractures by cautious passive stretching. During the following months the contractures diminished, but a kyphoscoliosis developed. At the age of 4 years the patient was supplied with a Milwaukee brace. On re-examination at age five, the hip, knee and elbow movements were normal. The

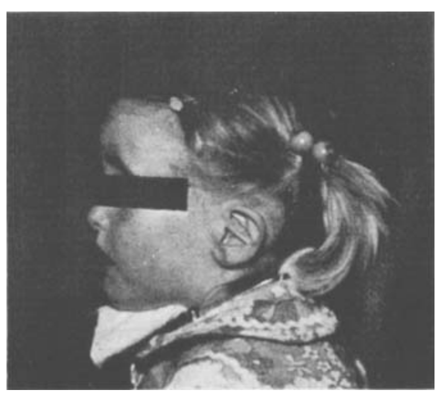

Figure 2. Case 4. The ears are deformed with flattened helix and crumpled antihelix.

extremities were long and thin, the feet slender with long toes, and there was a combination of arachnodactyly and camptodactyly (Figure 1). Her head was oval and her ears positioned lower down than normal with flattened helices and crumpled antihelices (Figure 2). She had developed a severe scoliotic double curve, 85° in the thoracic region and 62° in the lumbar region (Figure 3). No cardiovascular disorder or lens dislocation was found. The family investigation revealed no similar cases. Her parents, a brother and a sister were all healthy.

Case 5 (born 9 April, 1975). The third child of healthy and unrelated parents. He was admitted to our hospital at the age of 7 weeks because of multiple joint contractures and bilateral talipes equinovarus. Shortly after birth he was given the diagnosis arthrogryposis multiplex, and the foot deformities were treated by repeated plaster casts. His head was oval and a slight retrognathia was present. There were marked flexion contractures of both elbows, 90° on the right side and 70° on the left. Supination of the forearms was slightly reduced. The thumb was lying across the palm on both hands, and there was a flexion contracture of about 25° in the PIP joint of the third finger. He had flexion contractures of 15° in both knees and bilateral talipes equinovarus. No cardiovascular or ocular abnormities could be detected. Aminoacid chromatography of urine was normal. During the first 5 months of life the contractures of the fingers and knees disappeared, and the contractures of the elbows became somewhat reduced. His mental development seems to be normal. A sister, born 1972, is healthy, but the parents' first child, a son born in 1971, was stillborn, and had multiple joint contractures and talipes equinovarus. No other cases of malformation in the family could be traced.

Figure 3. Case 4. A severe thoraco-lumbar scoliosis at the age of 5 years, 85° in the thoracic region and 62° in the lumbar region.

DISCUSSION

Five cases of CCA, three belonging to one family, are reported. The pattern of inheritance in CCA appears to be autosomal dominant (Hecht & Beals 1972). The transmission in one of our kindreds may be in agreement with this pattern, whereas in two cases the disorder was present in one generation only. In these patients the disorder may have been caused by a mutant gen.

The contractures which are symmetrical, most often affect the knees, elbows, fingers and hips. The fingers are long with flexion contractures in the PIP joints, and a thumb-in-hand deformity is usual. Ulnar deviation of the hands and fingers is occasionally seen. The feet are long and slender and often talipes equinovarus or talipes calcaneovalgus is present. The toes may be bent. The limbs are long with poorly developed musculature. Scoliosis or kyphoscoliosis is frequently seen in CCA; this deformity is progressive in contrast to the joint contractures which frequently tend to improve with age. The deformity of the ears is often marked; a flattening of the helix and a crumpled antihelix with prominent crura. Although the clinical signs in CCA are usually distinctive, there may be great variations in the manifestation of the disorder (MacLeod & Fraser 1973). The youngest of our patients showed only some of the characteristic signs.

Congenital contractural arachnodactyly has many features in common with Marfan's syndrome, arthrogryposis multiplex congenita, and more uncommon disorders such as homocystinuria and

Achard's syndrome. Several patients who have been considered to have Marfan's syndrome or arthrogryposis, probably have CCA; even Marfan's original case had no abnormality of the eye or heart (Hecht & Beals 1972). The absence of lens dislocation and cardiovascular involvement usually rule out Marfan's syndrome. On the other hand CCA with congenital heart disease has been reported (Lipson et al. 1974). In arthrogryposis the contractures are more rigid and do not improve spontaneously. In homocystinuria, due to cystathionine synthesis deficiency, joint contractures are rare, the deformities develop after birth and mental retardation is frequent. Chromatography of urine and quantitation of serum aminoacide may be desirable in these patients in order to rule out or confirm the diagnosis of homocystinuria. In Achard's syndrome the fingers are slender and somewhat elongated, but contractures, dolichostenomelia and external ear deformities are not present.

Because CCA has a more favourable prognosis, and also in order that more accurate genetic counselling can be given, it is very important to differentiate this syndrome from Marfan's disease and arthrogryposis multiplex congenita.

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