CONGENITAL CONTRACTURAL ARACHNODACTYLY

REPORT OF A CASE AND OF AN OPERATION FOR KNEE CONTRACTURE

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A patient with typical congenital contractural arachnodactyly followed up from the age of 12 years to the age of 48 is reported. He had spiderlike fingers and toes and marked contractures of the knees, ankles, toes, shoulders, elbows and fingers; the mobility of the hips and wrists was almost normal. Persistent knee contractures prevented him from walking. In his twenties he had an osteotomy of both femora and shortening of the patellar tendons which enabled him to walk for the first time in an upright position without external support. When seen at the age of 48 the range of movement of his knees was 0 to 90°. He was working as a turner and had been doing so for almost 20 years.

In 1968 Epstein et al. described a father and a son with musculoskeletal anomalies, including severe kyphoscoliosis, slender long bones, arachnodactyly, congenital contractures, and abnormally-formed ears. They concluded that, although similar in many respects to Marfan’s syndrome, the condition seemed to represent a distinct genetic entity. For this condition Beals and Hecht (1971) introduced the term congenital contractual arachnodactyly (generally abbreviated to CCA). They identified 12 cases of this disorder in the literature, including a case reported by Marfan in 1896, and described two additional cases (Hecht and Beals 1972). By 1983 35 cases of congenital contractual arachnodactyly had been reported (Meinecke, Schaefer and Passarge 1983).

This paper reports a patient with congenital contractual arachnodactyly who had been followed up by the author from the age of 12 years to 48 years and who had been rendered able to walk at the age of 25 years after operations for knee contractures.

CASE REPORT

A boy aged 12 years was admitted to the Orthopaedic Hospital of the Invalid Foundation in Helsinki in 1947, when the author saw him for the first time. His limbs had been deformed since birth. He had never been able to walk upright and moved around on the floor with his hips and knees flexed, supporting himself with his hands (Fig. 1). There was no history of congenital deformities in relatives and the boy’s intelligence was normal. There were marked contractures of his knees, elbows, fingers, ankles and toes while mobility of his hips and wrists was almost normal. His hands and feet were long and slender, and spiderlike fingers and toes led to the diagnosis of arachnodactyly (Figs 2 to 4).

His most disabling deformity was contracture of the knees (Fig. 5). Webbing of the skin and contracture of the knee flexors prevented extension beyond 60°. Knee and ankle reflexes were normal. The muscles were small but were not paralysed.

The mobility of his elbows had improved gradually after birth, but at the shoulders it was limited by webbing of the skin of the axillae and contracture of latissimus dorsi and pectoralis muscles. There was a thoracolumbar kyphosis but no scoliosis. His ears were flattened at the helix and there was slight crumpling of the antihelix. A marked funnel-chest was present. No abnormality of his heart, aorta or eyes was found. No treatment was suggested in 1947.

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In 1957 he still could not walk in an upright position because of flexion contracture of the knees; indoors he still moved around in the bent position illustrated in Figure 1. His kyphosis had progressed and kyphoscoliosis had developed. Supracondylar osteotomy of the femora was planned to enable him to walk. The right knee was operated on in 1957 (Fig. 6) and the left knee in 1958. The operative technique is described below. A second operation was necessary in 1959 for pseudarthrosis of the left femur. At the age of 25 years the patient was able to walk in an upright position without external support for the first time in his life.

After vocational training, he was able to take up employment as a turner in 1964. He was still employed when last seen in 1983. Examination then showed that the left leg was medially rotated, and that plantarflexion of the ankles was not possible. His right leg was 2.5 cm shorter than the left (Fig. 7). The power of the quadriceps femoris muscle was good on both sides, but the calf muscles were very weak. Despite these residual deformities his gait was not grossly abnormal. There was still contracture of several joints. The range of movement of both knees was 0 to 90°. Although there were osteophytes in the knees (Fig. 8) there had been no pain.

The vital capacity of his lungs was 3.0 litres; his blood count and ECG were normal; and his height was 159 cm. His chromosomes were examined at the Department of Medical Genetics, University of Helsinki, using a high-resolution banding technique on prometaphase chromosomes; the karyotype was normal.

OPERATIVE TECHNIQUE

The purpose of the operations on the knees was to change the pre-operative range of movement of 60°–150° to 0°–90°. This could be expected to make standing, walking and sitting possible.

The distal third of the femur and the patellar tendon were exposed through a lateral incision. The femoral diaphysis was cut transversely about 2 cm proximal to the condyles (Fig. 9); and the synovial membrane of the suprapatellar pouch was detached from the underlying bone. An opening (with the same diameter as the diaphysis) was made in the cortical bone of the anterior aspect of the femur, proximal to the joint surface. About 5 cm of the distal end of the proximal fragment of the femoral diaphysis was sawn off and the proximal end placed in the hole in the condylar fragment (Fig. 10). The knee was then extended to the straight position. When the tibial and femoral shafts were in line the fragments were fixed to each other using a blade plate. The elongated patellar tendon was shortened by kilt-pleating it at the tibial tuberosity (Fig. 10). A plaster spica was applied for three months.
DISCUSSION

When the patient was seen in 1947, and when he was treated for contracture of the knees between 1957 and 1959, it was obvious that he had arachnodactyly, but his condition did not correspond to the so-called Marfan's syndrome—through a series of mistakes, Marfan's name was apparently applied to "the Marfan syndrome" despite the fact that he never reported it (Hecht 1981). However, when Epstein et al. (1968) and Beals and Hecht (1971) defined congenital contractural arachnodactyly it was obvious that our patient belonged to this group; Marfan had first described this condition in 1896.

Although CCA has been found to be a dominantly inherited disorder, at least four other cases have been reported in which no other family member had been affected (Lowry and Guichon 1972; Lipson, Viseskul and Herrmann 1974; Bjerkreim, Skogland and Trygstad 1976; Hernandez, Poznanski and Hensinger 1977).

Congenital contractural arachnodactyly has a more favourable prognosis than Marfan's syndrome (Beals and Hecht 1971) and spontaneous improvement is common. However, the course of contractures in CCA and the severity of the changes can show great variation (Meinecke et al. 1983).

In the original case of Marfan (1896) the contractures did not resolve and the child was never able to walk because of persistent contracture of the knees. Without osteotomy of the femur and shortening of the patellar tendons our patient would also have been confined to a wheelchair. Similar types of osteotomy for contracture of the knee have been described previously by Osgood (1913), Lange (1917) and Albee (1921).

REFERENCES


