A CASE OF BEALS' SYNDROME
(Congenital Contractural Arachnodactyly)

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SUMMARY

Beals' syndrome, also known as congenital contractural arachnodactyly, is a recent recognized disorder characterized by multiple joint contractures, arachnodactyly, dolichostenomelia, camptodactyly, kyphoscoliosis, crumpled appearance of the ears and an autosomal dominant transmision with variable expressivity. Fibrillin- 2 mutations lead to this syndrome and linkage to a gene coding for fibrillin on chromosome 5q 23- 31 has been shown in several kindreds. In this article we report an 11-year-old girl who presents all clinical features of congenital contractural arachnodactyly.

Key words: *Beals syndrome, congenital contractural arachnodactyly*

ÖZET

Beals sendromu (konjenital kontraktürel araknodaktili) eklem kontraktürleri, araknodaktili, dolikostenomeli, kamptodaktili, kifoskolyoz, kulakların buruşuk görünümü ile karakterize, otozomal dominant geçiş gösteren bir sendromdur. Fibrillin-2 mutasyonları bu sendroma neden olmakta ve kromozom 5q23-31 bölgesinde yer alan ve fibrillini kodlayan bir gende mutasyon saptanmaktadır. Bu olgu sunumunda Beals sendromunun tüm klinik bulgularının saptandığı 11 yaşındaki bir kız hasta literatür bilgileri ile tartışılmaktadır.

Anahtar sözcükler: *Beals sendromu, konjenital kontraktürel araknodaktili*

Beals' syndrome (congenital contractural arachnodactyly) is characterized by autosomal dominant inheritance, multiple joint contractures, arachnodactyly, dolichostenomelia, camptodactyly, kyphoscoliosis and crumpled appearance of the ears. It was first described by Beals and Hecht in 1971, who reported two kindreds and reviewed the literature concluding that some of the earlier diagnosed cases of Marfan syndrome were in fact cases of congenital contractural arachnodactyly, including the original Marfan report (1,2). Since then few cases have been reported as Beals' syndrom in literature (3-16). Recently it has been shown that fibrillin-2 mutations lead to this syndrome, and a linkage to a gene coding for fibrillin on chromosome 5q 23- 31 has been demonstrated in several kindreds (17). In this article, we describe an 11 - year - old girl who presents all clinical features of congenital contractural arachnodactyly.

CASE REPORT

An 11-year-old female patient was admitted to the Orthopedics Department of Dokuz Eylül University Hospital with the complaints of thoracic and lumbar scoliosis. She was the third child of healthy unrelated parents. She had one healthy brother and a sister. One male child of the parents had similar clinical features at birth and he died on the first day of life due to an unknown reason. The patient had been operated for surgical correction of pes equinovarus at the age of one and left patellar subluxation at the age of five years. On physical examination the crown-to-heel length was 141 cm and the arm span was 148 cm. She had crumpled appearance

of ears (Figure 1a), right thoracic and left lumbar scoliosis, dolichostenomelia, flexion contractures of the knees and elbows, and pes adductus deformity of the left foot (Figure 1b). She also had arachnodactyly and camptodactyly (Figure 1c). The physical examinations of the eye and the cardiovascular system were normal.

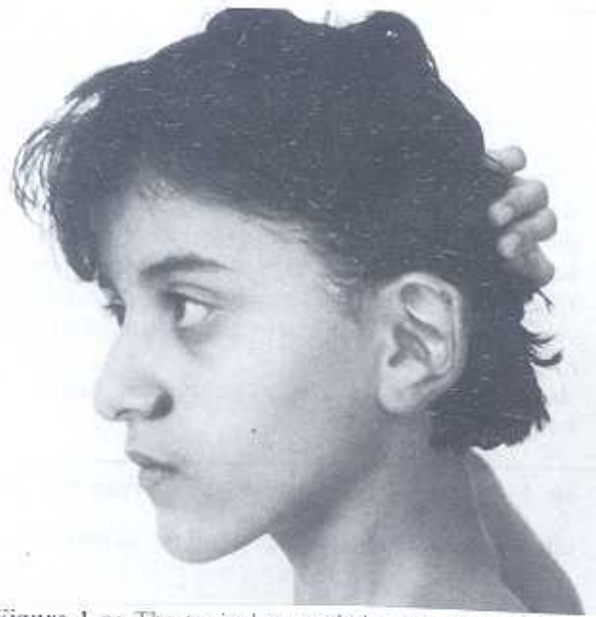

Figure 1 a: The typical crumpled appearance of the ear.

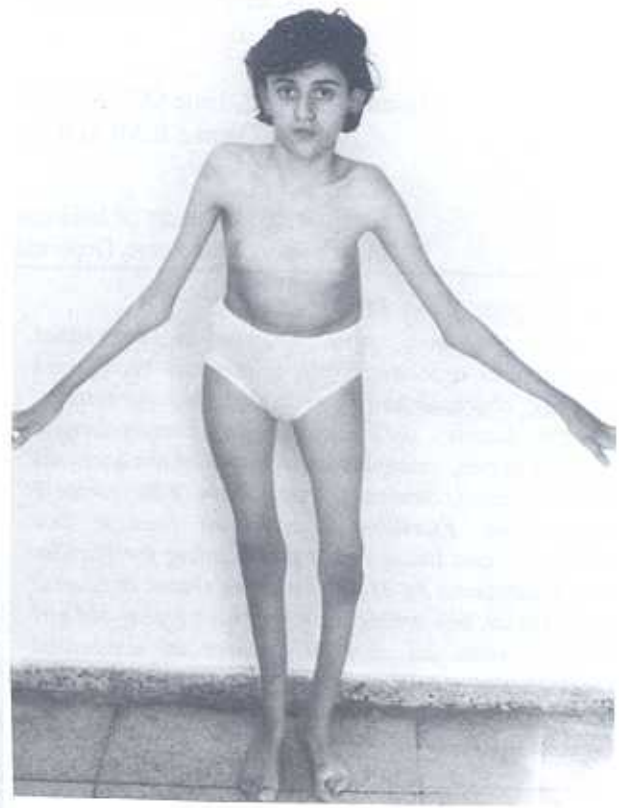

Figure 1 b: General appearance of the patient.

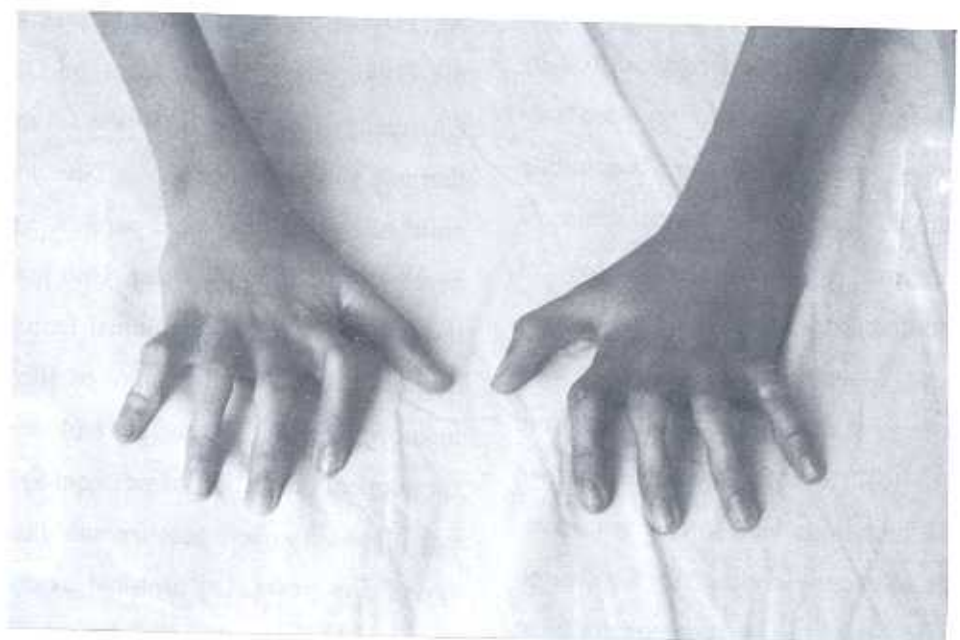

Figure 1c: Bilateral camptodactyly of the fingers and arachnodactyly of the hands. Written permission was obtained from the parents to publish the patient' s photo

Roentgenographic skeletal survey showed severe thoracolumbar scoliosis, left patellar subluxation, arachnodactyly and joint contractures. Chromosome studies and urinary homocystine concentrations were normal. She had no heart defects on echocardiography. Electromyographic findings were in normal limits.

The patient was diagnosed as Beals' syndrome depending on her clinical and laboratory findings. She was discharged from the hospital after the surgical correction of thoracolumbar scoliosis.

DISCUSSION

The most frequent clinical findings of congenital contractural arachnodactyly are multiple contractures at birth that resolve spontaneously (94%), dolicostenomelia, arachnodactyly (85%), camptodactyly (89%), abnormalities of the external ears (65%) and kyphosis/scoliosis (50%) (16). Less frequent clinical features are adducted thumbs, clubfoot deformity, bowed long bones, hypoplasia of calf muscles, unusually shaped head, micrognathia, high arched palate, heart defects and osteoporosis. The joint contractures are congenital, symmetrical and usually maximal at the knees and they improve gradually with age, but kyphoscoliosis may be progressive. The intelligence is generally normal (13-17). In our patient, Beals' syndrome was diagnosed according to the presence of typical clinical findings such as dolichostenomelia, arachnodactyly, camptodactyly, crumpled appearance of ears, right thoracal and left lumbar scoliosis, limited movement of elbows and knees, left patellar subluxation, hypoplasia of calf muscles and left clubfoot.

Differential diagnosis of congenital contractural arachnodactyly includes Marfan syndrome, Achard syndrome, homocystinuria, osteogenesis imperfecta, Stickler's syndrome and distal arthrogryposis. The differential diagnosis in our patient is based upon clinical findings and laboratory results.

Marfan syndrome is the most important condition to differentiate from Beals' syndrome since these two conditions are similar phenotypically. Both are characterized by autosomal dominant inheritance, arachnodactyly and dolichostenomelia, but there are frequently serious ocular and cardiovascular complications which lead to significant morbidity and early death in Marfan syndrome (16). The cardiovascular system is not usually affected in Beals' syndrome but sometimes congenital structural heart defects and mitral valve prolapsus can be detected (14). Significant ocular problems are very rare in Beals' syndrome but eighty percent of induviduals affected with Marfan syndrome have characteristic eye abnormalities including ectopia lentis, severe myopia, retinal detachment, iritis and glaucoma (17,18). Recently, Marfan syndrome and Beals' syndrome have been separated on a molecular basis as two distinct entities. It is identified that genetic cause of this syndrome is a mutation of

fibrillin-2 gene in contrast to fibrillin-1 gene mutation in Marfan syndrome (17). Shortly after the finding of linkage of Marfan syndrome to the fibrillin gene on chromosome 15q 15-21, Lee et al (19) and later, other investigators showed linkage of Beals' syndrome families to a second fibrillin locus on 5q 23-31 (17,20-22). Intragenic heterogeneity within the fibrillin 2 gene is likely to be responsible for the wide phenotypic differences in patients with Beals' syndrome. Mutational analysis could not be done in our patient because of technical insufficiency.

This case of Beals' syndrome is found interesting as a rare occuring malformation and is discussed under the light of recent literature.

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