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A rare presentation of Beals syndrome; a newly recognized connective tissue disorder

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Abstract

Beals syndrome is an autosomal-dominant connective tissue disorder, similar in many respects to Marfan syndrome, characterized by multiple flexion contractures, arachnodactyly, severe kyphoscoliosis, abnormal pinnae, and muscular hypoplasia. This relatively new syndrome contrasted with Marfan by much less incidences of eye and heart anomalies and the congenital presence of contractures. It was found by Beals and Hecht in 1972 when they identified 2 cases of Beals syndrome; it was also thought that the original case described by Marfan in 1896 was actually a case of contractural arachnodactyly rather than a case of Marfan syndrome. Beals syndrome has distinct features, however, and is caused by a mutation in the fibrillin-2 gene (FBN2) in 5q23; Marfan syndrome is caused by mutations in fibrillin-1. We present a case of a patient with Beals syndrome who presented to the emergency department with a history of fractures; and on this presentation with a fracture of the distal tibia and fibula. To our knowledge, this is not documented in the literature and is a characteristic not documented in relation to Beals syndrome.

Keywords

Beals syndrome; Beals-Hecht syndrome; congenital contractural arachnodactyly (CCA); distal arthrogryposis; type 9(2).

Introduction

Beals syndrome, also known as congenital contractural arachnodactyly (CCA), is an autosomal-dominant connective tissue disorder, similar in many respects to Marfan syndrome, characterized by multiple flexion contractures, arachnodactyly, severe kyphoscoliosis, abnormal pinnæ and muscular hypoplasia[1,2]. This relatively new syndrome differs from Marfan syndrome in that there is a much lower incidence of eye and heart anomalies and the congenital presence of contractures. It was described by Beals and Hecht in 1971 when they identified 2 cases[1] and it was also thought that the original case described by Marfan in 1896 was actually a case of CCA rather than a case of Marfan syndrome[1]. Beals syndrome has distinct features, however, and is

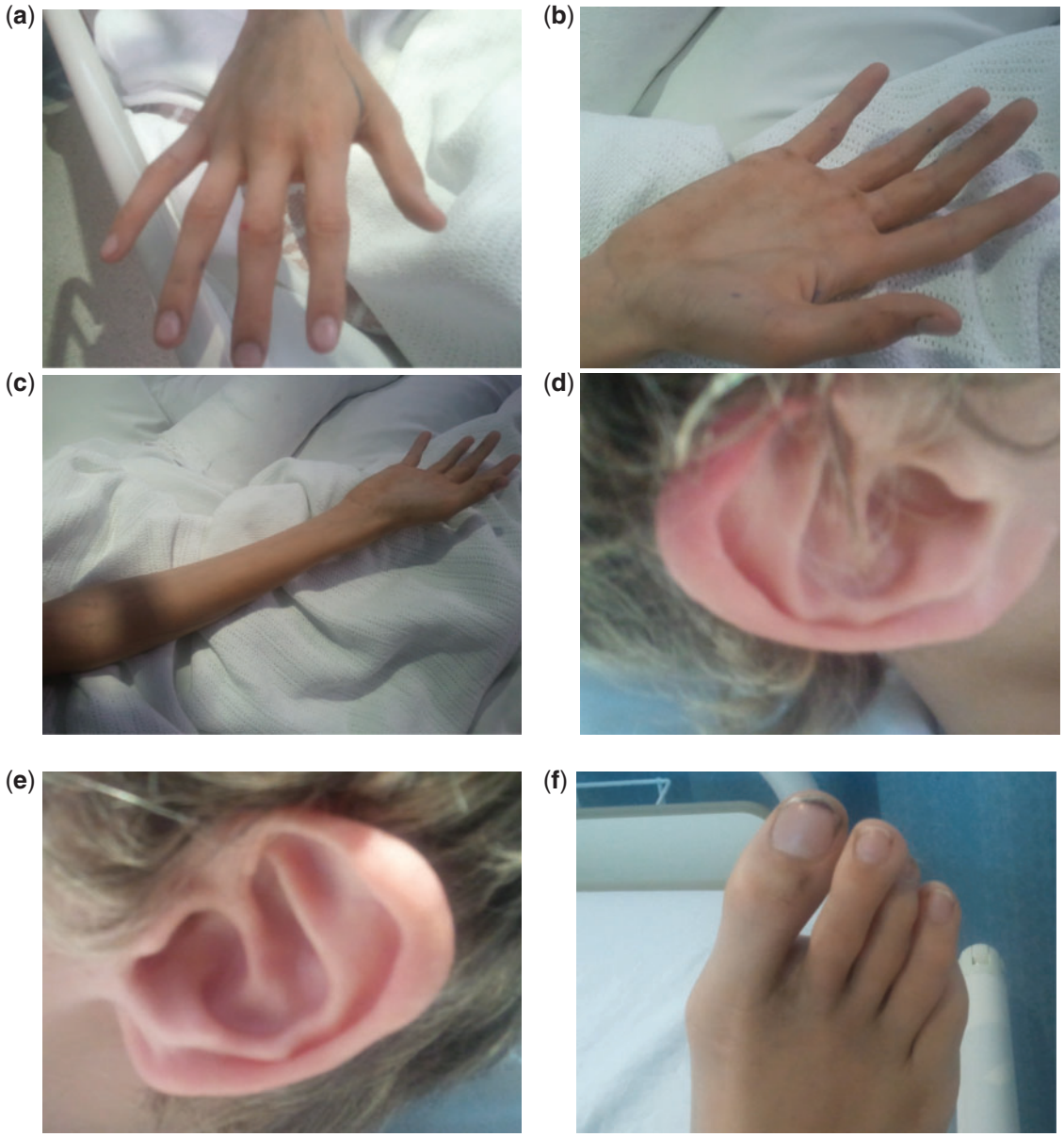

Fig. 1. (a) Right hand (dorsum); (b) right hand (palm); (c) right forearm; (d) right ear; (e) left ear; (f) right toes.

caused by a mutation in the fibrillin-2 gene (FBN2) in 5q23; Marfan syndrome is caused by mutations in fibrillin-1[2–4].

We present a case of a patient with Beals syndrome who presented to the emergency department with a history of fractures; and on this presentation with a fracture of the distal tibia and fibula. To our knowledge this is not documented in the literature and is a characteristic not documented in relation to Beals syndrome.

Case report

We present a case of a 13-year-old boy who has a medical history of Beals syndrome, diagnosed in 2000 at the age of 2 years. He came to his doctor's attention because of stridor and he was also found to have a number of dysmorphic features, which raised the possibility of infantile Marfan syndrome. This was later excluded by a normal echocardiogram and a normal ophthalmologic examination. He was diagnosed formally to have Beals syndrome after review by a geneticist. He has a strong family history of musculoskeletal problems. His mother and maternal grandmother reported fixed flexion deformity of the elbows and changes to the helix of the ear, his maternal great grandfather had knee and arm problems and one of his maternal aunts has had a surgical procedure for scoliosis.

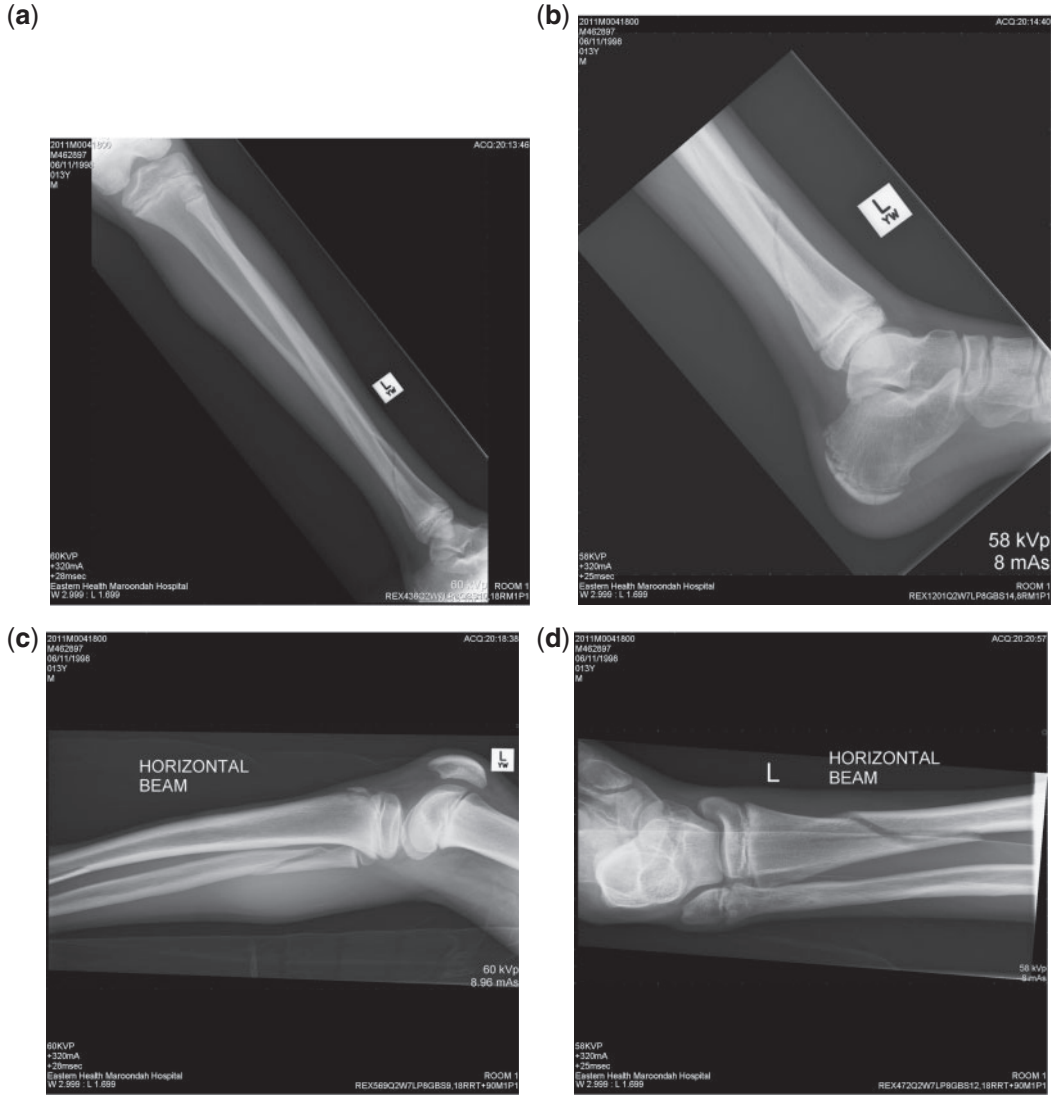

Fig. 2. Radiographs of (a) left tibia and fibula, (b) left distal tibia and fibula showing spiral fracture of distal tibia, (c) left proximal tibia fibula showing spiral fracture of proximal fibula, (d) left distal tibia fibula showing spiral fracture of distal tibia.

He presented to our emergency department after a collision with another player during a football match, resulting in his falling onto his left leg. He does not have a past history of fractures. Clinical examination revealed a tall, thin patient with a Marfanoid appearance, with easily foldable helixes of the ears, limited extension of the elbows and knees and no gross cardiovascular abnormalities. There was no camptodactyly. Physical characteristics are shown in Fig. 1. As a result of this injury he sustained multiple fractures to his left tibia and fibula; the radiographs are shown in Fig. 2.

The patient underwent closed reduction of his fractures in theatre and was discharged home and reviewed in our outpatient clinic. There was good bone alignment on the postoperative radiographs.

Discussion

Although Marfan syndrome has been recognized for more than a century, Beals syndrome has only recently been accepted as a distinct entity because its features are quite similar to Marfan syndrome. After the demonstration that CCA is linked to mutations in FBN2, the 2 syndromes were considered as truly separate entities. The incidence of CCA is unknown and its prevalence is difficult to estimate considering the overlap in phenotype with MFS[2]. The number of patients reported has increased following the identification of a mutation in the FBN2 gene[3]. The

prevalence of CCA in the general population may be estimated with more realistic figures in the near feature.

Joint contractures are the important features of this disorder. These contractures are present at birth and tend to improve spontaneously with age. The most frequently involved joints are those of the fingers, elbows, knees and hips. Kyphoscoliosis, especially of the thoracic spine, is another frequent feature, as noted in our patient. It was initially thought that Beals syndrome, unlike Marfan syndrome, is not associated with ocular or cardiac abnormalities, but some reports have recently shown that serious cardiac abnormalities can occur in CCA[2]. Aortic aneurysm, aortic regurgitation and mitral valve prolapse are the common cardiac anomalies seen in Marfan syndrome[3,4]. However, individuals with CCA are more likely to have structural heart defects such as ventricular and atrial septal defects, and patent ductus arteriosus[2]. Mitral valve prolapse observed in some of these patients caused no significant problems. Crumpled ears are commonly present in these individuals. Eye anomalies and joint laxity are commonly found in Marfan syndrome[2–4], whereas they are very rare in Beals syndrome[2].

An association between Beals syndrome and a predisposition to long bone fracture is not documented in the literature. This is a new finding that may be a characteristic of Beals syndrome. There is a theoretical clinical predisposition to fracture because of the increased connective tissue abnormalities and there is no doubt that the fracture sustained by our patient was an unusual one. There is currently no literature discussing the prevalence or incidence of fracture in patients with Beals syndrome.

Teaching points

- To our knowledge there is no described increased incidence of fractures associated with Beals syndrome
- Our patient had most of the characteristics of the syndrome as denoted above
- Unusually long bones associated with a tall and thin stature would theoretically increase the physical vulnerability to fractures
- Future studies in relation to bone density, bone cortical index may provide an appropriate conclusion in relation to fractures associated with Beals syndrome

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