Congenital Contractural Arachnodactyly (Beals-Hecht Syndrome): A Case Report

Konjenital Kontraktural Araknodaktili (Beals-Hecht Sendromu): Bir Olgu Sunumu

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Abstract

Congenital contractural arachnodactyly (Beals–Hecht syndrome) is an autosomal dominantly inherited connective tissue disorder characterized by crumpled ears, multiple flexion contractures, arachnodactyly, and camptodactyly. It is caused by a mutation in FBN2 gene on chromosome 5q23. We report the case of a 25-day-old male infant with mild crumpled ears, hooked nose, micrognathia, short neck, long slim limbs with arachnodactyly, camptodactyly and joint contractures. *(The Medical Bulletin of Haseki 2011; 49: 88-9)*

Key Words: Beals–Hecht syndrome, arachnodactyly, joint contractures

Özet

Konjenital kontraktural araknodaktili (Beals–Hecht sendromu) buruşuk kulak, çoklu fleksiyon kontraktürleri, araknodaktili ve kamptodaktili ile karakterize otozomal dominant geçişli bir bağ dokusu bozukluğudur. Kromozom 5q23 üzerindeki FBN2 genindeki bir mutasyon ile oluşur. Biz bu yazıda hafif buruşuk kulakları, gaga burunu, mikrognatisi, kısa boyunu, araknodaktilili uzun ince ekstremiteleri, kamptodaktilisi ve eklem kontraktürleri olan 25 günlük erkek bir hastayı sunmayı amaçladık. *(Haseki Tıp Bülteni 2011; 49: 88-9)*

Anahtar Kelimeler: Beals–Hecht sendromu, araknodaktili, eklem kontraktürleri

Introduction

Beals–Hecht syndrome, also known as congenital contractural arachnodactyly (CCA) is an autosomal dominantly inherited connective tissue disorder caused by a mutation in the fibrillin-2 gene (FBN2) in 5q23 (1). It has some similar clinical features with Marfan syndrome (MFS), however, multiple joint contractures and crumpled ears that are characteristic findings of Beals syndrome are rarely seen in MFS, for which aortic root dilatation is specific (2). General ocular complications are estimated to be present in 20% of patients with CCA (3). The typical ophthalmic features are blue sclera, axial myopia, cataract, lens coloboma, cilliary body hyoplasia, and glaucoma, while ectopia lentis is very rare. Here, we present a rare case of a patient with joint contractions and long slim limbs with arachnodactyly.

Case Report

The mother was a 19-year-old gravida 1, abortus 0, married to a 20-year-old man. The parents were nonconsanguineous. The patient was a 25-day-old male infant with weight of 2450g. He was born at a gestational age 39 weeks after a normal pregnancy. The boy had mild crumpled ears, hooked nose, micrognathia, short neck, long slim limbs with arachnodactyly, clenched position of hands, camptodactyly, contractions of the elbows, knees and fingers, and pectus carinatum (Figure 1A-D). Ophthalmologic examination did not show lens displacement. The results of blood and urine examinations were normal. Karyotype analysis was normal. Ultrasound examination revealed no abnormalities in internal organs. Tricuspit insufficiency, mitral insufficiency and patent foramen ovale were detected during echocardiographic examination. Family history was unremarkable.

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Discussion

Congenital contractural arachnodactyly (CCA) was first described by Beals and Hecht in 1971 (4). It is a connective tissue disorder that has autosomal dominant pattern of inheritance and shares common phenotypic features with MFS. However, Beals syndrome has distinct features as it is caused by a mutation in the fibrillin-2 gene (FBN2) in 5q23, while MFS is caused by mutations in fibrillin-1 gene (1). The incidence of CCA is unknown.

Patients with CCA typically have a marfanoid habitus, multiple congenital contractures, camptodactyly, arachnodactyly, kyphoscoliosis, muscular hypoplasia, and external ear malformations (crumpled ears). Although some clinical features are similar to those of MFS, multiple joint contractures and crumpled ears, which are typical findings of Beals syndrome, are rarely observed in MFS (2,5). On the other hand, while aortic dilatation is progressive in MFS, it is mild in CCA (5). Other congenital heart defects have also been reported in cases with CCA, but one of the most-well established cardiac features of the syndrome is mitral regurgitation. There has been increasing number of reports demonstrating cardiovascular involvement in CCA patients with molecularly proven FBN2 mutations (6). Our case had tricuspit insufficiency, mitral insufficiency and patent foramen ovale detected during echocardiographic examination.

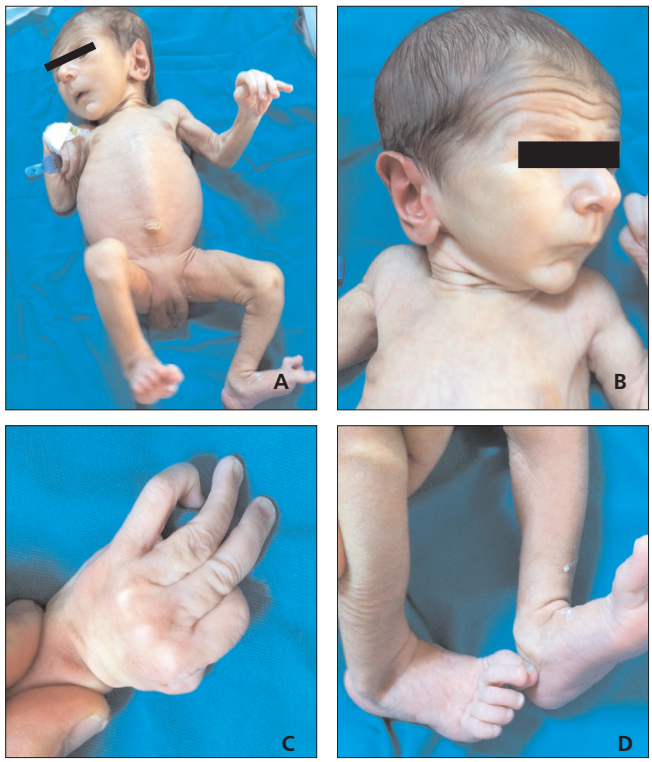

Figure 1: Appearance of the case: Contraction of the elbows, knees and fingers, and pectus carinatum (A). Facial appearance of the case: Hooked nose , mild crumpled ears, note micrognathia and short neck (B). The hand appearance of the case: Long digitis with contractures and clenched position of hands (C). The lower limb appearance of the case (D).

General ocular complications are estimated to be present in 20% of patients with CCA (3). The typical ophthalmic features include blue sclera, axial myopia, cataract, lens coloboma, cilliary body hyoplasia, and glaucoma (3). While ectopia lentis is very rare in Beals syndrome, it is present in approximately half of patients with MFS (5). Our case had no lens displacement on ophthalmologic examination.

Affected infants usually present with clenched position of hands and crumpled ears (a irregular superior helix and prominent antihelix and root of helix), as well as long fingers demonstrated by X-rays as elongation of the phalanges (5-7). Thus, CCA syndrome can be easily identified at birth. In addition, the patients may have pectus carinatum, striae and highly arched palate. All affected children exhibit contractures of varying degrees in the large joints (elbows, knees and fingers). The contractures may be mild and their severity tends to decrease, but camptodactyly always remains present. Our case had mild crumpled ears, hooked nose, micrognathia, short neck, long slim limbs with arachnodactyly, clenched position of hands, camptodactyly, pectus carinatum, contractions of the elbows, knees and fingers.

CCA is an autosomal dominantly inherited disorder with an estimated recurrence rate of 50% (8). Therefore, the non-invasive prenatal diagnosis of CCA by ultrasonography is an important advance for families at risk for this disease. Early diagnosis may help to prepare parents for the birth of an affected child. Ultrasound imaging may be used to demonstrate joint contractures and hypokinesia in suspected cases.

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