

Case Report:

Beals syndrome (Congenital contractural arachnodactyly) with choroid plexus cyst

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Abstract

Beals syndrome or Congenital contractural arachnodactyly (CCA) is an autosomal dominantly inherited connective tissue disorder caused by a mutation in *FBN2* gene on chromosome 5q23 and is characterized by multiple flexion contractures, arachnodactyly, severe kyphoscoliosis, abnormal pinna and muscular hypoplasia. Multiple joint contractures (especially finger joints), and crumpled ears in the absence of significant aortic root dilatation help to differentiate it from Marfan syndrome with which it bears close clinical resemblance. We describe Beals syndrome in a newborn baby with choroid plexus cyst .

Key words: Beals syndrome , Congenital contractural arachnodactyly , Crumpled ears, Marfanoid features, Choroid plexus cyst.

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Introduction

Congenital contractural arachnodactyly(CCA) also known as Beals syndrome is a very rare malformation. The incidence of CCA is unknown and its prevalence is difficult to estimate considering the overlap in phenotype with Marfan syndrome (MFS) [1,2]. Only one case has been described in Indian literature to the best of our knowledge. Choroid plexus cyst has not been reported in this condition earlier.

Case report

A term male baby was born by spontaneous vaginal delivery to a primigravida mother in our hospital with Apgar of 8 and 9 at 1 and 5 minutes of life respectively. He developed seizures at 6 hours of life. The parents were third degree consanguineous and the antenatal period was normal. On examination his limbs were tight and flexed at elbow, wrist, hip, knee and finger joints (Fig. 1a). Baby had an abnormal facies with micrognathia (Fig. 1a) and soft and crumpled pinna (Fig. 1b). Ophthalmological evaluation was normal. He had long fingers and toes (Fig. 1c, 1d) with contractures of joints along with early

contractures of knee joints. The shape of the chest was normal. The muscles of the baby were relatively underdeveloped. The vital parameters and other systemic examinations were within normal limits. . Anthropometry revealed a length of 49 cm, weight of 2.3 Kg, head circumference of 33 cms, upper segment / lower segment ratio of 1.5:1 with arm span of 50 cm.

Initial work up for cause of seizure showed normal septic screen, blood glucose and electrolytes. Baby was symptomatically managed with phenytoin and did not have any further seizures. He was maintained initially on intravenous fluids and by day 2 of admission started on breast feeds. Cranial ultrasound showed evidence of bilateral choroid plexus cyst of 4 mm size with normal brain parenchyma and no evidence of any bleed. Echocardiography of the heart showed situs solitus with normal aortic arch and normal valves, with small PDA and ASD. Ultrasonography of the abdomen was normal. Karyotyping showed 46XY.

Figure 1.a Retrognathia with flexion contractures of fingers, wrist, elbow, hip and Knee joints. b. Crumpled pinna c. Long fingers with contractures and d. Long toes with contractures



Fig. 1

Discussion

Beals syndrome (CCA) is an autosomal dominantly inherited connective tissue disorder that shares phenotypical features with Marfan syndrome [1,2]. Beals syndrome is caused by a mutation in the fibrillin-2 gene (*FBN2*) in 5q23, while Marfan syndrome (MFS) is caused by mutations in fibrillin-1 [3,4]. Even though Marfan syndrome has been recognized for more than a century, Beals syndrome started gaining recognition as a separate entity only recently. The incidence of CCA is unknown and its prevalence is difficult to estimate considering the overlap in phenotype with MFS.

The individuals affected by CCA can be easily identified at birth with long fingers with contractures and crumpled ears with irregular superior helix and prominent antihelix and root of helix. There will be elongation of phalanges on X-rays. Contractures of varying degrees of elbows, knees and fingers are present at birth. The contractures may be mild and tend to reduce in severity, but residual camptodactyly always persists. The arm span exceeds body height but the discrepancy may be underestimated due to contractures of elbows and fingers. The same holds for the lower body portion with knee contractures. The most serious complication in CCA is scoliosis and sometimes kyphoscoliosis mandating surgery [1,2].

Lens subluxation is present in approximately half of patients with MFS but it is very rare in CCA but general ocular complications are estimated to be present in 20% of patients with CCA [5]. Babies with CCA do not have the common cardiovascular complications of Marfan syndrome such as dilatation of aortic root and mitral valve prolapse. Mitral regurgitation is a well established feature of CCA. Other congenital heart defects also have been reported, but aortic dilatation is mild in CCA and stationary [6]. Our case had VSD and PDA. Individuals with CCA are expected to be mentally normal. But they can have motor developmental delay due to presence of contractures.

Children with CCA are managed symptomatically. Camptodactyly and contractures involving other joints may resolve with time, but residual camptodactyly always remains. The growing child should be followed up for deformities in the axial skeleton. Kyphoscoliosis is often present at birth or in early childhood [1,2]. Routine physical examination for spinal deformity and early intervention for scoliosis can prevent morbidity later in life. An initial cardiac evaluation with serial echocardiography is indicated if abnormal findings are present. Although ocular involvement is yet unclear, a thorough ophthalmologic

evaluation is recommended. So far no cases have been reported with Beals syndrome with choroid plexus cyst to the best of our knowledge.

Genetic counseling should be given to parents of a child with CCA as recurrence rate is estimated to be 50% as it is an autosomal dominant disorder. When counseling families with sporadic cases germline mosaicism should always be kept in mind. During the next pregnancy screening can be done by ultrasound by looking for joint contractures and hypokinesia. In suspected cases molecular diagnosis can be made antenatally by detecting mutations in fibrillin 2 gene [3,4].

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