Residência Pediátrica
Publicação Oficial da Sociedade Brasileira de Pediatria
ISSN-Online: 2236-6814

Submitted on: 09/22/2019
Approved on: 11/09/2019

CASE REPORT

Early diagnosis of steochondrodysplasia: a case report

Pâmilly Bruna de Araújo Barzzotto[1], Cristina de Oliveira[1], Luana Jung[1], Malcon Natan Panisson[1], Marília Steffanello de Moraes[1]

Keywords:
Dwarfism,
Genetics,
Osteochondrodysplasias.

Abstract

Skeletal dysplasias or osteochondrodysplasia are a heterogeneous group of conditions which affect bone development. Patients affected by this condition present a growth retardation, developing disproportionate short stature. The intelligence is preserved. This case report describes a 3-month-old male patient with atypical face, appropriate neuropsychomotor development, short stature for his age (z-score between -2 and -3) and disproportion between limbs; associated with early radiological manifestations that fall within the spectrum of osteochondrodysplasia, specifically the multiple epiphyseal dysplasia or pseudoachondroplasia subtypes. The importance of providing an early diagnosis lies in the possibility of following adverse events, such as delay of motor development, spinal stenosis, premature osteoarthritis, limited range of motion, kyphoscoliosis and ligaments limpness.

[1] Hospital de Clínicas de Passo Fundo, Pediatrics - PASSO FUNDO - RS - Brazil.

Correspondence to:
Pâmilly Bruna de Araújo Barzzotto.
Hospital de Clínicas de Passo Fundo. Rua Tiradentes, nº 295, Passo Fundo, RS, Brasil. CEP: 99010-260. E-mail: pamilly.bruna@hotmail.com

DOI: 10.25060/residpediatr-2021.v11n3-199

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INTRODUCTION

Skeletal dysplasias (SD) are a heterogeneous group of conditions that affect bone development, with more than 400 disorders described. Most cases result from genetic defects[1,2]. There are two distinct forms of ED: those with prenatal onset and those with postnatal onset[2]. Prenatal cases are often suspected of fetal ultrasound findings[2]. However, most EDs are suspected during childhood due to manifestations such as short stature, which occurs when linear growth is beyond two standard deviations below the mean, bone deformities, recurrent fractures and abnormal findings on radiographs[2]. Patients with suspected ED require a complete medical history, family history, physical examination, and radiographic studies. Parents should look for short stature, presence of recurrent fractures, limb bowing, retinal detachment, polydactyly and kidney disease[2].

Family history can help identify other similarly affected family members and can suggest specific diagnoses[2]. It is important to establish whether short stature is a result of prenatal or postnatal growth failure. As an example, achondroplasia is easily recognized at birth. However, other conditions, such as pseudoachondroplasia, may not be suspected until 2 to 3 years of age, when the patient has a failure to thrive and knee deformities[2].

OBJECTIVES

To report the case of a male patient with early manifestation of osteochondrodysplasia, most likely of the pseudoachondroplasia or multiple epiphyseal dysplasia subtypes, with clinically and radiographically unaltered parents, highlighting the most prevalent aspects of this condition as well as its clinical repercussions.

CASE REPORT

Male patient, born by cesarean section indicated for acute fetal distress, at 39 weeks and 2 days of gestation. He presented APGAR 3/8, weight of 3,100 kg and 47 cm in length. Mother reports prenatal care without complications. No morphological ultrasound (US) was performed. At 3 months of age, an infant was hospitalized for treatment of acute viral bronchiolitis. At hospital admission, it was noticed during ectoscopy that he had short and disproportionate limbs (figure 1). She has a negative family history for skeletal dysplasias and chronic diseases. Non-consanguineous parents, of medium height. During analysis of neuropsychomotor development, social smile, guttural noises and support of the cephalic pole were observed. Growth curve with adequate height at birth and low at the third month of life (figure 2). During the investigation, a transfontanellar US was performed , without alteration, and a radiography of the skeletal bone inventory, whose radiographic report showed deformities of the rib cage, highlighting the enlargement of the anterior portions of the ribs, hypoplasia of the caudal portions of the iliac bones, thoracolumbar kyphosis, diffuse shortening of the rhizomelic -like limbs , with marked delay in metaphyseal- epiphyseal ossification and enlarged “cup-shaped” metaphyses, with tapering corners, falling within the spectrum of osteochondrodysplasias, more specifically the Multiple Epiphyseal Dysplasia or Pseudochondroplasia subtypes.



Figure 1. Male patient, with short stature and disproportion between the limbs.

After the resolution of the respiratory condition, the patient was discharged from the hospital and was referred for follow-up with physiotherapy and orthopedics services, in order to minimize possible pain and gait alterations; pediatric outpatient clinic of the same institution and to medical genetics for specific diagnosis.

DISCUSSION

Pseudochondroplasia (PSACH), described in 1959, is a generalized bone dysplasia, with a prevalence of 4 cases per 1,000,000 inhabitants[3,4], which presents few signs or symptoms at birth, but its growth and gross motor development are affected. around 2 to 3 years of age[4]. It is caused by heterozygous mutations in the cartilage oligomeric protein (*COMP*)[1,3] gene on chromosome 19 and is closely related to dyspasia. multiple epiphyseal[3,5]. The COMP mutation causes

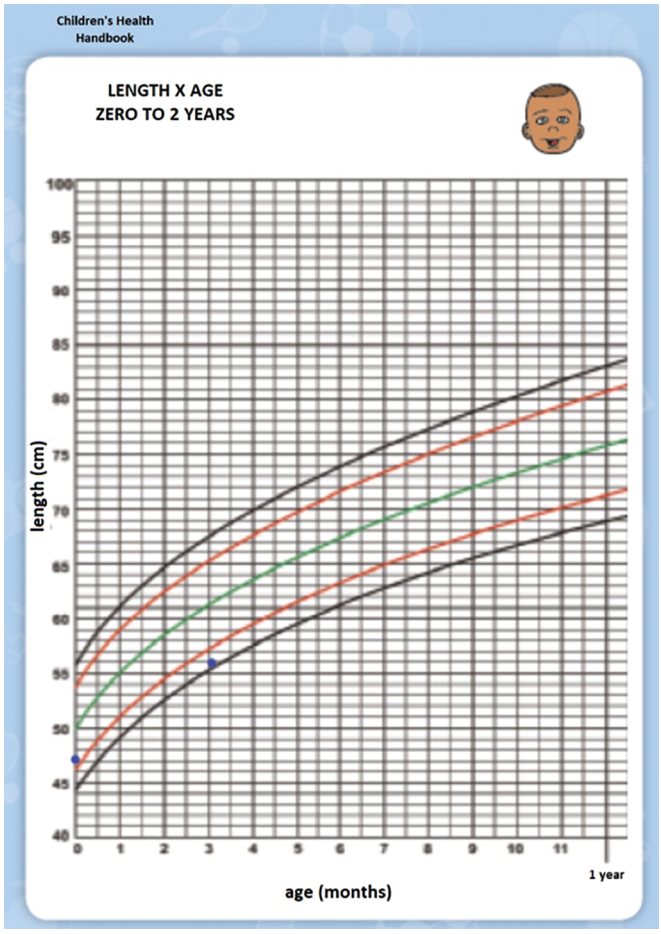


Figure 2. Transition from adequate height for age at birth to short stature at three months of age.

a defect in the cartilage protein matrix, interfering with the stratification of proteins and leading to their accumulation in the endoplasmic reticulum, causing intense oxidative stress, inflammation and premature cell death of chondrocytes in the growth plate, which corroborates a decrease in viable bone matrix cells and a reduction in their size[6].

These patients have atypical facies and normal intelligence, however, they develop disproportionate short stature, gait disturbance, prominent knees and kyphoscoliosis[1,3]. Generally, the height of the affected patient is limited to 90 and 145 cm as an adult[4]. The most debilitating complication of the disorder is joint pain since childhood, the extent and severity of which are poorly defined[7]. According to studies, 47% of patients report that pain was the initial symptom of PSACH presentation, and that it mainly affects weight-bearing joints, such as knees, hips and back[7]. Radiographic features are typical of rhizomelic dwarfism and include oval vertebral bodies, anterior osteophyte formation, platyspondyly, odontoid dysplasia, disc enlargement, short long bones with enlarged metaphyses, small, irregular and enlarged epiphyses with developmental delay[3] and brachydactyly[1].

The diagnosis of PSACH is mainly based on personal and family history and characteristic physical and radiographic findings[5]. The finding of decreased serum concentration of COMP can be an additional diagnostic marker, and whenever possible, genetic verification of the disorder should be performed[5]. Systematic trials with the therapeutic use of growth hormone (GH) in skeletal dysplasias are scarce, but the relationship between the length of the trunk and the legs does not seem to depend on the degree of GH deficiency, nor does it change with its treatment[8]. Thus, according to studies, the use of GH in these children does not produce benefits and further alters body proportions[6]. The treatment of PSACH is based on physical therapy, management of spinal deformities and corrective orthopedic surgery; the latter should be scheduled after the end of the growing period[5].

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