

Chondrodysplasia Punctata in a Newborn; Rhizomelic Type: A Case Report

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Introduction

Chondrodysplasia punctata refers to a heterogeneous group of conditions which share calcific stippling of cartilage and periarticular soft tissues. Particularly punctate calcification is seen in the heel in infancy. These disorders differ in clinical features, severity, inheritance pattern and radiological features. Various types like rhizomelic type (autosomal recessive), X linked dominant type, X linked recessive and Conradi-Hunerman syndrome are found.

The rhizomelic type: an autosomal recessive disorder of peroxisomal function is usually lethal in infancy and consists of proximal limb shortening, short stature, metaphyseal splaying and cupping, multiple joint contracture, flat facies, cataracts, mental retardation, microcephaly and ichthyosiform rash as occasional abnormality (28%)¹. Spranger et al clearly distinguished the rhizomelic type of chondrodysplasia punctata as a separate entity from the other types. Besides the nine personal cases, Spranger et al were able to find 33 additional cases from the literature².

Punctate calcifications may result in delayed endochondral ossification process, growth deficiency and deformity of the bones involved³. Chondrodysplasia punctata can be diagnosed by ultrasound during antenatal period⁴. An association with fetal ascites and polyhydramnios has been reported⁵. Other causes of calcific epiphyseal stippling include warfarin⁶, phenytoin exposure in pregnancy, several peroxisomal disorders including Zellweger syndrome, Smith Lemli Opitz syndrome⁷, trisomy 18 and 21⁸.

Case Report

A 2 days old male baby, out born, 3rd issue of a non consanguineous parents was delivered by LUCS at 41 weeks of gestation on 19/07/08 at ICMH, Matuail and was referred to the NICU of BSMMU, Dhaka.

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was admitted with the complaints of delayed cry and small size of the body and shortening of 4 limbs.

Mother was 28 years old and was on regular antenatal checkup. She took 2 doses of TT. Her pregnancy was uneventful up to 22 weeks of gestation. Then she developed leaking membrane which stopped spontaneously by taking bed rest. She has history of fall 2 times during 23 weeks and 32 weeks of pregnancy. She also complains of decreased volume of liquor in the last month of pregnancy. She gave no history of medication other than vitamins and iron tablets. She was neither hypertensive nor diabetic. There was no history of fever or rash. Her first issue is alive and healthy. She had one abortion. There is no family history of such deformity of limbs but one of her cousins is mentally retarded.

Clinically the baby was mildly icteric and had flat facies. His OFC was 31 cm which fell below the 3rd centile and length was 40 cm. Thus he was microcephalic and of short stature. His birth weight was 1700 gm. So there was symmetric IUGR. His respiratory rate was 58/min and regular, heart rate was 138 beats/min. Baby was acyanotic. On auscultation, breath sound was normal, there was no added sound. 1st and 2nd heart sounds were normal but systolic murmur was heard over the precordium mostly marked on the left parasternal region. Abdomen was soft, meconium passed and urinary bladder evacuated. Proximal shortening of upper and lower limbs was obvious (Fig.-1). There was mild flexion deformity of all limbs. No cataract or skin rash was found. The provisional diagnosis was Achondroplasia



Fig.-1: Photograph showing short upper limb

with perinatal asphyxia with hypoxic ischaemic encephalopathy (HIE) with symmetric IUGR and low birth weight baby.

Routine laboratory tests were done. Hb% was 16 gm/dL, WBC - 14000/cmm, neutrophil-40%, lymphocyte-50%, monocyte-2%, eosinophil-4% and basophil-4%. Urine R/E was normal, serum bilirubin was 9.5 mg/dL. On radiographic studies we have observed a) symmetrical bilateral proximal shortening of upper and lower limbs (rhizomelic pattern), b) multiple calcification in the epiphyseal cartilage of long bones and ankles, c) metaphyseal splaying, d) punctuate calcification in vertebral bodies, pedicles including the sacrum (Fig-2). Doppler echocardiography revealed small VSD (Ventricular Septal Defect) which was in the membranous part of the septum.

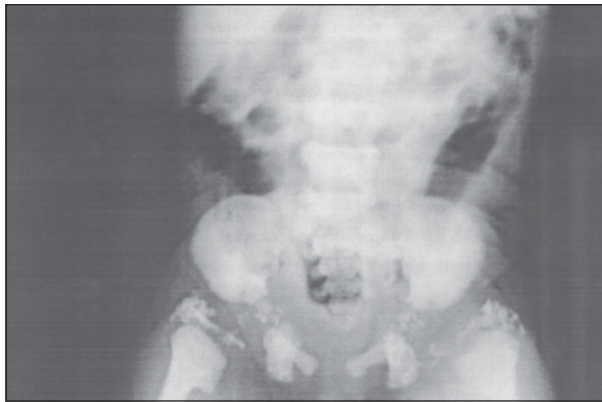


Fig.-2: X-Ray of pelvis showing multiple calcification in the epiphyseal cartilage

Discussion

Chondrodysplasia punctata (CDP) rhizomelic type is a disorder characterized by skeletal manifestation of proximal shortening of upper and lower limbs with short humerus and femur, short stature, microcephaly, flat facies and cataract¹. This condition is associated with a loss of specific peroxisomal function. Peroxisomes are subcellular organelles that play an important role in several metabolic processes. Deficiencies in the peroxisomal enzymes involved in phospholipids synthesis have been found and there is an impairment of phytanic acid oxidation.

The X linked dominant type⁹ thought to be lethal in male, is characterized by asymmetrical skeletal abnormalities with short stature, shortening of long bones, contractures of joints and scoliosis together with alopecia of scalp, abnormal hair and cataracts. An X linked recessive form has been described¹⁰ in

association with a deletion of the terminal short arm of an X chromosome in brothers with epiphyseal stippling, nasal hypoplasia, ichthyosis and mental retardation.

The term Conradi Hunermand syndrome has been applied to an apparently heterogeneous group of conditions whose features include asymmetrical short stature, scoliosis, cataract, ichthyotic rash and flat facies with nasal hypoplasia¹¹. The patients include some with X linked dominant, autosomal dominant¹² and sporadic form¹³.

The autosomal form results from a peroxisomal metabolic disorder^{14,15}, the X linked dominant by defects in cholesterol biosynthesis pathway and the X linked recessive results from defective arylsulphatase E¹⁴.

Today the CDP diagnosis is made by means of a clinical analysis concomitant with biochemical and radiological findings. Currently rhizomelic type of CDP is diagnosed through clinical features compatible with the syndrome and associated biochemical findings including phytanic acid serum level, screening of plasmalogen synthesis on cultured fibroblast¹⁶.

It is important to note that patients with the diagnosis of rhizomelic CDP should undergo ambulatorial follow-up in spite of the current inexistence of specific treatment. Many clinical manifestations, like alopecia, cataract might not be present at the moment of the diagnosis. Other manifestation like punctuate calcifications tend to disappear with aging¹⁶.

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