

**Case Report: Short Limb Dwarfism Rhizomelic
Chondrodysplasia Punctata**

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ABSTRACT

Rhizomelic chondrodysplasia punctata is a rare form of the peroxisomal disorder. It is an autosomal recessive disorder with a distinct clinical phenotype of dwarfism due to symmetrical shortening of the proximal long bones (rhizomelia), cataracts and specific radiological abnormality like punctate epiphyseal calcification (Braverman *et al.*, 2001). A Hindu male baby, born at term (39 weeks+6days) by vaginal delivery, had a weak cry at birth referred to the Neonatal Intensive Care Unit for fast breathing. Baby had proximal shortening of upper limbs and lower limbs. Other dysmorphic features included depressed nasal bridge, broad nose, coarse facial features, long philtrum, and macrostomia. Baby had contractures at thigh and elbow. On ophthalmological examination, there was bilateral megalocornea and near mature cataract.

Keywords: Short Limb Dwarfism Rhizomelic Chondrodysplasia Punctata and PEX7 gene mutation

INTRODUCTION

Rhizomelic chondrodysplasia punctata (RCDP) is one of the rare peroxisomal disorders. It is autosomal recessive and characterized by dwarfism due to symmetrical shortening of long bones (rhizomelia), cataracts, periarticular calcifications, multiple joint contractures, specific radiological abnormalities and psychomotor retardation (Braverman *et al.*, 2001). Specific radiological abnormalities include shortening of the proximal limb bones, stippled foci of calcification within hyaline cartilage, metaphyseal cupping and vertebral bodies having coronal clefts filled with cartilage (Braverman *et al.*, 2001). A biochemical profile is characteristic for different types of peroxisomal disorders and is confirmatory (Phadke *et al.*, 2010). This is a case report of a neonate with characteristic features of RCDP.

Case Report

A male baby, born at term (39 weeks+6days) by normal vaginal delivery, had a weak cry at birth. Baby had fast breathing on Neonatal Intensive Care Unit admission. There was no history of consanguinity. Mother's age was 24 years, and father's was 28 years. There was no history of abortions or exposure to a teratogen during pregnancy. His birth weight was 2100 g (<3rd centile for gestational age), length was 41 cm (<3rd centile) and head circumference was 33.6 cm (50th centile) The upper segment to lower segment ratio was 1.8:1. Baby had proximal shortening of upper limbs and lower limbs. Other dysmorphic features included depressed nasal bridge, broad nose, coarse facial features, long philtrum, and macrostomia. Baby had contractures at thigh and elbow. On ophthalmological examination, there was bilateral megalocornea and near mature cataract. A skeletal survey showed bilateral symmetrical shortening of humerus and femur with Punctate epiphysis due to stippled calcification. Diaphyseal thickening with metaphyseal splaying and fraying were noted. Bilateral acetabular erosion was present. In cervico-thoracic vertebral region, multiple paravertebral calcific foci were noted. Abdominal and cranial ultrasonography was normal. Two-dimensional echocardiography was normal. Biochemical profile and genetic assay could not be done due to financial constraints. Genetic counseling of parents was done.

Discussion

Chondrodysplasia punctata (CDP) is one of the peroxisomal disorders that are genetically determined disorders. They are either due to failure to form or maintain peroxisome or defect in function of a single enzyme that is normally located in peroxisome (Kliegman *et al.*, 2012). CDP is one of the peroxisome import disorders while others are Zellweger syndrome, neonatal

adrenoleukodystrophy, and infantile refsum disease. CDP has four main types autosomal dominant (conradi-Hunermann's type), autosomal recessive (rhizomelic type), X-linked dominant form (Happle) and the X-linked recessive form (Irving *et al.*, 2008). There are three types of RCDP. Type 1 involves PEX7 gene mutation. Type 2 and 3 are phenotypically similar to RCDP type 1 but result from dihydroxyacetone phosphate acyltransferase and alkyldihydroxyacetone phosphate synthase deficiencies, respectively (Steinberg *et al.*, 2006). Our patient had characteristic proximal limb shortening with cataract with joint contractures and physical parameters less than the normal centile values for gestational age. Typical radiological findings further strengthened the diagnosis of RCDP. RCDP is a radiological diagnosis with the specific finding of stippled calcification and shortening of proximal bones with biochemical parameters only confirming it. Biochemical assays which are confirmatory for diagnosis includes plasma phytanic acid levels, which are increased. However, during infancy, plasma phytanic acid levels are normal because the neonate has not ingested phytanic acid and red blood cells' plasmalogen levels, which are decreased. Plasma very long chain fatty acid levels remain normal unlike in Zellweger syndrome and infantile refsum disease. DNA analysis shows PEX7 gene defect (Hoefler *et al.*, 1988). There have been case reports of maternal autoimmune diseases like systemic lupus erythematosus (SLE) and phenylketonuria with CDP babies (Costa *et al.*, 1993). However, in this case, no such association was found.

Author contribution

Sushma, Nandipatti, Mary Chandrika, Aljin V encouraged and supervised the findings of this work. All authors discussed the results and contributed to the final manuscript.

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Conflict of interest: Nil

Study significance

The study is the Rhizomelic chondrodysplasia punctata (RCDP), a rare peroxisomal disorders.

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