Rhizomelic Chondrodysplasia Punctata: A Case Report
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Citation

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Abstract
Rhizomelic Chondrodysplasia Punctata (RCDP), a rare autosomal recessive disorder due to defective peroxisome metabolism, is characterized by symmetrical shortening of the proximal long bones (Rhizomelia), radiological evidence of punctate calcifications in cartilage (chondrodysplasia punctata), congenital cataract, ichthyotic skin changes and dysmorphic facial features. We report a case of neonate with clinical and radiological features suggestive of RCDP.

BACKGROUND
Rhizomelic Chondrodysplasia Punctata (RCDP), is a rare autosomal recessive, peroxisome biogenesis disorder with an estimated incidence of 1 in 100000. There are 3 genetic subtypes based upon biochemical or molecular genetic testing. RCDP type 1 (OMIM 215100), the most common type is caused by mutations in the PEX7 gene, RCDP type 2 (OMIM 222765) and 3 (OMIM 600121) are single enzyme defects in the plasmalogen biosynthesis pathway.1,2

“Classical features of the RCDP are proximal symmetrical shortening of the humerus and to a lesser degree the femur, radiologically evident punctate calcifications in cartilage with epiphyseal and metaphyseal abnormalities (chondrodysplasia punctata), coronal clefts of the vertebral bodies 3, multiple joint contractures, ichthyotic skin changes, cataract, psychomotor retardation and dysmorphic facial features like depressed nasal bridge, broad nose, long philtrum and macrostomia”.1

Diagnosis of is based on clinical and radiologic findings and can be confirmed by molecular analysis.4 Prenatal diagnosis is feasible when the causative mutation has already been identified in the family.5 Specific treatment is not available for the enzyme defect. RCPD has a very poor prognosis with death occurring mainly due to respiratory complications, during the first decade of life.6

We present a case of RCDP with characteristic clinical and radiological features.

CASE REPORT
A full term female neonate was born of 3rd degree consanguineous marriage by spontaneous vaginal delivery to a 25 year old third gravida mother. Baby was admitted at birth with complaints of respiratory distress and abnormal extremities.

Mother was diagnosed hypothyroidism three years back and was receiving L-Thyroxine since then. Mother had spontaneous abortion 3 years back in first trimester and an ectopic pregnancy 2 years back, for which exploratory laparotomy was done. There was no history of teratogen exposure particularly warfarin therapy or alcohol use during pregnancy. Prenatal ultrasonographic assessments reported proximal limb shortening.

On Clinical examination baby weight was 3000 gms (15th - 50th percentile), Head circumference 36 cm (85th – 97th percentile), Length 48 cm (15th - 50th percentile). Baby had proximal symmetrical shortening of the humerus and to a lesser degree the femur with flexion contractures in all extremities, midfacial hypoplasia with a depressed nasal bridge and anteverted nares with a short neck with nuchal fullness and ichthyotic skin changes. (Figure 1)

Ophthalmological examination showed cataract and megalocornea (Figure 2) in both eyes.

Radiographic studies showed symmetrical, bilateral, proximal shortening of upper and lower limbs with multiple punctate calcifications in the epiphyseal cartilage in shoulder, elbow, hip and knee joints. (Figure 3)
Biochemical profile and genetic assay could not be done due to financial constraints. Based on clinical and radiological findings, baby was diagnosed as Rhizomelic Chondrodysplasia Punctata. Baby died on day five of life due to respiratory complications.

**Figure 1**
Depicting Rhizomelia and characteristic facial features

**Figure 2**
Showing megalocornea and cataract

**Figure 3**
Showing Epiphyseal Stippling at Shoulder, Elbow and Hip Joints

**DISCUSSION**

“RCDP Type 1 involves mutations in the PEX7 gene, which encodes enzymes for peroxisome function. RCDP Types 2 and 3 are similar to RCDP Type 1, but result from deficiencies of peroxisomal enzymes dihydroxyacetone phosphate acyltransferase and alkyl dihydroxyacetone phosphate synthase respectively”1. RCDP can be diagnosed through clinical and classical radiological features as described, along with biochemical findings of low levels of plasmalogens in red blood cells, increased plasma levels of phytanic acid, and normal plasma levels of very long chain fatty acids.1

Our patient had characteristic proximal limb shortening with cataract with joint contractures and typical radiological findings supporting the diagnosis of RCDP. Biochemical profile and genetic assay were not done due to financial constraints.7

“Other causes of calcific epiphysial stippling include fetal warfarin syndrome (due to maternal exposure) 8, maternal
autoimmune disease like systemic lupus erythematosus 9 several peroxisomal disorders including Zellweger syndrome spectrum, Smith Lemli Opitz syndrome, Trisomy 18 and 21”1. In our case, there was no history or clinical features suggestive of above mentioned disorders.

Other malformations with RCDP like cleft palate, congenital heart disease 10 and ureteropelvic junction obstruction, were not observed in our case.

Prognosis is very poor, most of the affected children die within first decade of life due to respiratory complications. There is no specific therapy for the gene defect. Management is supportive which includes cataract surgery, seizure control, vision & hearing assessment, growth monitoring, physiotherapy, occupational therapy and genetic counseling.

Genetic counseling is important as this disorder can be diagnosed prenatally during the first trimester by demonstrating the peroxisomal dysfunction in cultured chorialnic villous or amniotic fluid cells and in the second trimester of pregnancy by detecting rhizomelic shortenings of humeri & femur and punctate stippling of epiphysis.

References


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