An Atypical form Rhizomelic Chondrodysplasia Punctata in a Newborn

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ABSTRACT

Rhizomelic Chondrodysplasia punctata (RCDP) is an autosomal recessive metabolic disorder affecting mainly peroxisomal function. We describe a case of RCDP in a 12 days old newborn based on the clinical and radiological ground without any major systemic structural or functional abnormalities.

Key words:
Autosomal recessive disorder, chondrodysplasia punctata, peroxisomal disorder, Rhizomelic chondrodysplasia punctata

INTRODUCTION

RCDP is a lethal inherited disease and is very much rare (incidence 1 in 100000).[1] This is due to deficiency of plasmalogens and deficient activity of the peroxisomal enzyme acyl-CoA dihydroxy-acetone-phosphate acyltransferase (DHAP-AT)[2] and is characterized by symmetric rhizomelic shortening of limbs, dwarfism, foot deformities, bowing of proximal limbs, flat face, microcephaly, micrognathia, cleft palate, ichthyosis, congenital heart disease, seizures, repeated respiratory infections, congenital cataracts, deafness, and joint contracture.[2] The characteristics radiologic findings include symmetric shortening of proximal bones, punctuate epiphyseal calcifications, metaphyseal abnormalities, and coronal clefts in the vertebral bodies.[3]

A 12 days old female neonate born out of non-consanguinous marriage admitted with poor sucking and cough, since last 3 days without any history of fever, respiratory distress, and convulsion. Mother had an uneventful perinatal period and there was no history of any exposure to teratogenic drugs and no family history of any autoimmune or peroxisomal disorder.

On examination, the baby was active alert and pink with normal reflexes and stable vitals, the face was round with full cheeks, hypertelorism, blunt nose with depressed nasal bridge, large forehead and high arched palate without any cleft (5-10%) or ear anomaly.[3] There was no alopecia or icthiosiform dermatitis. Rhizomelic shortening of all four limbs with contructure of both hips and knee joints were noted [Figure 1]. Examination of chest and abdomen was normal. There was a dorsal cleft in the lumber region 3 cm above the anal opening. Opthalmoscopy revealed no cataract or any disk changes.

Anthropometry revealed weight 2 kg (<3rd centile), length 46 cm (~15th centile), head circumference 30 cm (<3rd centile), chest circumference 28 cm, upper segment and lower segment ratio 1.43:1, proximal segment and distal segment ratio 1:2.16 in upper limb and 1:1.37 in the lower limb.

Investigation revealed mild leucocytosis in hemogram. Although specific biochemical work up (RBC plasmalogen,

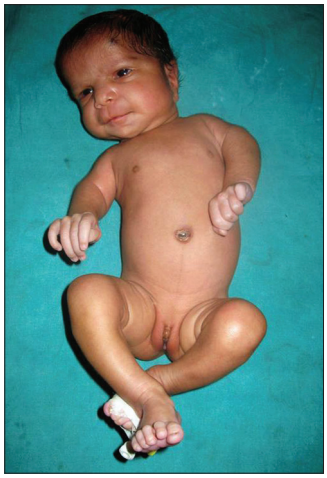

Figure 1: Typical facies of chondrodysplasia punctata along with rhizomelia of upper limbs and contracture of lower limbs (crossed leg position)

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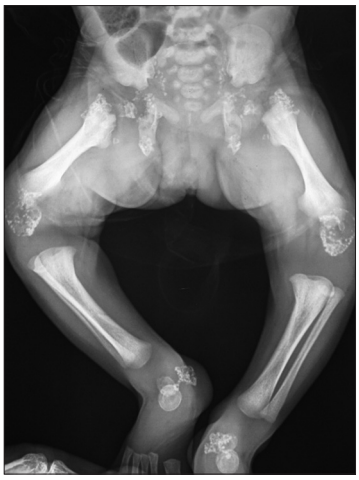

Figure 2: Typical punctuate calcification of epiphyseal region of femur, SI joints, and tarsal bones

phytanic acid level etc.) could not be done, diagnosis is supported by typical radiological changes in X-ray of limbs and vertebra [Figure 2]. Ultrasonography of abdomen, MRI of brain, echocardiography, and organ function tests (e.g., liver, kidney, and lung) including maternal autoimmune workup were found to normal.

Chondrodysplasia punctata is genetic disorder affecting children of every ethnicity and is due to an abnormality at the level of a receptor or transport protein in the peroxisomal membrane resulting abnormal peroxisomal function especially in lipids and hydrogen peroxide metabolism.[1]

Various differential diagnosis excluded are warfarin and phenytoin embryopathy, several peroxisomal disorders, including Zellweger syndrome, Smith Lemli Opitz syndrome, trisomy 18, 21, classical and neonatal Refsum disease, fetal alcohol syndrome, and maternal SLE.[2,3]

The only available treatment for RCDP is supportive. The disease carries a poor prognosis with approximately 60% and 39% cases surviving the first and second year, respectively, very few survive beyond 10 years.[3]

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