Rhizomelic Chondro-Dysplasia Punctate (RCDP)

An 8-month-old boy presented with developmental delay and decreased vision. On examination, a length of 62 cm (-4.44 WHO-Z-score) with an upper: lower segment ratio of 1.81:1, micrognathia, malar hypoplasia, flattened bridge, bulbous nose, rhizomelia, and bilateral cataract were noted (Fig. 1). The clinical diagnosis of skeletal dysplasia was made, and differentials of Zellweger syndrome, rhizomelic short stature with punctate calcification (RCDP), and Marinesco-Sjogren syndrome (MSS) were differentials considered. Zellweger syndrome was ruled out as no characteristic facies, cortical malformation, and MSS as there is no cerebellar atrophy. A skeletal survey showed flared rounded iliac wings, flattened acetabula, bilateral coxa vara, and stippled calcification in epiphysis with metaphyseal flaring (Fig. 2). A novel homozygous variation; c.500A>C (p.Asp167Ala) in exon 4 of the GNPAT gene was detected suggesting RCDP type 2.

RCDP is characterized by rhizomelia and stippled calcifications, and cataracts. Neuroimaging for brain malformation, skeletal survey, phytic acid levels, and exome sequencing aided the diagnosis. We managed the child with multi-disciplinary symptomatic care, cataract extraction, and a phytic acid restricted diet.

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Fig. 1 Clinical photographs of the child showing a) bilateral cataract; b) shortening of bilateral humerus and thighs.

Fig. 2 a) Chest radiograph showing bilateral short humerus (asterisk) with metaphyseal flaring (thin white arrow) and punctate cartilaginous calcifications in epiphysis (thick long white arrow). b) Frontal radiograph of bilateral hip joints, thigh, and knee joints showing bilateral short femurs (asterisk) with distal metaphyseal flaring (thin white arrow). Punctate cartilaginous calcifications in the femoral epiphysis and patella regions (thick long white arrow). Flattening of acetabulum and squaring of iliac bones seen (white arrowhead). c) Lateral radiograph of the thoracic and lumbar spine shows mild coronal clefting of vertebral bodies (thick short white arrows).