A case of rhizomelic chondrodysplasia punctata

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Rhizomelic chondrodysplasia punctata (RCDP) is a rare autosomal recessive syndrome characterized by punctuate calcifications of the epiphyseal cartilage with proximal limb shortening, joint contractures, cataracts, failure to thrive, and severe mental-motor retardation[1].

Case report:

A male infant born at 39 weeks of gestation to unrelated parents had a birth weight of 2300g, a birth length of 41 cm and a head circumference of 31 cm. At birth, facial dysmorphism comprising prominent forehead, upslanting palpebral fissures, broad nasal bridge, long philtrum, thin upper lip, short neck and shortened proximal limbs of all four extremities were noted (figure 1).

Figure 1: *Rhizomelic chondrodysplasia punctata*

Limb radiographs showed shortening of humeri and femora, multiple punctate calcific stippling of joints in shoulders, elbows (figure 2) ribs, hips, knees (figure 3) and posterior segments of vertebra with coronal fissures of cervical and thoracic vertebral bodies.

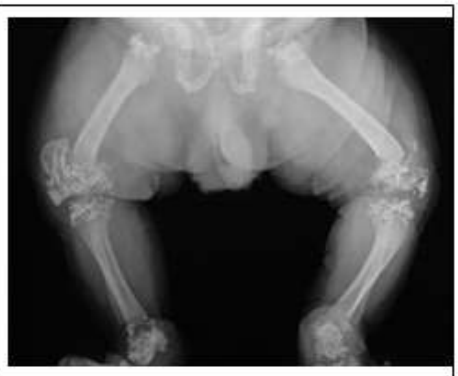

Figure 2: *Multiple punctate calcific stippling around lower limb joints*

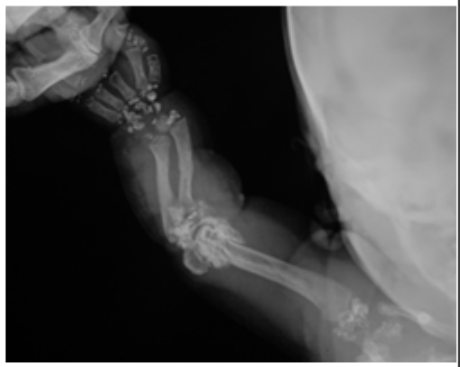

Figure 3: Stippled calcifications around joints of upper limb

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