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# Rhizomelic chondrodysplasia punctata: a classic 'spot' diagnosis

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## DESCRIPTION

Figures 1 and 2 are radiographs from a neonate admitted to our neonatal intensive care unit. He was delivered by ventouse, at term, requiring no resuscitation. Detailed antenatal ultrasound had shown disproportionately shortened humeri with indistinct metaphyses suggesting a significant skeletal dysplasia.

Examination confirmed proximally shortened limbs, hip and knee contractures, bilateral lens opacities and dysmorphic facial features (upslanting palpebral fissures, prominent philtrum, depressed nasal bridge). A chest radiograph (figure 1) showed short humeri with broad metaphyses, with punctuate calcification in and around the joints and vertebrae. Skeletal survey confirmed sagittal and coronal vertebral clefting, short long-bones with broad metaphyses, extensive punctuate calcification and absence of pubic ossification (figure 2).

These appearances are typical of rhizomelic chondrodysplasia punctata (RCDP). This condition (incidence <1:100 000)[1] is due to an abnormality of peroxisome biosynthesis. It is inherited in an autosomal recessive manner. Diagnosis is based on the presence of skeletal changes, cataracts (present at birth or developing in infancy) and biochemical changes secondary to peroxisome dysfunction (normal very long chain fatty acids, increased phytanic acid levels and reduced red-cell plasmalogens). The underlying molecular defect is mutation in the PEX7 gene resulting in abnormal peroxisome biosynthesis. Our patient was homozygous for a mutation in this gene (c.875T>A) and his parents were found to be carriers.

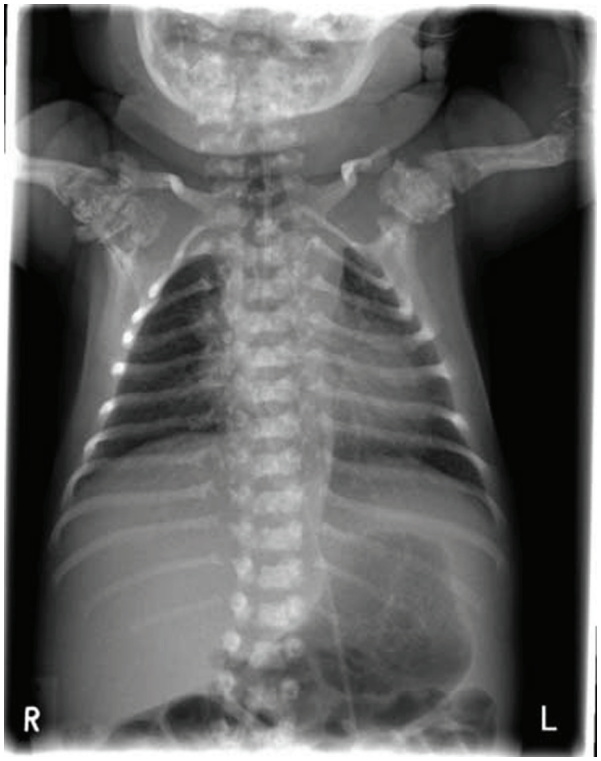


**Figure 1** Chest radiograph showing short humeri with broad epiphyses and marked, punctuate, joint and vertebral calcification.

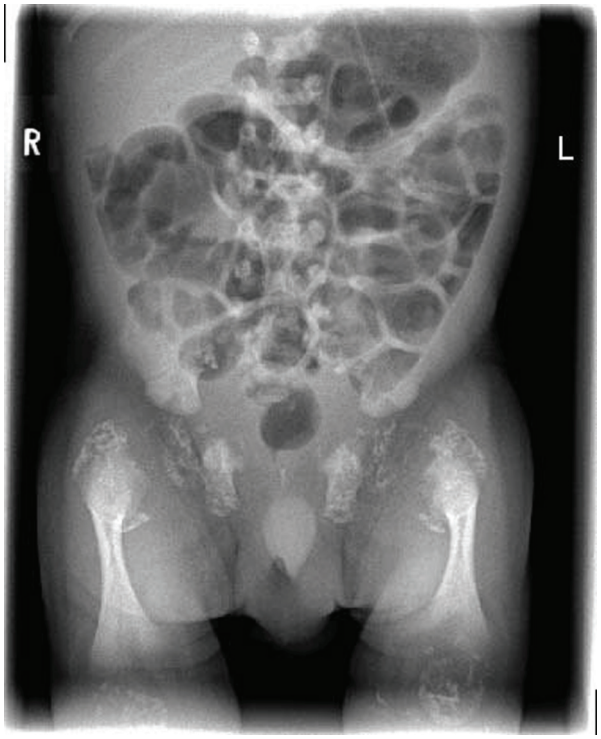


**Figure 2** Abdominal radiograph showing absent pubic ossification and calcification of and around the femoral head.

Affected children have a reduced lifespan (usually from respiratory complications), increased risk of seizures and marked neurodevelopmental delay. The radiographic appearances of this rare condition are characteristic and should alert the neonatologist to the need for targeted biochemical and genetic investigation.

**Competing interests** None.

**Patient consent** Obtained.

## REFERENCES

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