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Dysmorphism and Rhizomelia: Clues to Diagnose a Rare Genetic Disease

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Clinical Image description

A 4-year-old girl was brought with history of global developmental delay, failure to thrive, and generalised tonic seizures and myoclonic jerks from age of 1 year. She had bilateral cataracts aspirated at the age of six months and an atrial septal defect detected at the age of 1 year. On examination, she had microcephaly with severe wasting and stunting. Dysmorphic facies with low set ears, short nasal bridge with upturned nostrils, long philtrum and short neck were noted. Short upper extremities with disproportionately short arms, small chest and contractures at shoulder, knee and ankle were observed (Figure 1a). Neurological examination revealed spasticity, brisk deep tendon reflexes, clonus and extensor plantars. MRI brain at the age of two years showed cerebral atrophy. Early onset cataracts, rhizomelia (proximal shortening of long bones), microcephaly, failure to thrive, neurodevelopmental delay and seizures raised a clinical suspicion of rhizomelic chondrodysplasia punctata (RCDP). Skeletal survey confirmed the same with characteristic epiphyseal stippling of femur and tibia (Figure 1b). EEG of this child showed recurrent high amplitude generalised spike and polyspike discharges followed by generalised voltage attenuation pattern (Figure 1c). Plasma had high phytanic acid and low plasmalogen levels. Clinical, biochemical and radiological features satisfied the diagnosis of RCDP1.

RCDP is a rare autosomal recessive peroxisomal disorder with an estimated incidence of 1 in 100,000 [1]. It has 3 genetic subtypes. RCDP1 is due to a mutation in PEX7 gene and presents as classical or mild form. The classical form is characterised by cataracts, chondrodysplasia punctata with punctate calcifications in cartilage with epiphyseal and metaphyseal abnormalities, rhizomelia, postnatal growth retardation, microcephaly and congenital heart disease [2]. Differential diagnosis of chondrodysplasia punctata are RCDP, Conradi-Hünermann syndrome, spondyloepiphyseal dysplasia, diastrophic dysplasia, fetal warfarin syndrome and fetal alcohol syndrome. Children with RCDP have delayed development, seizures, intellectual disability and majority do not survive beyond the first decade [3].

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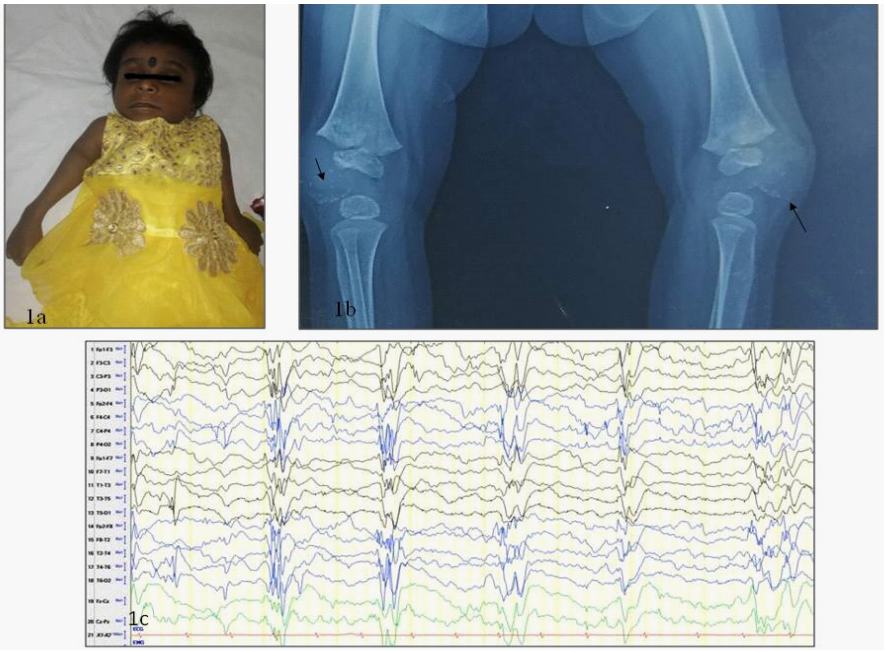

Figure 1: 4-year-old girl with rhizomelic chondrodysplasia punctata type 1
(a) dysmorphic facies with rhizomelia.
(b) Radiograph demonstrating epiphyseal stippling (arrows) of femur.
(c) EEG showing recurrent bursts of high amplitude generalised spike and poly-spike discharges with burst attenuation pattern.