Cono-spondylar dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cono-spondylar dysplasia. ORPHA:420794

Cono-spondylar dysplasia is a rare genetic primary bone dysplasia disorder characterized by early-onset severe lumbar kyphosis, marked brachydactyly and irregular, pronounced cone epiphyses of the metacarpals and phalanges. Additional reported features include developmental delay, intellectual disability, hypotonia, epileptic seizures and mild facial dysmorphism (incl. long and thin or square-shaped face, slight mid-face hypoplasia, hypertelorism, epicanthic folds, low-set ears, anteverted nostrils). Radiographic findings also reveal hypoplasia of iliac wings and anterior defect of vertebral bodies.