Holoprosencephaly-craniosynostosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Holoprosencephaly-craniosynostosis syndrome. ORPHA:2163

Holoprosencephaly-craniosynostosis syndrome is a rare developmental defect during embryogenesis syndrome characterized by the association of primary craniosynostosis (usually involving the coronal and metopic sutures) with holoprosencephaly (ranging from alobar to, most commonly, semilobar) and various skeletal anomalies (typically, hand and feet anomalies including fifth digit clinodactyly, hypoplastic phalanges and cone-shaped epiphyses, small vertebral bodies, scoliosis, coxa valga and/or flexion deformities of hips). Craniofacial asymmetry, microcephaly, brachy/plagiocephaly, short stature and psychomotor delay are additional common features.