Epiphyseal dysplasia-hearing loss-dysmorphism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Epiphyseal dysplasia-hearing loss-dysmorphism syndrome. ORPHA:1825

Epiphyseal dysplasia-hearing loss-dysmorphism syndrome is a rare multiple congenital anomalies/dysmorphic syndrome characterized by developmental delay, intellectual disability, short stature, sensorineural hearing impairment, facial dysmorphism (incl. epicanthus, broad, depressed nasal bridge, broad, fleshy nasal tip, mildly anteverted nares, deep nasolabial folds, broad mouth with thin upper lip) and skeletal anomalies (incl. abnormally placed thumbs, brachydactyly, scoliosis, dysplastic carpal bones). Patients also present severe behavior disturbances (aggression, hyperactivity), as well as hypopigmented skin lesions and hypoplastic digital patterns. There have been no further descriptions in the literature since 1992.