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Intellectual disability-polydactyly-uncombable hair syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Intellectual disability-polydactyly-uncombable hair syndrome. ORPHA:3082*

Intellectual disability-polydactyly-uncombable hair syndrome is a multiple congenital anomalies/dysmorphic syndrome characterized by intellectual disability, postaxial polydactyly, phalangeal hypoplasia, 2-3 toe syndactyly, uncombable hair and facial dysmorphism (including frontal bossing, hypotelorism, narrow palpebral fissures, nasal bridge and lips, prominent nasal root, large abnormal ears with prominent antihelix, poorly folded helix, underdeveloped lobule and antitragus, and micrognathia evolving into prognathism). Cryptorchidism, conductive hearing loss and progressive thoracic kyphosis were also reported.