

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych Kozłowski-Krajewska syndrome	Zespół Kozłowskiego i Krajewskiej

Kod ORPHA	Kod OMIM	Kod ICD10
3082	-	Q87.2

Kod ICD11
-

*Źródło

orphanet