

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Epiphyseal dysplasia-deafness-dysmorphism syndrome Zespół Finucane, Kurtz i Scotta Finucane-Kurtz-Scott syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
1825	-	Q87.0

Kod ICD11
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*Źródło

orphanet