

Dysplazja nasadowa - utrata słuchu - dysmorfizm

Kod Orpha: 1825 Kod OMIM:

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

Klasifikacja

Zespół wad
wrodzonych

Synonimy

Epiphyseal dysplasia-deafness-dysmorphism
syndrome
Zespół Finucane, Kurtz i Scotta
Finucane-Kurtz-Scott syndrome

Kod ORPHA

1825

Kod OMIM

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Kod ICD10

Q87.0

Kod ICD11

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[*Źródło](#)

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Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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