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

Klasyfikacja
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

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.