Opis choroby *

Definicja

Orofaciodigital syndrome type 13 is a rare subtype of orofaciodigital syndrome, with sporadic occurrence, characterized by cardiac (mitral and tricuspid valve dysplasia) and neuropsychiatric manifestations (epilepsy, depression), in addition to oral, facial and digital malformations (lingual hamartomas, cleft lip, brachydactyly, clinodactyly, syndactyly of hands and feet). Leukoaraiosis, on brain MRI examination, is also associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Degner syndrome
OFD13
OFD13
Oral-facial-digital syndrome type 13

Kod ORPHA
141330

Kod OMIM
-

Kod ICD10
Q87.0

Kod ICD11
LD25.00

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*Źródło

orphanet