

Opis choroby *

Definicja

Oral-facial-digital syndrome, type 9 is characterized by highly arched palate with bifid tongue and bilateral supernumerary lower canines, hamartomatous tongue, multiple frenula, hypertelorism, telecanthus, strabismus, broad and/or bifid nasal tip, short stature, bifid halluces, forked metatarsal, poly- and syndactyly, mild intellectual deficit and specific retinal abnormalities (bilateral optic disc coloboma and retinal dysplasia with partial detachment).

Dane

Klasyfikacja

Zespół wad wrodzonych OFD9

Synonimy

OFD9

Zespół ustno-twarzowo-palcowy z nieprawidłowościami siatkówki

Oral-facial-digital syndrome type 9

Oral-facial-digital syndrome with retinal abnormalities

Orofaciodigital syndrome with retinal abnormalities

Kod ORPHA

141007

Kod OMIM

258865

Kod ICD10

Q87.0

Kod ICD11

LD25.00

[*Źródło](#)

orphanet