

Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability characterized by short stature, sparse hair, characteristic coarse face, brachydactyly with prominent interphalangeal joints, seizures and intellectual disability. Facial characteristics include triangular shaped face, dense and prominent eyelashes, rounded premaxilla, broad nasal base, thick alae nasi, upturned nasal tip, broad philtrum, thin upper vermillion, thick and everted lower vermillion and wide mouth.

Dane

Klasyfikacja

Zespół wad wrodzonych
Intellectual disability-sparse hair-brachydactyly syndrome
Zespół Nicolaidesa i Baraitsera

Kod ORPHA

3051

Kod OMIM

601358

Kod ICD10

Q87.1

Kod ICD11

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*Źródło

orphanet