

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 vermilion, thick and everted lower vermilion and wide mouth.

Dane

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

Kod ORPHA	Kod OMIM	Kod ICD10
3051	601358	Q87.1

Kod ICD11

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*Źródło

orphanet