Opis choroby *

Definicja

A rare genetic disease characterized by sparse scalp hair, lateral thinning of eyebrows, mild facial dysmorphism (bulbous tip of the nose, long flat philtrum, thin upper lip vermilion, and protruding ears), and skeletal anomalies including cone-shaped phalangeal epiphyses, hip dysplasia, and short stature. Type 3 can be differentiated by the presence of severe brachydactyly due to short metacarpals. Cartilaginous exostoses are not present in both types.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
77258

Kod OMIM
190351

Kod ICD10
Q87.1

Kod ICD11
LD27.0Y

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*Źródło

orphanet