

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

*Źródło

orphanet