Opis choroby *

Definicja

A malformation syndrome that is characterized by facial dysmorphism, severe hypoplasia of the nasal bones and frontal sinuses, ocular involvement, early-onset hearing loss, skeletal and anhidrotic ectodermal anomalies and short stature with spondyloepiphyseal dysplasia and early-onset osteoarthritis.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
560

Kod OMIM
154780

Kod ICD10
Q87.0

Kod ICD11
LD27.0Y

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*Źródło

orphanet