

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

*Źródło

orphanet