

## 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