

## Opis choroby \*

### Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by the association of dysplastic external ears, nail hypoplasia, and variable skeletal malformations, such as hypoplastic or absent fibulae, abnormalities of the scapula, clavicle, and acromioclavicular joint, and talipes equinovarus, among others. Joint contractures and mild facial dysmorphism have also been reported.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

2793

#### Kod OMIM

259780

#### Kod ICD10

Q87.5

#### Kod ICD11

LD27.4

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### \*Źródło

orphanet