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