Opis choroby *

Definicja

Omodysplasia is a rare skeletal dysplasia characterized by severe limb shortening and facial dysmorphism. Two types of omodysplasia have been described: an autosomal recessive or generalized form (also referred to as micromelic dysplasia with dislocation of radius) marked by severe micromelic dwarfism with predominantly rhizomelic shortening of both the upper and lower limbs, and an autosomal dominant form in which stature is normal and shortening is limited to the upper limbs.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2733

Kod OMIM
258315

Kod ICD10
Q78.8

Kod ICD11
LD24.A

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*Źródło

orphanet