Opis choroby *

Definicja

A rare syndrome characterised by severe reduction or absence of the fibula and complex brachydactyly. Less than 30 cases have been described in the literature so far. The syndrome is inherited in an autosomal recessive manner and is caused by mutations in the cartilage-derived morphogenetic protein-1 gene (GDF5).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Du Pan syndrome
Zespół Du Pan

Kod ORPHA
2639

Kod OMIM
228900

Kod ICD10
Q73.8

Kod ICD11
LD26.0

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*Źródło

orphanet