

Opis choroby *

Definicja

A rare, lethal skeletal dysplasia characterized by short limbed dwarfism, osteogenesis imperfecta, and punctate calcification within cartilage. It has been described in less than ten cases.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

85175

Kod OMIM

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Kod ICD10

Q77.3

Kod ICD11

LD24.04

*Źródło

orphanet