

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	Kod OMIM	Kod ICD10
85175	-	Q77.3
Kod ICD11		
LD24.04		

*Źródło

orphanet