Opis choroby *

Definicja

An extremely rare mostly lethal congenital disorder characterized by absence of all four limbs and frequent associated major malformations involving the head, face, eyes, skeleton, heart, lungs, anus, urogenital, and central nervous systems. The syndrome has been described in fewer than 20 patients mainly of middle Eastern descent.

Q87.8

Dane

Klasyfikacja Synonimy

Zespół wad wrodzonych Zimmer phocomelia

Fokomelia Zimmera

Kod ORPHA Kod OMIM Kod ICD10 3301 618021

Kod ICD11 LD2F.1Y

*Źródło

orphanet