

Opis choroby *

Definicja

A rare, genetic, developmental defect with connective tissue involvement syndrome characterized by neonatal cutis laxa, marfanoid habitus with arachnodactyly, pulmonary emphysema, cardiac anomalies, and diaphragmatic hernia. Mild contractures of the elbows, hips, and knees, with bilateral hip dislocation may also be associated. There have been no further descriptions in the literature since 1991.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

171719

Kod OMIM

614100

Kod ICD10

Q87.8

Kod ICD11

LD28.2

*Źródło

orphanet