

Opis choroby *

Definicja

A rare ciliopathy with major skeletal involvement characterized by short ribs and extremely narrow thorax, severely shortened tubular bones with round metaphyseal ends and lateral spikes, and anomalies of multiple organs such as the heart, kidneys, liver, pancreas, intestine, and genitalia, with occasional occurrence of situs inversus totalis. Cleft lip/palate and polydactyly may also be present. The syndrome is fatal prenatally or in the perinatal period.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Short rib-polydactyly syndrome type 3
Zespół krótkie żebro-polidaktylia typu 3

Kod ORPHA

93271

Kod OMIM

615633

Kod ICD10

Q77.2

Kod ICD11

LD24.B0

*Źródło

orphanet