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
Zespół wad wrodzonych

Synonimy
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

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*Źródło

orphanet