Opis choroby *

Definicja

A rare ciliopathy with major skeletal involvement characterized by a hypoplastic thorax with short ribs and protuberant abdomen, micromelia with particularly short tibiae with ovoid configuration, pre- and postaxial polydactyly, brachydactyly, hypoplasia or aplasia of nails, and dysmorphic craniofacial features (such as prominent forehead, low-set and malformed ears, short and flat nose, lobulated tongue, micrognathia, and cleft lip/palate). Additional reported manifestations include urogenital, gastrointestinal, cardiovascular, and cerebral malformations, among others. The condition is fatal in the neonatal period.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Short rib-polydactyly syndrome type 2
Zespół krótkie żebro-polidaktylia typu 2

Kod ORPHA
93269

Kod OMIM
613091

Kod ICD10
Q77.2

Kod ICD11
LD24.B0

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*Źródło

orphanet