

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

Synonimy

Kod ORPHA

93269

Kod OMIM

613091

Kod ICD10

Q77.2

Kod ICD11

LD24.B0

[*Źródło](#)

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