Opis choroby *

Definicja

A rare ciliopathy with major skeletal involvement characterized by short ribs and hypoplastic thorax, small iliac bones, short tubular bones with smooth metaphyseal margins, and bowed radii and ulnae. The tibiae are relatively well tubulated and longer than the fibulae. There is a high frequency of brain defects, while post-axial polydactyly is rare. Additional features may include cleft lip, absence of internal genitalia, and renal, biliary, and pancreatic cysts, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Short rib-polydactyly syndrome type 4
Zespół krótkie żebro-polidaktylia typu 4

Kod ORPHA
93268

Kod OMIM
269860

Kod ICD10
Q77.2

Kod ICD11
LD24.B0

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*Źródło

orphanet