Opis choroby *

Definicja

A rare ciliopathy with major skeletal involvement characterized by short ribs with an extremely narrow thorax, very short limbs, absent or very small fibulae, severe metaphyseal dysplasia of tubular bones, post-axial polydactyly, and defective ossification in the calvaria, vertebrae, pelvis, and bones of the hands and feet. Congenital anomalies of multiple other organs have also been described, such as polycystic kidneys, transposition of the great vessels, and atretic lesions of the gastrointestinal and genitourinary tract. Hydrops fetalis may be observed at an early gestational age.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Short rib-polydactyly syndrome type 1
Zespół krótkie żebro-polidaktylia typu 1

Kod ORPHA
93270

Kod OMIM
613091

Kod ICD10
Q77.2

Kod ICD11
LD24.B0

---

*Źródło

orphanet