

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