Opis choroby *

Definicja

An extremely rare lethal autosomal recessive disorder characterized by massive birth weight, swollen globular body, generalized edema, short limbs, postaxial polydactyly, thick skin, facial dysmorphism (slanted palpebral fissures, hypertelorism, epicanthic folds, dysplastic ears), excessive connective tissue, renal dysplasia, and in some patients, organomegaly, craniosynostosis with acrocephaly, omphalocele, cleft palate, and cryptorchidism. Fewer than 10 cases have been reported to date.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Acrocephalopolydactylous dysplasia
Zespół Elejalde
Elejalde syndrome

Kod ORPHA
221054

Kod OMIM
200995

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet