

Dysplazja kostna typu Larsena - niski wzrost

Kod Orpha: 2370 Kod OMIM: 608545

Opis choroby *

Definicja

Larsen-like osseous dysplasia-short stature syndrome is a rare primary bone dysplasia characterized by a Larsen-like phenotype including multiple, congenital, large joint dislocations, craniofacial abnormalities (i.e. macrocephaly, flat occiput, prominent forehead, hypertelorism, low-set, malformed ears, flat nose, cleft palate), spinal abnormalities, cylindrical fingers, and talipes equinovarus, as well as growth retardation (resulting in short stature) and delayed bone age. Other reported clinical manifestations include severe developmental delay, hypotonia, clinodactyly, congenital heart defect and renal dysplasia.

Dane

Klasyfikacja

Zespół wad
wrodzonych

Kod ORPHA
2370

Kod OMIM
608545

Kod ICD10
Q74.8

Kod ICD11
LD24.E

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl