

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