

# 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

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\*Źródło

orphanet

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie [www.orphanet.pl](http://www.orphanet.pl)