

Opis choroby *

Definicja

Multiple epiphyseal dysplasia, Lowry type is a rare primary bone dysplasia characterized by small, flat epiphyses (esp. the capital femoral epiphyses), rhizomelic shortening of limbs, cleft of secondary palate, micrognathia, mild joint contractures and facial dysmorphism (incl. mildly upward-slanting palpebral fissures, hypertelorism, broad nasal tip). Additionally reported features include scoliosis, genu valgum, mild pectus excavatum, platyspondyly, dislocated radial heads, brachydactyly, hypoplastic fibulae and talipes equinovarus.

Dane

Klasyfikacja

Choroba

Synonimy

Multiple epiphyseal dysplasia with Robin phenotype

Dysplazja wielonasadowa z fenotypem Robina

Kod ORPHA

166016

Kod OMIM

601560

Kod ICD10

Q78.8

Kod ICD11

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*Źródło

orphanet