Opis choroby *

Definicja

Larsen-like syndrome, B3GAT3 type is a rare, genetic, primary bone dysplasia characterized by laxity, dislocations and contractures of the joints, short stature, foot deformities (e.g. clubfeet), broad tips of fingers and toes, short neck, dysmorphic facial features (hypertelorism, downslanting palpebral fissures, upturned nose with anteverted nares, high arched palate) and various cardiac malformations. Severe disease is associated with multiple fractures, osteopenia, arachnodactyly and blue sclerae. A broad spectrum of additional features, including scoliosis, radio-ulnar synostosis, mild developmental delay, and various eye disorders (glaucoma, amblyopia, hyperopia, astigmatism, ptosis), are also reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Multiple joint dislocations-short stature-craniofacial dysmorphism-congenital heart defects syndrome
Wielokrotne przemieszczenia stawów - niski wzrost - dysmorfia czaszkowo-twarzowa - wrodzone wady serca

Kod ORPHA
284139

Kod OMIM
245600

Kod ICD10
Q74.8

Kod ICD11
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*Źródło

orphanet