Larsen-like syndrome, B3GAT3 type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Larsen-like syndrome, B3GAT3 type. ORPHA:284139

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.