Reunion Island Larsen-like syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Reunion Island Larsen-like syndrome. ORPHA:294049

A rare, genetic, congenital disorder of glycosylation characterized by severe, pre- and post-natal short stature, joint hyperlaxity with multiple dislocations (elbows, fingers, hips, knees), and facial dysmorphism (round flat face, high forehead, hypertelorism, prominent bulging eyes with under-eye shadows, hypoplastic midface, microstomia, protruding lips). Other associated features may include cutaneous hyperextensibility, learning difficulties, and ocular abnormalities. Advanced carpal ossification, widened metaphyses, and, occasionally, radioulnar synostosis, scoliosis and a Swedish key appearance of the proximal femora, is observed on imaging.