Emery-Nelson syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Emery-Nelson syndrome. ORPHA:1927

Emery-Nelson syndrome is a rare congenital limb malformation syndrome characterized by facial dysmorphism (high forehead, depressed nasal bridge, long philtrum, flat malar region, high arched palate), short stature and deformities of the hands and feet (small hands/feet, flexion contractures of the first three metacarpophalangeal joints, extension contractures of the thumbs at the interphalangeal joints, clawed toes, mild pes cavus). Additional features include neonatal hypotonia, thin and shiny skin of the hands/feet, ridged nails, dry and coarse hair, mild weakness of the orbicularis oculi muscles and occasional ventricular extrasystoles. Intellectual disability may be present. There have been no further descriptions in the literature since 1970.