

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Hand and foot deformity-flat facies syndrome
	Deformacja ręki i stopy - płaska twarz

Kod ORPHA	Kod OMIM	Kod ICD10
1927	139750	Q87.8

Kod ICD11

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*Źródło

orphanet