

Opis choroby *

Definicja

Neurofaciodigitorenal syndrome is a rare multiple developmental anomalies syndrome characterized by neurological abnormalities (including megalencephaly, hypotonia, intellectual disability, abnormal EEG), dysmorphic facial features (high prominent forehead, grooved nasal tip, ptosis, ear anomalies) and acrorenal defects (such as triphalangism, broad halluces, unilateral renal agenesis). Additionally, intrauterine growth restriction, short stature and congenital heart defects may be associated. There have been no further descriptions in the literature since 1997.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Freire Maia-Pinheiro-Opitz syndrome Zespół Freire Maia, Pinheiro i Opitza

Kod ORPHA	Kod OMIM	Kod ICD10
2673	256690	Q87.8

Kod ICD11

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*Źródło

orphanet