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
Zespół wad wrodzonych

Synonimy
Freire Maia-Pinheiro-Opitz syndrome
Zespół Freire Maia, Pinheiro i Opitza

Kod ORPHA
2673

Kod OMIM
256690

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet