Opis choroby *

Definicja

An extremely rare genetic syndromic intellectual disability described in less than 20 families to date and characterized by total or partial alopecia associated with intellectual deficit. The syndrome can be associated with other anomalies such as seizures, sensorineural hearing loss, delayed psychomotor development, and/or hypertonia.

Dane

Klasyfikacja
Choroba

Synonimy
Perniola-Krajewska-Carnevale syndrome
Zespół Perniola, Krajewskiej i Carnevale

Kod ORPHA
2850

Kod OMIM
613930

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet