

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