Opis choroby *

Definicja

A rare, X-linked syndromic intellectual disability disorder characterized by severe intellectual disability, psychomotor developmental delay, generalized seizures, and psoriasis. Mild craniofacial dysmorphism, such as hypertelorism, broad nasal bridge, anteverted nares, macrostomia, highly arched palate and large ears, is also associated. There have been no further descriptions in the literature since 1988.

Dane

Klasyfikacja
Choroba

Synonimy
Tranebjaerg-Svejgaard syndrome
Zespół Tranebjaerga i Svejgaarda

Kod ORPHA
3052

Kod OMIM
309480

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet