

## 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