

## Opis choroby \*

### Definicja

A rare X-linked syndromic intellectual disability characterized by intellectual deficit, microcephaly, short stature, and ectodermal anomalies (including alopecia, spontaneous formation of bullae without evident trauma, hyper- or hypopigmented maculae, acrocyanosis, and dystrophic nails) in male patients. Additional reported features are short, tapering fingers, ocular anomalies (such as corneal opacities and cataract), and hypogenitalism. There have been no further descriptions in the literature since 1995.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

1867

#### Kod OMIM

302000

#### Kod ICD10

Q81.8

#### Kod ICD11

-

---

### \*Źródło

orphanet