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