## Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by intellectual deficit, growth retardation with short stature, deafness and ophthalmoplegia. Choreoathetosis with muscle spasticity generally appears during childhood.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

**Kod ORPHA**
85285

**Kod OMIM**
312840

**Kod ICD10**
Q87.8

**Kod ICD11**
LD90

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*Źródło

orphanet