

## Opis choroby \*

### Definicja

A rare, syndromic intellectual disability characterized by hypotonia, global developmental delay, limited or absent speech, intellectual disability, macrocephaly, mild dysmorphic features, seizures and autism spectrum disorder. Associated ophthalmologic, heart, skeletal and central nervous system anomalies have been reported.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

457279

#### Kod OMIM

616355

#### Kod ICD10

Q87.8

#### Kod ICD11

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### \*Źródło

orphanet