

## Opis choroby \*

### Definicja

A rare, genetic intellectual disability syndrome characterized by severe global developmental delay with intellectual disability, microcephaly, growth retardation, ocular defects such as congenital cataract, and nevus flammeus simplex on the forehead. Cardiac, urogenital, and skeletal abnormalities, as well as seizures are present in most patients. Dysmorphic craniofacial features include sparse hair, downslanting palpebral fissures, hypertelorism, broad and overhanging nasal tip and short philtrum, among others.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

464738

#### Kod OMIM

616449

#### Kod ICD10

Q87.8

#### Kod ICD11

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### \*Źródło

orphanet