

## Opis choroby \*

### Definicja

A rare neuro-ophthalmological disease characterized by nonprogressive cerebellar ataxia, delayed motor and language development and intellectual disability, in addition to ophthalmological abnormalities (e.g. oculomotor apraxia, strabismus, amblyopia, retinal dystrophy and myopia). Cerebellar cysts, cerebellar dysplasia and cerebellar vermis hypoplasia, seen on magnetic resonance imaging, are also characteristic of the disease.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Poretti-Boltshauser syndrome

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#### Kod ORPHA

370022

#### Kod OMIM

615960

#### Kod ICD10

G11.1

#### Kod ICD11

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#### \*Źródło

orphanet