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
Poretti-Boltshauser syndrome

Kod ORPHA
370022

Kod OMIM
615960

Kod ICD10
G11.1

Kod ICD11
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*Źródło

orphanet