

# Izolowana agenezja mózdzku

Kod Orpha: 1398 Kod OMIM:

## Opis choroby \*

### Definicja

A rare non-syndromic central nervous system malformation characterized by complete or near-complete absence of the cerebellum with a normal sized posterior fossa, possibly accompanied by hypoplasia of the brainstem. The clinical picture is highly variable, but typically includes ataxia, dysarthria, tremor, dysmetria, dysdiadochokinesia, and oculomotor abnormalities, in addition to impaired mental, motor, and language development and intellectual disability.

### Dane

#### Klasyfikacja

Wada morfologiczna

#### Synonimy

Near total absence of cerebellum  
Subtotal absence of cerebellum

#### Kod ORPHA

1398

#### Kod OMIM

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#### Kod ICD10

Q04.3

#### Kod ICD11

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[\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.