

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.