

Izolowana agnezja robaka mózdzku

Kod Orpha: 269203 Kod OMIM:

Opis choroby *

Definicja

A rare, congenital, cerebellar malformation disorder characterized by complete or partial cerebellar vermis agenesis, with no other associated malformations or anomalies. Patients may be asymptomatic, although psychomotor delay, hypotonia and incoordination are usually associated. Additional variable manifestations include intellectual disability, oculomotor abnormalities (such as nystagmus, impaired smooth pursuit, impaired saccades, strabismus, ptosis, and oculomotor apraxia), retinopathy, abnormal visual evoked potentials, ataxia, episodic hyperpnea, and delayed gait acquisition, as well as delayed speech and language development.

Dane

Klasyfikacja

Wada morfologiczna

Kod ORPHA

269203

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.

Dostępna na stronie www.orphanet.pl