

Opis choroby *

Definicja

A rare cerebellar malformation characterized by congenital complete or partial fusion of the cerebellar hemispheres, dentate nuclei, and middle cerebellar peduncles, and complete or partial absence of the vermis. It may occur as an isolated anomaly or together with other malformations of the brain and is associated with variable clinical manifestations including developmental delay, ataxia, dysarthria, oculomotor abnormalities, seizures, and involuntary head movements, among others.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

59315

Kod OMIM

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Kod ICD10

Q04.3

Kod ICD11

LA06.Y

*Źródło

orphanet