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
-

Kod ICD10
Q04.3

Kod ICD11
LA06.Y

---

*Źródło

orphanet