Opis choroby *

Definicja

A rare, non-syndromic, posterior fossa malformation characterized by a cisterna magna that measures above 15 mm in length, 5 mm in height and 20 mm in width (or greater than 10 mm in fetuses) associated with a normal cerebellar vermis and absence of hydrocephalus. The majority of patients are asymptomatic; however, variable neurodevelopmental outcomes, including delayed speech and language development, motor development delay, visiospatial perception difficulties, and attention problems, has been observed in some patients.

Dane

Klasyfikacja

Wada morfologiczna

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 97252
 Q07.8

Kod ICD11

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*Źródło

orphanet