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