Opis choroby *

Definicja

A rare group of neurodegenerative disorders with a prenatal onset characterized by hypoplasia and/or atrophy of the cerebellum and pons. Involvement of supratentorial structures is variable. Multiple forms have been described based on severity, age of onset and clinical presentation.

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
PCH
Atrofia mostowo-móżdżkowa
Hipoplazja mostowo-móżdżkowa
PCH
Pontoneocerebellar atrophy
Pontoneocerebellar hypoplasia

Kod ORPHA
98523

Kod OMIM
-

Kod ICD10
Q04.3

Kod ICD11
LD20.01

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*Źródło

orphanet