Opis choroby *

Definicja

Pontocerebellar hypoplasia type 8 (PCH8) is a novel very rare form of pontocerebellar hypoplasia (see this term) characterized clinically by progressive microencephaly, feeding difficulties, severe developmental delay, although walking may be achieved, hypotonia often associated with increased muscle tone of lower extremities and deep tendon reflexes, joint deformities in the lower extremities, and occasionally complex seizures. PCH8 is caused by a loss-of-function mutation in the CHMP1A gene. MRI demonstrates a pontocerebellar hypoplasia with vermis and hemispheres equally affected and mild to severely reduced cerebral white matter volume with a fully formed very thin corpus callosum.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
PCH8
Hipoplazja mostowo-móżdżkowa z powodu mutacji CHMP1A
PCH8
Pontocerebellar hypoplasia due to CHMP1A mutation

Kod ORPHA
324569

Kod OMIM
614961

Kod ICD10
Q04.3

Kod ICD11
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*Źródło

orphanet