## 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 <i>CHMP1A</i> 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** Synonimy Zespół wad wrodzonych 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** 

-

## \*Źródło

orphanet