Opis choroby *

Definicja

Pontocerebellar hypoplasia type 7 (PCH7) is a novel very rare form of pontocerebellar hypoplasia (see this term) with unknown etiology and poor prognosis reported in four patients and is characterized clinically during the neonatal period by hypotonia, no palpable gonads, micropenis and from infancy by progressive microcephaly, apneic episodes, poor feeding, seizures and regression of penis. MRI demonstrates a pontocerebellar hypoplasia. PCH7 is expressed as PCH with 46,XY disorder of sex development (see this term) in individuals with XY karyotype, and may be expressed as PCH only in individuals with XX karyotype.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
PCH7
Hipoplazja mostowo-móżdżkowa-zaburzenie różnicowania płci o kariotypie 46,XY
PCH7
Pontocerebellar hypoplasia-46,XY disorder of sex development syndrome

Kod ORPHA
284339

Kod OMIM
614969

Kod ICD10
Q04.3

Kod ICD11
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*Źródło

orphanet