Opis choroby *

Definicja

A severe, genetic form of pontocerebellar hypoplasia (PCH) characterized by spinal cord anterior horn cell degeneration in addition to pontocerebellar hypoplasia. Clinically, patients manifest with a severe global development deficit that is evident early on from difficulties in feeding and swallowing

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Norman disease
Choroba Normana
PCH1
PCH1

Kod ORPHA
2254

Kod OMIM
616081

Kod ICD10
Q04.3

Kod ICD11
LD20.01

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*Źródło

orphanet