Opis choroby *

Definicja

X-linked intellectual deficit-cerebellar hypoplasia, also known as OPHN1 syndrome, is a rare syndromic form of cerebellar dysgenesis characterized by moderate to severe intellectual deficit and cerebellar abnormalities.

Dane

Klasyfikacja
Choroba

Synonimy
OPHN1 syndrome
Zespół oligofreniny-1
Zespół OPHN1
Oligophrenin-1 syndrome

Kod ORPHA
137831

Kod OMIM
300486

Kod ICD10
Q04.3

Kod ICD11
LD90

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*Źródło

orphanet