

Opis choroby *

Definicja

Najm type X-linked intellectual deficit is a rare cerebellar dysgenesis syndrome characterized by variable clinical manifestations ranging from mild intellectual deficit with or without congenital nystagmus, to severe cognitive impairment associated with cerebellar and pontine hypoplasia/atrophy and abnormalities of cortical development.

Dane

Klasyfikacja

Choroba

Synonimy

MICPCH

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Niepełnosprawność intelektualna sprzężona z chromosomem X - małogłowie - hipoplazja mostu i mózdzku

X-linked intellectual disability-microcephaly-pontocerebellar hypoplasia syndrome

Kod ORPHA

163937

Kod OMIM

300749

Kod ICD10

Q04.3

Kod ICD11

LD90.Y

*Źródło

orphanet