Opis choroby *

Definicja

A rare central nervous system malformation characterized by an abnormally large brain, accompanied by abnormal head circumference measurements evident at birth or developing over the first years of life. The condition can be unilateral or bilateral and affects males more often than females. There is no typical pattern of symptoms, but mental retardation, seizures, and other neurologic abnormalities have been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Macroencephaly
Macrencephaly

Kod ORPHA
2477

Kod OMIM
248000

Kod ICD10
Q04.5

Kod ICD11
LA05.1

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*Źródło

orphanet