Opis choroby *

Definicja

A rare syndrome with a central nervous system malformation as a major feature characterized by macrocephaly, megalencephaly, bilateral perisylvian polymicrogyria, variable degrees of ventriculomegaly/hydrocephalus, developmental delay and intellectual disability, oromotor dysfunction, hypotonia, seizures, and dysmorphic facial features (such as frontal bossing, low-set ears, a flat nasal bridge, and high-arched palate). Postaxial polydactyly of one or more extremities is also common.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
MPPH syndrome
Zespół MPPH

Kod ORPHA
83473

Kod OMIM
615937

Kod ICD10
Q04.8

Kod ICD11
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*Źródło

orphanet