## 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