

Opis choroby *

Definicja

Macrocephaly-developmental delay syndrome is a rare, intellectual disability syndrome characterized by macrocephaly, mild dysmorphic features (frontal bossing, long face, hooded eye lids with small, downslanting palpebral fissures, broad nasal bridge, and prominent chin), global neurodevelopmental delay, behavioral abnormalities (e.g. anxiety, stereotyped movements) and absence or generalized tonic-clonic seizures. Additional features reported in some patients include craniosynostosis, fifth finger clinodactyly, recurrent pneumonia, and hepatosplenomegaly.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

397612

Kod OMIM

615637

Kod ICD10

Q75.3

Kod ICD11

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*Źródło

orphanet