Opis choroby *

Definicja

Glucose transporter type 1 (GLUT1) deficiency syndrome is characterized by an encephalopathy marked by childhood epilepsy that is refractory to treatment, deceleration of cranial growth leading to microcephaly, psychomotor retardation, spasticity, ataxia, dysarthria and other paroxysmal neurological phenomena often occurring before meals. Symptoms appear between the age of 1 and 4 months, following a normal birth and gestation.

Dane

Klasyfikacja
Choroba

Synonimy
Classic GLUT1 deficiency syndrome
Choroba De Vivo
Glut1-DS
Niedobór transportera glukozy typu 1
Zespół niedoboru Glut-1
Classic GLUT1-DS
De Vivo disease
Encephalopathy due to GLUT1 deficiency

Kod ORPHA
71277

Kod OMIM
606777

Kod ICD10
G40.4

Kod ICD11
5C61.5

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*Źródło

orphanet