

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

[*Źródło](#)

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