

Opis choroby *

Definicja

A rare disorder of gamma-aminobutyric acid (GABA) metabolism characterized by a severe neonatal-infantile epileptic encephalopathy (manifesting with symptoms such as seizures, hypotonia, hyperreflexia and developmental delay) and growth acceleration.

Dane

Klasyfikacja

Choroba

Synonimy

GABA transaminase deficiency

Niedobór transaminazy GABA

Kod ORPHA

2066

Kod OMIM

613163

Kod ICD10

E72.8

Kod ICD11

5C59.1

[*Źródło](#)

orphanet