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