Opis choroby *

Definicja

A rare genetic neurometabolic disease characterized by early neonatal refractory seizures, hypotonia, and respiratory failure. Brain imaging reveals simplified gyral pattern of the frontal lobes, white matter abnormalities, gliosis and volume loss in various brain regions, and vasogenic edema. Serum glutamine levels are significantly elevated. Death occurs within weeks after birth.

Dane

Klasyfikacja
Choroba

Kod ORPHA
557064

Kod OMIM
618328

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet