Opis choroby *

Definicja

A rare neurometabolic disease characterized by neonatal onset of severe epileptic encephalopathy with brain malformations (including cerebral and cerebellar atrophy, white matter abnormalities, delayed gyration or complete agyria, and thin corpus callosum), generalized hypotonia, and lack of normal development. Additional features include facial dysmorphism and necrolytic erythema of the skin. Biochemical hallmarks are decreased levels of glutamine in body fluids and chronic hyperammonemia. Death may occur in the early post-natal period due to multiple organ failure.

Dane

Klasyfikacja
Choroba

Synonimy
Inherited GS deficiency
Dziedziczny Niedobór GS
Dziedziczny Niedobór syntetazy glutaminy
Inherited glutamine synthetase deficiency

Kod ORPHA
71278

Kod OMIM
610015

Kod ICD10
E72.8

Kod ICD11
5C50.Y

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*Źródło

orphanet