

## 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