

Opis choroby *

Definicja

A rare disorder of urea cycle metabolism causing a deficit of ammonia detoxification and arginine synthesis, and characterized by hyperammonemia of variable severity. Manifestations range from neonatal presentation of poor feeding, vomiting, lethargy, tachypnea, convulsions and coma to adult-onset headaches, hazy gastrointestinal symptoms, seizures, behavioral/psychiatric problems, confusion and lethargy.

Dane

Klasyfikacja

Choroba

Synonimy

NAGS deficiency

Niedobór NAGS

Kod ORPHA

927

Kod OMIM

237310

Kod ICD10

E72.2

Kod ICD11

5C50.AY

*Źródło

orphanet