

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	Synonimy
Choroba	NAGS deficiency Niedobór NAGS
Kod ORPHA	Kod OMIM
927	237310
Kod ICD11	Kod ICD10
5C50.AY	E72.2

*Źródło

orphanet