Opis choroby *

Definicja

A rare autosomal recessive amino acid metabolism disorder characterized clinically by variable degrees of hyperammonemia, developing from about 3 years of age, and leading to progressive loss of developmental milestones and spasticity in the absence of treatment.

Dane

Klasyfikacja
Choroba

Synonimy
Arginase deficiency
Hyperargininemia
Niedobór arginazy
Hyperargininemia

Kod ORPHA
90

Kod OMIM
207800

Kod ICD10
E72.2

Kod ICD11
5C50.A2

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*Źródło

orphanet