

## 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

---

#### \*Źródło

orphanet