

Opis choroby *

Definicja

Dicarboxylicaminoaciduria is characterised by infantile-onset hypoglycaemia and hyperprolinaemia associated, in certain cases, with intellectual deficit.

Dane

Klasyfikacja

Choroba

Synonimy

Glutamate-aspartate transport defect

Defekt transportu glutaminianu i aspartanianu

Kod ORPHA

2195

Kod OMIM

222730

Kod ICD10

E72.0

Kod ICD11

5C60.Y

*Źródło

orphanet