

Opis choroby *

Definicja

Beta-ureidopropionase deficiency is a very rare pyrimidine metabolism disorder described in fewer than 10 patients to date with an extremely wide clinical picture ranging from asymptomatic cases to neurological (epilepsy, autism) and developmental disorders (urogenital, colorectal).

Dane

Klasyfikacja

Choroba

Synonimy

Beta-alanine synthase deficiency
Niedobór syntazy beta-alaninowej

Kod ORPHA

65287

Kod OMIM

613161

Kod ICD10

E79.8

Kod ICD11

5C55.1

[*Źródło](#)

orphanet