

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	Synonimy
Choroba	Beta-alanine synthase deficiency Niedobór syntazy beta-alaninowej

Kod ORPHA	Kod OMIM	Kod ICD10
65287	613161	E79.8

Kod ICD11
5C55.1

*Źródło

orphanet