

Opis choroby *

Definicja

Propionic acidemia (PA) is an organic aciduria caused by the deficient activity of the propionyl Coenzyme A carboxylase and is characterized by life threatening episodes of metabolic decompensation, neurological dysfunction and that may be complicated by cardiomyopathy.

Dane

Klasyfikacja	Synonimy
Choroba	Ketotic hyperglycinemia
	Acyduria propionowa
	Hiperglycinemia ketonowa
	Niedobór karboksylazy propionylo-CoA
	Propionic aciduria
	Propionyl-CoA carboxylase deficiency

Kod ORPHA	Kod OMIM	Kod ICD10
35	606054	E71.1

Kod ICD11
5C50.E0

*Źródło

orphanet