

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

Choroba

Synonimy

Ketotic hyperglycinemia

Acyduria propionowa

Hiperglicynemia ketonowa

Niedobór karboksylazy propionyl-CoA

Propionic aciduria

Propionyl-CoA carboxylase deficiency

Kod ORPHA

35

Kod OMIM

606054

Kod ICD10

E71.1

Kod ICD11

5C50.E0

*Źródło

orphanet