

## 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

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#### \*Źródło

orphanet