

Opis choroby *

Definicja

A rare inborn error of amino acid metabolism characterized by elevated blood phenylalanine and low levels or absence of phenylalanine hydroxylase enzyme. If not detected early or left untreated, the disorder manifests with mild to severe mental disability.

Dane

Klasyfikacja

Choroba

Synonimy

PAH deficiency

Niedobór hydroksylazy fenyloalaninowej

Niedobór PAH

PKU

Phenylketonuria

Phenylalanine hydroxylase deficiency

Kod ORPHA

716

Kod OMIM

261600

Kod ICD10

E70.1

Kod ICD11

5C50.0

*Źródło

orphanet