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
PKU
Phenylalanine hydroxylase deficiency

Kod ORPHA
716

Kod OMIM
261600

Kod ICD10
E70.1

Kod ICD11
5C50.0

---

*Źródło

orphanet