Opis choroby *

Definicja

A mild to moderate form of phenylketouria (PKU), an inborn error of amino acid metabolism, characterized by blood phenylalanine concentrations of 600-1,200 micromol/L and manifests with reduced cognitive function and behavioral and developmental disorders. Dietary phenylalanine tolerance is 400-600 mg/day.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Mild PKU
mPKU
PKU łagodna
Wariant fenylketonurii
Wariant PKU
Variant PKU
Variant phenylketonuria
mPKU

Kod ORPHA
79253

Kod OMIM
-

Kod ICD10
E70.1

Kod ICD11
-

---

*Źródło

orphanet