

Opis choroby *

Definicja

A form of phenylketonuria (PKU), an inborn error of amino acid metabolism, characterized by mild to moderate symptoms of PKU including impaired cognitive function, seizures, and behavioral and developmental disorders, and a marked reduction of elevated phenylalanine concentrations after oral loading with tetrahydrobiopterin (BH4), an essential cofactor of phenylalanine hydroxylase.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

BH4-responsive HPA/PKU

Hiperfenyloalaninemia/fenyloketonuria wrażliwa na BH4

HPA/PKU wrażliwa na BH4

HPA/PKU wrażliwa na tetrahydrobiopterynę

BH4-responsive

hyperphenylalaninemia/phenylketonuria

Tetrahydrobiopterin-responsive HPA/PKU

Kod ORPHA

293284

Kod OMIM

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Kod ICD10

E70.1

Kod ICD11

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*Źródło

orphanet