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