Opis choroby *

Definicja

A rare form of phenylketonuria, an inborn error of amino acid metabolism, characterized by blood phenylalanine (Phe) concentrations of 120-600 micromol/L with or without clinical manifestations of impaired cognitive function, and behavioral and developmental disorders.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Mild HPA
Łagodna HPA
mHPA
Non-PKU HPA
Non-PKU HPA
mHPA

Kod ORPHA
79651

Kod OMIM
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Kod ICD10
E70.1

Kod ICD11
5C50.0Y

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*Źródło

orphanet