

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

Kod ORPHA 79651	Kod OMIM -	Kod ICD10 E70.1
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Kod ICD11
5C50.0Y

*Źródło

orphanet