

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Podtyp kliniczny	Mild HPA Łagodna HPA mHPA Non-PKU HPA Non-PKU HPA mHPA

<b>Kod ORPHA</b> 79651	<b>Kod OMIM</b> -	<b>Kod ICD10</b> E70.1
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**Kod ICD11**  
5C50.0Y

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### \*Źródło

orphanet