

## Opis choroby \*

### Definicja

An amino acid disorder with neonatal onset that is clinically characterized by the classic manifestations of phenylketonuria (PKA) and that later on is clinically differentiated by neurologic symptoms such as microcephaly, intellectual disability, central hypotonia, delayed motor development, peripheral spasticity and seizures, that develop and persist despite an established metabolic control of plasma phenylalanine.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Hyperphenylalaninemia due to BH4 deficiency

Hiperfenyloalaninemia nie związana z fenyloketonurią

Hiperfenyloalaninemia z powodu niedoboru BH4

Hiperfenyloalaninemia z powodu niedoboru tetrahydrobiopteryny

Non-phenylketonuric hyperphenylalaninemia

#### Kod ORPHA

238583

#### Kod OMIM

261640

#### Kod ICD10

E70.1

#### Kod ICD11

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#### \*Źródło

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