

## Opis choroby \*

### Definicja

Dihydropteridine reductase (DHPR) deficiency is a severe form of hyperphenylalaninemia (HPA) due to impaired regeneration of tetrahydrobiopterin (BH4) (see this term), leading to decreased levels of neurotransmitters (dopamine, serotonin) and folate in cerebrospinal fluid, and causing neurological symptoms such as psychomotor delay, hypotonia, seizures, abnormal movements, hypersalivation, and swallowing difficulties.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Hyperphenylalaninemia due to dihydropteridine reductase deficiency  
Fenyloketonuria typu 2  
Hiperfenyloalaninemia spowodowana niedoborem reduktazy dihydropterydyny  
PKU typu 2  
PKU type 2  
Phenylketonuria type 2

#### Kod ORPHA

226

#### Kod OMIM

261630

#### Kod ICD10

E70.1

#### Kod ICD11

5C59.01

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#### [\\*Źródło](#)

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