

## Opis choroby \*

### Definicja

6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency is one of the causes of malignant hyperphenylalaninemia due to tetrahydrobiopterin deficiency. Not only does tetrahydrobiopterin deficiency cause hyperphenylalaninemia, it is also responsible for defective neurotransmission of monoamines because of malfunctioning tyrosine and tryptophan hydroxylases, both tetrahydrobiopterin-dependent hydroxylases.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Hyperphenylalaninemia due to 6-pyruvoyltetrahydropterin synthase deficiency  
Hyperphenylalaninemia due to 6-pyruvoyltetrahydropterin synthase deficiency

#### Kod ORPHA

13

#### Kod OMIM

261640

#### Kod ICD10

E70.1

#### Kod ICD11

5C59.01

---

#### [\\*Źródło](#)

orphanet