

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	Synonimy
Podtyp kliniczny	Hyperphenylalaninemia due to 6-pyruvoyltetrahydropterin synthase deficiency Hyperphenylalaninemia due to 6-pyruvoyltetrahydropterin synthase deficiency

Kod ORPHA	Kod OMIM	Kod ICD10
13	261640	E70.1

Kod ICD11
5C59.01

[*Źródło](#)

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