

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