

DISEASE:
6-pyruvoyl-tetrahydropterin synthase deficiency

NAME:	6-pyruvoyl-tetrahydropterin synthase deficiency
DESCRIPTION:	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.
ORPHACODE:	13
SYNONYMS:	Hyperphenylalaninemia due to 6-pyruvoyltetrahydropterin synthase deficiency
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u> <u>MeSH</u>
ANALYTE(S):	<u>PTS</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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- 6-pyruvoyltetrahydropterin synthase

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