

Opis choroby *

Definicja

GTP-cyclohydrolase I deficiency, an autosomal recessive genetic disorder, 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

Klasifikacja	Synonimy
Podtyp kliniczny	GTPCH deficiency Hiperfenylalaninemia spowodowana niedoborem cyklohydrolazy GTP Niedobór GTPCH Hyperphenylalaninemia due to GTP cyclohydrolase deficiency

Kod ORPHA	Kod OMIM	Kod ICD10
2102	233910	E70.1

Kod ICD11
5C59.01

*Źródło

orphanet