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

Klasyfikacja
Podtyp kliniczny

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

Kod ORPHA
2102

Kod OMIM
233910

Kod ICD10
E70.1

Kod ICD11
5C59.01

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*Źródło

orphanet