# DISEASE:
# GTP cyclohydrolase I deficiency

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| **NAME:** | GTP cyclohydrolase I deficiency |
| **DESCRIPTION:** | 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. |
| **ORPHACODE:** | 2102 |
| **SYNONYMS:** | GTPCH deficiency<br>Hyperphenylalaninemia due to GTP cyclohydrolase deficiency |
| **XREF(S):** | Orphanet<br>OMIM<br>ICD-10 |
| **ANALYTE(S):** | GCH1 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Parkinson (gene panel)
- Segawa syndrome (GCH1 gene)

## Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- GTP cyclohydrolase 1

## Related Gene Panels

- Parkinson (18 genes) - KUL