GTP Cyclohydrolase I Deficiency

National Cancer Institute

Source

National Cancer Institute. GTP Cyclohydrolase I Deficiency. NCI Thesaurus. Code C141442.

An autosomal recessive condition caused by mutation(s) in the GCH1 gene, encoding GTP cyclohydrolase 1. It is characterized by hyperphenylalaninemia and GTP cyclohydrolase 1-deficient dopa-responsive dystonia.