

Recombinant Full Length Human GCH1 Protein, GST-tagged

Cat. No. GCH1-5170HF **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human GCH1 full-length ORF (AAH25415.1, 1 a.a. - 250 a.a.) recombinant protein with GST-tag at N-terminal.
Species	Human
Source	In Vitro Cell Free System
ProteinLength	250 amino acids
Description	This gene encodes a member of the GTP cyclohydrolase family. The encoded protein is the first and rate-limiting enzyme in tetrahydrobiopterin (BH4) biosynthesis, catalyzing the conversion of GTP into 7,8-dihydroneopterin triphosphate. BH4 is an essential cofactor required by aromatic amino acid hydroxylases as well as nitric oxide synthases. Mutations in this gene are associated with malignant hyperphenylalaninemia and dopa-responsive dystonia. Several alternatively spliced transcript variants encoding different isoforms have been described; however, not all variants give rise to a functional enzyme. [provided by RefSeq]
Molecular Mass	53.24 kDa
AA Sequence	MEKGPVRAPAEKPRGARCSNGFPERDPPRPGPSRPAEKPPRPEAKSAQPADGWK GERPRSEEDNELNLPNLAAAYSSILSSLGENPQRQGLLKTPWRAASAMQFFTKGYQ ETISDVLNDAIFDEDHDEMIVKIDIDMFMSCEHHLVPFVGKVVHIGYLPNKQVLGLSKL ARIVEIYSRRLQVQERLTKQIAVAITEALRPAGVGVVVEATHMCMVMRGVQKMNSKT VTSTMLGVFREDPKTREEFLTLRS

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Applications	Enzyme-linked Immunoabsorbent Assay Western Blot (Recombinant protein) Antibody Production Protein Array
Notes	Best use within three months from the date of receipt of this protein.
Storage	Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.
Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

GENE INFORMATION

Gene Name	GCH1 GTP cyclohydrolase 1 [Homo sapiens]
Official Symbol	GCH1
Synonyms	GCH1; GTP cyclohydrolase 1; dystonia 14, DYT5, DYT14, GCH; dopa responsive dystonia; DYT5a; GTPCH1; GTP-CH-I; dystonia 14; GTP cyclohydrolase I; guanosine 5-triphosphate cyclohydrolase I; GCH; DYT5; DYT14; HPABH4B; GTP-CH-1;
Gene ID	2643
mRNA Refseq	NM_001024070
Protein Refseq	NP_001019241
MIM	600225
UniProt ID	P30793

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