

Recombinant Full Length Human GCH1 Protein

Cat. No. GCH1-183HF **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant full length Human GTP cyclohydrolase 1 with N-terminal proprietary tag. Mol Wt 53.61 kDa inclusive of tag.
Species	Human
Source	In Vitro Cell Free System
Protein Length	251 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.
Form	Liquid
Molecular Mass	53.610kDa inclusive of tags
AA Sequence	MEKGPVRAPAEKPRGARCSNGFPERDPPRPGPSRPAEKPP RPEAKSAQPADGWK GERPRSEEDNELNLPNLAAAYSSILS SLGENPQRQGLLKTPWRAASAMQFFTKGY QETISDVLNDA IFDEDHDEMIVVKDIDMFMSMCEHHLVPFVGKVHIGYLPNK QVLGLS

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KLARIVEIYSRRLQVQERLTQIAVAITEALRPA GVGVVVEATHMCMVMRGVQKMN
SKTVTSTMLGVFREDPKT REEFLTLIRS

Purity Proprietary Purification

Storage Shipped on dry ice. Upon delivery aliquot and store at -80 centigrade. Avoid freeze / thaw cycles.

Storage Buffer pH: 8.00. Constituents:0.79% Tris HCl, 0.31% Glutathione.

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

Gene ID 2643

mRNA Refseq NM_000161

Protein Refseq NP_000152

MIM 600225

UniProt ID P30793

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