Recombinant Human GCH1

Cat. No. GCH1-28280TH **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	Wheat Germ
ProteinLength	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.
Molecular Weight	53.610kDa inclusive of tags
Tissue specificity	In epidermis, expressed predominantly in basal undifferentiated keratinocytes and in some but not all melanocytes (at protein level).
Form	Liquid

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Purity	Proprietary Purification
Storage buffer	pH: 8.00Constituents:0.3% Glutathione, 0.79% Tris HCl
Storage	Shipped on dry ice. Upon delivery aliquot and store at -80oC. Avoid freeze / thaw cycles.
Sequences of amino acids	MEKGPVRAPAEKPRGARCSNGFPERDPPRPGPSRPAEKPP RPEAKSAQPADGWK GERPRSEEDNELNLPNLAAAYSSILS SLGENPQRQGLLKTPWRAASAMQFFTKGY QETISDVLNDA IFDEDHDEMVIVKDIDMFSMCEHHLVPFVGKVHIGYLPNK QVLGLS KLARIVEIYSRRLQVQERLTKQIAVAITEALRPA GVGVVVEATHMCMVMRGVQKMN SKTVTSTMLGVFREDPKT REEFLTLIRS
Sequence Similarities	Belongs to the GTP cyclohydrolase I family.

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

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MIM	600225
Uniprot ID	P30793
Chromosome Location	14q22.1-q22.2
Pathway	Folate biosynthesis, organism-specific biosystem; Folate biosynthesis, conserved biosystem; Metabolic pathways, organism-specific biosystem; Metabolism, organism-specific biosystem; Metabolism of nitric oxide, organism-specific biosystem;
Function	GTP binding; GTP cyclohydrolase I activity; NOT GTP cyclohydrolase I activity; GTP-dependent protein binding; calcium ion binding;

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