Recombinant Human GCH1 Protein, His-tagged

Cat. No. GCH1-801H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human GCH1 fused with His tag at N-terminal was expressed in E. coli.
Species	Human
Source	E.coli
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	25mM Tris, pH8.0, 150 mM NaCl, 10% glycerol, 1 % Sarkosyl.
Molecular Mass	27.7 kDa
Purity	> 80% as determined by SDS-PAGE and Coomassie blue staining
Concentration	>50 ug/mL as determined by microplate BCA method

GENE INFORMATION

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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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