

## 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      | <p>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.</p> |
| 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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|                        |                                                                                                                                                                                                                                   |
|------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Gene Name</b>       | GCH1 GTP cyclohydrolase 1 [ Homo sapiens ]                                                                                                                                                                                        |
| <b>Official Symbol</b> | GCH1                                                                                                                                                                                                                              |
| <b>Synonyms</b>        | 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; |
| <b>Gene ID</b>         | 2643                                                                                                                                                                                                                              |
| <b>mRNA Refseq</b>     | NM_001024070                                                                                                                                                                                                                      |
| <b>Protein Refseq</b>  | NP_001019241                                                                                                                                                                                                                      |
| <b>MIM</b>             | 600225                                                                                                                                                                                                                            |
| <b>UniProt ID</b>      | P30793                                                                                                                                                                                                                            |

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