

# Recombinant Human GCH1

**Cat. No.** GCH1-28283TH    **Lot. No.** (See product label)

## SPECIFICATION

|                           |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
|---------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Product Overview</b>   | Recombinant fragment of Human GTP cyclohydrolase 1 with N-terminal proprietary tag. Mol Wt 35.42 kDa inclusive of tag.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
| <b>Species</b>            | Human                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |
| <b>Source</b>             | Wheat Germ                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
| <b>ProteinLength</b>      | 89 amino acids                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |
| <b>Description</b>        | <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> |
| <b>Molecular Weight</b>   | 35.420kDa inclusive of tags                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            |
| <b>Tissue specificity</b> | In epidermis, expressed predominantly in basal undifferentiated keratinocytes and in some but not all melanocytes (at protein level).                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |
| <b>Form</b>               | Liquid                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |

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|                                 |                                                                                                |
|---------------------------------|------------------------------------------------------------------------------------------------|
| <b>Purity</b>                   | Proprietary Purification                                                                       |
| <b>Storage buffer</b>           | pH: 8.00Constituents:0.3% Glutathione, 0.79% Tris HCl                                          |
| <b>Storage</b>                  | Shipped on dry ice. Upon delivery aliquot and store at -80oC. Avoid freeze / thaw cycles.      |
| <b>Sequences of amino acids</b> | ENPQRQGLLKTPWRAASAMQFFTKGYQETISDVLNDAIFDEDHDEMIVVKDIDMFSM<br>CEHHLVPFVGKVVHIGYLPNKQVLGLSKLARIV |
| <b>Sequence Similarities</b>    | Belongs to the GTP cyclohydrolase I family.                                                    |

## GENE INFORMATION

|                        |                                                                                                      |
|------------------------|------------------------------------------------------------------------------------------------------|
| <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; |
| <b>Gene ID</b>         | 2643                                                                                                 |
| <b>mRNA Refseq</b>     | NM_000161                                                                                            |
| <b>Protein Refseq</b>  | NP_000152                                                                                            |
| <b>MIM</b>             | 600225                                                                                               |
| <b>Uniprot ID</b>      | P30793                                                                                               |

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|----------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Chromosome Location</b> | 14q22.1-q22.2                                                                                                                                                                                                                                  |
| <b>Pathway</b>             | 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; |
| <b>Function</b>            | GTP binding; GTP cyclohydrolase I activity; NOT GTP cyclohydrolase I activity; GTP-dependent protein binding; calcium ion binding;                                                                                                             |

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