

# 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>	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.
<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

 Tel: 1-631-559-9269    1-516-512-3133

 Email: [info@creative-biomart.com](mailto:info@creative-biomart.com)     Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

<b>Purity</b>	Proprietary Purification
<b>Storage buffer</b>	pH: 8.00 Constituents: 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>	ENPQRQGLLKTPWRAASAMQFFTKGYQETISDVLNDAIFDEHDDEMIVVKDIDMFMSM 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

 Tel: 1-631-559-9269 1-516-512-3133

 Email: [info@creative-biomart.com](mailto:info@creative-biomart.com)  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

<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;

 Tel: 1-631-559-9269 1-516-512-3133

 Email: [info@creative-biomart.com](mailto:info@creative-biomart.com)  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA