

Recombinant Human GCH1

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

SPECIFICATION

Product Overview	Recombinant fragment of Human GTP cyclohydrolase 1 with N-terminal proprietary tag. Mol Wt 35.42 kDa inclusive of tag.
Species	Human
Source	Wheat Germ
ProteinLength	89 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	35.420kDa 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

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

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

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

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	ENPQRQGLLKTPWRAASAMQFFTKGYQETISDVLNDAIFDEDHDEMIVVKDIDMFSM CEHHLVPFVGKVVHIGYLPNKQVLGLSKLARIV
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
MIM	600225
Uniprot ID	P30793

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

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

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

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;

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

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

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