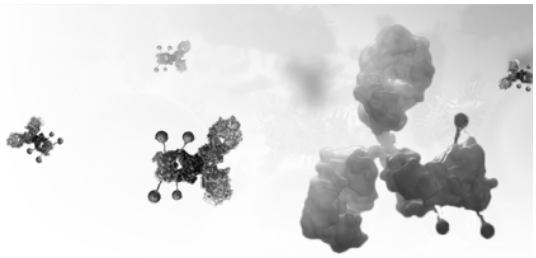

Human GCH1 blocking peptide (CDBP1344)

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

Product Overview	Blocking/Immunizing peptide for anti-GCH1 antibody
Antigen 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.
Species	Human
Conjugate	Unconjugated
Applications	Apuri, BL, ELISA
Format	Lyophilized powder
Size	100 µg
Preservative	None
Storage	Shipped at ambient temperature, store at -20°C.

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;

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	DYT5a; GTPCH1; GTP-CH-I; dystonia 14; GTP cyclohydrolase I; guanosine 5-triphosphate cyclohydrolase I; GCH; DYT5; DYT14; HPABH4B; GTP-CH-1;
Entrez Gene ID	2643
mRNA Refseq	NM_001024070
Protein Refseq	NP_001019241
UniProt ID	P30793
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; Tetrahydrobiopterin (BH4) synthesis, recycling, salvage and regulation, organism-specific biosystem; eNOS activation and regulation, organism-specific biosystem;
Function	GTP binding; GTP cyclohydrolase I activity; NOT GTP cyclohydrolase I activity; GTP-dependent protein binding; calcium ion binding; coenzyme binding; hydrolase activity; nucleotide binding; protein binding; protein homodimerization activity; zinc ion bindi

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