



Anti-GCH1 (internal region) polyclonal antibody (DPAB-DC1303)

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

PRODUCT INFORMATION

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.
Specificity	This antibody is expected to recognize all four isoforms (NP_000152.1; NP_001019195.1; NP_001019241.1; NP_001019242.1).
Immunogen	A synthetic peptide corresponding to internal region of human GCH1. The sequence is C-GKVHIGYLPNKQ
Source/Host	Goat
Species Reactivity	Human
Purification	Antigen affinity purification
Conjugate	Unconjugated
Applications	WB (Tissue lysate), ELISA,
Format	Liquid
Concentration	0.5 mg/mL
Size	100 µg
Buffer	In Tris saline, pH 7.3 (0.5% BSA, 0.02% sodium azide)

Preservative	0.02% Sodium Azide
Storage	Store at -20°C. Aliquot to avoid repeated freezing and thawing.

GENE INFORMATION

Gene Name	GCH1 GTP cyclohydrolase 1 [Homo sapiens (human)]
Official Symbol	GCH1
Synonyms	GCH1; GTP cyclohydrolase 1; GCH; DYT5; DYT14; DYT5a; GTPCH1; HPABH4B; GTP-CH-1; GTP-CH-I; dystonia 14; GTP cyclohydrolase I; guanosine 5-triphosphate cyclohydrolase I;
Entrez Gene ID	2643
Protein Refseq	NP_000152
UniProt ID	A0A024R642
Chromosome Location	14q22.1-q22.2
Pathway	Folate biosynthesis; Metabolism; Tetrahydrobiopterin (BH4) synthesis, recycling, salvage and regulation; tetrahydrobiopterin biosynthesis I
Function	GTP binding; GTP cyclohydrolase I activity; NOT GTP cyclohydrolase I activity; GTP-dependent protein binding