



Rabbit Anti-GTP cyclohydrolase 1 antibody

SL0136R

Product Name:	GTP cyclohydrolase 1
Chinese Name:	三磷酸鸟苷环水解酶抗体
Alias:	GTP-CH-1; DYT 5; DYT5; GCH 1; GCH; GCH1; GTP CH 1; GTP CH I; GTP cyclohydrolase 1 (dopa responsive dystonia); GTP cyclohydrolase 1; GTP cyclohydrolase I; GTPCH 1; GTPCH1; Guanosine 5' triphosphate cyclohydrolase I; DYT14; DYT5a; HPABH4B.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	27kDa
Cellular localization:	The nucleuscytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human GTP-CH-1:34-110/250
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	GTP cyclohydrolase I (also designated dopa-responsive dystonia) catalyzes the conversion of GTP to D-erythro-7,8-dihydroneopterin triphosphate, the first and rate-limiting step in tetrahydrobiopterin (BH4) biosynthesis. Tetrahydrobiopterin is an essential cofactor for 3 aromatic amino acid monooxygenases: phenylalanine, tyrosine,

and tryptophan hydroxylases. Animals can synthesize tetrahydrobiopterin in vivo from GTP through several enzymatic reactions.

Function:

Positively regulates nitric oxide synthesis in umbilical vein endothelial cells (HUVECs). May be involved in dopamine synthesis. May modify pain sensitivity and persistence. Isoform GCH-1 is the functional enzyme, the potential function of the enzymatically inactive isoforms remains unknown.

Subcellular Location:

Cytoplasm. Nucleus.

Tissue Specificity:

In epidermis, expressed predominantly in basal undifferentiated keratinocytes and in some but not all melanocytes (at protein level).

Post-translational modifications:

Phosphorylated by casein kinase II at Ser-81 in HAECs during oscillatory shear stress; phosphorylation at Ser-81 results in increased enzyme activity.

DISEASE:

Defects in GCH1 are the cause of GTP cyclohydrolase 1 deficiency (GCH1D) [MIM:233910]; also known as atypical severe phenylketonuria due to GTP cyclohydrolase I deficiency;. GCH1D is one of the causes of malignant hyperphenylalaninemia due to tetrahydrobiopterin deficiency. It is also responsible for defective neurotransmission due to depletion of the neurotransmitters dopamine and serotonin. The principal symptoms include: psychomotor retardation, tonic disorders, convulsions, drowsiness, irritability, abnormal movements, hyperthermia, hypersalivation, and difficulty swallowing. Some patients may present a phenotype of intermediate severity between severe hyperphenylalaninemia and mild dystonia type 5 (dystonia-parkinsonism with diurnal fluctuation). In this intermediate phenotype, there is marked motor delay, but no mental retardation and only minimal, if any, hyperphenylalaninemia.

Defects in GCH1 are the cause of dystonia type 5 (DYT5) [MIM:128230]; also known as progressive dystonia with diurnal fluctuation, autosomal dominant Segawa syndrome or dystonia-parkinsonism with diurnal fluctuation. DYT5 is a DOPA-responsive dystonia. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DYT5 typically presents in childhood with walking problems due to dystonia of the lower limbs and worsening of the dystonia towards the evening. It is characterized by postural and motor disturbances showing marked diurnal fluctuation. Torsion of the trunk is unusual. Symptoms are alleviated after sleep and aggravated by fatigue and exercise. There is a favorable response to L-DOPA without side effects.

Similarity:

Belongs to the GTP cyclohydrolase I family.

SWISS:
P30793

Gene ID:
2643

Database links:

[Entrez Gene: 2643](#) Human

[Entrez Gene: 14528](#) Mouse

[Entrez Gene: 100286831](#) Rabbit

[Entrez Gene: 29244](#) Rat

[Omim: 600225](#) Human

[SwissProt: P30793](#) Human

[SwissProt: Q05915](#) Mouse

[SwissProt: P51596](#) Rabbit

[SwissProt: P22288](#) Rat

[Unigene: 624900](#) Human

[Unigene: 86724](#) Human

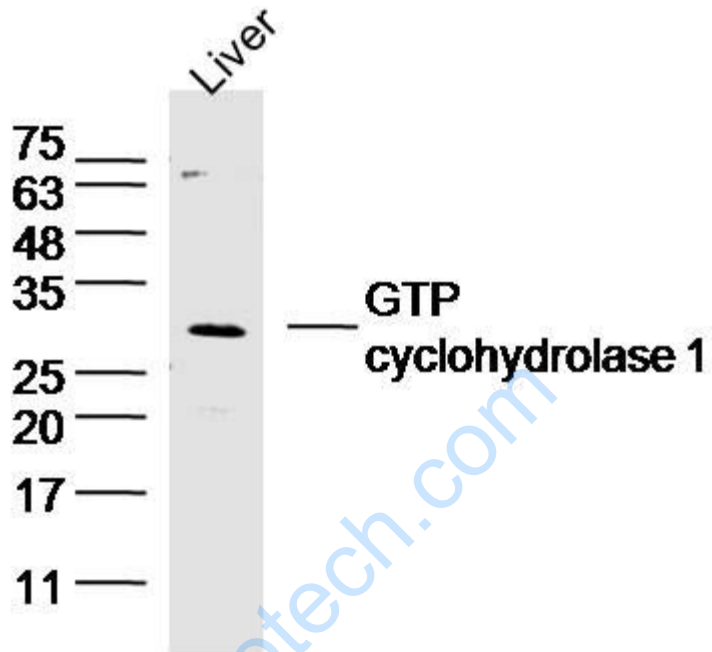
[Unigene: 10651](#) Mouse

[Unigene: 28195](#) Rat

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

Picture:



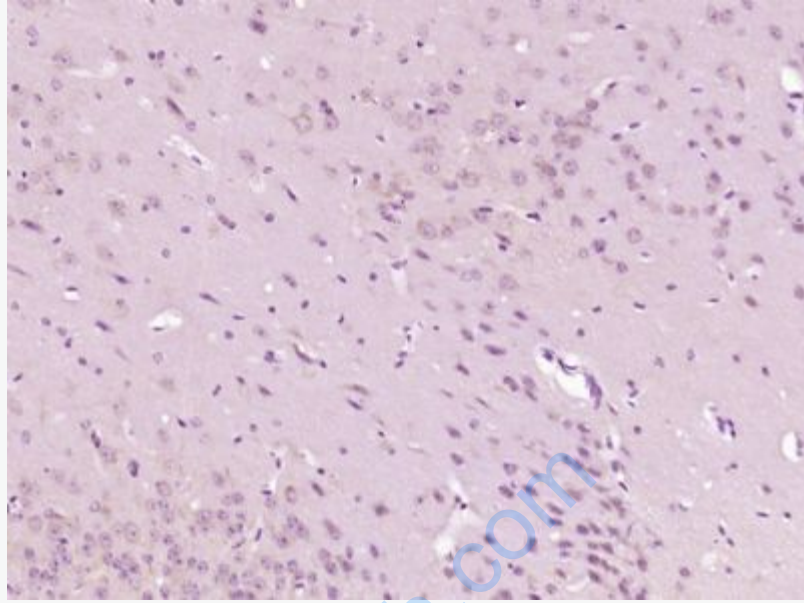
Sample: Liver (Mouse) Lysate at 40 ug

Primary: Anti-GTP cyclohydrolase 1 (SL0136R) at 1/300 dilution

Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution

Predicted band size: 27 kD

Observed band size: 27 kD



Paraformaldehyde-fixed, paraffin embedded (Mouse brain); Antigen retrieval by boiling in sodium citrate buffer (pH6.0) for 15min; Block endogenous peroxidase by 3% hydrogen peroxide for 20 minutes; Blocking buffer (normal goat serum) at 37°C for 30min; Antibody incubation with (GTP cyclohydrolase 1) Polyclonal Antibody, Unconjugated (SL0136R) at 1:400 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructions and DAB staining.