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GTP cyclohydrolase 1 Rabbit pAb

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Catalog # AP54184

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	P30793
Reactivity	Mouse
Predicted	Rat, Human
Host	Rabbit
Clonality	Polyclonal
Calculated MW	27903
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human GTP-CH-1
Epitope Specificity	34-110/250
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Cytoplasm. Nucleus.
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, tonicity 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 excercise. There is a favorable response to L-DOPA without side effects.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	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.

Additional Information

Gene ID	2643
Other Names	GTP cyclohydrolase 1, 3.5.4.16, GTP cyclohydrolase I, GTP-CH-I, GCH1, DYT5, GCH
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

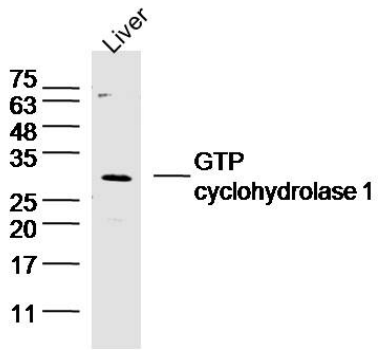
Protein Information

Name	GCH1
Synonyms	DYT5, GCH
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.
Cellular Location	Cytoplasm. Nucleus
Tissue Location	In epidermis, expressed predominantly in basal undifferentiated keratinocytes and in some but not all melanocytes (at protein level).

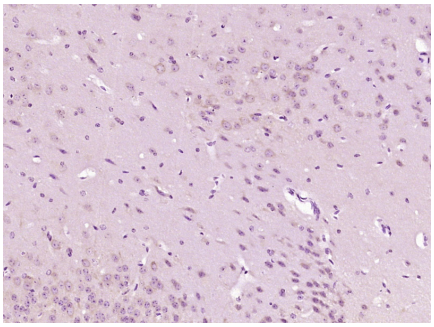
Background

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Images



Sample:Liver (Mouse) Lysate at 40 ug
Primary: Anti-GTP cyclohydrolase 1 (AP54184) 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 (AP54184) at 1:400 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructionsand DAB staining.

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.