abcam

Product datasheet

Anti-GCH1 antibody - N-terminal ab186633

1 Image

Overview

Product name	Anti-GCH1 antibody - N-terminal
Description	Rabbit polyclonal to GCH1 - N-terminal
Host species	Rabbit
Tested applications	**Suitable for:** WB
Species reactivity	**Reacts with:** Human **Predicted to work with:** Rat, Rabbit, Cow, Dog, Pig
Immunogen	Synthetic peptide within Human GCH1 aa 39-88 (N terminal). The exact sequence is proprietary. Sequence: PPRPEAKSAQPADGWKGERPRSEEDNELNLPNLAAA YSSILSSLGENPQR Database link: P30793 Run BLAST with Run BLAST with
Positive control	Human fetal heart tissue
General notes	This product was previously labelled as GTP cyclohydrolase 1

Properties

Form	Liquid
Storage instructions	Shipped at 4°C. Store at +4°C short term (1-2 weeks). Upon delivery aliquot. Store at -20°C long term. Avoid freeze / thaw cycle.
Storage buffer	Preservative: 0.09% Sodium azide Constituents: 2% Sucrose, PBS
Purity	Immunogen affinity purified
Clonality	Polyclonal
Isotype	IgG

Applications

Our Abpromise guarantee covers the use of **ab186633** in the following tested applications.

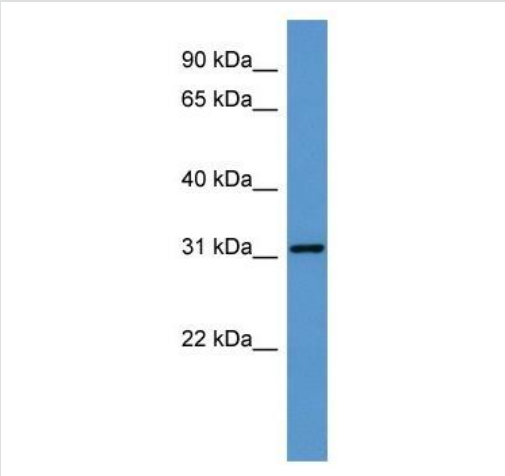
The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Application	Abreviews	Notes
WB		Use a concentration of 1 µg/ml. Predicted molecular weight: 28 kDa.

Target

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.
Tissue specificity	In epidermis, expressed predominantly in basal undifferentiated keratinocytes and in some but not all melanocytes (at protein level).
Pathway	Cofactor biosynthesis; 7,8-dihydroneopterin triphosphate biosynthesis; 7,8-dihydroneopterin triphosphate from GTP: step 1/1.
Involvement in 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.
Sequence similarities	Belongs to the GTP cyclohydrolase I family.
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.
Cellular localization	Cytoplasm. Nucleus.

Images



Western blot - Anti-GCH1 antibody - N-terminal (ab186633)

Anti-GCH1 antibody - N-terminal (ab186633) at 1 µg/ml + Human fetal heart tissue lysate at 10 µg

Developed using the ECL technique.

Predicted band size: 28 kDa

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