abcam

Product datasheet

Anti-GCH1 antibody ab69962

1 References 2 Images

Overview

Product name	Anti-GCH1 antibody
Description	Mouse polyclonal to GCH1
Host species	Mouse
Tested applications	**Suitable for:** WB, ICC/IF
Species reactivity	**Reacts with:** Human
Immunogen	Recombinant full length protein within Human GCH1. The exact immunogen sequence used to generate this antibody is proprietary information. If additional detail on the immunogen is needed to determine the suitability of the antibody for your needs, please **contact** our Scientific Support team to discuss your requirements. Database link: **NP_000152.1**
Positive control	GCH1 transfected 293T cell lysate.
General notes	The Life Science industry has been in the grips of a reproducibility crisis for a number of years. Abcam is leading the way in addressing this with our range of recombinant monoclonal antibodies and knockout edited cell lines for gold-standard validation. Please check that this product meets your needs before purchasing. If you have any questions, special requirements or concerns, please send us an inquiry and/or contact our Support team ahead of purchase. Recommended alternatives for this product can be found below, along with publications, customer reviews and Q&As

Properties

Form	Liquid
Storage instructions	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.
Storage buffer	pH: 7.40 Constituent: 100% PBS
Purity	Protein G purified
Clonality	Polyclonal
Isotype	IgG

Applications

The Abpromise guarantee Our **Abpromise guarantee** covers the use of ab69962 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		1/500 - 1/1000. Detects a band of approximately 28 kDa (predicted molecular weight: 28 kDa).
ICC/IF		Use a concentration of 10 µg/ml.

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.

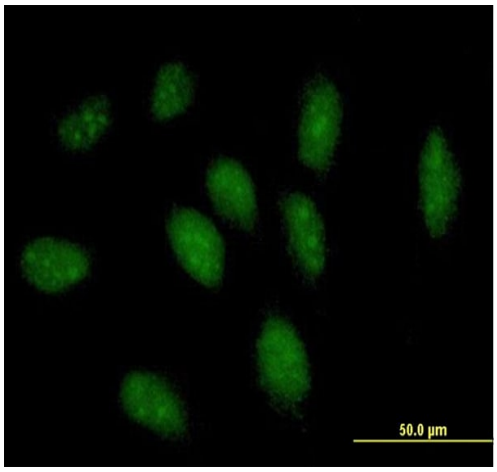
Involvement in disease Defects in GCH1 are the cause of GTP cyclohydrolase 1 deficiency (GCH1D) [MIM:233910]; also known as atypical severe phenylketonuria due to GTP cyclohydrolase I deficiency;. GCH1D is one of the causes of malignant hyperphenylalaninemia due to tetrahydrobiopterin deficiency. It 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.

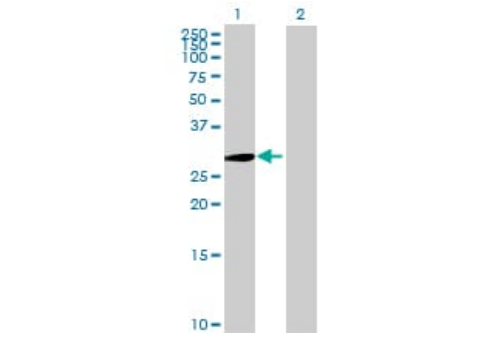
Cellular localization Cytoplasm. Nucleus.

Images



Immunocytochemistry/ Immunofluorescence - Anti-GCH1 antibody (ab69962)

Immunocytochemistry of HeLa cells staining GCH1 using ab69962 at 10µg/ml.



Western blot - Anti-GCH1 antibody (ab69962)

All lanes : Anti-GCH1 antibody (ab69962) at 1/500 dilution

Lane 1 : GCH1 transfected 293T cell lysate
Lane 2 : Non-transfected 293T cell lysate

Lysates/proteins at 25 µg per lane.

Secondary
All lanes : Goat Anti-Mouse IgG (H&L)-HRP Conjugate at 1/2500 dilution

Predicted band size: 28 kDa
Observed band size: 28 kDa

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