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Product datasheet

Anti-GCH1 antibody ab69962

1 References 画像数 2

製品の概要

製品名	Anti-GCH1 antibody
製品の詳細	Mouse polyclonal to GCH1
由来種	Mouse
アプリケーション	**適用あり:** WB, ICC/IF
種交差性	**交差種:** Human
免疫原	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**
ポジティブ・コントロール	GCH1 transfected 293T cell lysate.
特記事項	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

製品の特性

製品の状態	Liquid
保存方法	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.
バッファー	pH: 7.40 Constituent: 100% PBS
精製度	Protein G purified
ポリ/モノ	ポリクローナル
アイソタイプ	IgG

アプリケーション

The Abpromise guarantee **Abpromise保証は、** 次のテスト済みアプリケーションにおけるab69962の使用に適用されます

アプリケーションノートには、推奨の開始希釈率がありますが、適切な希釈率につきましてはご検討ください。

アプリケーション	Abreviews	特記事項
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.

ターゲット情報

機能

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.

組織特異性

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

パスウェイ

Cofactor biosynthesis; 7,8-dihydroneopterin triphosphate biosynthesis; 7,8-dihydroneopterin triphosphate from GTP: step 1/1.

関連疾患

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.

配列類似性

Belongs to the GTP cyclohydrolase I family.

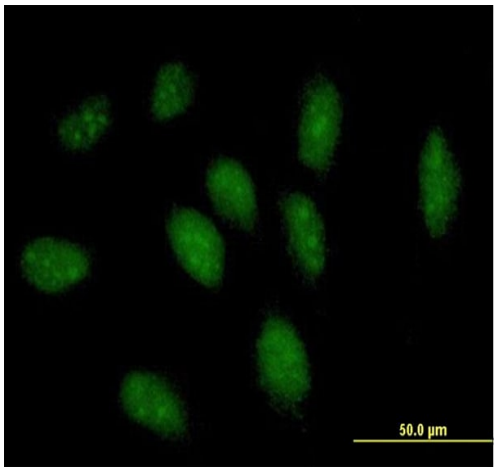
翻訳後修飾

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

細胞内局在

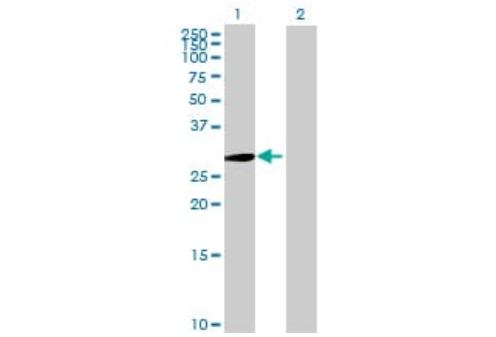
Cytoplasm. Nucleus.

画像



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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