abcam

Product datasheet

Anti-GCH1 antibody ab69962

1 References 2 图像

概述

产品名称	Anti-GCH1**抗体**
描述	**小鼠多克隆抗体**to GCH1
宿主	Mouse
经测试应用	**适用于:** WB, ICC/IF
种属反应性	**与反应:** 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**
阳性对照	GCH1 transfected 293T cell lysate.
常规说明	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

性能

形式	Liquid
存放说明	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.
存储溶液	pH: 7.40 Constituent: 100% PBS
纯度	Protein G purified
克隆	**多克隆**
同种型	IgG

应用

The Abpromise guarantee **Abpromise™承诺保证使用ab69962于以下的经测试应用**

"应用说明"部分 下显示的仅为推荐的起始稀释度;实际最佳的稀释度/浓度应由使用者检定。

应用	Ab评论	说明
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.

靶标

功能 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.

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

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

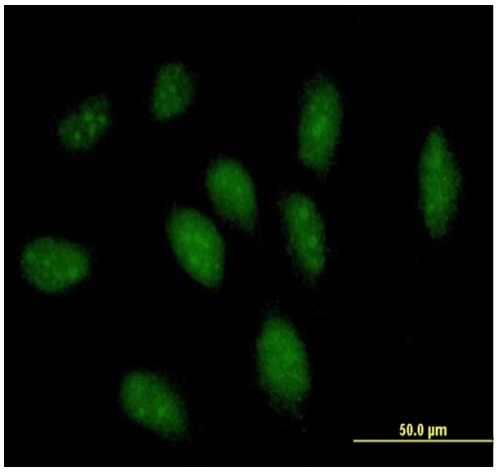
疾病相关 Defects in GCH1 are the cause of GTP cyclohydrolase 1 deficiency (GCH1D) [MIM:233910]; also known as atypical severe phenylketonuria due to GTP cyclohydrolase I deficiency;. GCH1D is one of the causes of malignant hyperphenylalaninemia due to tetrahydrobiopterin deficiency. It is also responsible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

序列相似性 Belongs to the GTP cyclohydrolase I family.

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

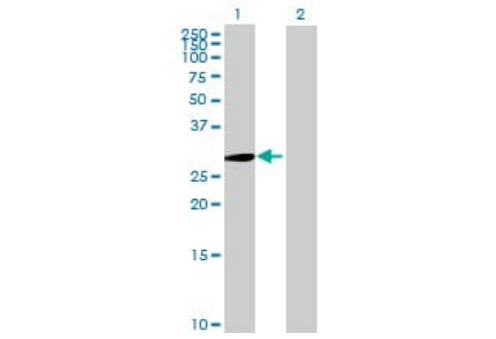
细胞定位 Cytoplasm. Nucleus.

图片



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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