abcam

Product datasheet

Recombinant Human GTP cyclohydrolase 1 protein ab114820

画像数 1

製品の概要

製品名	Recombinant Human GTP cyclohydrolase 1 protein
タンパク質長	Full length protein

製品の詳細

由来	Recombinant
由来	Wheat germ
アミノ酸配列	
アクセッション番号	P30793
生物種	Human
配列	MEKGPVRAPAEKPRGARCSNGFPERDPPRPGPSRPAEKPPRPEAKSAQPA DGWKGERPRSEEDNELNLPNLAAAYSSILSSLGENPQRQGLLKTPWRAAS AMQFFTKGYQETISDVLNDAIFDEDHDEMVIVKDIDMFSMCEHHLVPFVG KVHIGYLPNKQVLGLSKLARIVEIYSRRLQVQERLTKQIAVAITEALRPA GVGVVVEATHMCMVMRGVQKMNSKTVTSTMLGVFREDPKTREEFLTLIRS
分子量	54 kDa including tags
領域	1 to 251

特性

Our Abpromise guarantee covers the use of **ab114820** in the following tested applications.

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

アプリケーション	ELISA SDS-PAGE Western blot
製品の状態	Liquid
備考	Protein concentration is above or equal to 0.05 mg/ml. Best used within three months from the date of receipt.

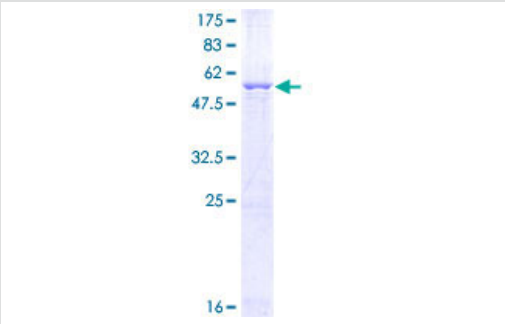
前処理および保存

保存方法および安定性	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.3% Glutathione, 0.79% Tris HCl

関連情報

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



SDS-PAGE - GTP cyclohydrolase 1 protein (ab114820)

12.5% SDS-PAGE Stained with Coomassie Blue

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- We investigate all quality concerns to ensure our products perform to the highest standards

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