abcam

Product datasheet

# Human GCH1 knockout A549 cell lysate ab258878

2 Images

## Overview

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|---|---|
| **Product name** | Human GCH1 knockout A549 cell lysate |
| **Product overview** | Knockout cell lysate achieved by CRISPR/Cas9. |
| **Parental Cell Line** | A549 |
| **Organism** | Human |
| **Mutation description** | Knockout achieved by using CRISPR/Cas9, 1 bp insertion in exon1 and 2 bp deletion in exon1. |
| **Passage number** | <20 |
| **Knockout validation** | Sanger Sequencing |
| **Reconstitution notes** | To use as WB control, resuspend the lyophilizate in 50 µL of LDS* Sample Buffer to have a final concentration of 2 mg/ml. For reducing conditions, we recommend a final concentration of 0.1 M DTT.<br>**Usage of SDS sample buffer is not recommended with these lyophilized lysates.* |
| **Notes** | **Lysate preparation:** Our lysates are made using RIPA buffer to which we add a protease inhibitor cocktail and phosphatase inhibitor cocktail (ratio: 300:100:10). *This means that the protein of interest is denatured.* If you require a native form of the protein please use the live cell version - found **here**. Please refer to our lysis protocol for further details on how our lysates are prepared.<br><br>**User storage instructions:** Lyophilizate may be stored at 4°C. After reconstitution, store at -20°C for short-term storage or -80°C for long-term storage.<br><br>Access thousands of knockout cell lysates, generated from commonly used cancer cell lines.<br>**See here for more information on knockout cell lysates.**<br><br>Abcam has not and does not intend to apply for the REACH Authorisation of customers' uses of products that contain European Authorisation list (Annex XIV) substances.<br>It is the responsibility of our customers to check the necessity of application of REACH Authorisation, and any other relevant authorisations, for their intended uses.<br><br>This product is subject to limited use licenses from The Broad Institute, ERS Genomics Limited and Sigma-Aldrich Co. LLC, and is developed with patented technology. For full details of the licenses and patents please refer to our **limited use license** and **patent pages**. |

## Properties

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**Storage instructions** Store at -80°C. Please refer to protocols.

| Components | 1 kit |
|---|---|
| ab262569 - Human GCH1 knockout A549 cell lysate | 1 x 100µg |
| ab255554 - Human wild-type A549 cell lysate | 1 x 100µg |

**Cell type** epithelial

**Disease** Carcinoma

**STR Analysis** Amelogenin X,Y D5S818: 11 D13S317: 11 D7S820: 8, 11 D16S539: 11, 12 vWA: 14 TH01: 8,9.3 TPOX: 8,11 CSF1PO: 10, 12

## 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


Sanger Sequencing - Human GCH1 knockout A549 cell lysate (ab258878)

Allele-1: 2 bp deletion in exon1


Sanger Sequencing - Human GCH1 knockout A549 cell lysate (ab258878)

Allele-2: 1 bp insertion in exon1

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