# abcam

## Product datasheet

## Recombinant Human GPD1L protein ab113595

## 1 Image

**Description** 

Product name Recombinant Human GPD1L protein

Purity > 95 % SDS-PAGE.

ab113595 was purified using conventional chromatography.

**Expression system** Escherichia coli

Accession Q8N335

Protein length Full length protein

Animal free No

**Nature** Recombinant

**Species** Human

Sequence MGSSHHHHHHSSGLVPRGSHMAAAPLKVCIVGSGNWG

SAVAKIIGNNVKK

 ${\tt LQKFASTVKMWVFEETVNGRKLTDIINNDHENVKYLPGHK}$ 

**LPENVVAMSN** 

LSEAVQDADLLVFVIPHQFIHRICDEITGRVPKKALGITLIKGI

**DEGPEG** 

LKLISDIIREKMGIDISVLMGANIANEVAAEKFCETTIGSKVM

ENGLLFK

ELLQTPNFRITVVDDADTVELCGALKNIVAVGAGFCDGLR

**CGDNTKAAVI** 

RLGLMEMIAFARIFCKGQVSTATFLESCGVADLITTCYGGR

NRRVAEAFA

RTGKTIEELEKEMLNGQKLQGPQTSAEVYRILKQKGLLDKF

**PLFTAVYQI CYESRPVQEMLSCLQSHPEHT** 

Predicted molecular weight 41 kDa including tags

Amino acids 1 to 351

Tags His tag N-Terminus

#### **Specifications**

Our Abpromise guarantee covers the use of ab113595 in the following tested applications.

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

Applications SDS-PAGE

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Mass Spectrometry

Mass spectrometry

**MALDI-TOF** 

**Form** 

Liquid

#### **Preparation and Storage**

#### Stability and Storage

Shipped at 4°C. Store at +4°C short term (1-2 weeks). Upon delivery aliquot. Store at -20°C or -

80°C. Avoid freeze / thaw cycle.

pH: 8.00

Constituents: 0.02% DTT, 0.32% Tris HCl, 20% Glycerol (glycerin, glycerine)

#### **General Info**

Function Play a role in regulating cardiac sodium current; decreased enzymatic activity with resulting

increased levels of glycerol 3-phosphate activating the DPD1L-dependent SCN5A

phosphorylation pathway, may ultimately lead to decreased sodium current; cardiac sodium current may also be reduced due to alterations of NAD(H) balance induced by DPD1L.

Tissue specificity

Most highly expressed in heart tissue, with lower levels in the skeletal muscle, kidney, lung and

other organs.

Involvement in disease Defects in GPD1L are the cause of Brugada syndrome type 2 (BRS2) [MIM:611777]. BRS2 is an

autosomal dominant tachyarrhythmia characterized by right bundle branch block and ST segment elevation on an electrocardiogram (ECG). It can cause the ventricles to beat so fast that the blood is prevented from circulating efficiently in the body. When this situation occurs (called ventricular

fibrillation), the individual will faint and may die in a few minutes if the heart is not reset.

Defects in GPD1L are a cause of sudden infant death syndrome (SIDS) [MIM:272120]. SIDS is the sudden death of an infant younger than 1 year that remains unexplained after a thorough case investigation, including performance of a complete autopsy, examination of the death scene, and review of clinical history. Pathophysiologic mechanisms for SIDS may include respiratory

 $dys function, cardiac\ dysrhythmias,\ cardiorespiratory\ instability,\ and\ inborn\ errors\ of\ metabolism,$ 

but definitive pathogenic mechanisms precipitating an infant sudden death remain elusive.

Sequence similarities Belongs to the NAD-dependent glycerol-3-phosphate dehydrogenase family.

**Cellular localization** Cytoplasm. Localized to the region of the plasma membrane.

## **Images**



15% SDS-PAGE analysis of ab113595 (3µg)

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