abcam

Product datasheet

Recombinant Human GFPT1 protein ab152423

1 Image

Description

Product name	Recombinant Human GFPT1 protein
Expression system	Wheat germ
Accession	Q06210-2
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MCGIFAYLNYHVPRTRREILETLIKGLQRLEYRGYDSAGVGF DGGNDKDW EANACKIQLIKKKGKVKALDEEVHKQQDMDLDIEFDVHLGI AHTRWATHG EPSPVNSHPQRSDKNNEFIVIHNGIITNYKDLKKFLESKGYD FESETDTE TIAKLVKYMYDNRESQDTSFTTLVERVIQQLEGAFALVFKS VHFPGQAVG TRRGSPLLIGVRSEHKLSTDHIPILYRTGKDKKGSCNLSRV DSTTCLFPV EEKAVEYYFASDASAVIEHTNRVIFLEDDDVAAVVDGRLSI HRIKRTAGD HPGRAVQTLQMELQQIMKGNFSSFMQKEIFEQPESVVNT MRGRVNFDDYT VNLGGLKDHIKEIQRCRRLILIACGTSYHAGVATRQVLEELT ELPVMVEL ASDFLDRNTPVFRDDVCFFLSQSGETADTLMGLRYCKER GALTVGITNTV GSSISRETDCGVHINAGPEIGVASTKAYTSQFVSLVMFALM MCDDRISMQ ERRKEIMLGLKRLPDLIKEVLSMDDEIQKLATELYHQKSVLI MGRGYHYA TCLEGALKIKEITYMHSEGILAGELKHGPLALVDKLMPVIMII MRDHTYA KCQNALQQVVARQGRPVVICDKEDTETIKNTKRTIKVPHS VDCLQGILSV IPLQLLAFHLAVLRGYDVDFPRNLAKSVTVE
Predicted molecular weight	103 kDa including tags

Amino acids	1 to 681

Specifications

Our **Abpromise guarantee** covers the use of **ab152423** in the following tested applications.

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

Applications	ELISA SDS-PAGE Western blot
Form	Liquid
Additional notes	

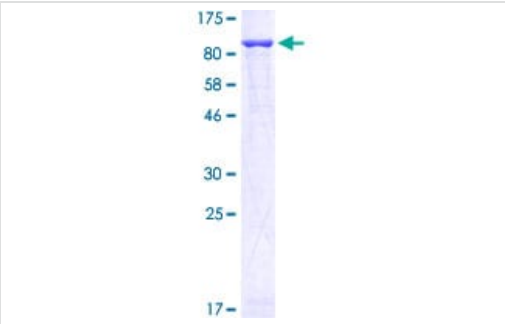
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Function	Controls the flux of glucose into the hexosamine pathway. Most likely involved in regulating the availability of precursors for N- and O-linked glycosylation of proteins.
Tissue specificity	Isoform 1 is predominantly expressed in skeletal muscle. Not expressed in brain. Seems to be selectively expressed in striated muscle.
Pathway	Nucleotide-sugar biosynthesis; UDP-N-acetyl-alpha-D-glucosamine biosynthesis; alpha-D-glucosamine 6-phosphate from D-fructose 6-phosphate: step 1/1.
Involvement in disease	Defects in GFPT1 are the cause of limb-girdle myasthenia with tubular aggregates (LGMTA) [MIM:610542]. A congenital myasthenic syndrome characterized by onset of proximal muscle weakness in the first decade. Individuals with this condition have a recognizable pattern of weakness of shoulder and pelvic girdle muscles, and sparing of ocular or facial muscles. EMG classically shows a decremental response to repeated nerve stimulation, a sign of neuromuscular junction dysfunction. Affected individuals show a favorable response to acetylcholinesterase (AChE) inhibitors.
Sequence similarities	Contains 1 glutamine amidotransferase type-2 domain. Contains 2 SIS domains.

Images



SDS-PAGE - Recombinant Human GFPT1 protein (ab152423)

12.5% SDS-PAGE analysis of ab152423 stained with Coomassie Blue.

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