

Recombinant Human GFPT1 protein ab152423

1 Image

Description

Product name	Recombinant Human GFPT1 protein
Expression system	Wheat germ
Accession	<u>Q06210-2</u>
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	<p>MCGIFAYLNYHVPRTREILETLIKGLQRLEYRGYDSAGVGF DGGNDKDW EANACKIQLIKKKGKVKALDEEVHKQQDMDLDIEFDVHLGI AHTRWATHG EPSPVNSHPQRSDDKNNEFVIHNGIITNYKDLKKFLESKGYD FESETDTE TIAKLVKMYDNRESQDTSFTTLVERVIQQLEGAFALVFKS VHFPGQAVG TRRGSPLLIGVRSEHKLSTDHIPILYRTGKDKKGCNLSRV DSTTCLFPV EEKAVEYYFASDASAVIEHTNRVIFLEDDDDVAAVVDGRLSI HRIKRTAGD HPGRAVQTLQMELQQIMKGNFSSFMQKEIFEQPESVVNT MRGRVNFDDYT VNLGGLKDHKEIQRRCRLILIACGTSYHAGVATRQVLEELT ELPVMVEL ASDFLDRNTPVFRDDVCFFLSQSGETADTLMGLRYCKER GALTVGITNTV GSSISRETDCGVHINAGPEIGVASTKAYTSQFVSLVMFALM MCDDRISMQ ERRKEIMLGLKRLPDLIKEVLSMDDEIQKLATELYHQKSVLI MGRGYHYA TCLEGALKIKEITYMHSEGILAGELKHGPLALVDKLMPVIMII MRDHTYA KCQNALQQVVARQGRPVVICDKEDTETIKNTKRTIKVPHS VDCLQGILSV IPLQLLAFHLAVLRGYDVDFPRNLAKSVTVE</p>
Predicted molecular weight	103 kDa including tags

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	

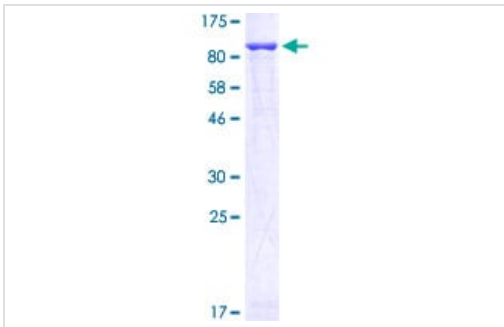
Preparation and Storage

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



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

SDS-PAGE - Recombinant Human GFPT1 protein
(ab152423)

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