abcam

Product datasheet

Recombinant Human GFPT1 protein ab152423

1 图像

描述

产品名称	重组人GFPT1蛋白
表达系统	Wheat germ
Accession	Q06210-2
蛋白长度	Full length protein
无动物成分	No
性质	Recombinant
种属	Human
序列	MCGIFAYLNYHVPRTRREILETLIKGLQRLEYRGYDSAGVGF DGGNDKDW EANACKIQLIKKKGKVKALDEEVHKQQDMDLDIEFDVHLGIA HTRWATHG EPSPVNSHPQRSDKNNEFIVIHNGIITNYKDLKKFLESKGYD FESETDTE TIAKLVKYMYDNRESQDTSFTTLVERVIQQLEGAFALVFKSV HFPGQAVG TRRGSPLLIGVRSEHKLSTDHIPILYRTGKDKKGSCNLSRVD STTCLFPV EEKAVEYYFASDASAVIEHTNRVIFLEDDDVAAVVDGRLSIH RIKRTAGD HPGRAVQTLQMELQQIMKGNFSSFMQKEIFEQPESVVNTMRG RVNFDDYT VNLGGLKDHIKEIQRCRRLILIACGTSYHAGVATRQVLEELT ELPVMVEL ASDFLDRNTPVFRDDVCFFLSQSGETADTLMGLRYCKERGAL TVGITNTV GSSISRETDCGVHINAGPEIGVASTKAYTSQFVSLVMFALMM CDDRISMQ ERRKEIMLGLKRLPDLIKEVLSMDDEIQKLATELYHQKSVLI MGRGYHYA TCLEGALKIKEITYMHSEGILAGELKHGPLALVDKLMPVIMI IMRDHTYA KCQNALQQVVARQGRPVVICDKEDTETIKNTKRTIKVPHSVD CLQGILSV IPLQLLAFHLAVLRGYDVDFPRNLAKSVTVE
预测分子量	103 kDa including tags

氨基酸	1 to 681

技术指标

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.

应用	ELISA SDS-PAGE Western blot
形式	Liquid
补充说明	

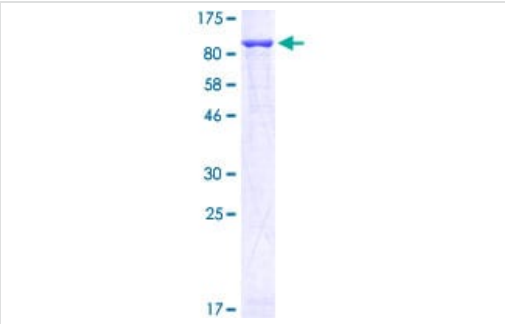
制备和贮存

稳定性和存储	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

常规信息

功能	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.
组织特异性	Isoform 1 is predominantly expressed in skeletal muscle. Not expressed in brain. Seems to be selectively expressed in striated muscle.
通路	Nucleotide-sugar biosynthesis; UDP-N-acetyl-alpha-D-glucosamine biosynthesis; alpha-D-glucosamine 6-phosphate from D-fructose 6-phosphate: step 1/1.
疾病相关	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.
序列相似性	Contains 1 glutamine amidotransferase type-2 domain. Contains 2 SIS domains.

图片



SDS-PAGE - Recombinant Human GFPT1 protein (ab152423)

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

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