abcam

Product datasheet

Recombinant Human GBA protein ab235716

1 Image

Description

Product name	Recombinant Human GBA protein
Purity	85 % SDS-PAGE.
Expression system	Escherichia coli
Accession	P04062-1
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	ARPCIPKSFGYSSVVCVCNATYCDSFDPPTFPALGTFSRY ESTRSGRRME LSMGPIQANHTGTGLLLTLQPEQKFQKVKGFGGAMTDAA ALNILALSPPA QNLLLKSYFSEEGIGYNIIRVPMASCDFSIRTYTYADTPDDF QLHNFSLP EEDTKLKIPLIHRALQLAQRPVSLLASPWTSPTWLKTNGAV NGKGSLKGQ PGDIYHQTWARYFVKFLDAYAEHKLQFWAVTAENEPSAG LLSGYPFQCLG FTPEHQRDFIARDLGPTLANSTHHNVRLLMLDDQRLLLPH WAKVVLTDPE AAKYVHGIAVHWYLDFLAPAKATLGETHRLFPNTMLFASE ACVGSKFWEQ SVRLGSWDRGMQYSHSIITSLLYHVVGWTDWNLALNPEG GPNWVRNFVDS PIIVDITKDTFYKQPMFYHLGHFSKFIPEGSQRVGLVASQKN DLDAVALM HPDGSAVVVVLNRSSKDVPLTIKDPAVGFLETISPGYSIHT YLWRRQ
Predicted molecular weight	56 kDa
Amino acids	40 to 536
Additional sequence information	Full length mature protein without the signal peptide (1-39aa)

Specifications

Our Abpromise guarantee covers the use of **ab235716** 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
Form	Liquid

Preparation and Storage

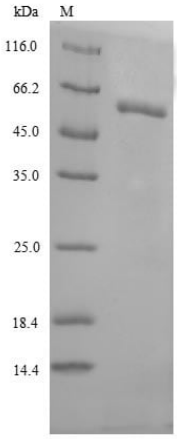
Stability and Storage	Shipped at 4°C. Store at -20°C or -80°C. Avoid freeze / thaw cycle. Constituents: Tris buffer, 50% Glycerol (glycerin, glycerine)

General Info

Involvement in disease	Defects in GBA are the cause of Gaucher disease (GD) [MIM:230800]; also known as glucocerebrosidase deficiency. GD is the most prevalent lysosomal storage disease, characterized by accumulation of glucosylceramide in the reticulo-endothelial system. Different clinical forms are recognized depending on the presence (neuronopathic forms) or absence of central nervous system involvement, severity and age of onset. Defects in GBA are the cause of Gaucher disease type 1 (GD1) [MIM:230800]; also known as adult non-neuronopathic Gaucher disease. GD1 is characterized by hepatosplenomegaly with consequent anemia and thrombopenia, and bone involvement. The central nervous system is not involved. Defects in GBA are the cause of Gaucher disease type 2 (GD2) [MIM:230900]; also known as acute neuronopathic Gaucher disease. GD2 is the most severe form and is universally progressive and fatal. It manifests soon after birth, with death generally occurring before patients reach two years of age. Defects in GBA are the cause of Gaucher disease type 3 (GD3) [MIM:231000]; also known as subacute neuronopathic Gaucher disease. GD3 has central nervous manifestations. Defects in GBA are the cause of Gaucher disease type 3C (GD3C) [MIM:231005]; also known as pseudo-Gaucher disease or Gaucher-like disease. Defects in GBA are the cause of Gaucher disease perinatal lethal (GDPL) [MIM:608013]. It is a distinct form of Gaucher disease type 2, characterized by fetal onset. Hydrops fetalis, in utero fetal death and neonatal distress are prominent features. When hydrops is absent, neurologic involvement begins in the first week and leads to death within 3 months. Hepatosplenomegaly is a major sign, and is associated with ichthyosis, arthrogryposis, and facial dysmorphism. Note=Perinatal lethal Gaucher disease is associated with non-immune hydrops fetalis, a generalized edema of the fetus with fluid accumulation in the body cavities due to non-immune causes. Non-immune hydrops fetalis is not a diagnosis in itself but a symptom, a feature of many genetic disorders, and the end-stage of a wide variety of disorders. Defects in GBA contribute to susceptibility to Parkinson disease (PARK) [MIM:168600]. A complex neurodegenerative disorder characterized by bradykinesia, resting tremor, muscular rigidity and postural instability. Additional features are characteristic postural abnormalities, dysautonomia, dystonic cramps, and dementia. The pathology of Parkinson disease involves the loss of dopaminergic neurons in the substantia nigra and the presence of Lewy bodies (intraneuronal accumulations of aggregated proteins), in surviving neurons in various areas of the brain. The disease is progressive and usually manifests after the age of 50 years, although early-onset cases (before 50 years) are known. The majority of the cases are sporadic suggesting a multifactorial etiology based on environmental and genetic factors. However, some patients

	present with a positive family history for the disease. Familial forms of the disease usually begin at earlier ages and are associated with atypical clinical features.
Sequence similarities	Belongs to the glycosyl hydrolase 30 family.
Cellular localization	Lysosome membrane. Interaction with saposin-C promotes membrane association.

Images



SDS-PAGE - Recombinant Human GBA protein (ab235716)

(Tris-Glycine gel) Discontinuous SDS-PAGE (reduced) with 5% enrichment gel and 15% separation gel using ab235716.

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