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GBA Rabbit pAb

Catalog # AP98952

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	P04062
Other Accession	P04062
Reactivity	Human
Predicted	Rat, Mouse, Rabbit
Host	Rabbit
Clonality	Polyclonal
Calculated MW	59716
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human GBA
Epitope Specificity	141-240/536
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the glycosyl hydrolase 30 family.
SUBUNIT	Interacts with saposin-C. Interacts with SCARB2.
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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a lysosomal membrane protein that cleaves the beta-glucosidic linkage of glycosylceramide, an intermediate in glycolipid metabolism. Mutations in this gene cause Gaucher disease, a lysosomal storage disease characterized by an accumulation of glucocerebrosides. A related pseudogene is approximately 12 kb downstream of this gene on chromosome 1. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Jan 2010]

Additional Information

Gene ID	2629
Other Names	GBA; GCB; GLUC; GBA1_HUMAN; GBA1; Lysosomal acid GCase; Acid beta-glucosidase; Alglucerase; Beta-glucocerebrosidase (Beta-GC); Beta-glucosylceramidase 1; Cholesterol glucosyltransferase (SGTase); Cholesteryl-beta-glucosidase; D-glucosyl-N-acylsphingosine glucohydrolase; Glucosylceramidase beta 1; Imiglucerase; Lysosomal cholesterol glycosyltransferase; Lysosomal galactosylceramidase; Lysosomal glycosylceramidase; 3.2.1.45; GC;
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	GBA1 (HGNC:4177)
Synonyms	GBA, GC, GLUC
Function	Glucosylceramidase that catalyzes, within the lysosomal compartment, the hydrolysis of glucosylceramides/GlcCers (such as beta-D-glucosyl-(11')-N-acylsphing-4-enine) into free ceramides (such as N-acylsphing-4-enine) and glucose (PubMed:15916907, PubMed:24211208, PubMed:32144204, PubMed:39395789, PubMed:9201993). Plays a central role in the degradation of complex lipids and the turnover of cellular membranes (PubMed:27378698). Through the production of ceramides, participates in the PKC-activated salvage pathway of ceramide formation (PubMed:19279011).

	Catalyzes the glucosylation of cholesterol, through a transglucosylation reaction where glucose is transferred from GlcCer to cholesterol (PubMed:24211208, PubMed:26724485, PubMed:32144204). GlcCer containing mono-unsaturated fatty acids (such as beta-D-glucosyl-N-(9Z-octadecenoyl)-sphing-4-enine) are preferred as glucose donors for cholesterol glucosylation when compared with GlcCer containing same chain length of saturated fatty acids (such as beta-D-glucosyl-N-octadecanoyl-sphing-4-enine) (PubMed:24211208). Under specific conditions, may alternatively catalyze the reverse reaction, transferring glucose from cholesteryl 3-beta-D-glucoside to ceramide (Probable) (PubMed:26724485). Can also hydrolyze cholesteryl 3-beta-D- glucoside producing glucose and cholesterol (PubMed:24211208, PubMed:26724485, PubMed:39395789). Catalyzes the hydrolysis of galactosylceramides/GalCers (such as beta-D-galactosyl-(11')-N- acylsphing-4-enine), as well as the transfer of galactose between GalCers and cholesterol in vitro, but with lower activity than with GlcCers (PubMed:32144204). Contrary to GlcCer and GalCer, xylosylceramide/XylCer (such as beta-D-xyosyl-(11')-N-acylsphing-4- enine) is not a good substrate for hydrolysis, however it is a good xylose donor for transxylosylation activity to form cholesteryl 3-beta- D-xyloside (PubMed:33361282). Can also metabolize plant glycosyl phytosterols such as glucosylstigmasterol (PubMed:39395789).
Cellular Location	Lysosome membrane; Peripheral membrane protein; Lumenal side. Note=Interaction with saposin-C promotes membrane association (PubMed:10781797). Targeting to lysosomes occurs through an alternative MPR-independent mechanism via SCARB2 (PubMed:18022370).

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