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Rabbit Anti-GBA/AF350 Conjugated antibody

SL11779R-AF350

Product Name	Anti-GBA/AF350
Chinese Name	AF350 标记的 β-葡萄糖脑苷脂酶抗体
Alias	Glucosidase beta; Acid beta glucosidase; Acid beta-glucosidase; Alglucerase; Beta glucocerebrosidase; BETA GLUCOSIDASE, ACID; Beta-glucocerebrosidase; betaGC; D glucosyl N acylsphingosine glucohydrolase; D-glucosyl-N-acylsphingosine glucohydrolase; EC 3.2.1.45 ; GBA; Gba protein; GBA1; GC antibody GCase; GCB; GLCM_HUMAN; GLUC; Glucocerebrosidase (alt.); Glucocerebrosidase; GLUCOCEREBROSIDASE PSEUDOGENE; Glucosidase beta; Glucosidase, beta, acid; Glucosidase, beta; acid (includes glucosylceramidase); Glucosylceramidase; Imiglucerase; Lysosomal glucocerebrosidase.
Research Area	Tumour Cell biology Neurobiology Signal transduction The new supersedes the old
Immunogen Species	Rabbit
Clonality	Polyclonal
React Species	Human(predicted:Mouse,Rat,Rabbit)
Applications	IF=1:100-500 not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight	56kDa
Form	Lyophilized or Liquid
Concentration	1mg/ml
immunogen	KLH conjugated synthetic peptide derived from human Glucosidase beta
Lsotype	IgG
Purification	affinity purified by Protein A
Storage Buffer	1M TBS(pH7.4) with 1% BSA, 3% Proclin300 and 50% Glycerol
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 1M PBS or diluent of antibody the antibody is stable for at least two weeks

at 2-4 °C.

Product Detail

background:
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]

Subunit:
Interacts with saposin-C. Interacts with SCARB2.

Subcellular Location:
Lysosome membrane. Interaction with saposin-C promotes membrane association.

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.

Similarity:
Belongs to the glycosyl hydrolase 30 family.

Database links:

Entrez Gene: 2629 Human

Entrez Gene: 14466 Mouse

Entrez Gene: 684536 Rat

Omim: 606463 Human

SwissProt: P04062 Human

SwissProt: P17439 Mouse

Unigene: 282997 Human

Unigene: 719930 Human

Unigene: 5031 Mouse

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Unigene: 162606 Rat

Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.