10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

GBE1 Rabbit pAb

Catalog # AP97528

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Primary Accession	Q04446
Other Accession	Q04446
Predicted	Rat, Human, Mouse, Rabbit
Host	Rabbit
Clonality	Polyclonal
Calculated MW	80474
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human GBE1
Epitope Specificity	101-200/702
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 13 family.
DISEASE	Defects in GBE1 are the cause of glycogen storage disease type 4 (GSD4) [MIM:232500]; also known as Andersen disease. GSD4 is a metabolic disorder characterized by the accumulation of an amylopectin-like polysaccharide. The typical clinical manifestation is liver disease of childhood, progressing to lethal hepatic cirrhosis. Most children with this condition die before two years of age. However, the liver disease is not always progressive. No treatment apart from liver transplantation has been found to prevent progression of the disease. There is also a neuromuscular form of GSD4 that varies in onset (perinatal, congenital, juvenile, or adult) and severity. Note=Neuromuscular perinatal glycogen storage disease type 4 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 GBE1 are the cause of adult polyglucosan body disease (APBD) [MIM:263570]. APBD is a late-onset, slowly progressive disorder affecting the central and peripheral nervous systems. Patients typically present after age 40 years with a variable combination of cognitive impairment, pyramidal tetraparesis, peripheral neuropathy, and neurogenic bladder. Other manifestations include cerebellar dysfunction and extrapyramidal signs. The pathologic hallmark of APBD is the widespread accumulation of round, intracellular polyglucosan bodies throughout the nervous system, which are confined to neuronal and astrocytic processes.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	GBE1 is a 702 amino acid protein that is expressed at high levels in muscle and liver and is involved in glycogen biosynthesis. Existing as a monomer, GBE1 catalyzes the transfer of alpha-1,4-linked glucosyl units from the outer end of a glycogen chain to an alpha-1,6 position on a neighboring glycogen chain and, via this catalytic activity, plays an essential role in glycogen accumulation. Defects in the gene encoding GBE1 are the cause of glycogen

storage disease type 4 (GSD4) and adult polyglucosan body disease (APBD), the first of which is a metabolic disorder that is associated with the accumulation of polysaccharides and is characterized by liver disease during childhood. Unlike GSD4, APBD is a late-onset disorder that affects the central and peripheral nervous systems and is characterized by cognitive impairment, pyramidal tetraparesis and peripheral neuropathy.

Additional Information

Gene ID	2632
Other Names	GLGB_HORSE; GBE1; Brancher enzyme; Glycogen-branching enzyme; 2.4.1.18; GLGB_HUMAN; GLGB_MOUSE; 1, 4-alpha-glucan branching enzyme 1; glucan (1, 4-alpha-), branching enzyme 1; glycogen branching enzyme; Andersen disease; glycogen storage disease type IV
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	GBE1
Function	Glycogen-branching enzyme participates in the glycogen biosynthetic process along with glycogenin and glycogen synthase. Generates alpha-1,6-glucosidic branches from alpha-1,4-linked glucose chains, to increase solubility of the glycogen polymer (PubMed:26199317, PubMed:8463281, PubMed:8613547).

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.