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Rabbit Anti-GBE1 antibody

SL13300R

Product Name:	GBE1
Chinese Name:	分支酶GBE1抗体
Alias:	1,4 alpha glucan branching enzyme; 4-alpha-glucan-branching enzyme; amylo (1,4 to 1,6) transglucosidase; amylo (1,4 to 1,6) transglycosylase; Andersen disease; Brancher enzyme; GBE 1; GBE; GBE1; gGlucan (1,4 alpha), branching enzyme 1; GLGB_HUMAN; Glucan (1,4 alpha) branching enzyme; Glycogen branching enzyme; Glycogen storage disease type IV; Glycogen-branching enzyme; OTTHUMP00000213788; OTTHUMP00000213833.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Rabbit,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	80kDa
Cellular localization:	cytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human GBE1:101-200/702
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% 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 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	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.

Function:
Required for sufficient glycogen accumulation. The alpha 1-6 branches of glycogen play an important role in increasing the solubility of the molecule and, consequently, in reducing the osmotic pressure within cells.

Tissue Specificity:
Highest levels found in liver and muscle.

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.

Similarity:
Belongs to the glycosyl hydrolase 13 family.

SWISS:
Q04446

	Gene ID: 2632 **Database links:** Entrez Gene: 100034152Horse Entrez Gene: 2632Human Entrez Gene: 74185Mouse Omim: 607839Human SwissProt: Q6EAS5Horse SwissProt: Q04446Human SwissProt: Q9D6Y9Mouse **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.