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GAA Rabbit pAb

Catalog # AP99641

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	P10253
Other Accession	P10253
Reactivity	Mouse
Predicted	Rat, Pig, Human, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	105324
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human GAA/Glucosidase alpha
Epitope Specificity	541-640/952
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 31 family. Contains 1 P-type (trefoil) domain.
Post-translational modifications	The different forms of acid glucosidase are obtained by proteolytic processing. Phosphorylation of mannose residues ensures efficient transport of the enzyme to the lysosomes via the mannose 6-phosphate receptor.
DISEASE	Defects in GAA are the cause of glycogen storage disease type 2 (GSD2) [MIM:232300]; also called acid alpha-glucosidase (GAA) deficiency or acid maltase deficiency (AMD). GSD2 is a metabolic disorder with a broad clinical spectrum. The severe infantile form, or Pompe disease, presents at birth with massive accumulation of glycogen in muscle, heart and liver. Cardiomyopathy and muscular hypotonia are the cardinal features of this form whose life expectancy is less than two years. The juvenile and adult forms present as limb-girdle muscular dystrophy beginning in the lower limbs. Final outcome depends on respiratory muscle failure. Patients with the adult form can be free of clinical symptoms for most of their life but finally develop a slowly progressive myopathy.
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 acid alpha-glucosidase, which is essential for the degradation of glycogen to glucose in lysosomes. Different forms of acid alpha-glucosidase are obtained by proteolytic processing. Defects in this gene are the cause of glycogen storage disease II, also known as Pompe's disease, which is an autosomal recessive disorder with a broad clinical spectrum. Three transcript variants encoding the same protein have been found for this gene. [provided by RefSeq, Jul 2008].

Additional Information

Gene ID	2548
Other Names	IOPD; LOPD; LYAG; E430018M07Rik; LYAG_HUMAN; GAA; Acid maltase; Aglucosidase alfa; 3.2.1.20; LYAG_MOUSE; LYAG_RAT; alpha glucosidase; glucosidase, alpha; acid; glucosidase alpha, acid; lysosomal alpha-glucosidase; Pompe disease; glycogen storage disease type II
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:200-800
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	GAA
Function	Essential for the degradation of glycogen in lysosomes (PubMed: 14695532, PubMed:18429042, PubMed:1856189, PubMed:7717400). Has highest activity on alpha-1,4-linked glycosidic linkages, but can also hydrolyze alpha-1,6-linked glucans (PubMed:29061980).
Cellular Location	Lysosome. Lysosome membrane

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