Active Recombinant Human Acid α-glucosidase (GAA)

Cat. No. GAA-002H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant human GAA protein was expressed in HEK293T.
Species	Human
Source	HEK293
Description	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.
Form	Lyophilized from sterile PBS (pH7.4), 300 mM NaCl, 1 mM DTT.
Bio-activity	Hydrolysis of terminal, non-reducing (1->4)-linked alpha-D-glucose residues with release of alpha-D-glucose.
Molecular Mass	The recombinant α-glucosidase comprises 952 amino acids with a predicted MW of 105 kDa. Then it is cleaved into the following 2 chains: 76 kDa or 70 kDa during post-translational modification.
Storage	Store it at +4ºC for short term (4 weeks). For long term storage(12 months), store it at -20ºC~-70ºC from date of receipt. Avoid freeze-thaw cycles.

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Reconstitution	It is recommended that sterile water be added to the vial to prepare a stock solution of 0.2 ug/ul. Centrifuge the vial at 4℃ before opening to recover the entire contents.

GENE INFORMATION

Gene Name	GAA glucosidase, alpha; acid [Homo sapiens]
Official Symbol	GAA
Synonyms	GAA; glucosidase, alpha; acid; lysosomal alpha-glucosidase; glycogen storage disease type II; Pompe disease; acid maltase; aglucosidase alfa; LYAG;
Gene ID	2548
mRNA Refseq	NM_000152
Protein Refseq	NP_000143
MIM	606800
UniProt ID	P10253
Chromosome Location	17q25.2-q25.3
Pathway	Galactose metabolism, organism-specific biosystem; Galactose metabolism, conserved biosystem; Lysosome, organism-specific biosystem; Lysosome, conserved biosystem; Metabolic pathways, organism-specific biosystem; Notch-mediated HES/HEY network, organism-specific biosystem; Starch and sucrose metabolism, organism-specific biosystem;

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Function	alpha-glucosidase activity; carbohydrate binding; hydrolase activity, hydrolyzing O-glycosyl compounds; maltose alpha-glucosidase activity;

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