

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 +4°C for short term (4 weeks). For long term storage(12 months), store it at -20°C~-70°C 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°C 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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