

## Active Recombinant Human Acid $\alpha$ -glucosidase (GAA)

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

### SPECIFICATION

|                         |                                                                                                                                                                                                                                                                                                                                                                                                                                                                       |
|-------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Product Overview</b> | Recombinant human GAA protein was expressed in HEK293T.                                                                                                                                                                                                                                                                                                                                                                                                               |
| <b>Species</b>          | Human                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
| <b>Source</b>           | HEK293                                                                                                                                                                                                                                                                                                                                                                                                                                                                |
| <b>Description</b>      | 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. |
| <b>Form</b>             | Lyophilized from sterile PBS (pH7.4), 300 mM NaCl, 1 mM DTT.                                                                                                                                                                                                                                                                                                                                                                                                          |
| <b>Bio-activity</b>     | Hydrolysis of terminal, non-reducing (1->4)-linked alpha-D-glucose residues with release of alpha-D-glucose.                                                                                                                                                                                                                                                                                                                                                          |
| <b>Molecular Mass</b>   | The recombinant $\alpha$ -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.                                                                                                                                                                                                                                                              |
| <b>Storage</b>          | 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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