

Active Recombinant Human GAA protein, His-tagged

Cat. No. GAA-173H **Lot. No.** (See product label)

SPECIFICATION

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| Product Overview | Recombinant Human GAA(Ala70-Cys952) fused with His tag at N-terminal was expressed in HEK293. |
| Species | Human |
| Source | HEK293 |
| ProteinLength | 70-952 a.a. |
| Description | <p>Acid alpha-glucosidase (GAA) is an enzyme that is essential in the degradation of glycogen to glucose in the lysosome. Defects in GAA are the cause of glycogen storage disease II, also known as Pompe's disease, which is a rare autosomal recessive metabolic disorder that damages muscle and nerve cells throughout the body, primarily due to the accumulation of glycogen in the lysosome. Pompe disease occurs in babies, children, and adults who inherit a defective GAA gene and affects an estimated 5,000 to 10,000 people worldwide. Enzyme replacement therapy (ERT) is used to treat patients with this disease.</p> |
| Predicted N Terminal | His |
| Form | Supplied as a 0.2 µm filtered solution in Tris, NaCl and Glycerol. |
| Bio-activity | Measured by its ability to release glucose from starch. The specific activity is >7,500 pmol/min/ug. |

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|-----------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Molecular Mass | Predicted Molecular Mass: 99 kDa;SDS-PAGE: 95-105 kDa, reducing conditions |
| Endotoxin | <1.0 EU per 1 µg of the protein by the LAL method. |
| Purity | >95%, by SDS-PAGE under reducing conditions and visualized by Colloidal Coomassie® Blue stain at 5 µg per lane. |
| Storage | Avoid repeated freeze-thaw cycles.6 months from date of receipt, -20 to -70 centigrade as supplied.3 months, -20 to -70 centigrade under sterile conditions after opening. |

GENE INFORMATION

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|------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------|
| 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 | 17q25.2-q25.3 |

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Location**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;

Function

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

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