

## Recombinant Human GAA protein, MYC/DDK-tagged

Cat. No. GAA-177H    Lot. No. (See product label)

### SPECIFICATION

|                  |                                                                                                                                                                                                                                                                                                                                                                                                                                                                       |
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| Product Overview | Recombinant Human GAA, transcript variant 2, fused with MYC/DDK tag at C-terminal was expressed in HEK293.                                                                                                                                                                                                                                                                                                                                                            |
| 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             | 25 mM Tris.HCl, pH 7.3, 100 mM glycine, 10% glycerol.                                                                                                                                                                                                                                                                                                                                                                                                                 |
| Molecular Mass   | 102.5 kDa                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
| Purity           | > 80% as determined by SDS-PAGE and Coomassie blue staining.                                                                                                                                                                                                                                                                                                                                                                                                          |
| Concentration    | >50 ug/mL as determined by microplate BCA method                                                                                                                                                                                                                                                                                                                                                                                                                      |

### GENE INFORMATION

|           |                                               |
|-----------|-----------------------------------------------|
| Gene Name | GAA glucosidase, alpha; acid [ Homo sapiens ] |
|-----------|-----------------------------------------------|

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| <b>Official Symbol</b>     | <a href="#">GAA</a>                                                                                                                                                                                                                                                                                                                           |
| <b>Synonyms</b>            | GAA; glucosidase, alpha; acid; lysosomal alpha-glucosidase; glycogen storage disease type II; Pompe disease; acid maltase; aglucosidase alfa; LYAG;                                                                                                                                                                                           |
| <b>Gene ID</b>             | <a href="#">2548</a>                                                                                                                                                                                                                                                                                                                          |
| <b>mRNA Refseq</b>         | <a href="#">NM_001079803</a>                                                                                                                                                                                                                                                                                                                  |
| <b>Protein Refseq</b>      | <a href="#">NP_001073271</a>                                                                                                                                                                                                                                                                                                                  |
| <b>MIM</b>                 | <a href="#">606800</a>                                                                                                                                                                                                                                                                                                                        |
| <b>UniProt ID</b>          | <a href="#">P10253</a>                                                                                                                                                                                                                                                                                                                        |
| <b>Chromosome Location</b> | 17q25.2-q25.3                                                                                                                                                                                                                                                                                                                                 |
| <b>Pathway</b>             | 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; |
| <b>Function</b>            | alpha-glucosidase activity; carbohydrate binding; hydrolase activity, hydrolyzing O-glycosyl compounds; maltose alpha-glucosidase activity;                                                                                                                                                                                                   |