Recombinant Human Acid-glucosidase (GAA)

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

<table>
<thead>
<tr>
<th>Specification</th>
<th>Details</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Product Overview</strong></td>
<td>Recombinant human GAA protein was expressed in HEK293T.</td>
</tr>
<tr>
<td><strong>Description</strong></td>
<td>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.</td>
</tr>
<tr>
<td><strong>Source</strong></td>
<td>HEK293T</td>
</tr>
<tr>
<td><strong>Species</strong></td>
<td>Human</td>
</tr>
<tr>
<td><strong>Form</strong></td>
<td>Lyophilized from sterile PBS (pH7.4), 300 mM NaCl, 1 mM DTT.</td>
</tr>
<tr>
<td><strong>Bio-activity</strong></td>
<td>Hydrolysis of terminal, non-reducing (1-&gt;4)-linked alpha-D-glucose residues with release of alpha-D-glucose.</td>
</tr>
<tr>
<td><strong>Molecular Mass</strong></td>
<td>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.</td>
</tr>
<tr>
<td><strong>Storage</strong></td>
<td>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.</td>
</tr>
<tr>
<td><strong>Reconstitution</strong></td>
<td>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.</td>
</tr>
</tbody>
</table>

**Gene Information**

<table>
<thead>
<tr>
<th>Gene Name</th>
<th>GAA glucosidase, alpha; acid [ Homo sapiens ]</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Official Symbol</strong></td>
<td>GAA</td>
</tr>
<tr>
<td><strong>Synonyms</strong></td>
<td>GAA; glucosidase, alpha; acid; lysosomal alpha-glucosidase; glycogen storage disease type II; Pompe disease; acid maltase; aglucosidase alfa; LYAG;</td>
</tr>
<tr>
<td><strong>Gene ID</strong></td>
<td>2548</td>
</tr>
<tr>
<td><strong>mRNA Refseq</strong></td>
<td>NM_000152</td>
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<tr>
<td><strong>Protein Refseq</strong></td>
<td>NP_000143</td>
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<tr>
<td><strong>MIM</strong></td>
<td>606800</td>
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<tr>
<td><strong>UniProt ID</strong></td>
<td>P10253</td>
</tr>
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<table>
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<tr>
<th>Chromosome Location</th>
<th>17q25.2-q25.3</th>
</tr>
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<tbody>
<tr>
<td>Pathway</td>
<td>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;</td>
</tr>
<tr>
<td>Function</td>
<td>Alpha-glucosidase activity; carbohydrate binding; hydrolase activity, hydrolyzing O-glycosyl compounds; maltose alpha-glucosidase activity;</td>
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