Recombinant Human GAA protein, MYC/DDK-tagged

Cat. No. GAA-176H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human GAA, transcript variant 1, 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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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;
Function	alpha-glucosidase activity; carbohydrate binding; hydrolase activity, hydrolyzing O-glycosyl compounds; maltose alpha-glucosidase activity;

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