

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

**Cat. No.** GBA-179H    **Lot. No.** (See product label)

### SPECIFICATION

<b>Product Overview</b>	Recombinant Human GBA, transcript variant 3, fused with MYC/DDK tag at C-terminal was expressed in HEK293.
<b>Species</b>	Human
<b>Source</b>	HEK293
<b>Description</b>	This gene encodes a lysosomal membrane protein that cleaves the beta-glucosidic linkage of glycosylceramide, an intermediate in glycolipid metabolism. Mutations in this gene cause Gaucher disease, a lysosomal storage disease characterized by an accumulation of glucocerebrosides. A related pseudogene is approximately 12 kb downstream of this gene on chromosome 1. Alternative splicing results in multiple transcript variants.
<b>Form</b>	25 mM Tris.HCl, pH 7.3, 100 mM glycine, 10% glycerol.
<b>Molecular Mass</b>	55.5 kDa
<b>Purity</b>	> 80% as determined by SDS-PAGE and Coomassie blue staining.
<b>Concentration</b>	>50 ug/mL as determined by microplate BCA method

### GENE INFORMATION

<b>Gene Name</b>	GBA glucosidase, beta, acid [ Homo sapiens ]
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<b>Official Symbol</b>	<a href="#">GBA</a>
<b>Synonyms</b>	GBA; glucosidase, beta, acid; GLUC, glucosidase, beta; acid (includes glucosylceramidase) , glucosylceramidase; glucosylceramidase; GBA1; alglucerase; imiglucerase; acid beta-glucosidase; beta-glucocerebrosidase; lysosomal glucocerebrosidase; D-glucosyl-N-acylsphingosine glucohydrolase; GCB; GLUC;
<b>Gene ID</b>	<a href="#">2629</a>
<b>mRNA Refseq</b>	<a href="#">NM_001005742</a>
<b>Protein Refseq</b>	<a href="#">NP_001005742</a>
<b>MIM</b>	<a href="#">606463</a>
<b>UniProt ID</b>	<a href="#">P04062</a>
<b>Chromosome Location</b>	1q22
<b>Pathway</b>	Glycosphingolipid metabolism, organism-specific biosystem; Lysosome, organism-specific biosystem; Lysosome, conserved biosystem; Metabolic pathways, organism-specific biosystem; Metabolism, organism-specific biosystem; Metabolism of lipids and lipoproteins, organism-specific biosystem; Other glycan degradation, organism-specific biosystem;
<b>Function</b>	cation binding; glucosylceramidase activity; hydrolase activity, acting on glycosyl bonds; receptor binding;

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