

Recombinant Human GBA protein, MYC/DDK-tagged

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

SPECIFICATION

Product Overview	Recombinant Human GBA, transcript variant 3, fused with MYC/DDK tag at C-terminal was expressed in HEK293.
Species	Human
Source	HEK293
Description	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.
Form	25 mM Tris.HCl, pH 7.3, 100 mM glycine, 10% glycerol.
Molecular Mass	55.5 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	GBA glucosidase, beta, acid [Homo sapiens]
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Official Symbol	GBA
Synonyms	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;
Gene ID	2629
mRNA Refseq	NM_001005742
Protein Refseq	NP_001005742
MIM	606463
UniProt ID	P04062
Chromosome Location	1q22
Pathway	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;
Function	cation binding; glucosylceramidase activity; hydrolase activity, acting on glycosyl bonds; receptor binding;