Active Recombinant Human GBA, MYC/DDK-tagged

Cat. No. GBA-020H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human GBA, fused with C-terminal MYC/DDK, was expressed in HEK293 cells.
Species	Human
Source	HEK293
ProteinLength	1-536 aa
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.
Bio-activity	The enzymatic activity of GBA was measured by its ability to hydrolyze a fluorescent substrate 4-methylumbelliferyl-ß-D-glucopyranoside. The specific activity is > 70,000 pmol/hour/μg, as measured under the following conditions: 27 ng of GBA was incubated with 10 mM 4-methylumbelliferyl- ß-D-glucopyranoside in the following buffer at 37°C for 40 min: 150 mM citrate-phosphate buffer, pH 5.4, 0.25% (w/w) sodium taurocholate, 0.25% (w/w) Triton X-100, and 1% bovine serum albumin. The reaction was terminated by adding 0.5 volume of 1M glycine buffer, pH 12.5. The hydrolyzed product of reaction, 4-methylumbelliferone (4-MU), was measured using a FlexStation 3 microplat

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	e reader (Ex365/Em445). Specific activity of GBA was calculated based on a standar d curve of known concentration of 4-MU.
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
Storage Buffer	25 mM Tris.HCl, pH 7.3, 100 mM glycine, 10% glycerol.

GENE INFORMATION

Gene Name	GBA glucosidase, beta, acid [Homo sapiens]
Official Symbol	GBA
Synonyms	GBA; GCB; GBA1; GLUC; glucosidase, beta, acid; glucosylceramidase; beta-GC; alglucerase; imiglucerase; acid beta-glucosidase; beta-glucocerebrosidase; lysosomal glucocerebrosidase; D-glucosyl-N-acylsphingosine glucohydrolase; NP_000148.2; EC 3.2.1.45; NP_001005741.1; NP_001005742.1; NP_001165282.1; NP_001165283.1
Gene ID	2629
mRNA Refseq	NM_000157
Protein Refseq	NP_000148
MIM	606463
UniProt ID	P04062

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Chromosome Location	1q21
Pathway	Glycosphingolipid metabolism; Other glycan degradation; Sphingolipid metabolism
Function	glucosylceramidase activity; protein binding; receptor binding

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