

Recombinant Human GBA therapeutic protein

Cat. No. GBA-P047H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human Beta-glucocerebrosidase or Beta-D-glucosyl-N-acylsphingosine glucosylhydrolase E.C. 3.2.1.45. 497 residue protein with N-linked carbohydrates, MW=59.3 kD. It is prepared by modification of the oligosaccharide chains of human Beta-glucocerebrosidase. The modification alters the sugar residues at the non-reducing ends of the oligosaccharide chains of the glycoprotein so that they are predominantly terminated with mannose residues
Species	Human
ProteinLength	497 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. Our expression product is the active ingredient of Ceredase and Cerezyme.
Molecular Mass	55.6 Kda
AA Sequence	ARPCIPKSFGYSSVVCVCNATYCDSFDPPTFPALGTFSTRYESTRSGRRMELSMGPI QANHTGTGLLLTLQP EQKFQKVKGFGGAMTDAAALNILALSPPAQNLLKSYFSEE GIGYNIIRVPMASCDFSIRTYTYADTPDDF QLHNFSLPEEDTKLKIPLIHRALQLAQR

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VSLASPWTSPTWLKTNGAVNGKGS�KGQPGDIYHQTWARYF VKFLDAYAEHKLQ
FWAVTAENEPSAGLLSGYPFQCLGFTPEHQRFIARDLGPTLANSTHHNVRLMLD
DQ RLLLPHWAKVVLTDPEAAKYVHGIAVHWYLDFLAPAKATLGETHRLFPNTMLFA
SEACVGSKFWEQSVRLG SWDRGMQYSHSIITNLLYHVVGWTDWNLALNPEGGP
WVRNFVDSPiIVDITKDTFYKQPMFYHLGH FSKFIPEGSSQRVGLVASQKNDLDAVAL
MHPDGSASVVVVLNRSSKDVPLTIKDPVGFLETISPGYSIHTYL WRRQ

Endotoxin < 1.0 EU per µg of the protein

Purity >95%

Storage Can be stored at +4 centigrade short term (1-2weeks). For long term storage, aliquot and store at -20 centigrade or -70 centigrade. Avoid repeated freezing and thawing cycles.

GENE INFORMATION

Gene Name GBA glucosidase, beta, acid [Homo sapiens]

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 glucosylhydrolase; GCB; GLUC;

Gene ID 2629

mRNA Refseq NM_000157

Protein Refseq NP_000148

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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;

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