

## 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 glucohydrolase 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**

ARPCIPKSFYSSVVCNATYCDSFDPPTFPALGTFSTRYESTRSGRRMELSMGPI  
QANHTGTGLLLTLQP EQKFQKVKGFGGAMTDAALNILALSPPAQNLLLSYFSEE  
GIGYNIIRVPMASCDFSIRTYTYADTPDDF QLHNFSLPEEDTKLKIPLIHRALQLAQRP

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VSLLASPWTSPWLKTNGAVNGKGS�KGQPGLDIYHQTWARYF VKFLDAYAEHLQ  
 FWAVTAENEPSAGLLSGYPFQCLGFTPEHQRFIARDLGPTLANSTHHNVRLMLD  
 DQ RLLPHWAKVVLTDPEAAKYVHGIAVHWYLDLFLAPAKATLGETHRLFPNTMLFA  
 SEACVGSKFWEQSVRLG SWDRGMQYSHSIITNLLYHVVGWTDWNLALNPEGGPN  
 WVRNFVDSPIIVDITKDTFYKQPMFYHLGH FSKFIPEGSQRVGLVASQKNDLDAVAL  
 MHPDGSVVVVLNRSSKDVPLTIKDPVGFLETISPGYSIHTYL 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 glucohydrolase; GCB; GLUC;

**Gene ID** 2629

**mRNA Refseq** NM\_000157

**Protein Refseq** NP\_000148

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<b>MIM</b>	606463
<b>UniProt ID</b>	P04062
<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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