

# Recombinant Human GBA therapeutic protein(Velaglucerase alfa)

**Cat. No.** GBA-P024H    **Lot. No.** (See product label)

## SPECIFICATION

**Product Overview**      The therapeutic protein is a hydrolytic lysosomal glucocerebroside-specific enzyme, which is a recombinant form of glucocerebrosidase indicated as a long-term enzyme replacement therapy for those suffering of Gaucher disease Type 1. It has an identical amino acid sequence to the naturally occurring enzyme. It is used for the treatment of Type 1 Gaucher disease, caused by a deficiency of the lysosomal enzyme glucocerebrosidase. Additionally, Velaglucerase alfa has also been investigated for use in Type 3 Gaucher disease.

**Species**      Human

**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.

**Molecular Mass**      ~63 kDa

**AA Sequence**      ARPCIPKSFYSSVVCVCNATYCDSFDPPTFPALGTFSTRYESTRSGRRMELSMGPI  
 QANHTGTGLLLTLQP EQKFQKVKGFGGAMTDAAALNILALSPPAQNLLKSYFSEE  
 GIGYNIIRVPMASCDFSIRTYTYADTPDDF QLHNFSLPEEDTKLKIPLIHRALQLAQRP  
 VLLASPWTSPTWLKTNGAVNGKGS�KGQPGDIYHQTWARYF VKFLDAYAEHKLQ

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FWAVTAENEPSAGLLSGYPFQCLGFTPEHQRFIARDLGPTLANSTHHNVRLMLD  
 DQ RLLPHWAKVVLTDPEAAKYVHGIAVHWYLDLFLAPAKATLGETHRLFPNTMLFA  
 SEACVGSKFWEQSVRLG SWDRGMQYSHSIITNLLYHVVGWTDWNLALNPEGGPN  
 WVRNFVDSPPIVDITKDTFYKQPMFYHLGH FSKFIPEGSQRVGLVASQKNDLDAVAL  
 MHPDGSAVVVVVLRSSKDVPLTIKDPAVGFLETISPGYSIHTYL WRRQ

**Endotoxin** < 0.1 EU per µg of the protein

**Purity** >96%

**Alias** GBA; GBA1; GCB; GLUC; Velaglucerase alfa

## 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](#)

**MIM** [606463](#)

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<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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