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	ARPCIPKSFGYSSVVCVCNATYCDSFDPPTFPALGTFSRYESTRSGRRMELSMGPI QANHTGTGLLLTLQP EQKFQKVKGFGGAMTDAAALNILALSPPAQNLLLKSYFSEE GIGYNIIRVPMASCDFSIRTYTYADTPDDF QLHNFSLPEEDTKLKIPLIHRALQLAQRP VSLLASPWTSPTWLKTNGAVNGKGSLKGQPGDIYHQTWARYF VKFLDAYAEHKLQ

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	FWAVTAENEPSAGLLSGYPFQCLGFTPEHQRDFIARDLGPTLANSTHHNVRLLMLD DQ RLLLPHWAKVVLTDPEAAKYVHGIAVHWYLDFLAPAKATLGETHRLFPNTMLFA SEACVGSKFWEQSVRLG SWDRGMQYSHSIITNLLYHVVGWTDWNLALNPEGGPN WVRNFVDSPIIVDITKDTFYKQPMFYHLGH FSKFIPEGSQRVGLVASQKNDLDAVAL MHPDGSAVVVVLNRSSKDVPLTIKDPAVGFLETISPGYSIHTYL 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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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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