

Recombinant Human GBA

Cat. No. GBA-28984TH **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant full length Human GBA with a N terminal proprietary tag: predicted molecular weight 84.70 kDa.
Species	Human
Source	Wheat Germ
ProteinLength	536 amino acids
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 Weight	84.700kDa inclusive of tags
Form	Liquid
Purity	Proprietary Purification
Storage buffer	pH: 8.00 Constituents: 0.3% Glutathione, 0.79% Tris HCl
Storage	Shipped on dry ice. Upon delivery aliquot and store at -80oC. Avoid freeze / thaw

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cycles.

Sequences of amino acids

MEFSSPSREECPKPLSRVSIAGSLTGLLLLQAVSWASGA RPCIPKSFSGYSSVVCVC
 NATYCDSDPPTFPALGTFSRYE STRSGRRMELSMGPIQANHTGTLLLLTLQPEQK
 FQKVKGFF GGAMTDAALNILALSPPAQNLLLKSYFSEEGIGYNIIRV PMASCDFSIRT
 YTYADTPDDFQLHNFSLPEEDTKLKIPLI HRALQLAQRPVSLASPWTSPWLKTNB
 AVNGKGSLSKGGP GDIYHQTWARYFVKFLDAYAEHKLQFWAVTAENEPSAGLL SGY
 PFQCLGFTPEHQDFIARDLGPSTLANSTHHNVRLLML DDQRLLLPHWAKVVLTDPE
 AAKYVHGIADVHWYLDLAPAK ATLGETHRLFPNTMLFASEACVGSKFWEQSVRLGS
 WDRGM QYSHSIITNLLYHVVGWTDWNLALNPEGGPNWVRNFVDSP IIVDITKDIFY
 KQPMFYHLGHFSKFIPEGSQRVGLVASQK NDLDAVALMHPDGSVVVVVLRSSKD
 VPLTIKDPVGFLE TISPGYSIHTYLWRRQ

Sequence Similarities

Belongs to the glycosyl hydrolase 30 family.

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;

Gene ID

2629

mRNA Refseq

NM_000157

Protein Refseq

NP_000148

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MIM	606463
Uniprot ID	P04062
Chromosome Location	1q22
Pathway	Lysosome, organism-specific biosystem; Lysosome, conserved biosystem; Metabolic pathways, organism-specific biosystem; Other glycan degradation, organism-specific biosystem; Other glycan degradation, conserved biosystem;
Function	cation binding; glucosylceramidase activity; hydrolase activity, acting on glycosyl bonds; receptor binding;

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