

# Recombinant Full Length Human GBA Protein

**Cat. No.** GBA-186HF    **Lot. No.** (See product label)

## SPECIFICATION

|                         |                                                                                                                                                                                                                                                                                                                                                                                                                                            |
|-------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Product Overview</b> | Recombinant full length Human GBA with a N terminal proprietary tag: predicted molecular weight 84.70 kDa.                                                                                                                                                                                                                                                                                                                                 |
| <b>Species</b>          | Human                                                                                                                                                                                                                                                                                                                                                                                                                                      |
| <b>Source</b>           | In Vitro Cell Free System                                                                                                                                                                                                                                                                                                                                                                                                                  |
| <b>ProteinLength</b>    | 536 amino acids                                                                                                                                                                                                                                                                                                                                                                                                                            |
| <b>Description</b>      | 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. |
| <b>Form</b>             | Liquid                                                                                                                                                                                                                                                                                                                                                                                                                                     |
| <b>Molecular Mass</b>   | 84.700kDa inclusive of tags                                                                                                                                                                                                                                                                                                                                                                                                                |
| <b>AA Sequence</b>      | MEFSSPSREECPKPLSRVSIAGSLTGLLLLQAVSWASGA RPCIPKSFSGYSSVVCVC<br>NATYCDSFDPPTFPALGTFSRYE STRSGRRMELSMGPIQANHTGTGLLLTLQPEQK<br>FQKVKGFFGGAMTDAAALNILALSPPAQNLLLSYFSEEGIGYNIIRV PMASCDIFSIRT<br>YTYADTPDDFQLHNFSLPEDTKLKIPLI HRALQLAQRPVSLASPWTSPWLKTNG<br>AVNGKGSLSKGQP GDIYHQTWARYFVKFLDAYAEHKLQFWAVTAENEPSAGLL SGY                                                                                                                           |

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PFQCLGFTPEHQQRDFIARDLGPTLANSTHHNVRLML DDQRLLPHWAKVVLTDPE  
AAKYVHGIADVHWYLDLAPAK ATLGETHRLFPNTMLFASEACVGSKFWEQSVRLGS  
WDRGM QYSHSIITNLLYHVVGWTDWNLALNPEGGPNNWRNFVDSP IVDITKDTFY  
KQPMFYHLGHFSKFIPEGSQRVGLVASQK NDLDVALMHPDGSAVVVVLNRSSKD  
VPLTIKDPVGFLE TISPGYSIHTYLWRRQ

**Purity** Proprietary Purification

**Storage** Shipped on dry ice. Upon delivery aliquot and store at -80 centigrade. Avoid freeze / thaw cycles.

**Storage Buffer** pH: 8.00. Constituents: 0.79% Tris HCl, 0.31% Glutathione.

## 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

**MIM** 606463

**UniProt ID** P04062

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