

Recombinant Human GBA therapeutic protein, Imiglucerase

Cat. No. GBA-P028H **Lot. No.** (See product label)

SPECIFICATION

Product Overview

Recombinant Human β -glucocerebrosidase therapeutic protein is the recombinant active form of the human lysosomal enzyme, β -glucocerebrosidase for use in patients with type 1 Gaucher's disease. It is a modified form of glucocerebrosidase and is provided to counter this enzyme deficiency, resulting in smaller liver and spleen size, and improved thrombocytopenia and anemia. This recombinant enzyme is different from human glucocerebrosidase by two amino acids at the N terminal and up to 7 amino acids at the C terminal. It allows the hydrolysis reaction of glucocerebroside to glucose and ceramide that naturally occurs in healthy individuals.

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 56637.9397

Endotoxin < 0.1 EU per μ g of the protein

Purity >98%

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Alias GBA; GBA1; GCB; GLUC; Imiglucerase

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

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;

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Function

cation binding; glucosylceramidase activity; hydrolase activity, acting on glycosyl bonds; receptor binding;

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