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Product Information

Anti-Glucocerebrosidase
produced in rabbit, affinity isolated antibody

Product Number **G4046**

Product Description

Anti-Glucocerebrosidase is produced in rabbit using as immunogen a synthetic peptide corresponding to a fragment of human glucocerebrosidase (GBA) (GeneID: 2629), conjugated to KLH. This sequence is highly conserved in mouse and rat GBA (78% identity). The antibody is affinity-purified using the immunizing peptide immobilized on agarose.

Anti-Glucocerebrosidase specifically recognizes human GBA by immunoblotting (~60 kDa). Staining of the GBA band is specifically inhibited by the GBA immunizing peptide.

Gaucher disease (GD) is the most prominent inherited lysosomal storage disorder. The disease is caused by defects in the activity of the lysosomal hydrolase glucocerebrosidase (D-glucosyl-N-acylsphingosine glucohydrolase, GBA, GlcCerase) resulting in the accumulation of glucosylceramide (GlcCer) in lysosomes of cells of the reticuloendothelial system.[1-3] Mutations in the human GBA gene cause a reduction in the GBA activity and accumulation of GlcCer.

Significant heterogeneity is observed in GD with three main types known: non-neuronopathic (type-1), infantile (type- 2), and juvenile (type-3) neuronopathic types. The acute neuronopathic type of the disease is characterized by severe loss of neurons in the central nervous system and early onset of the disease. GlcCer accumulation in neurons causes changes in axonal morphology.

In addition, it leads to changes in neuronal functionality including increased levels of tubular endoplasmic reticulum (ER) elements, a large increase in Ca^{2+} release from the ER in response to glutamate, and an increased sensitivity to glutamate-induced neurotoxicity. Fibroblasts from patients with defined GBA mutations show either retarded or blocked transport of GBA in the ER.[3] Mutations in the human GBA gene may contribute to the development of common age-related dementia known as dementia with Lewy bodies or DLB. Several studies indicate that mutations in the human GBA gene are associated with early-onset Parkinson disease.[4-5]

Reagent

Supplied as a solution in 0.01 M phosphate buffered saline, pH 7.4, containing 15 mM sodium azide.

Antibody concentration: ~1.0 mg/mL

Precautions and Disclaimer

For R&D use only. Not for drug, household, or other uses. Please consult the Safety Data Sheet for information regarding hazards and safe handling practices.

Storage/Stability

For continuous use, store at 2–8 °C for up to one month. For extended storage, freeze in working aliquots. Repeated freezing and thawing, or storage in "frost-free" freezers, is not recommended. If slight turbidity occurs upon prolonged storage, clarify the solution by centrifugation before use. Working dilutions should be discarded if not used within 12 hours.

Product Profile

Immunoblotting: a working concentration of 1-2 μg/mL is recommended using HEK-293Tcell lysate expressing human glucocerebrosidase (GBA) or using SH-SY-5Y cell lysate.

Note: In order to obtain best results in various techniques and preparations, it is recommended to determine optimal working dilutions by titration.

References

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3. Schmitz, M. et al., *Int. J. Biochem. Cell Biol.*, **37**, 2310-2320 (2005).
4. Clark, L.N. et al., *Neurology*, **69**, 1270-1277 (2007).
5. Ziegler, S.G. et al., *Mol. Genet. Metab.*, **91**, 195-200 (2007).

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