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# GM2A Rabbit pAb

Catalog # AP98239

## Product Information

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| **Application** | IHC-P, IHC-F, IF |
| **Primary Accession** | P17900 |
| **Other Accession** | P17900 |
| **Reactivity** | Rat, Human |
| **Host** | Rabbit |
| **Clonality** | Polyclonal |
| **Calculated MW** | 20838 |
| **Physical State** | Liquid |
| **Immunogen** | KLH conjugated synthetic peptide derived from human GM2A/SAP3 |
| **Epitope Specificity** | 131-193/193 |
| **Isotype** | IgG |
| **Purity** | affinity purified by Protein A |
| **Buffer** | 0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol. |
| **Post-translational modifications** | The serines in positions 32 and 33 are absent in 80% of the sequenced protein. |
| **DISEASE** | Defects in GM2A are the cause of GM2-gangliosidosis type AB (GM2GAB) [MIM:272750]; also known as Tay-Sachs disease AB variant. GM2-gangliosidosis is an autosomal recessive lysosomal storage disease marked by the accumulation of GM2 gangliosides in the neuronal cells. GM2GAB is characterized by GM2 gangliosides accumulation in the presence of both hexosaminidase A and B. |
| **Important Note** | This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications. |
| **Background Descriptions** | This gene encodes a small glycolipid transport protein which acts as a substrate specific co-factor for the lysosomal enzyme beta-hexosaminidase A. Beta-hexosaminidase A, together with GM2 ganglioside activator, catalyzes the degradation of the ganglioside GM2, and other molecules containing terminal N-acetyl hexosamines. Mutations in this gene result in GM2-gangliosidosis type AB or the AB variant of Tay-Sachs disease. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Nov 2009]. |

## Additional Information

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| **Gene ID** | 2760 |
| **Other Names** | GM2-AP; GM2AP; SAP-3; SAP3_HUMAN; GM2A; Cerebroside sulfate activator protein; Sphingolipid activator protein 3 (SAP-3); SAP3_MOUSE; ganglioside GM2 activator; GM2 ganglioside activator protein; GM2 ganglioside activator; sphingolipid activator protein 3; GM2-activator protein |
| **Dilution** | IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500 |
| **Storage** | Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When |

reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

## Protein Information

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| **Name** | GM2A ( HGNC:4367) |
| **Function** | Lipid-binding and transfer protein essential for lysosomal degradation of ganglioside GM2. Extracts single GM2 molecules from intralysosomal luminal vesicle (ILV) membranes and presents them in soluble form to beta-hexosaminidase A (HEXA) for cleavage of N-acetyl- D-galactosamine and conversion to GM3 (PubMed:30988135, PubMed:8900233). Forms a stoichiometric GM2-GM2A complex that serves as the substrate for HEXA (PubMed:30988135, PubMed:8900233). Also stimulates the breakdown of glycolipid GA2 by HEXA (By similarity). Possesses dual function: membrane lipid mobilization/solubilization and lipid substrate presentation to hydrolases (PubMed:30988135). The large binding pocket can accommodate several single-chain phospholipids and fatty acids (By similarity). Exhibits some calcium-independent phospholipase activity (By similarity). Participates in cholesterol transfer (PubMed:17552909). |
| **Cellular Location** | Lysosome. |

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