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Acid sphingomyelinase Rabbit pAb

Catalog # AP98446

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	P17405
Other Accession	P17405
Reactivity	Rat, Human
Predicted	Pig, Mouse, Rabbit, Dog
Host	Rabbit
Clonality	Polyclonal
Calculated MW	69936
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human Acid sphingomyelinase
Epitope Specificity	201-300/629
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the acid sphingomyelinase family.Contains 1 saposin B-type domain.
SUBUNIT	Monomer.
DISEASE	Defects in SMPD1 are the cause of Niemann-Pick disease type A (NPDA) [MIM:257200]; also known as Niemann-Pick disease classical infantile form. It is an early-onset lysosomal storage disorder caused by failure to hydrolyze sphingomyelin to ceramide. It results in the accumulation of sphingomyelin and other metabolically related lipids in reticuloendothelial and other cell types throughout the body, leading to cell death. Niemann-Pick disease type A is a primarily neurodegenerative disorder characterized by onset within the first year of life, mental retardation, digestive disorders, failure to thrive, major hepatosplenomegaly, and severe neurologic symptoms. The severe neurological disorders and pulmonary infections lead to an early death, often around the age of four. Clinical features are variable. A phenotypic continuum exists between type A (basic neurovisceral) and type B (purely visceral) forms of Niemann-Pick disease, and the intermediate types encompass a cluster of variants combining clinical features of both types A and B.Defects in SMPD1 are the cause of Niemann-Pick disease type B (NPDB) [MIM:607616]; also known as Niemann-Pick disease visceral form. It is a late-onset lysosomal storage disorder caused by failure to hydrolyze sphingomyelin to ceramide. It results in the accumulation of sphingomyelin and other metabolically related lipids in reticuloendothelial and other cell types throughout the body, leading to cell death. Clinical signs involve only visceral organs. The most constant sign is hepatosplenomegaly which can be associated with pulmonary symptoms. Patients remain free of neurologic manifestations. However, a phenotypic continuum exists between type A (basic neurovisceral) and type B (purely visceral) forms of Niemann-Pick disease, and the intermediate types encompass a cluster of variants combining clinical features of both types A and B. In Niemann-Pick disease type B, onset of the first symptoms occurs in early childhood and patients can survive into adulthood.

Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Converts sphingomyelin to ceramide. Also has phospholipase C activities toward 1,2-diacylglycerolphosphocholine and 1,2-diacylglycerolphosphoglycerol. Isoform 2 and isoform 3 have lost catalytic activity. Involvement in disease: Defects in SMPD1 are the cause of Niemann-Pick disease type A (NPDA) ; also known as Niemann-Pick disease classical infantile form. It is an early-onset lysosomal storage disorder caused by failure to hydrolyze sphingomyelin to ceramide. It results in the accumulation of sphingomyelin and other metabolically related lipids in reticuloendothelial and other cell types throughout the body, leading to cell death. Niemann-Pick disease type A is a primarily neurodegenerative disorder characterized by onset within the first year of life, mental retardation, digestive disorders, failure to thrive, major hepatosplenomegaly, and severe neurologic symptoms. The severe neurological disorders and pulmonary infections lead to an early death, often around the age of four. Clinical features are variable. A phenotypic continuum exists between type A (basic neurovisceral) and type B (purely visceral) forms of Niemann-Pick disease, and the intermediate types encompass a cluster of variants combining clinical features of both types A and B.

Additional Information

Gene ID	6609
Other Names	ASM; ASMASE; NPD; A-SMase; Zn-SMase; ASM_BOVIN; SMPD1; Acid sphingomyelinase (aSMase); 3.1.4.12; 3.1.4.3; ASM_HUMAN; ASM_MOUSE; sphingomyelin phosphodiesterase 1; sphingomyelin phosphodiesterase 1, acid lysosomal; acid sphingomyelinase; Niemann-Pick type A/B
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500,Flow-Cyt=2ug /Test
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

Name	SMPD1 (HGNC:11120)
Function	Converts sphingomyelin to ceramide (PubMed: 12563314, PubMed:1840600, PubMed:18815062, PubMed:25339683, PubMed:25920558, PubMed:27659707, PubMed:33163980). Exists as two enzymatic forms that arise from alternative trafficking of a single protein precursor, one that is targeted to the endolysosomal compartment, whereas the other is released extracellularly (PubMed:20807762, PubMed:21098024, PubMed:9660788). However, in response to various forms of stress, lysosomal exocytosis may represent a major source of the secretory form (PubMed:12563314, PubMed:20530211, PubMed:20807762, PubMed:22573858, PubMed:9393854).
Cellular Location	Lysosome. Lipid droplet. Secreted. Note=The secreted form is induced in a time- and dose-dependent by IL1B and TNF as well as stress and viral infection. This increase of the secreted form seems to be due to exocytosis of the lysosomal form and is Ca(2+)-dependent (PubMed:20530211,

PubMed:20807762, PubMed:22573858). Secretion is dependent of phosphorylation at Ser-510 (PubMed:17303575). Secretion is induced by inflammatory mediators such as IL1B, IFNG or TNF as well as infection with bacteria and viruses (PubMed:12563314, PubMed:20807762)

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