

Recombinant Protein Technical Manual Recombinant Human SMPD1/ASM Protein (His Tag)

RPES2220

Product Data:

Product SKU: RPES2220 **Size:** 10μg

Species: Human Expression host: Baculovirus-Insect Cells

Uniprot: BAD93012.1

Protein Information:

Molecular Mass: 66.3 kDa

AP Molecular Mass:

Tag: C-His

Bio-activity: Measured by its ability to cleave 2-N-Hexadecanoylamino-4-

nitrophenylphosphorylcholine (HNPPC). The specific activity is >1000

pmol/min/µg.

Purity: > 95 % as determined by reducing SDS-PAGE.

Endotoxin: $< 1.0 \text{ EU per } \mu\text{g}$ as determined by the LAL method.

Storage: Lyophilized proteins are stable for up to 12 months when stored at -20 to -80°C.

Reconstituted protein solution can be stored at 4-8°C for 2-7 days. Aliquots of

reconstituted samples are stable at < -20°C for 3 months.

Shipping: This product is provided as lyophilized powder which is shipped with ice packs.

Formulation: Lyophilized from sterile 20 mM Tris, 500 mM NaCl, 25 % glycerol, pH 7.5

Reconstitution: Please refer to the printed manual for detailed information.

Application:

Synonyms: ASM;ASMASE;NPD

Immunogen Information:

Sequence: Met 1-Pro628

Background:

Sphingomyelin phosphodiesterase 1 (SMPD1), also known as ASM (acid sphingomyelinase), is a member of the acid sphingomyelinase family of enzymes. Three isoforms have been identified, isoform 1 is 631 amino acids (aa) in length as the pro form, while Isoform 2 and isoform 3 have lost catalytic activity. The active SMPD1 isoform 1 contains one saposin B-type domain that likely interacts with sphingomyelin, and a catalytic region. Human SMPD1 is 86% aa identical to mouse SMPD1. SMPD1 is a monomeric lysosomal enzyme that converts sphingomyelin (a plasma membrane lipid) into ceramide through the removal of phosphorylcholine. This generates second messenger components that participate in signal transduction. Defects in SMPD1 are the cause of Niemann-Pick disease type A (NPA) and type B (NPB), also known as Niemann-Pick disease classical infantile form and Niemann-Pick disease visceral form. Niemann-Pick disease is a clinically and genetically heterogeneous recessive disorder. NPB has little if any neurologic involvement and patients may survive into adulthood.