

## Recombinant Human SMPD1/ASM Protein (His Tag)

**Catalog Number:** PKSH030434

**Note:** Centrifuge before opening to ensure complete recovery of vial contents.

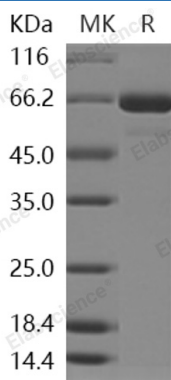
### Description

|                     |  |
|---------------------|--|
| <b>Species</b>      | Human  |
| <b>Source</b>       | Baculovirus-Insect Cells-derived Human SMPD1/ASM protein Met 1-Pro628, with an C-terminal His  |
| <b>Mol_Mass</b>     | 66.3 kDa   |
| <b>Accession</b>    | NP_000534.3  |
| <b>Bio-activity</b> | Measured by its ability to cleave 2-N-Hexadecanoylamino-4-nitrophenylphosphorylcholine (HNPPC). The specific activity is > 1000 pmol/min/μg. |

### Properties

|                       |   |
|-----------------------|---|
| <b>Purity</b>         | > 90 % as determined by reducing SDS-PAGE.  |
| <b>Endotoxin</b>      | < 1.0 EU per μg of the protein as determined by the LAL method.   |
| <b>Storage</b>        | Store at < -20°C, stable for 6 months. Please minimize freeze-thaw cycles.  |
| <b>Shipping</b>       | This product is provided as liquid. It is shipped at frozen temperature with blue ice/gel packs. Upon receipt, store it immediately at < -20°C. |
| <b>Formulation</b>    | Supplied as sterile 20 mM Tris, 500 mM NaCl, 25 % glycerol, pH 7.5.   |
| <b>Reconstitution</b> | Not Applicable  |

### Data



> 90 % as determined by reducing SDS-PAGE.

### 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.

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