



# Recombinant Protein Technical Manual

## Recombinant Human OSTM1 Protein (His Tag)

RPES2500

### Product Data:

**Product SKU:** RPES2500

**Size:** 20µg

**Species:** Human

**Expression host:** HEK293 Cells

**Uniprot:** NP\_054747.2

### Protein Information:

**Molecular Mass:** 29.7 kDa

**AP Molecular Mass:** 40-50 kDa

**Tag:** C-His

**Bio-activity:**

**Purity:** > 97 % as determined by reducing SDS-PAGE.

**Endotoxin:** < 1.0 EU per µg 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 PBS, pH 7.4

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

**Application:**

**Synonyms:** GIPN;GL;HSPC019;OPTB5

## Immunogen Information:

**Sequence:** Met 1-Pro 284

## Background:

Osteopetrosis-associated transmembrane protein 1 (OSTM1) is a Single-pass type I membrane protein. It is expressed in many hematopoietic cells of the myeloid and lymphoid B- and T-lineages. The analysis of OSTM1 association with CLCN7 demonstrated that OSTM1 requires CLCN7 to localize to lysosomes, whereas the formation of a CLCN7-OSTM1 complex is required to stabilize CLCN7. The researches found that OSTM1 plays a major role in myelopoiesis and lymphopoiesis and provided evidence of a crosstalk mechanism between hematopoietic cells for osteoclast activation. Thus, OSTM1 has an important role in osteoclast function and activation. The loss of function of OSTM1 results in deregulation of multiple hematopoietic lineages in addition to osteoclast lineage, OSTM1-defect patients display the most severe recessive osteopetrotic phenotype and die at early ages. Furthermore, it is suggested that OSTM1 has a primary role in neural development not related to lysosomal dysfunction. The canonical Wnt/beta-catenin signaling pathway may be a molecular basis for OSTM1 mutations and severe autosomal recessive osteopetrosis (ARO).