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OSTM1 Rabbit pAb

Catalog # AP96864

Product Information

Application	WB
Primary Accession	Q86WC4
Other Accession	Q86WC4
Reactivity	Rat, Human, Mouse
Predicted	Pig
Host	Rabbit
Clonality	Polyclonal
Calculated MW	37257
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human OSTM1
Epitope Specificity	21-120/334
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
DISEASE	Defects in OSTM1 are the cause of osteopetrosis autosomal recessive type 5 (OPTB5) [MIM:259720]; also called infantile malignant osteopetrosis 3. Osteopetrosis is a rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. The disorder occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. OPTB5 patients manifest primary central nervous system involvement in addition to the classical stigmata of severe bone sclerosis, growth failure, anemia, thrombocytopenia and visual impairment with optic atrophy.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	OSTM1 (osteopetrosis associated transmembrane protein 1), also known as gl (gray-lethal) or HSPC019, is a 338 amino acid single-pass type I membrane protein that is expressed primarily in osteoclasts and melanocytes as well as brain, kidney and spleen. Bone autosomal recessive osteopetrosis (ARO) is the most severe form of hereditary bone disease whose cellular basis is in the osteoclast and is characterized by abnormally dense bone, due to defective resorption of immature bone. ARO is suggested to be caused by mutations in the OSTM1 gene. The disorder occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. Defects in the OSTM1 gene are also the cause of the spontaneous gl mutant, which is responsible for a coat color defect in mice.

Additional Information

Gene ID	28962

Other Names	GIPN; GL; HSPC019; OPTB5; 1200002H13Rik; OSTM1_HUMAN; OSTM1; Chloride channel 7 beta subunit; OSTM1_MOUSE; Grey-lethal protein; osteoclastogenesis associated transmembrane protein 1; osteopetrosis associated transmembrane protein 1; CLCN7 accessory beta subunit; grey-lethal
Dilution	WB=1:500-2000,Flow-Cyt=1 μg/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	OSTM1
Synonyms	GL
Function	Required for osteoclast and melanocyte maturation and function.
Cellular Location	Lysosome membrane; Single-pass type I membrane protein Note=Requires CLCN7 to travel to lysosomes

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.