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SUMF1 Rabbit pAb

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Catalog # AP54839

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	Q8NBK3
Predicted	Rat, Pig, Dog, Human, Mouse, Rabbit, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	40556
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SUMF1
Epitope Specificity	301-374/374
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Endoplasmic reticulum lumen.
DISEASE	Defects in SUMF1 are the cause of multiple sulfatase deficiency (MSD) [MIM:272200]. MSD is a clinically and biochemically heterogeneous disorder caused by the simultaneous impairment of all sulfatases, due to defective post-translational modification and activation. It combines features of individual sulfatase deficiencies such as metachromatic leukodystrophy, mucopolysaccharidosis, chondrodysplasia punctata, hydrocephalus, ichthyosis, neurologic deterioration and developmental delay. Inheritance is autosomal recessive.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	SUMF1 is a 374 amino acid alternatively spliced protein that localizes to the lumen of the endoplasmic reticulum and belongs to the sulfatase-modifying factor family. Expressed ubiquitously with highest expression in liver, kidney and pancreas, SUMF1 exists as either a monomer, a homodimer or a heterodimer (with SUMF2) and functions to oxidize sulfatase cysteine residues to an active FGIy residue, thereby playing an important role in sulfatase activity. Defects in the gene encoding SUMF1 are the cause of multiple sulfatase deficiency (MSD), a heterogeneous disorder characterized by metachromatic leukodystrophy, mucopolysaccharidosis, chondrodysplasia punctata, hydrocephalus, ichthyosis, neurologic deterioration and developmental delay.

Additional Information

Gene ID	285362
Other Names	Formylglycine-generating enzyme, FGE, 1.8.3.7, C-alpha-formylglycine-generating enzyme 1, Sulfatase-modifying factor 1,

	SUMF1 {ECO:0000303|PubMed:12757706, ECO:0000312|HGNC:HGNC:20376}
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	SUMF1 {ECO:0000303|PubMed:12757706, ECO:0000312|HGNC:HGNC:20376}
Function	Oxidase that catalyzes the conversion of cysteine to 3- oxoalanine on target proteins, using molecular oxygen and an unidentified reducing agent (PubMed:12757706, PubMed:15657036, PubMed:15907468, PubMed:16368756, PubMed:21224894, PubMed:25931126). 3- oxoalanine modification, which is also named formylglycine (fGly), occurs in the maturation of arylsulfatases and some alkaline phosphatases that use the hydrated form of 3-oxoalanine as a catalytic nucleophile (PubMed:12757706, PubMed:15657036, PubMed:15907468, PubMed:16368756, PubMed:25931126). Known substrates include GALNS, ARSA, STS and ARSE (PubMed:12757706, PubMed:15657036, PubMed:15907468).
Cellular Location	Endoplasmic reticulum lumen
Tissue Location	Ubiquitous. Highly expressed in kidney, pancreas and liver. Detected at lower levels in leukocytes, lung, placenta, small intestine, skeletal muscle and heart

Background

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