

SUMF1 Polyclonal Antibody

Purified Rabbit Polyclonal Antibody (Pab) Catalog # AP54839

Product Information

Application WB, IHC-P, IHC-F, IF, ICC, E

Primary Accession Q8NBK3

Reactivity Rat, Pig, Dog, Bovine

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.

SIMILARITY

Belongs to the sulfatase-modifying factor family.

Monomer, homodimer and heterodimer with SUMF2.

Post-translational N-glycosylated. Contains high-mannose-type oligosaccharides. **modifications**

DISEASEDefects 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 DescriptionsSUMF1 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}

Target/Specificity Ubiquitous. Highly expressed in kidney, pancreas and liver. Detected at lower

levels in leukocytes, lung, placenta, small intestine, skeletal muscle and heart.

Dilution WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC=1:100-500,IF=1:100-50

0,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

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