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Site 2 protease Rabbit pAb

Catalog # AP99020

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	O43462
Other Accession	O43462
Reactivity	Rat, Mouse
Predicted	Human, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	57444
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human Site 2 protease
Epitope Specificity	1-100/519
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the peptidase M50A family.
DISEASE	Defects in MBTPS2 are the cause of ichthyosis follicularis-atrichia-photophobia syndrome (IFAPS) [MIM:308205]. A syndrome characterized by a peculiar triad of follicular ichthyosis, total or subtotal atrichia, and photophobia of varying degree. Histopathologically, the epidermal granular layer is generally well-preserved or thickened at the infundibulum. Hair follicles are poorly developed and tend to be surrounded by an inflammatory infiltrate. A subgroup of patients is described with lamellar rather than follicular ichthyosis. Non-consistent features may include growth and psychomotor retardation, aganglionic megacolon, seizures and nail dystrophy. Defects in MBTPS2 are a cause of keratosis follicularis spinulosa decalvans X-linked (KFSDX) [MIM:308800]. A rare disorder affecting the skin and the eye. Affected men show thickening of the skin of the neck, ears, and extremities, especially the palms and soles, loss of eyebrows, eyelashes and beard, thickening of the eyelids with blepharitis and ectropion, and corneal degeneration.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a intramembrane zinc metalloprotease, which is essential in development. This protease functions in the signal protein activation involved in sterol control of transcription and the ER stress response. Mutations in this gene have been associated with ichthyosis follicularis with atrichia and photophobia (IFAP syndrome); IFAP syndrome has been quantitatively linked to a reduction in cholesterol homeostasis and ER stress response.[provided by RefSeq, Aug 2009]

Additional Information

Gene ID	51360

Other Names	BRESEK; IFAP; KFSD; KFSDX; OI19; OLMSX; S2P; 9630032G22Rik; LRRGT00063; MBTP2_HUMAN; MBTPS2; Endopeptidase S2P; Sterol regulatory element-binding proteins intramembrane protease (SREBPs intramembrane protease); 3.4.24.85; MBTP2_MOUSE; membrane bound transcription factor peptidase, site 2; membrane-bound transcription factor protease, site 2; keratosis follicularis spinulosa decalvans; site-2 protease; sterol regulatory element-binding proteins intramembrane protease
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
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	MBTPS2 {ECO:0000303|PubMed:19361614, ECO:0000312|HGNC:HGNC:15455}
Function	Zinc metalloprotease that mediates intramembrane proteolysis of proteins such as ATF6, ATF6B, SREBF1/SREBP1 and SREBF2/SREBP2 (PubMed:10805775, PubMed:11163209). Catalyzes the second step in the proteolytic activation of the sterol regulatory element-binding proteins (SREBPs) SREBF1/SREBP1 and SREBF2/SREBP2: cleaves SREBPs within the first transmembrane segment, thereby releasing the N- terminal segment with a portion of the transmembrane segment attached (PubMed:10805775, PubMed:27380894, PubMed:9659902). Mature N-terminal SREBP fragments shuttle to the nucleus and activate gene transcription (PubMed:10805775, PubMed:27380894, PubMed:9659902). Also mediates the second step in the proteolytic activation of the cyclic AMP-dependent transcription factor ATF-6 (ATF6 and ATF6B) (PubMed:11163209). Involved in intramembrane proteolysis during bone formation (PubMed:27380894). In astrocytes and osteoblasts, upon DNA damage and ER stress, mediates the second step of the regulated intramembrane proteolytic activation of the transcription factor CREB3L1, leading to the inhibition of cell- cycle progression (PubMed:16417584).
Cellular Location	Membrane; Multi- pass membrane protein. Cytoplasm. Golgi apparatus membrane; Multi-pass membrane protein
Tissue Location	Expressed in heart, brain, placenta, lung, liver, muscle, kidney and pancreas.

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