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SMAD6 Rabbit pAb

Catalog # AP100265

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	O43541
Other Accession	O43541
Reactivity	Mouse
Predicted	Rat, Pig, Human, Rabbit, Chicken, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	53497
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SMAD6
Epitope Specificity	351-450/469
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 dwarfin/SMAD family.
SUBUNIT	Interacts with NEDD4L (By similarity). Interacts with WWP1 (By similarity). Interacts with STAMBP and PRKX. Interacts with RNF111 and AXIN1. Interacts with TGF-beta type I receptor superfamily members, including ACVR1B, BMPR1B and TGFBR1. In response to BMP2, but not to TGFB treatment, interacts with SMAD1, but not with SMAD2, nor with SMAD4; this interaction may inhibit SMAD1 binding to SMAD4. Interacts with HOXC8 and HOXC9. Interacts with PELI1; this interaction interferes with PELI1 complex formation with TRAF6, IRAK1, IRAK4 and MYD88 in response to IL1B and hence negatively regulates IL1R-TLR signaling.
Post-translational modifications	Phosphorylated by BMP type 1 receptor kinase and by PRKX. Monoubiquitinated at Lys-173 by the E2/E3 hybrid ubiquitin-protein ligase UBE2O, leading to reduced binding affinity for the activated BMP type I receptor ACVR1/ALK2, thereby enhancing BMP7 and regulating adipocyte differentiation (PubMed:23455153). Ubiquitinated by WWP1 (By similarity). Ubiquitinated by RNF165, promoting proteasomal degradation, leading to enhance the BMP-Smad signaling (By similarity). Arginine methylation by PRMT1, which is recruited by BMPR2, initiates BMP-Induced signaling and induces dissociation from the BMPR1B receptor at the cell surface leading to derepress downstream Smad1/Smad5 signaling.
DISEASE	Aortic valve disease 2 (AOVD2): The disease is caused by mutations affecting the gene represented in this entry. SMAD6 variants may contribute to increased risk of congenital cardiovascular malformations (CVM). CVM is a major cause of mortality and morbidity in childhood. In most sporadic cases that cannot be attributed to particular malformation syndromes or teratogenic exposures, there remains a substantial excess familial risk, indicating a significant genetic contribution to disease susceptibility (PubMed:22275001). Disease descriptionA common defect in the aortic valve in which two rather than three leaflets are present. It is often associated with aortic valve calcification, stenosis and insufficiency. In extreme cases, the blood flow may be so restricted that the left ventricle fails to grow, resulting in

	hypoplastic left heart syndrome.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	The protein encoded by this gene belongs to the SMAD family of proteins, which are related to Drosophila 'mothers against decapentaplegic' (Mad) and C. elegans Sma. SMAD proteins are signal transducers and transcriptional modulators that mediate multiple signaling pathways. This protein functions in the negative regulation of BMP and TGF-beta/activin-signalling. Multiple transcript variants have been found for this gene.[provided by RefSeq, Sep 2014]

Additional Information

Gene ID	4091
Other Names	AOVD2; HsT17432; MADH6; MADH7; b2b390Clo; SMAD6_HUMAN; SMAD6; MAD homolog 6; Mothers against DPP homolog 6; SMAD family member 6 (SMAD 6 | Smad6 | hSMAD6); SMAD6_MOUSE; Mad homolog 7; SMAD family member 6 (SMAD 6 | Smad6); Msmad6; SMAD family member 6; MAD, mothers against decapentaplegic homolog 6 (Drosophila); SMAD, mothers against DPP homolog 6 (Drosophila)
Dilution	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	SMAD6 (HGNC:6772)
Synonyms	MADH6
Function	Acts as an inhibitory regulator of signaling mediated by the TGF-beta superfamily, with strong selectivity toward BMP-dependent pathways (PubMed:10647776, PubMed:10708948, PubMed:10708949, PubMed:16951688, PubMed:22275001, PubMed:30848080, PubMed:9436979, PubMed:9759503). Suppresses IL1R-TLR signaling through its direct interaction with PEL1, preventing NF-kappa-B activation, nuclear translocation and NF-kappa-B-mediated expression of pro-inflammatory genes (PubMed:16951688). Blocks the BMP-SMAD1 signaling pathway by competing with SMAD4 for receptor-activated SMAD1-binding (PubMed:30848080, PubMed:9436979). Associates with regulatory elements in target promoter regions (PubMed:16491121). Functions as an adapter protein that recruits ubiquitin ligases, including SMURF1 and, in some contexts, SMURF2, promoting proteasomal degradation of signaling components and transcription factors such as RUNX2, TBX6, BMPR1A and MYD88 (PubMed:16299379, PubMed:19561075, PubMed:28847510).
Cellular Location	Nucleus. Cytoplasm {ECO:0000250|UniProtKB:O35182}
Tissue Location	[Isoform B]: Expressed in the brain, heart, ovary, peripheral blood leukocytes, small intestine, spleen, thymus, bone marrow, fetal liver and lymph nodes.

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