

Patched Rabbit pAb

Catalog # AP100260

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q13635
Other Accession	Q13635
Host	Rabbit
Clonality	Polyclonal
Calculated MW	160545
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human Patched
Epitope Specificity	1231-1330/1447
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 patched family. Contains 1 SSD (sterol-sensing) domain.
SUBUNIT	Interacts with SNX17. Interacts with IHH.
Post-translational modifications	Glycosylation is necessary for SHH binding.
DISEASE	Defects in PTCH1 are probably the cause of basal cell nevus syndrome (BCNS) [MIM:109400]; also known as Gorlin syndrome or Gorlin-Goltz syndrome. BCNS is an autosomal dominant disease characterized by nevoid basal cell carcinomas (NBCCS) and developmental abnormalities such as rib and craniofacial alterations, polydactyly, syndactyly, and spina bifida. In addition, the patients suffer from a multitude of tumors like basal cell carcinomas (BCC), fibromas of the ovaries and heart, cysts of the skin, jaws and mesentery, as well as medulloblastomas and meningiomas. PTCH1 is also mutated in squamous cell carcinoma (SCC). Could also be associated with large body size observed in BCNS patients. Defects in PTCH1 are a cause of sporadic basal cell carcinoma (BCC) [MIM:605462]. Defects in PTCH1 are the cause of holoprosencephaly type 7 (HPE7) [MIM:610828]. Holoprosencephaly (HPE) [MIM:236100] is the most common structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	PTCH (Patched protein homolog 1) is a receptor for sonic hedgehog (SHH), indian hedgehog (IHH) and desert hedgehog (DHH). PTCH associates with the smoothened protein (SMO) to transduce the hedgehog's proteins signal. PTCH has a tumor suppressor function, as inactivation of this protein is probably a necessary, if not sufficient step for tumorigenesis. PTCH is expressed in the adult brain, lung, liver, heart, placenta, skeletal muscle, pancreas and kidney. It is also expressed in tumor cells but not in normal skin. During development PTCH is found in all major target tissues of sonic hedgehog, such as the ventral neural tube, somites, and tissues surrounding the zone of polarizing activity of the limb bud. Defects in PTCH are probably the cause of basal cell nevus syndrome also known as Gorlin syndrome or Gorlin-Goltz syndrome.

Additional Information

Gene ID	5727
Other Names	BCNS; BCNS1; NBCCS; PTC; PTC1; PTCH; SLC65B1; A230106A15Rik; mes; wig; Ptch2; PTC1_HUMAN; PTCH1; Q86XG7_HUMAN; PTC1_MOUSE; patched 1; patched (Drosophila) homolog; patched homolog (Drosophila); patched homolog 1 (Drosophila)
Dilution	IHC-P=1:400-800,IHC-F=1:400-800,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	PTCH1 {ECO:0000303 PubMed:31555730, ECO:0000312 HGNC:HGNC:9585}
Function	Negative regulator of the smoothened signaling pathway, which acts as a receptor for hedgehog (SHH, IHH and DHH) morphogens (PubMed: 21537345 , PubMed: 29954986 , PubMed: 29995851 , PubMed: 30139912 , PubMed: 31127104 , PubMed: 31555730 , PubMed: 37611095 , PubMed: 40128518 , PubMed: 9811851). In absence of hedgehog, acts as an inhibitor of smoothened protein (SMO) by preventing SMO access to cholesterol: mechanistically, acts as a cholesterol transporter, which depletes cholesterol from the outer leaflet of plasma membrane, thereby preventing cholesterol-binding to the extracellular CRD region of SMO (PubMed: 31555730 , PubMed: 37611095). Hedgehog-binding inhibits PTCH1 cholesterol transporter activity, promoting SMO activation by cholesterol and transduction of the smoothened signaling pathway (PubMed: 31548691). Molecular mechanism governing cholesterol transport are still unclear: may move cholesterol from the outer to the inner leaflet of the membrane in exchange for potassium or sodium ion export in the opposite direction (PubMed: 37611095). Seems to have a tumor suppressor function, as inactivation of this protein is probably a necessary, if not sufficient step for tumorigenesis (PubMed: 8658145).
Cellular Location	Cell membrane; Multi-pass membrane protein. Cell projection, cilium membrane {ECO:0000250 UniProtKB:Q61115}; Multi-pass membrane protein Note=Localizes to primary cilium and prevents cilium localization of SMO (PubMed:29954986, PubMed:29995851, PubMed:30139912, PubMed:31127104, PubMed:31555730). Delocalizes from primary cilium following hedgehog-binding (By similarity) {ECO:0000250 UniProtKB:Q61115, ECO:0000269 PubMed:29954986, ECO:0000269 PubMed:29995851, ECO:0000269 PubMed:30139912, ECO:0000269 PubMed:31127104, ECO:0000269 PubMed:31555730}
Tissue Location	In the adult, expressed in brain, lung, liver, heart, placenta, skeletal muscle, pancreas and kidney. Expressed in tumor cells but not in normal skin.

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