

Total Patched Cell-Based Colorimetric ELISA Kit

Catalog No :	KA3354C
Reactivity :	Human;Mouse
Applications :	ELISA
Gene Name :	PTCH1
Human Gene Id :	5727
Human Swiss Prot No :	Q13635
Mouse Swiss Prot No :	Q61115
Storage Stability :	2-8 °C/6 months
Detection Method :	Colorimetric

Background : developmental stage:In the embryo, 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.,disease:Defects in PTCH1 are a cause of sporadic basal cell carcinoma (BCC) [MIM:605462].,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.,disease: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.,function:Acts as a receptor for sonic hedgehog (SHH), indian hedgehog (IHH) and desert hedgehog (DHH). Associates with the smoothened protein (SMO) to transduce the hedgehog's proteins signal. Seems to have a tumor suppressor function, as inactivation of this protein is probably a necessary, if not sufficient step for tumorigenesis.,PTM:Glycosylation is necessary for SHH

binding.,similarity:Belongs to the patched family.,similarity:Contains 1 SSD (sterol-sensing) domain.,subunit:Interacts with SNX17.,tissue specificity:In the adult, expressed in brain, lung, liver, heart, placenta, skeletal muscle, pancreas and kidney. Expressed in tumor cells but not in normal skin.,

Function :

embryonic epithelial tube formation, neural tube formation, neural tube closure, morphogenesis of an epithelium,regionalization, heart morphogenesis, cell surface receptor linked signal transduction, smoothened signaling pathway,pattern specification process, ectoderm development, heart development, response to nutrient, cell proliferation,negative regulation of cell proliferation, epidermis development, regulation of smoothened signaling pathway,response to mechanical stimulus, response to abiotic stimulus, response to endogenous stimulus, response to hormone stimulus, embryonic development ending in birth or egg hatching, dorsal/ventral pattern formation, response to extracellular stimulus, response to organic substance, primary neural tube formation, response to organic cyclic substance, morphogenesis of embryonic epithelium, protein processing, neural tube patterning, dorsal/ventral

Subcellular**Location :**

Cell membrane ; Multi-pass membrane protein .

Expression :

In the adult, expressed in brain, lung, liver, heart, placenta, skeletal muscle, pancreas and kidney. Expressed in tumor cells but not in normal skin.

Sort :

23069

No4 :

1

Modifications :

Unmodified

Products Images