

Rabbit Anti-TCTN1 antibody

SL12319R

Product Name:	TCTN1
Chinese Name:	结构蛋白家族1抗体
Alias:	TCTN1; TECT1 HUMAN; Tectonic-1.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Pig,Cow,Sheep,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100- 500IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	61kDa
Cellular localization:	Secretory protein
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human TCTN1:481-587/587
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. 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.
PubMed:	PubMed
Product Detail:	Regulator of Hedgehog (Hh), required for both activation and inhibition of the Hh pathway in the patterning of the neural tube. During neural tube development, it is required for formation of the most ventral cell types and for full Hh pathway activation. Functions in Hh signal transduction to fully activate the pathway in the presence of high Hh levels and to repress the pathway in the absence of Hh signals. Modulates Hh signal transduction downstream of SMO and RAB23.

Function:

Component of the tectonic-like complex, a complex localized at the transition zone of primary cilia and acting as a barrier that prevents diffusion of transmembrane proteins between the cilia and plasma membranes. Regulator of Hedgehog (Hh), required for both activation and inhibition of the Hh pathway in the patterning of the neural tube. During neural tube development, it is required for formation of the most ventral cell types and for full Hh pathway activation. Functions in Hh signal transduction to fully activate the pathway in the presence of high Hh levels and to repress the pathway in the absence of Hh signals. Modulates Hh signal transduction downstream of SMO and RAB23 (By similarity).

Subunit:

Part of the tectonic-like complex (also named B9 complex) (By similarity).

Subcellular Location: Secreted.

DISEASE:

Defects in TCTN1 are the cause of Joubert syndrome type 13 (JBTS13) [MIM:614173]. JBTS13 is a disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease.

Similarity: Belongs to the tectonic family.

SWISS: Q2MV58

Gene ID: 79600

Database links:

Entrez Gene: 79600Human

Entrez Gene: 654470Mouse

Omim: 609863Human

SwissProt: Q2MV58Human

SwissProt: Q8BZ64Mouse

Unigene: 211511Human

