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[Marker Primary Antibody]

Rabbit Anti-TCTN1/FITC Conjugated antibody

Catalog Number: SL12319R-FITC

FOR RESEARCH USE ONLY

[DATASHEET]

Host:	Rabbit
Isotype:	IgG
Clonality:	Polyclonal
Target:	Anti-TCTN1/FITC
Immunogen:	KLH conjugated synthetic peptide derived from human TCTN1
Purification:	affinity purified by Protein A
Concentration:	1mg/ml
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 1M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
Applications:	IF=1:100-500 not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Reactivity:	Rat(predicted:Human,Mouse,Pig,Cow,Sheep)
Background:	background: 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

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	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. **Database links:** **Entrez Gene:** 79600 Human **Entrez Gene:** 654470 Mouse Omim: 609863 Human **SwissProt:** Q2MV58 Human **SwissProt:** Q8BZ64 Mouse **Unigene:** 211511 Human **Unigene:** 375934 Mouse Important **Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.