

TCTN3 Rabbit pAb

TCTN3 Rabbit pAb
Catalog # AP54822

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	Q6NUS6
Predicted	Rat, Dog, Human, Mouse, Rabbit, Chicken, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	66157
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human TCTN3/TECT3
Epitope Specificity	201-300/607
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Membrane; Single-pass type I membrane protein (Potential).
DISEASE	Defects in TCTN3 are the cause of orofacioidigital syndrome 4 (OFD4) [MIM:258860]. A form of orofacioidigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD4 patients have tongue nodules, multiple frenulae, broad flat nose, hypertelorism, and short rib polydactyly with tibial dysplasia (Majewski syndrome). The presence of severe tibial aplasia differentiates OFD4 from OFD1. Additional features of cystic dysplastic kidneys and brain malformation, including occipital encephalocele, are observed in severely affected patients. Defects in TCTN3 are the cause of Joubert syndrome 18 (JBTS18) [MIM:614815]. A form of Joubert syndrome, a disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermal 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. JBTS18 patients have vermis agenesis and the molar tooth sign as well as severe kyphoscoliosis. Other features include intrauterine growth retardation, oral anomalies, micrognathism, polydactyly and camptodactyly, joint laxity, horseshoe kidney, and ventricular septal defect. Note=TCTN3-mutated fibroblasts from JBTS18 patients fail to respond to Shh agonists suggesting that at least some of the defects in affected individuals may be secondary to reduced Shh signaling (PubMed:22883145).
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a member of the tectonic gene family which functions in Hedgehog signal transduction and development of the neural tube. Mutations in this gene have been associated with Orofaciodigital Syndrome IV and Joubert Syndrom 18. Alternatively spliced transcript variants encoding

multiple isoforms have been observed for this gene. [provided by RefSeq, Sep 2012].

Additional Information

Gene ID	26123
Other Names	Tectonic-3, TCTN3, C10orf61, TECT3
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	TCTN3
Synonyms	C10orf61, TECT3
Function	Part of the tectonic-like complex which is required for tissue-specific ciliogenesis and may regulate ciliary membrane composition (By similarity). May be involved in apoptosis regulation. Necessary for signal transduction through the sonic hedgehog (Shh) signaling pathway.
Cellular Location	Membrane; Single-pass type I membrane protein

Background

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.