

Rabbit Anti-TCTN3/AF350 Conjugated antibody

SL12321R-AF350

Product Name	Anti-TCTN3/AF350
Chinese Name	AF350 标记的结构蛋白家族 3 抗体
Alias	C10orf61; Chromosome 10 open reading frame 61; DKFZP564D116; TCTN3; TECT3_HUMAN; Tectonic 3; Tectonic 3 precursor; Tectonic family member 3; Tectonic-3; TECT3; UNQ1881/PRO4324; PSEC0041.
Research Area	Cell biology Apoptosis Growth factors and hormones
Immunogen Species	Rabbit
Clonality	Polyclonal
React Species	(predicted:Human,Mouse,Rat,Chicken,Dog,Cow,Rabbit,Sheep) ICC/IF=1:50-200,IF=1:100-500
Applications	not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight	64kDa
Form	Lyophilized or Liquid
Concentration	1mg/ml
immunogen	KLH conjugated synthetic peptide derived from human TCTN3/TECT3
Lsotype	IgG
Purification	affinity purified by Protein A
Storage Buffer	1M TBS(pH7.4) with 1% BSA, 3% Proclin300 and 50% Glycerol 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.
Storage	
Product Detail	background: 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].

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

Subunit:

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

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 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. JBTS18 patients have vermian 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).

Similarity:

Belongs to the tectonic family.



Database links:

[Entrez Gene: 26123](#) Human

[Entrez Gene: 67590](#) Mouse

[Entrez Gene: 309486](#) Rat

[SwissProt: Q6NUS6](#) Human

[SwissProt: Q8R2Q6](#) Mouse

[Unigene: 438991](#) Human

[Unigene: 374056](#) Mouse

[Unigene: 60758](#) Mouse

Important Note:

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