



## Rabbit Anti-ALS2CR4 antibody

SL7958R

<b>Product Name:</b>	ALS2CR4
<b>Chinese Name:</b>	肌萎缩侧索硬化症相关蛋白4抗体
<b>Alias:</b>	ALS2CR4 protein, N terminus truncated; Amyotrophic lateral sclerosis 2 (juvenile) chromosome region, candidate 4; TM237_HUMAN.
<b>Organism Species:</b>	Rabbit
<b>Clonality:</b>	Polyclonal
<b>React Species:</b>	Human,Mouse,Rat,Pig,Horse,Rabbit,Sheep,
<b>Applications:</b>	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
<b>Molecular weight:</b>	45kDa
<b>Cellular localization:</b>	The cell membrane
<b>Form:</b>	Lyophilized or Liquid
<b>Concentration:</b>	1mg/ml
<b>immunogen:</b>	KLH conjugated synthetic peptide derived from human ALS2CR4:156-250/408
<b>Lsotype:</b>	IgG
<b>Purification:</b>	affinity purified by Protein A
<b>Storage Buffer:</b>	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
<b>Storage:</b>	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.
<b>PubMed:</b>	<a href="#">PubMed</a>
<b>Product Detail:</b>	The protein encoded by this gene is a tetraspanin protein that is thought to be involved in WNT signaling. Defects in this gene are a cause of Joubert syndrome-14. Two transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Jan 2012]. <b>Function:</b>

Component of the transition zone in primary cilia. Required for ciliogenesis.

**Subcellular Location:**

Membrane; Multi-pass membrane protein (Potential). Cell projection, cilium.

Note=Localizes at the proximal region of primary cilia were observed, consistent with localization to the transition zone. Anchored to the transition zone by RPGRIP1L.

**DISEASE:**

Defects in TMEM237 are the cause of Joubert syndrome type 14 (JBTS14) [MIM:614424]. An autosomal recessive disorder characterized by severe mental retardation, hypotonia, breathing abnormalities in infancy, and dysmorphic facial features. 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 JBTS14 features include renal disease, abnormal eye movements, and postaxial polydactyly.

**Similarity:**

Belongs to the TMEM237 family.

**SWISS:**

Q96Q45

**Gene ID:**

65062

**Database links:**

[Entrez Gene: 65062](#)Human

[Entrez Gene: 381259](#)Mouse

[SwissProt: Q96Q45](#)Human

[SwissProt: Q3V0J1](#)Mouse

**Important Note:**

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