

Rabbit Anti-Presenilin 1 + 2 antibody

SL11681R

Product Name:	Presenilin 1 + 2
Chinese Name:	早老素蛋白1+2抗体
Alias:	AD 3; AD4; AD5; Alzheimer disease 3; Alzheimer disease 4; FAD; Presenilin 1; Presenilin 2; Presenilin-1; Presenilin-2; Presenilin1; Presenilin1; PS1; PS2; PS-1; PS-2; PSEN1; PSEN2; STM2; PSN1_HUMAN; PSN2_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human, Mouse, Rat, Chicken,
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:	19/52kDa 💙
Cellular localization:	cytoplasmic The cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human Presenilin-1 NTF subunit:321-430/467
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:	 Alzheimer's disease (AD) patients with an inherited form of the disease carry mutations in the presenilin proteins (PSEN1; PSEN2) or the amyloid precursor protein (APP). These disease-linked mutations result in increased production of the longer form of amyloid-beta (main component of amyloid deposits found in AD brains). Presenilins are

postulated to regulate APP processing through their effects on gamma-secretase, an endoprotease complex that catalyzes the intramembrane cleavage of integral membrane proteins such as Notch receptors and APP.

Function:

Probable catalytic subunit of the gamma-secretase complex, an endoprotease complex that catalyzes the intramembrane cleavage of integral membrane proteins such as Notch receptors and APP (beta-amyloid precursor protein). Requires the other members of the gamma-secretase complex to have a protease activity. May play a role in intracellular signaling and gene expression or in linking chromatin to the nuclear membrane. Stimulates cell-cell adhesion though its association with the E-cadherin/catenin complex. Under conditions of apoptosis or calcium influx, cleaves E-cadherin promoting the disassembly of the E-cadherin/catenin complex and increasing the pool of cytoplasmic beta-catenin, thus negatively regulating Wnt signaling. May also play a role in hematopoiesis.

Subunit:

Homodimer. Component of the gamma-secretase complex, a complex composed of a presenilin homodimer (PSEN1 or PSEN2), nicastrin (NCSTN), APH1 (APH1A or APH1B) and PEN2. Such minimal complex is sufficient for secretase activity. Other components which are associated with the complex include SLC25A64, SLC5A7, PHB and PSEN1 isoform 3. Predominantly heterodimer of a N-terminal (NTF) and a C-terminal (CTF) endoproteolytical fragment. Associates with proteolytic processed C-terminal fragments C83 and C99 of the amyloid precursor protein (APP). Associates with NOTCH1. Associates with cadherin/catenin adhesion complexes through direct binding to CDH1 or CDH2. Interaction with CDH1 stabilizes the complex and stimulates cell-cell aggregation. Interaction with CDH2 is essential for trafficking of CDH2 from the endoplasmic reticulum to the plasma membrane. Interacts with CTNND2, CTNNB1, HERPUD1, FLNA, FLNB, MTCH1, PKP4 and PARL. Interacts through its N-terminus with isoform 3 of GFAP. Interacts with DOCK3.

Subcellular Location:

Endoplasmic reticulum membrane; Multi-pass membrane protein. Golgi apparatus membrane; Multi-pass membrane protein. Cell surface. Note=Bound to NOTCH1 also at the cell surface. Colocalizes with CDH1/2 at sites of cell-cell contact. Colocalizes with CTNNB1 in the endoplasmic reticulum and the proximity of the plasma membrane. Also present in azurophil granules of neutrophils.

Tissue Specificity:

Expressed in a wide range of tissues including various regions of the brain, liver, spleen and lymph nodes.

Post-translational modifications:

Heterogeneous proteolytic processing generates N-terminal (NTF) and C-terminal (CTF) fragments of approximately 35 and 20 kDa, respectively. During apoptosis, the C-terminal fragment (CTF) is further cleaved by caspase-3 to produce the fragment, PS1-

CTF12.

After endoproteolysis, the C-terminal fragment (CTF) is phosphorylated on serine residues by PKA and/or PKC. Phosphorylation on Ser-346 inhibits endoproteolysis.

DISEASE:

Defects in PSEN1 are a cause of Alzheimer disease type 3 (AD3) [MIM:607822]. AD3 is a familial early-onset form of Alzheimer disease. Alzheimer disease is a neurodegenerative disorder characterized by progressive dementia, loss of cognitive abilities, and deposition of fibrillar amyloid proteins as intraneuronal neurofibrillary tangles, extracellular amyloid plaques and vascular amyloid deposits. The major constituent of these plaques is the neurotoxic amyloid-beta-APP 40-42 peptide (s), derived proteolytically from the transmembrane precursor protein APP by sequential secretase processing. The cytotoxic C-terminal fragments (CTFs) and the caspase-cleaved products such as C31 derived from APP, are also implicated in neuronal death. Defects in PSEN1 are a cause of frontotemporal dementia (FTD) [MIM:600274]. [DISEASE] Defects in PSEN1 are the cause of cardiomyopathy dilated type 1U (CMD1U) [MIM:613694]. It is a disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death.

Defects in PSEN1 are the cause of familial acne inversa type 3 (ACNINV3) [MIM:613737]. A chronic relapsing inflammatory disease of the hair follicles characterized by recurrent draining sinuses, painful skin abscesses, and disfiguring scars. Manifestations typically appear after puberty.

Similarity:

Belongs to the peptidase A22A family.

SWISS: P49768

Gene ID: 5663

Database links:

Entrez Gene: 5663 Human

Entrez Gene: 19164 Mouse

Entrez Gene: 29192 Rat

<u>Omim: 104311</u> Human

SwissProt: P49768 Human

SwissProt: P49769 Mouse





