

Product name:	Presenilin 1 Rabbit Polyclonal Antibody
Cat number:	ABN16488
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human PSEN1. AA range:323-372
Reactivity:	Human,Mouse,Rat
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:5000-1:10000
Molecular Weight:	43kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

Alzheimer's disease (AD) patients with an inherited form of the disease carry mutations in the presenilin proteins (PSEN1; PSEN2) or in 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 enzyme that cleaves APP. Also, it is thought that the presenilins are involved in the cleavage of the Notch receptor, such that they either directly regulate gamma-secretase activity or themselves are protease enzymes. Several alternatively spliced transcript variants encoding different isoforms have been identified for this gene, the full-length nature of only some have been determined. [provided by RefSeq, Aug 2008],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.,disease:Defects in PSEN1 are a cause of frontotemporal dementia [MIM:600274].,domain:The PAL motif is required for normal active site conformation.,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 through 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.,online information:Presenilins mutations,PTM:After endoproteolysis, the C-terminal fragment (CTF) is phosphorylated on serine residues by PKA and/or PKC. Phosphorylation on Ser-346 inhibits endoproteolysis.,PTM: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.,similarity:Belongs to the peptidase A22A family.,subcellular location:Bound to NOTCH1 also at the cell surface. Colocalizes with CDH1/2 at sites of cell-cell contact. Colocalizes with CTNBN1 in the endoplasmic reticulum and the proximity of the plasma membrane. Also present in azurophil granules of neutrophils.,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. Component of 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, CTNBN1, HERPUD1, FLNA, FLNB, MTCH1, PKP4 and PARL. Interacts through its N-terminus with isoform 3 of GFAP. Interacts with DOCK3.,tissue specificity:Expressed in a wide range of tissues including various regions of the brain, liver, spleen and lymph nodes.,