abcam

Product datasheet

Human Presenilin 1/PS-1 peptide ab49413

Description

Product name	Human Presenilin 1/PS-1 peptide
Purity	> 50 % Mass-Spectrometry. Purified by Ion Spray Mass Spectrometry.
Animal free	No
Nature	Synthetic
Species	Human
Sequence	AQMSEDNHLSNTVRSQNDNR
Amino acids	14 to 33

Specifications

Our **Abpromise guarantee** covers the use of **ab49413** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Form	Liquid

Preparation and Storage

Stability and Storage	Shipped at 4°C. Store at +4°C short term (1-2 weeks). Upon delivery aliquot. Store at -20°C long term. Avoid freeze / thaw cycle.

General Info

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

Involvement in 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 [MIM:600274]. 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 acne inversa familial type 3 (ACNIF3) [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.
Sequence similarities	Belongs to the peptidase A22A family.
Domain	The PAL motif is required for normal active site conformation.
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.
Cellular localization	Endoplasmic reticulum membrane. Golgi apparatus membrane. Cell surface. 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.