abcam

Product datasheet

Human Filamin A protein fragment ab91686

1 Image

Description

Product name	Human Filamin A protein fragment
Expression system	Escherichia coli
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	VTALAGDQPSVQPPLRSQQLAPQYTYAQGGQQTWAPER PLVGVNGLDV TSLRPFDLVIPFTIKKGEITGEVRMPSGKVAQPTITDNKDGT VTVRYA PSEAGLHEMDIRYDNMHIPGSPLQFYVDYVNCGHVTAYGP GLTHGVVN KPATFTVNTKDAGEGGLSLAIEGPSKAEISCTDNQDGTCS VSYLPVLP GDYSILVKYNEQHVPGSPFTARVTGDDSMRMSHLKVGSA ADIPINISE TDLSLLTATVVPPSGREEPCLLKRLRNGHVGISFVPKETG EHLVHVKK NGQHVASSPIPVVISQSEIGDASRVRVSGQGLHEGHTFEP AEFIIDTR DAGYGGLSLSIEGPSKVDINTEDLEDGTCRVTYCPTEPGN YIINIKFA DQHVPGSPFSVKVTGEGRVKESITRRRRAPSVANVGSHC DLSLKIPEI SIQDMTAQVTSPSGKTHEAEIVEGENHTYCIRFVPAEMGTH TVSVKYK GQHVPGSPFQFTVGPLGEGGAHKVRAGGPGLERAEAGV PAEFSIWTRE AGAGGLAIAVEGPSKAEISFEDRKDGSCGVAYVVQEPGD YEVSVKFNE EHIPDSPFVVPVASPSGDARRLTVSSLQESGLKVNQPAS FAVSLNGAK GAIDAKVHSPSGALEECYVTEIDQDKYAVRFIPRENGVYLI DVKFNGT HIPGSPFKIRVGEPGHGGDPGLVSAYGAGLEGGVTGNPA

EFVVNTSNA
GAGALSVTIDGPSKVKMDCQECPEGYRVTYTPMAPGSYLI
SIKYGGPY
HIGGSPFKAKVTGPRLVSNHSLHETSSVFVDSLTKATCAP
QHGAPGPG
PADASKVVAKGLGLSKAYVGQKSSFTVDCSKAGNNMLL
VGVHGPRTPC
EEILVKHVGSRLYSVSYLLKDKGEYTLVVKWGDEHIPGSPY
RVVVP

Amino acids 1730 to 2639

Specifications

Our **Abpromise guarantee** covers the use of **ab91686** in the following tested applications.

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

Applications SDS-PAGE

Form Lyophilized

Preparation and Storage

Stability and Storage Shipped at 4°C. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles.

Preservative: None
Constituents: 0.5% Trehalose, 6M Urea, 100mM Sodium phosphate, 10mM Sodium chloride, pH 4.5

Reconstitution Reconstitute with 111 µl aqua dest.

General Info

Function Promotes orthogonal branching of actin filaments and links actin filaments to membrane glycoproteins. Anchors various transmembrane proteins to the actin cytoskeleton and serves as a scaffold for a wide range of cytoplasmic signaling proteins. Interaction with FLNA may allow neuroblast migration from the ventricular zone into the cortical plate. Tethers cell surface-localized furin, modulates its rate of internalization and directs its intracellular trafficking.

Tissue specificity Ubiquitous.

Involvement in disease Defects in FLNA are the cause of periventricular nodular heterotopia type 1 (PVNH1) [MIM:300049]; also called nodular heterotopia, bilateral periventricular (NHBP or BPNH). PVNH is a developmental disorder characterized by the presence of periventricular nodules of cerebral gray matter, resulting from a failure of neurons to migrate normally from the lateral ventricular proliferative zone, where they are formed, to the cerebral cortex. PVNH1 is an X-linked dominant form. Heterozygous females have normal intelligence but suffer from seizures and various manifestations outside the central nervous system, especially related to the vascular system. Hemizygous affected males die in the prenatal or perinatal period.
Defects in FLNA are the cause of periventricular nodular heterotopia type 4 (PVNH4) [MIM:300537]; also known as periventricular heterotopia Ehlers-Danlos variant. PVNH4 is characterized by nodular brain heterotopia, joint hypermobility and development of aortic dilation in early adulthood.
Defects in FLNA are the cause of otopalatodigital syndrome type 1 (OPD1) [MIM:311300]. OPD1

	is an X-linked dominant multiple congenital anomalies disease mainly characterized by a generalized skeletal dysplasia, mild mental retardation, hearing loss, cleft palate, and typical facial anomalies. OPD1 belongs to a group of X-linked skeletal dysplasias known as oto-palato-digital syndrome spectrum disorders that also include OPD2, Melnick-Needles syndrome (MNS), and frontometaphyseal dysplasia (FMD). Remodeling of the cytoskeleton is central to the modulation of cell shape and migration. FLNA is a widely expressed protein that regulates re-organization of the actin cytoskeleton by interacting with integrins, transmembrane receptor complexes and second messengers. Males with OPD1 have cleft palate, malformations of the ossicles causing deafness and milder bone and limb defects than those associated with OPD2. Obligate female carriers of mutations causing both OPD1 and OPD2 have variable (often milder) expression of a similar phenotypic spectrum. Defects in FLNA are the cause of otopalatodigital syndrome type 2 (OPD2) [MIM:304120]; also known as cranioorodigital syndrome. OPD2 is a congenital bone disorder that is characterized by abnormally modeled, bowed bones, small or absent first digits and, more variably, cleft palate, posterior fossa brain anomalies, omphalocele and cardiac defects. Defects in FLNA are the cause of frontometaphyseal dysplasia (FMD) [MIM:305620]. FMD is a congenital bone disease characterized by supraorbital hyperostosis, deafness and digital anomalies. Defects in FLNA are the cause of Melnick-Needles syndrome (MNS) [MIM:309350]. MNS is a severe congenital bone disorder characterized by typical facies (exophthalmos, full cheeks, micrognathia and malalignment of teeth), flaring of the metaphyses of long bones, s-like curvature of bones of legs, irregular constrictions in the ribs, and sclerosis of base of skull. Defects in FLNA are the cause of X-linked congenital idiopathic intestinal pseudoobstruction (CIIPX) [MIM:300048]. CIIPX is characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. Defects in FLNA are the cause of FG syndrome type 2 (FGS2) [MIM:300321]. FG syndrome (FGS) is an X-linked disorder characterized by mental retardation, relative macrocephaly, hypotonia and constipation. Defects in FLNA are the cause of terminal osseous dysplasia (TOD) [MIM:300244]. A rare X-linked dominant male-lethal disease characterized by skeletal dysplasia of the limbs, pigmentary defects of the skin and recurrent digital fibroma during infancy. A significant phenotypic variability is observed in affected females. Defects in FLNA are the cause of cardiac valvular dysplasia X-linked (CVDX) [MIM:314400]. A rare X-linked heart disease characterized by mitral and/or aortic valve regurgitation. The histologic features include fragmentation of collagenous bundles within the valve fibrosa and accumulation of proteoglycans, which produces excessive valve tissue leading to billowing of the valve leaflets.
Sequence similarities	Belongs to the filamin family. Contains 1 actin-binding domain. Contains 2 CH (calponin-homology) domains. Contains 24 filamin repeats.
Domain	Comprised of a NH2-terminal actin-binding domain, 24 internally homologous repeats and two hinge regions. Repeat 24 and the second hinge domain are important for dimer formation.
Post-translational modifications	Phosphorylated upon DNA damage, probably by ATM or ATR (By similarity). Phosphorylation extent changes in response to cell activation. The N-terminus is blocked.
Cellular localization	Cytoplasm > cell cortex. Cytoplasm > cytoskeleton.

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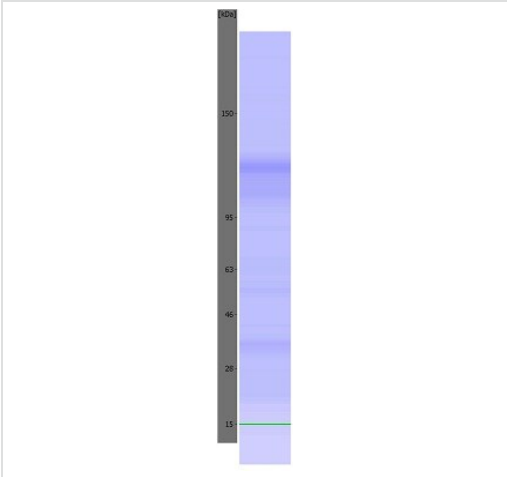
Images



SDS-PAGE - Human Filamin A protein fragment (ab91686)

The image shows an electrophoretic assay performed using an Agilent 5100 ALP. In some images coloured control bands can be seen at 15 kDa (green) and/or 240 kDa (purple). The protein-specific band is blue.

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