Filamin 1 Rabbit pAb

CatalogNo: YT1711

Key Features

Host Species
- Rabbit

Reactivity
- Human,Mouse,Rat

Applications
- WB,IHC,IF,ELISA

MW
- 280kD (Observed)

Isotype
- IgG

Recommended Dilution Ratios

WB 1:500-1:2000
IHC 1:100-1:300
ELISA 1:10000
IF 1:50-200

Storage

Storage*	-15°C to -25°C/1 year(Do not lower than -25°C)
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.

Basic Information

Clonality	Polyclonal

Immunogen Information

Immunogen	The antiserum was produced against synthesized peptide derived from human Filamin A. AA range:2121-2170
Specificity	Filamin 1 Polyclonal Antibody detects endogenous levels of Filamin 1 protein.

Target Information

Gene name FLNA

Protein Name Filamin-A

Organism	Gene ID	UniProt ID
Human	2316;	P21333;
Mouse	192176;	Q8BTM8;

Cellular Localization Cytoplasm, cell cortex . Cytoplasm, cytoskeleton . Perikaryon . Cell projection, growth cone . Colocalizes with CPMR1 in the central region of DRG neuron growth cone (By similarity). Following SEMA3A stimulation of DRG neurons, colocalizes with F-actin (By similarity). .

Tissue specificity Ubiquitous.

Function

Disease:Defects in FLNA are associated with cerebrofrontofacial syndrome [MIM:608578]. This syndrome consists of a phenotype of male PVNH, with relatively normal development, no epilepsy or other neurological abnormality, severe constipation, and facial dysmorphism and without a discernible skeletal phenotype.,Disease: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.,Disease: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.,Disease: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.,Disease: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.,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.,Disease: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 dilatation in early adulthood.,Disease: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.,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.,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.,PTM:Phosphorylated upon DNA damage, probably by ATM or ATR (By similarity). Phosphorylation extent changes in response to cell activation.,PTM:The N-terminus is blocked.,similarity:Belongs to the filamin family.,similarity:Contains 1 actin-binding domain.,similarity:Contains 2 CH (calponin-homology) domains.,similarity:Contains 24 filamin repeats.,subunit:Interacts with PDLIM2 (By similarity). Homodimer. Interacts with FCGR1A, FLNB, FURIN, HSPB7, INPPL1, KCND2, MYOT, MYOZ1, ARHGAP24, PSEN1, PSEN2 and ECSCR. Interacts also with various other binding partners in addition to filamentous actin.,tissue specificity:Ubiquitous.,

Validation Data

Contact information

Orders:	order@immunoway.com
Support:	tech@immunoway.com
Telephone:	877-594-3616 (Toll Free), 408-747-0185
Website:	http://www.immunoway.com
Address:	2200 Ringwood Ave San Jose, CA 95131 USA

Please scan the QR code to access additional product information: **Filamin 1 Rabbit pAb**