

BFSP2 Rabbit pAb

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Catalog # AP54342

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	Q13515
Predicted	Rat, Dog, Human, Mouse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	45880
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human BFSP2/Phakinin
Epitope Specificity	181-280/415
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Membrane. Cytoplasm. Cytoplasm, cytoskeleton. Membrane- and cytoskeleton-associated.
DISEASE	Defects in BFSP2 are the cause of cataract autosomal dominant BFSP2-related (ADC-BFSP2); also known as cataract autosomal dominant multiple types 1. Cataract is an opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. Cataract autosomal dominant BFSP2-related is characterized by a variable phenotype that may or may not be consistent within a family. The opacities can be nuclear, sutural, stellate cortical, lamellar, cortical, nuclear embryonic, Y-sutural, punctate cortical, congenital or with juvenile- and adult-onset.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Phakinin is a membrane-associated and cytoskeletal intermediate filament (IF) protein specific to the eye lens. IFs are cytoskeletal structures that typically contain a head, rod and tail domain. Unlike most IFs, Phakinin completely lacks the C-terminal tail domain thus contributing to the unique structure of the beaded filament that is specific to the lens. Phakinin is required for the assembly of beaded filaments and cytoskeletal networks that are important for the long-term maintenance of optical properties and transparency of the lens. Phakinin copolymerizes with Filensin, another IF protein, to form the 10-nm filamentous structures of the beaded filaments. Phakinin is also capable of self-assembling into filament-like structures that form thicker bundles. Mutations in the gene encoding Phakinin can result in lens cataract.

Additional Information

Gene ID	8419
Other Names	Phakinin, 49 kDa cytoskeletal protein, Beaded filament structural protein 2, Lens fiber cell beaded filament protein CP 47, CP47, Lens fiber cell beaded filament protein CP 49, CP49, Lens intermediate filament-like light, LIFL-L, BFSP2
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

Name	BFSP2
Function	Required for the correct formation of lens intermediate filaments as part of a complex composed of BFSP1, BFSP2 and CRYAA (PubMed: 28935373). Plays a role in maintenance of retinal lens optical clarity (By similarity).
Cellular Location	Cell membrane {ECO:0000250 UniProtKB:Q28177}; Peripheral membrane protein {ECO:0000250 UniProtKB:Q28177}; Cytoplasmic side {ECO:0000250 UniProtKB:Q28177}. Cytoplasm {ECO:0000250 UniProtKB:D3ZER2}. Cytoplasm, cytoskeleton {ECO:0000250 UniProtKB:Q28177}. Cytoplasm, cell cortex {ECO:0000250 UniProtKB:Q28177}. Note=Expressed primarily at the plasma membrane in peripheral lens fiber cells, however also localizes to the cytoplasm in mature lens fiber cells. {ECO:0000250 UniProtKB:Q28177}
Tissue Location	Lens.

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