

BFSP1 rabbit pAb

Catalog No :	YT6349
Reactivity :	Human;Mouse;Rat
Applications :	WB
Target :	BFSP1
Gene Name :	BFSP1
Protein Name :	BFSP1
Human Gene Id :	631
Human Swiss Prot No :	Q12934
Mouse Gene Id :	12075
Mouse Swiss Prot No :	A2AMT1
Rat Gene Id :	25394
Rat Swiss Prot No :	Q02435
Immunogen :	Synthesized peptide derived from human BFSP1 AA range: 494-544
Specificity :	This antibody detects endogenous levels of BFSP1 at Human/Mouse/Rat
Formulation :	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.
Source :	Polyclonal, Rabbit,IgG
Dilution :	WB 1:500-2000
Purification :	The antibody was affinity-purified from rabbit antiserum by affinity-chromatography using epitope-specific immunogen.

Concentration : 1 mg/ml

Storage Stability : -15°C to -25°C/1 year(Do not lower than -25°C)

Molecularweight : 73kD

Background : This gene encodes a lens-specific intermediate filament-like protein named filensin. The encoded protein is expressed in lens fiber cells after differentiation has begun. This protein functions as a component of the beaded filament which is a cytoskeletal structure found in lens fiber cells. Mutations in this gene are the cause of autosomal recessive cortical juvenile-onset cataract. Alternate splicing results in multiple transcript variants. [provided by RefSeq, Jul 2013],

Function : disease:Defects in BFSP1 are the cause of autosomal recessive cortical juvenile-onset cataract [MIM:611391]. Cataract is the most frequent cause of visual impairment and blindness worldwide. While congenital cataracts are less frequent than age related cataracts, if not treated promptly they can result in irreversible neural blindness. The frequency of non-syndromic congenital cataract is estimated to be 1-6 cases per 10'000 children with one additional case being diagnosed during childhood. Developmental or juvenile onset cataract is distinguished from congenital cataract by initial clarity of the lens at birth and development of opacities progressively with maturation during childhood or adolescence. Approximately 25% of non-syndromic cataracts are inherited, and they are phenotypically and genetically heterogeneous, with autosomal dominant generally considered to be more common than a

Subcellular Location : Cell membrane ; Peripheral membrane protein ; Cytoplasmic side . Cytoplasm . Cytoplasm, cytoskeleton . Cytoplasm, cell cortex .

Expression : Expressed in the cortex and nucleus of the retina lens (at protein level).

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