

CEP290 Rabbit pAb

Catalog # AP98133

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q15078
Other Accession	Q15078
Reactivity	Mouse
Predicted	Rat, Human, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	290386
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human CEP290
Epitope Specificity	131-230/2479
Isotype	IgG
Purity	affinity purified by Protein A

Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
DISEASE	Defects in CEP290 are a cause of Joubert syndrome type 5 (JBTS5) [MIM:610188]. Joubert syndrome is an autosomal recessive disease characterized by cerebellar vermis hypoplasia with prominent superior cerebellar peduncles (the 'molar tooth sign' on axial magnetic resonance imaging), psychomotor delay, hypotonia, ataxia, oculomotor apraxia and neonatal breathing abnormalities. JBTS5 shares the neurologic and neuroradiologic features of Joubert syndrome together with severe retinal dystrophy and/or progressive renal failure characterized by nephronophthisis. Defects in CEP290 are a cause of Senior-Loken syndrome type 6 (SLSN6) [MIM:610189]. Senior-Loken syndrome is also known as juvenile nephronophthisis with Leber amaurosis. It is an autosomal recessive renal-retinal disorder, characterized by progressive wasting of the filtering unit of the kidney, with or without medullary cystic renal disease, and progressive eye disease. Defects in CEP290 are the cause of Leber congenital amaurosis type 10 (LCA10) [MIM:611755]. LCA designates a clinically and genetically heterogeneous group of childhood retinal degenerations, generally inherited in an autosomal recessive manner. Affected infants have little or no retinal photoreceptor function as tested by electroretinography. LCA represents the most common genetic cause of congenital visual impairment in infants and children. Defects in CEP290 are the cause of Meckel syndrome type 4 (MKS4) [MIM:611134]. MKS4 is an autosomal recessive disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. Note=Antibodies against CEP290 are present in sera from patients with cutaneous T-cell lymphomas, but not in the healthy control population. Defects in CEP290 are the cause of Bardet-Biedl syndrome type 14 (BBS14) [MIM:209900]. A syndrome characterized by usually severe pigmentary retinopathy, early-onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Inheritance is autosomal

	recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for disease manifestation in some cases (triallelic inheritance).
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a protein with 13 putative coiled-coil domains, a region with homology to SMC chromosome segregation ATPases, six KID motifs, three tropomyosin homology domains and an ATP/GTP binding site motif A. The protein is localized to the centrosome and cilia and has sites for N-glycosylation, tyrosine sulfation, phosphorylation, N-myristoylation, and amidation. Mutations in this gene have been associated with Joubert syndrome and nephronophthisis and the presence of antibodies against this protein is associated with several forms of cancer. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	80184
Other Names	3H11Ag; BBS14; CT87; JBTS5; LCA10; MKS4; NPHP6; POC3; SLSN6; rd16; CE290_HUMAN; CEP290; Bardet-Biedl syndrome 14 protein; Cancer/testis antigen 87 (CT87); Nephrocystin-6; Tumor antigen se2-2; KIAA0373; centrosomal protein 290; centrosomal protein 290kDa; Joubert syndrome 5; cancer/testis antigen 87; POC3 centriolar protein homolog (Chlamydomonas); Meckel syndrome, type 4; Bardet-Biedl syndrome 14
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
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	CEP290
Synonyms	BBS14, KIAA0373, NPHP6
Function	Involved in early and late steps in cilia formation. Its association with CCP110 is required for inhibition of primary cilia formation by CCP110 (PubMed: 18694559). May play a role in early ciliogenesis in the disappearance of centriolar satellites and in the transition of primary ciliar vesicles (PCVs) to capped ciliary vesicles (CCVs). Required for the centrosomal recruitment of RAB8A and for the targeting of centriole satellite proteins to centrosomes such as of PCM1 (PubMed: 24421332). Required for the correct localization of ciliary and phototransduction proteins in retinal photoreceptor cells; may play a role in ciliary transport processes (By similarity). Required for efficient recruitment of RAB8A to primary cilium (PubMed: 17705300). In the ciliary transition zone is part of the tectonic-like complex which is required for tissue-specific ciliogenesis and may regulate ciliary membrane composition (By similarity). Involved in regulation of the BBSome complex integrity, specifically for presence of BBS2, BBS5 and BBS8/TTC8 in the complex, and in ciliary targeting of selected BBSome cargos. May play a role in controlling entry of the BBSome complex to cilia possibly implicating IQCB1/NPHP5 (PubMed: 25552655). Activates ATF4-mediated transcription (PubMed: 16682973).

Cellular Location	Cytoplasm, cytoskeleton, microtubule organizing center, centrosome. Cytoplasm, cytoskeleton, microtubule organizing center, centrosome, centriolar satellite. Nucleus {ECO:0000250 UniProtKB:Q6A078} Cell projection, cilium. Cytoplasm, cytoskeleton, cilium basal body {ECO:0000250 UniProtKB:Q6A078}. Cytoplasm, cytoskeleton, microtubule organizing center, centrosome, centriole. Cytoplasmic vesicle. Note=Displaced from centriolar satellites in response to cellular stress, such as ultraviolet light (UV) radiation or heat shock (PubMed:24121310). Found in the connecting cilium of photoreceptor cells, base of cilium in kidney intramedullary collecting duct cells (By similarity). Localizes at the transition zone, a region between the basal body and the ciliary axoneme (PubMed:23943788, PubMed:36606424). Localization at the ciliary transition zone as well as at centriolar satellites is BBSome-dependent (PubMed:23943788). {ECO:0000250 UniProtKB:Q6A078, ECO:0000269 PubMed:23943788, ECO:0000269 PubMed:24121310, ECO:0000269 PubMed:36606424}
Tissue Location	Ubiquitous. Expressed strongly in placenta and weakly in brain.

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