



CEP290 Polyclonal Antibody

Catalog No	YP-Ab-03759
Isotype	IgG
Reactivity	Human;Mouse
Applications	WB;ELISA
Gene Name	CEP290
Protein Name	Centrosomal protein of 290 kDa
Immunogen	The antiserum was produced against synthesized peptide derived from human CEP290. AA range:771-820
Specificity	CEP290 Polyclonal Antibody detects endogenous levels of CEP290 protein.
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.
Source	Polyclonal, Rabbit,IgG
Purification	The antibody was affinity-purified from rabbit antiserum by affinity-chromatography using epitope-specific immunogen.
Dilution	Western Blot: 1/500 - 1/2000. ELISA: 1/20000. Not yet tested in other applications.
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	CEP290; BBS14; KIAA0373; NPHP6; Centrosomal protein of 290 kDa; Cep290; Bardet-Biedl syndrome 14 protein; Cancer/testis antigen 87; CT87; Nephrocystin-6; Tumor antigen se2-2
Observed Band	290kD
Cell Pathway	Cytoplasm, cytoskeleton, microtubule organizing center, centrosome . Cytoplasm, cytoskeleton, microtubule organizing center, centrosome, centriolar satellite . Nucleus . Cell projection, cilium . Cytoplasm, cytoskeleton, cilium basal body . Cytoplasm, cytoskeleton, microtubule organizing center, centrosome, centriole . Cytoplasmic vesicle . 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). Localization at the ciliary transition zone as well as at centriolar sa
Tissue Specificity	Ubiquitous. Expressed strongly in placenta and weakly in brain.
Function	disease:Antibodies against CEP290 are present in sera from patients with cutaneous T-cell lymphomas, but not in the healthy control population..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. disease: 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 a

Background

centrosomal protein 290(CEP290) Homo sapiens 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],

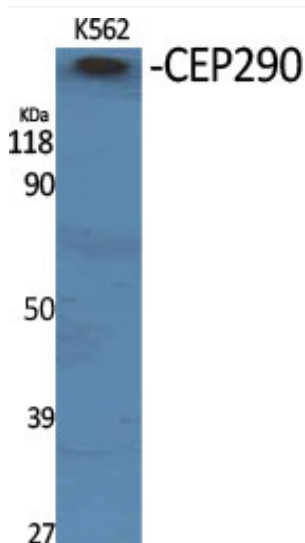
matters needing attention

Avoid repeated freezing and thawing!

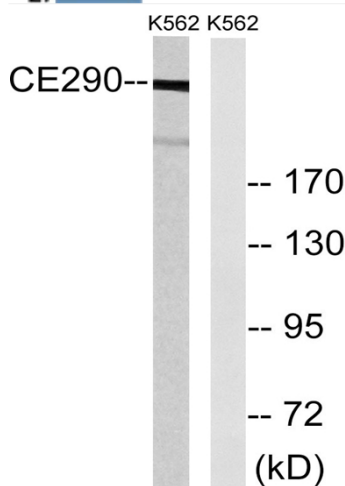
Usage suggestions

This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.

Products Images



Western Blot analysis of various cells using CEP290 Polyclonal Antibody



Western blot analysis of lysates from K562 cells, using CEP290 Antibody. The lane on the right is blocked with the synthesized peptide.