Product Name: CEP290 Rabbit Polyclonal Antibody
Catalog #: APRab08662

Summary

Production Name	CEP290 Rabbit Polyclonal Antibody
Description	Rabbit Polyclonal Antibody
Host	Rabbit
Application	WB,ELISA
Reactivity	Human,Mouse

Performance

Conjugation	Unconjugated
Modification	Unmodified
Isotype	IgG
Clonality	Polyclonal
Form	Liquid
Storage	Store at 4°C short term. Aliquot and store at -20°C long term. Avoid freeze/thaw cycles.
Buffer	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Purification	Affinity purification

Immunogen

Gene Name	CEP290
Alternative Names	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
Gene ID	80184.0
SwissProt ID	O15078.The antiserum was produced against synthesized peptide derived from human CEP290. AA range:771-820

Application

Dilution Ratio	WB 1:500 - 1:2000. ELISA: 1:20000
Molecular Weight	290kD

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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],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 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.,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.,disease: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.,function:Activates ATF4-mediated transcription. Required for the correct localization of ciliary and phototransduction proteins in retinal photoreceptor cells; may play a role in ciliary transport processes.,sequence caution:Contaminating sequence. Potential poly-A sequence.,subcellular location:Connecting cilium of photoreceptor cells, base of cilium in kidney intramedullary collecting duct cells.,subunit:Interacts with ATF4 via its N-terminal region. Part of selected centrosomal and microtubule-associated protein complexes. Interacts with CC2D2A.,tissue specificity:Ubiquitous. Expressed strongly in placenta and weakly in brain.,

Research Area

Image Data

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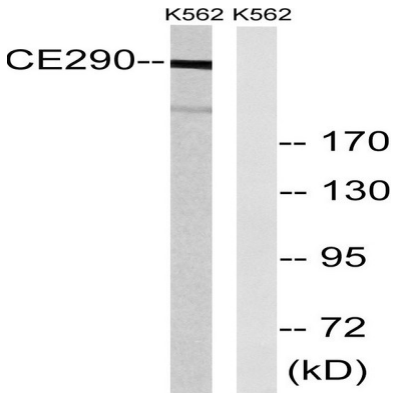
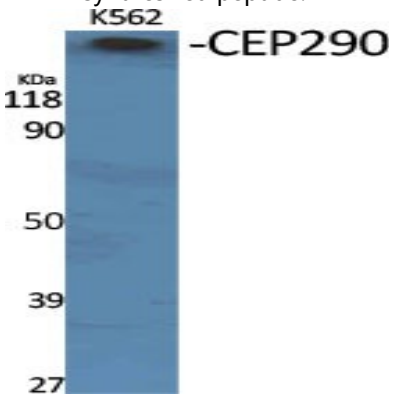


Western blot analysis of lysates from K562 cells, using CEP290 Antibody. The lane on the right is blocked with the synthesized peptide.



Western Blot analysis of various cells using CEP290 Polyclonal Antibody

Note

For research use only.