CEP290 Rabbit pAb

CatalogNo: YT0859

Key Features

Host Species
- Rabbit

Reactivity
- Human,Mouse

Applications
- WB,ELISA

MW
- 290kD (Observed)

Isotype
- IgG

Recommended Dilution Ratios

WB 1:500-1:2000
ELISA 1:20000
Not yet tested in other applications.

Storage

Storage*	-15°C to -25°C/1 year(Do not lower than -25°C)
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.

Basic Information

Clonality	Polyclonal

Immunogen Information

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.

Target Information

Gene name CEP290

Protein Name Centrosomal protein of 290 kDa

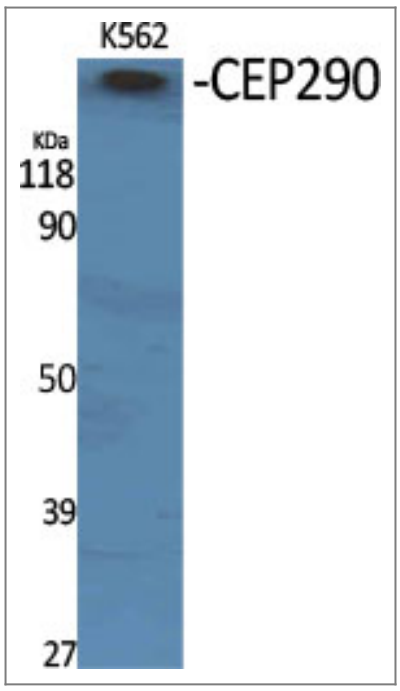
Organism	Gene ID	UniProt ID
Human	80184;	O15078;
Mouse	216274;	Q6A078;

Cellular Localization 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 satellites is BBsome-dependent (PubMed:23943788). .

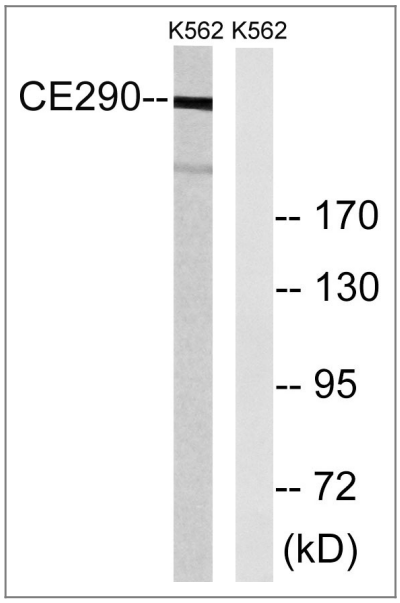
Tissue specificity Ubiquitous. Expressed strongly in placenta and weakly in brain.

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 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.,

Validation Data



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.

Contact information

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Please scan the QR code to access additional product information:
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