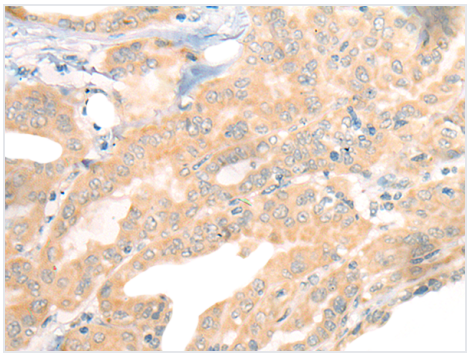
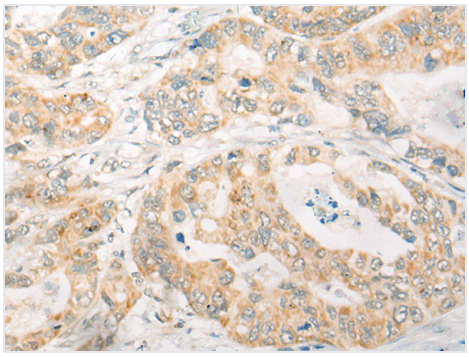
CEP290 Antibody – Rabbit Polyclonal

PRODUCT OVERVIEW

Product Name	CEP290, Polyclonal Antibody
Full Product Name	CEP290 Polyclonal Antibody
Product Gene Name	anti-CEP290 antibody
Product Synonym Names	3H11AG; Bardet-Biedl syndrome 14 protein; BBS14; Cancer/testis antigen 87; CE290; Centrosomal protein 290; Centrosomal protein 290kDa; Centrosomal protein of 290 kDa; Cep290; CT87; CTCL tumor antigen se2 2; FLJ13615; FLJ21979,JBTS5,JBTS6; KIAA0373; LCA10; Meckel syndrome; Type 4; MKS4; Monoclonal 3H11 antigen; Nephrocystin 6; Nephrocystin-6; NPHP6; POC3; POC3 centriolar protein homolog; Prostate cancer antigen T21; Rd16; SLSN6 1; 2; 5; SLSN6; Tumor antigen se2-2
Clonality	Polyclonal
Isotype	IgG
Host	Rabbit
Species Reactivity	Human, Mouse
Form/Format	PBS with 0.05% NaN3 and 40% Glycerol, pH7.4
Purity/Purification	Antigen Affinity Purification
Concentration	0.5mg/mL (lot specific)
Preparation and Storage	Store at -20 degree C. Avoid freeze/thaw cycles.
OMIM	209900
3D Structure	ModBase 3D Structure for O15078
Immunogen	Fusion protein of human CEP290
Conjugation	Unconjugated
Product Categories/Family for anti-CEP290 antibody	Cancer; Epigenetics and Nuclear Signaling; Neuroscience
ISO Certification	Manufactured in an ISO 9001:2015 Certified Laboratory.
Applications Tested/Suitable for anti-CEP290 antibody	Immunohistochemistry (IHC), ELISA (EIA)
Application Notes for anti-CEP290 antibody	IHC: 1:25-1:100 ELISA: 1:5000-1:10000
Related Product Information for anti-CEP290 antibody	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.
Other Notes	Small volumes of anti-CEP290 antibody vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.
Searchable Terms for CEP290 purchase	MBS2562440 is an antibody targeting CEP290. Also known as CEP290 Polyclonal Antibody. Reactive in Human, Mouse. Host: Rabbit.



Immunohistochemistry (IHC) of anti-CEP290 antibody
Immunohistochemistry of paraffin-embedded Human thyroid cancer tissue using CEP290 Polyclonal Antibody at dilution of 1:25(×200)



Immunohistochemistry (IHC) of anti-CEP290 antibody
Immunohistochemistry of paraffin-embedded Human gastric cancer tissue using CEP290 Polyclonal Antibody at dilution of 1:25(×200)

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for CEP290. It may not necessarily be applicable to this product.

NCBI GI #	109255234
NCBI GeneID	80184
NCBI Accession #	NP_079390.3
NCBI Related Accession #	BC008641
UniProt Primary Accession #	O15078
UniProt Related Accession #	O15078
GenBank Nucleotide #	NP_079390.3
NCBI Official Full Name	centrosomal protein of 290 kDa
NCBI Official Synonym Full Names	centrosomal protein 290
NCBI Official Symbol	CEP290
NCBI Official Synonym Symbols	CT87; MKS4; POC3; rd16; BBS14; JBTS5; LCA10; NPHP6; SLSN6; 3H11Ag
UniProt Protein Name	Centrosomal protein of 290 kDa
UniProt Synonym Protein Names	Bardet-Biedl syndrome 14 protein; Cancer/testis antigen 87; CT87; Nephrocystin-6; Tumor antigen se2-2
Protein Family	Centrosomal protein
UniProt Gene Name	CEP290
UniProt Synonym Gene Names	BBS14; KIAA0373; NPHP6; Cep290; CT87
UniProt Entry Name	CE290_HUMAN
NCBI Protein Information	centrosomal protein of 290 kDa

UniProt Comments for CEP290	CEP290: Part of the tectonic-like complex which is required for tissue-specific ciliogenesis and may regulate ciliary membrane composition. 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. Defects in CEP290 are a cause of Joubert syndrome type 5 (JBTS5). 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). 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). 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). 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. 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). 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). 2 isoforms of the human protein are produced by alternative splicing. Protein type: Cancer Testis Antigen (CTA) Chromosomal Location of Human Ortholog: 12q21.32 Cellular Component: centrosome; protein complex; membrane; cytoplasm; nucleus; cytosol; photoreceptor connecting cilium; gamma-tubulin complex Molecular Function: protein binding; microtubule minus-end binding Biological Process: regulation of cAMP metabolic process; organelle organization and biogenesis; positive regulation of transcription, DNA-dependent; establishment and/or maintenance of cell polarity; pronephros development; protein transport; eye photoreceptor cell development; retina development in camera-type eye; cilium biogenesis; mitotic cell cycle; otic vesicle formation; G2/M transition of mitotic cell cycle; hindbrain development Disease: Bardet-biedl Syndrome 14; Leber Congenital Amaurosis 10; Meckel Syndrome, Type 4; Joubert Syndrome 5; Senior-loken Syndrome 6
Research Articles on CEP290	**1.** Detailed analysis of the clinical phenotype of LCA-CEP290 in a large cohort confirms that there is a window of opportunity in childhood for therapeutic intervention based on relative structural preservation in the central cone-rich retina in a significant proportion of patients, with the majority harboring the deep intronic variant potentially tractable to several planned gene editing approaches.
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