




Product datasheet

Anti-CEP290 antibody ab190237

[2 Images](#)

Overview

Product name	Anti-CEP290 antibody
Description	Rabbit polyclonal to CEP290
Host species	Rabbit
Tested applications	Suitable for: WB, IHC-P
Species reactivity	Reacts with: Human Predicted to work with: Mouse 
Immunogen	Synthetic peptide within Human CEP290 aa 750-850. The exact immunogen sequence used to generate this antibody is proprietary information. If additional detail on the immunogen is needed to determine the suitability of the antibody for your needs, please contact our Scientific Support team to discuss your requirements. NP_079390.3. Database link: O15078 <div style="text-align: right;">Run BLAST with  Run BLAST with </div>
Positive control	Human pancreas tissue; K462 cell extracts.
General notes	<p>The Life Science industry has been in the grips of a reproducibility crisis for a number of years. Abcam is leading the way in addressing this with our range of recombinant monoclonal antibodies and knockout edited cell lines for gold-standard validation. Please check that this product meets your needs before purchasing.</p> <p>If you have any questions, special requirements or concerns, please send us an inquiry and/or contact our Support team ahead of purchase. Recommended alternatives for this product can be found below, along with publications, customer reviews and Q&As</p>

Properties

Form	Liquid
Storage instructions	Shipped at 4°C. Store at +4°C short term (1-2 weeks). Upon delivery aliquot. Store at -20°C long term. Avoid freeze / thaw cycle.
Storage buffer	pH: 7.40 Preservative: 0.02% Sodium azide Constituents: 49% PBS, 50% Glycerol (glycerin, glycerine), 0.87% Sodium chloride PBS (without Mg ²⁺ , Ca ²⁺)

Purity	Immunogen affinity purified
Clonality	Polyclonal
Isotype	IgG

Applications

The Abpromise guarantee Our [Abpromise guarantee](#) covers the use of ab190237 in the following tested applications. The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Application	Abreviews	Notes
WB		1/500 - 1/1000. Predicted molecular weight: 290 kDa.
IHC-P		Use a concentration of 10 µg/ml.

Target

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.

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

Involvement in 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.

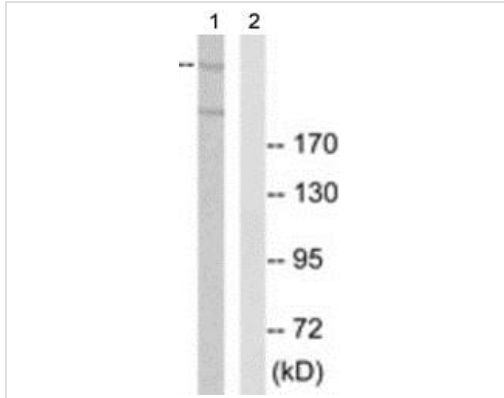
Defects in CEP290 are the cause of Bardet-Biedl syndrome type 14 (BBS14) [MIM:209900]. A syndrome characterized by usually severe pigmentary retinopathy, early-onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension 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 (trialelic inheritance).

Cellular localization

Cytoplasm > cytoskeleton > centrosome. Nucleus. Cell projection > cilium. Connecting cilium of photoreceptor cells, base of cilium in kidney intramedullary collecting duct cells.

Images



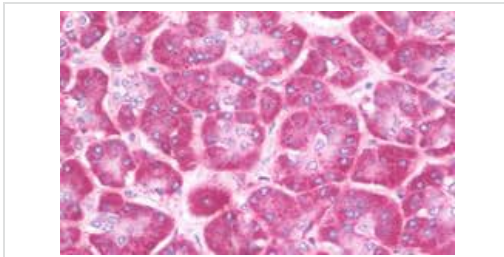
Western blot - Anti-CEP290 antibody (ab190237)

All lanes : Anti-CEP290 antibody (ab190237) at 1/500 dilution

Lane 1 : K562 cell extract

Lane 2 : K562 cell extract with synthesized peptide

Predicted band size: 290 kDa



Immunohistochemistry (Formalin/PFA-fixed paraffin-embedded sections) - Anti-CEP290 antibody (ab190237)

Immunohistochemical analysis of formalin-fixed, paraffin-embedded Human pancreas tissue labeling CEP290 with ab190237 at 10µg/ml.

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