

Product datasheet

Anti-CEP290 antibody ab85728

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Overview

<b>Product name</b>	Anti-CEP290 antibody
<b>Description</b>	Rabbit polyclonal to CEP290
<b>Host species</b>	Rabbit
<b>Tested applications</b>	<b>Suitable for:</b> ICC/IF, WB, IP, ICC
<b>Species reactivity</b>	<b>Reacts with:</b> Human <b>Predicted to work with:</b> Horse, Rhesus monkey, Gorilla, Orangutan, Elephant 
<b>Immunogen</b>	Synthetic peptide corresponding to a region between residue 2429 and 2479 of Human CEP290 (NP_079390.3).
<b>Positive control</b>	Whole cell lysate from HeLa and 293T cells. IF/ICC: HeLa cell line

Properties

<b>Form</b>	Liquid
<b>Storage instructions</b>	Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles.
<b>Storage buffer</b>	Preservative: 0.09% Sodium azide Constituent: Tris citrate/phosphate
<b>Purity</b>	Immunogen affinity purified
<b>Clonality</b>	Polyclonal
<b>Isotype</b>	IgG

Applications

Our [Abpromise guarantee](#) covers the use of **ab85728** 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
ICC/IF		Use a concentration of 5 µg/ml.
WB		1/2000 - 1/10000. Predicted molecular weight: 290 kDa.

Application	Abreviews	Notes
IP		Use at 2-5 µg/mg of lysate.
ICC		1/250 - 1/1000. Formaldehyde fixation with Triton-X 100 permeabilization is recommended.

## 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.

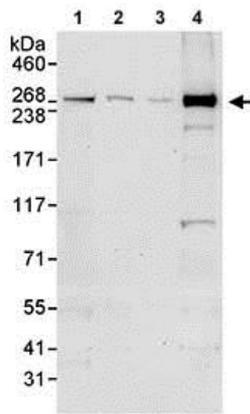
Defects in CEP290 are the cause of Meckel syndrome type 4 (MKS4) [MIM:611134]. MKS4 is an autosomal recessive disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal 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 (ab85728)

**All lanes :** Anti-CEP290 antibody (ab85728) at 0.1 µg/ml

**Lane 1 :** Whole cell lysate from HeLa cells at 50 µg

**Lane 2 :** Whole cell lysate from HeLa cells at 15 µg

**Lane 3 :** Whole cell lysate from HeLa cells at 5 µg

**Lane 4 :** Whole cell lysate from 293T cells at 50 µg

Developed using the ECL technique.

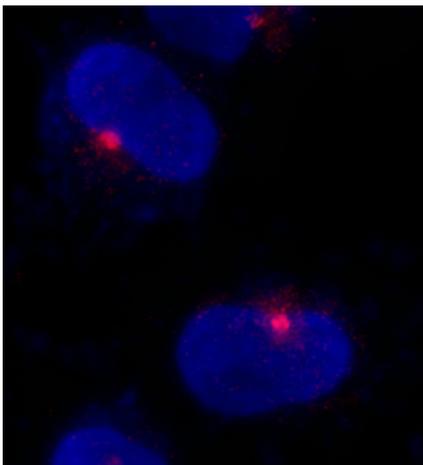
**Predicted band size:** 290 kDa

**Observed band size:** 270 kDa

[why is the actual band size different from the predicted?](#)

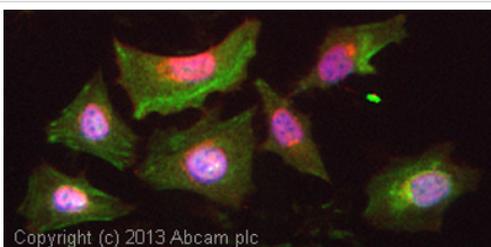
**Additional bands at:** 100 kDa, 200 kDa. We are unsure as to the identity of these extra bands.

**Exposure time:** 30 seconds



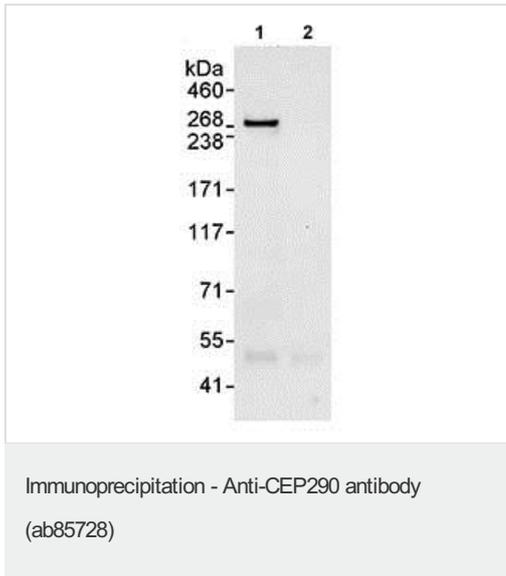
Immunocytochemistry/ Immunofluorescence - Anti-CEP290 antibody (ab85728)

Immunocytochemistry/Immunofluorescence analysis of NBF-fixed asynchronous HeLa cells labelling CEP290 with ab85728 at 1/500. A DyLight® 594-conjugated goat anti-rabbit IgG (1/100) was used as the secondary antibody.



Immunocytochemistry/ Immunofluorescence - Anti-CEP290 antibody (ab85728)

ICC/IF image of ab85728 stained HeLa cells. The cells were 4% formaldehyde fixed (10 min) and then incubated in 1%BSA / 10% normal goat serum / 0.3M glycine in 0.1% PBS-Tween for 1h to permeabilise the cells and block non-specific protein-protein interactions. The cells were then incubated with the antibody (ab85728, 5µg/ml) overnight at +4°C. The secondary antibody (green) was [ab96899](#), DyLight® 488 goat anti-mouse IgG (H+L) used at a 1/250 dilution for 1h. Alexa Fluor® 594 WGA was used to label plasma membranes (red) at a 1/200 dilution for 1h. DAPI was used to stain the cell nuclei (blue) at a concentration of 1.43µM.



Detection of Human CEP290 in Immunoprecipitates of Whole cell lysate from HeLa cells (1 mg for IP, 20% of IP loaded) using ab85728 at 3  $\mu\text{g}/\text{mg}$  lysate for IP (Lane 1) and at 1  $\mu\text{g}/\text{ml}$  for subsequent Western blot detection. Lane 2 represents control IgG IP. Detection: Chemiluminescence with an exposure time of 10 seconds.

**Please note:** All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

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