## abcam

#### Product datasheet

### Anti-CEP290 antibody ab85728

1 Abreviews 10 References 画像数 4

#### 製品の概要

製品名 Anti-CEP290 antibody

製品の詳細 Rabbit polyclonal to CEP290

由来種 Rabbit

**アプリケーション 適用あり:** ICC, WB, IP

種交差性 交差種: Human

交差が予測される動物種: Horse, Rhesus monkey, Gorilla, Orangutan, Elephant 4

免疫原 Synthetic peptide corresponding to a region between residue 2429 and 2479 of Human CEP290

(NP\_079390.3).

特記事項

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.

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

製品の特性

製品の状態 Liquid

保存方法 Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles.

**バッファー** pH: 7

Preservative: 0.09% Sodium azide Constituent: Tris citrate/phosphate

精製度 Immunogen affinity purified

**ポリ/モノ** ポリクローナル

アイソタイプ lgG

アプリケーション

The Abpromise guarantee Abpromise保証は、次のテスト済みアプリケーションにおけるab85728の使用に適用されます

#### アプリケーションノートには、推奨の開始希釈率がありますが、適切な希釈率につきましてはご検討ください。

アプリケーション	Abreviews	特記事項
ICC		Use at an assay dependent concentration.
WB		1/2000 - 1/10000. Predicted molecular weight: 290 kDa.
IP		Use at 2-5 µg/mg of lysate.

#### ターゲット情報

#### 機能

# 組織特異性関連疾患

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.

Ubiquitous. Expressed strongly in placenta and weakly in brain.

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.

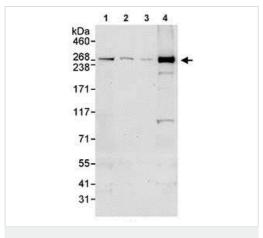
Note=Antibodies against CEP290 are present in sera from patients with cutaneous T-cell lymphomas, but not in the healthy control population.

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 (triallelic inheritance).

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

#### 細胞内局在

#### 画像



Western blot - Anti-CEP290 antibody (ab85728)



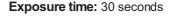
Lane 1: Whole cell lysate from HeLa cells at 50 μg Lane 2: Whole cell lysate from HeLa cells at 15 μg Lane 3: Whole cell lysate from HeLa cells at 5 μg Lane 4: Whole cell lysate from 293T cells at 50 μg

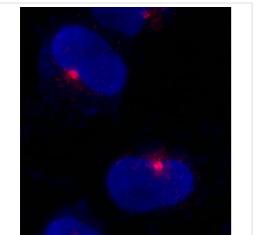
Developed using the ECL technique.

**Predicted band size:** 290 kDa **Observed band size:** 270 kDa

Additional bands at: 100 kDa, 200 kDa. We are unsure as to the

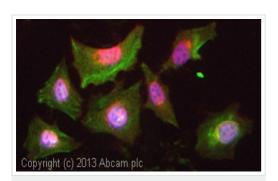
identity of these extra bands.





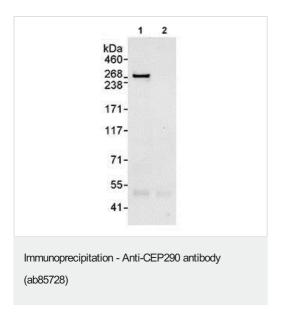
Immunocytochemistry - Anti-CEP290 antibody (ab85728)

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



Immunocytochemistry - 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 lgG (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  $\mu$ g/mg lysate for IP (Lane 1) and at 1  $\mu$ g/ml for subsequent Western blot detection. Lane 2 represents control lgG 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"

#### Our Abpromise to you: Quality guaranteed and expert technical support

- · Replacement or refund for products not performing as stated on the datasheet
- · Valid for 12 months from date of delivery
- Response to your inquiry within 24 hours
- We provide support in Chinese, English, French, German, Japanese and Spanish
- Extensive multi-media technical resources to help you
- · We investigate all quality concerns to ensure our products perform to the highest standards

If the product does not perform as described on this datasheet, we will offer a refund or replacement. For full details of the Abpromise, please visit <a href="https://www.abcam.co.jp/abpromise">https://www.abcam.co.jp/abpromise</a> or contact our technical team.

#### Terms and conditions

• Guarantee only valid for products bought direct from Abcam or one of our authorized distributors