

# CEP290 (H-300): sc-292138

## BACKGROUND

Centrosomes are the major microtubule-organizing centers of mammalian cells. They are composed of a centriole pair and surrounding microtubule-nucleating material, termed pericentriolar material (PCM). Bipolar mitotic spindle assembly relies on two intertwined processes: centriole duplication and centrosome maturation. Failure to properly orchestrate centrosome duplication and maturation is subsequently linked to spindle defects, which can result in aneuploidy and promote cancer progression. The CEP290 (centrosomal protein of 290 kDa) gene encodes a protein of 2,479 amino acids that activates CREB-2-mediated transcription. Specifically, CEP290 ensures the correct localization of ciliary and phototransduction proteins in retinal photoreceptor cells. Mutations in the CEP290 gene have been identified in several diseases, including Joubert syndrome type 5 (JBTS5), Senior-Løken syndrome type 6 (SLSN6), Leber congenital amaurosis type 10 (LCA10) and Meckel syndrome type 4 (MKS4).

## REFERENCES

- Lange, B.M., et al. 2000. Centriole duplication and maturation in animal cells. *Curr. Top. Dev. Biol.* 49: 235-249.
- Palazzo, R.E., et al. 2000. Centrosome maturation. *Curr. Top. Dev. Biol.* 49: 449-470.
- Pelletier, L., et al. 2006. Centriole assembly in *Caenorhabditis elegans*. *Nature* 444: 619-623.
- Valente, E.M., et al. 2006. Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. *Nat. Genet.* 38: 623-625.
- Sayer, J.A., et al. 2006. The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF-4. *Nat. Genet.* 38: 674-681.
- Helou, J., et al. 2007. Mutation analysis of NPHP6/CEP290 in patients with Joubert syndrome and Senior-Løken syndrome. *J. Med. Genet.* 44: 657-663.
- Leitch, C.C., et al. 2008. Hypomorphic mutations in syndromic encephalocoele genes are associated with Bardet-Biedl syndrome. *Nat. Genet.* 40: 443-448.
- den Hollander, A.I., et al. 2008. Leber congenital amaurosis: genes, proteins and disease mechanisms. *Prog. Retin. Eye Res.* 27: 391-419.
- Zhu, F., et al. 2008. The mammalian SPD-2 ortholog CEP192 regulates centrosome biogenesis. *Curr. Biol.* 18: 136-141.

## CHROMOSOMAL LOCATION

Genetic locus: CEP290 (human) mapping to 12q21.32; Cep290 (mouse) mapping to 10 D1.

## SOURCE

CEP290 (H-300) is a rabbit polyclonal antibody raised against amino acids 1441-1740 mapping within an internal region of CEP290 of human origin.

## PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

## APPLICATIONS

CEP290 (H-300) is recommended for detection of CEP290 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

CEP290 (H-300) is also recommended for detection of CEP290 in additional species, including equine, canine, bovine and porcine.

Suitable for use as control antibody for CEP290 siRNA (h): sc-72865, CEP290 siRNA (m): sc-72866, CEP290 shRNA Plasmid (h): sc-72865-SH, CEP290 shRNA Plasmid (m): sc-72866-SH, CEP290 shRNA (h) Lentiviral Particles: sc-72865-V and CEP290 shRNA (m) Lentiviral Particles: sc-72866-V.

Molecular Weight of CEP290: 290 kDa.

## RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

## STORAGE

Store at 4° C, **\*\*DO NOT FREEZE\*\***. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

## RESEARCH USE

For research use only, not for use in diagnostic procedures.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) or our catalog for detailed protocols and support products.


 MONOS  
 Satisfation  
 Guaranteed

Try **CEP290 (B-7): sc-390462** or **CEP290 (G-4): sc-390637**, our highly recommended monoclonal alternatives to CEP290 (H-300).