SANTA CRUZ BIOTECHNOLOGY, INC.

RD3 (D-11): sc-514692

BACKGROUND

Leber congenital amaurosis (LCA) is one of the most common causes of hereditary blindness or severe visual impairment in infants. Mutations in several genes with diverse functions mapping to two loci have been implicated in LCA causation. These proteins are involved in processes such as photoreceptor development and maintenance, phototransduction, vitamin A metabolism and protein trafficking. RD3 (retinal degeneration 3), also known as LCA12, is a 195 amino acid protein expressed in retina. RD3 is suggested to be part of the subnuclear protein complexes involved in diverse processes, such as transcription and splicing. Defects in the gene encoding RD3 are the cause of Leber congenital amaurosis type 12. Infants affected with Leber congenital amaurosis type 12 have little or no retinal photoreceptor function.

REFERENCES

1. Chang, B., et al. 2002. Retinal degeneration mutants in the mouse. Vision Res. 42: 517-525.
2. Mohamed, M.D., et al. 2003. Progression of phenotype in Leber's congenital amaurosis with a mutation at the LCA5 locus. Br. J. Ophthalmol. 87: 473-475.
3. Friedman, J.S., et al. 2006. Premature truncation of a novel protein, RD3, exhibiting subnuclear localization is associated with retinal degeneration. Am. J. Hum. Genet. 79: 1059-1070.
4. Gerber, S., et al. 2007. Mutations in LCA5 are an uncommon cause of Leber Congenital Amaurosis (LCA) type II. Hum. Mutat. 28: 1245.
5. Ramprasad, V.L., et al. 2008. Identification of a novel splice-site mutation in the Lebercilin (LCA5) gene causing Leber Congenital Amaurosis. Mol. Vis. 14: 481-486.
6. den Hollander, A.I., et al. 2008. Leber congenital amaurosis: genes, proteins and disease mechanisms. Prog. Retin. Eye Res. 27: 391-419.
7. Kukekova, A.V., et al. 2009. Canine RD3 mutation establishes rod-cone dysplasia type 2 (rcd2) as ortholog of human and murine RD3. Mamm. Genome 20: 109-123.

CHROMOSOMAL LOCATION

Genetic locus: RD3 (human) mapping to 1q32.3.

SOURCE

RD3 (D-11) is a mouse monoclonal antibody raised against amino acids 135-194 mapping near the C-terminus of RD3 of human origin.

PRODUCT

Each vial contains 200 µg IgG_1 kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

STORAGE

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

APPLICATIONS

RD3 (D-11) is recommended for detection of RD3 of human origin by Western Blotting (starting dilution 1:100, 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).

Suitable for use as control antibody for RD3 siRNA (h): sc-88397, RD3 shRNA Plasmid (h): sc-88397-SH and RD3 shRNA (h) Lentiviral Particles: sc-88397-V.

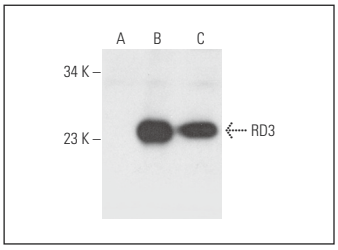
Molecular Weight of RD3: 23 kDa.

Positive Controls: RD3 (h): 293T Lysate: sc-117023 or Y79 cell lysate: sc-2240.

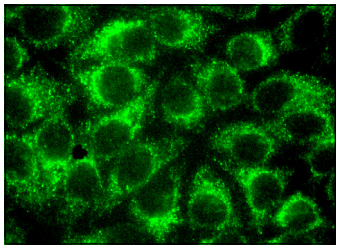
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 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 m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

DATA



RD3 (D-11): sc-514692. Western blot analysis of RD3 expression in non-transfected 293T: sc-117752 (**A**), human RD3 transfected 293T: sc-117023 (**B**) and Y79 (**C**) whole cell lysates.



RD3 (D-11): sc-514692. Immunofluorescence staining of methanol-fixed HeLa cells showing mitochondrial localization.

RESEARCH USE

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

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.