

RD3 (h): 293T Lysate: sc-117023

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.

PRODUCT

RD3 (h): 293T Lysate represents a lysate of human RD3 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

RD3 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive RD3 antibodies. Recommended use: 10-20 µl per lane.

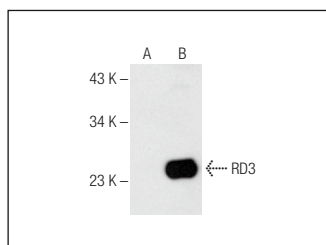
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

RD3 (B-10): sc-376800 is recommended as a positive control antibody for Western Blot analysis of enhanced human RD3 expression in RD3 transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

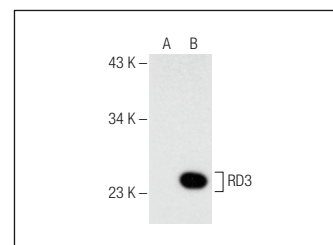
RECOMMENDED SUPPORT REAGENTS

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.

DATA



RD3 (B-10): sc-376800. Western blot analysis of RD3 expression in non-transfected: sc-117752 (A) and human RD3 transfected: sc-117023 (B) 293T whole cell lysates.



RD3 (B-8): sc-376516. Western blot analysis of RD3 expression in non-transfected: sc-117752 (A) and human RD3 transfected: sc-117023 (B) 293T whole cell lysates.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. 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 for detailed protocols and support products.