

RDH12 (h): 293 Lysate: sc-114062

BACKGROUND

Retinol dehydrogenase 11 (RDH12), also known as all-*trans* and 9-*cis* retinol dehydrogenase, LCA3, LCA13 or SDR7C2, is a 316 amino acid protein belonging to the short-chain dehydrogenases/reductases (SDR) family. Widely expressed, mostly in eye, kidney, brain, skeletal muscle and stomach, RDH12 exhibits an oxidoreductive catalytic activity towards retinoids. RDH12 is an efficient NADPH-dependent retinal reductase and displays high activity toward 9-*cis* and all-*trans*-retinol. RDH12 is involved in the metabolism of short-chain aldehydes and may be a key enzyme in the formation of 11-*cis*-retinol from 11-*cis*-retinol during regeneration of the cone visual pigments. Leber congenital amaurosis (LCA) type 3, an inherited autosomal recessive retinal disease, has been associated with defects of RDH12. LCA represents the most common genetic cause of congenital visual impairment in infants and children.

REFERENCES

1. Yzer, S., et al. 2006. Microarray-based mutation detection and phenotypic characterization of patients with Leber congenital amaurosis. *Invest. Ophthalmol. Vis. Sci.* 47: 1167-1176.
2. Lippmann, T., et al. 2006. Indirect exclusion of four candidate genes for generalized progressive retinal atrophy in several breeds of dogs. *J. Negat. Results Biomed.* 5: 19.
3. Maeda, A., et al. 2006. Retinol dehydrogenase (RDH12) protects photoreceptors from light-induced degeneration in mice. *J. Biol. Chem.* 281: 37697-37704.
4. Jacobson, S.G., et al. 2007. RDH12 and RPE65, visual cycle genes causing leber congenital amaurosis, differ in disease expression. *Invest. Ophthalmol. Vis. Sci.* 48: 332-338.
5. Schuster, A., et al. 2007. The phenotype of early-onset retinal degeneration in persons with RDH12 mutations. *Invest. Ophthalmol. Vis. Sci.* 48: 1824-1831.
6. Keller, B. and Adamski, J. 2007. RDH12, a retinol dehydrogenase causing Leber's congenital amaurosis, is also involved in steroid metabolism. *J. Steroid Biochem. Mol. Biol.* 104: 190-194.
7. Kurth, I., et al. 2007. Targeted disruption of the murine retinal dehydrogenase gene *Rdh12* does not limit visual cycle function. *Mol. Cell. Biol.* 27: 1370-1379.
8. Sun, W., et al. 2007. Novel RDH12 mutations associated with Leber congenital amaurosis and cone-rod dystrophy: biochemical and clinical evaluations. *Vision Res.* 47: 2055-2066.
9. Parés, X., et al. 2008. Medium- and short-chain dehydrogenase/reductase gene and protein families: Medium-chain and short-chain dehydrogenases/reductases in retinoid metabolism. *Cell. Mol. Life Sci.* 65: 3936-3949.

CHROMOSOMAL LOCATION

Genetic locus: RDH12 (human) mapping to 14q24.1.

PRODUCT

RDH12 (h): 293 Lysate represents a lysate of human RDH12 transfected 293 cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

RDH12 (h): 293 Lysate is suitable as a Western Blotting positive control for human reactive RDH12 antibodies. Recommended use: 10-20 µl per lane.

Control 293 Lysate: sc-110760 is available as a Western Blotting negative control lysate derived from non-transfected 293 cells.

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.