

## LCA5L (S-20): sc-83226

### 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. LCA5, also known as Lebercilin, is a ciliary protein that is widely expressed during development and localizes to the connecting cilia of photoreceptors and to the microtubules, centrioles and primary cilia of cultured mammalian cells. The Leber congenital amaurosis 5-like protein (LCA5L) is a 670 amino acid protein that belongs to the LCA5 family.

### REFERENCES

1. 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.
2. Gerber, S., et al. 2007. Mutations in LCA5 are an uncommon cause of Leber congenital amaurosis (LCA) type II. *Hum. Mutat.* 28: 1245.
3. den Hollander, A.I., et al. 2007. Mutations in LCA5, encoding the ciliary protein Lebercilin, cause Leber congenital amaurosis. *Nat. Genet.* 39: 889-895.
4. 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.
5. den Hollander, A.I., et al. 2008. Leber congenital amaurosis: genes, proteins and disease mechanisms. *Prog. Retin. Eye Res.* 27: 391-419.
6. Jacobson, S.G., et al. 2009. Leber congenital amaurosis caused by Lebercilin (LCA5) mutation: retained photoreceptors adjacent to retinal disorganization. *Mol. Vis.* 15: 1098-1106.
7. Seong, M.W., et al. 2009. LCA5, a rare genetic cause of leber congenital amaurosis in Koreans. *Ophthalmic Genet.* 30: 54-55.

### CHROMOSOMAL LOCATION

Genetic locus: LCA5L (human) mapping to 21q22.2.

### SOURCE

LCA5L (S-20) is an affinity purified rabbit polyclonal antibody raised against a peptide mapping within an internal region of LCA5L of human origin.

### PRODUCT

Each vial contains 100 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-83226 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

### RESEARCH USE

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

### APPLICATIONS

LCA5L (S-20) is recommended for detection of LCA5L of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), 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 LCA5L siRNA (h): sc-91417, LCA5L shRNA Plasmid (h): sc-91417-SH and LCA5L shRNA (h) Lentiviral Particles: sc-91417-V.

Molecular Weight of LCA5L: 77 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) 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.

### PROTOCOLS

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