

# ALMS1 (K-20): sc-54506

## BACKGROUND

ALMS1 (Alström syndrome protein 1), or ALSS, is a widely expressed protein that localizes to centrosomes and the basal bodies of cilia. It consists of a putative leucine zipper, a tandem repeat domain and a stretch of 17 glutamine residues followed by 7 alanine residues near the N-terminal. Three ALMS1 isoforms exist due to splicing variation. The first isoform is the full length ALMS1. Isoform 2 lacks amino acids 4,121-4,167. The third isoform is only 3,858 amino acids long and it contains an alternate sequence for amino acids 3,850-3,858. ALMS1 may play a role in intracellular transport, microtubule organization and basal body and cilia function. A mutation in the gene encoding ALMS1 results in the dysfunction of basal bodies and/or cilia. This dysfunction is suggested to be the cause of Alström syndrome, a rare autosomal-recessive condition. Symptoms include Insulin resistance, type 2 diabetes and obesity.

## REFERENCES

- Tai, T.S., et al. 2003. Metabolic effects of growth hormone therapy in an Alström syndrome patient. *Horm. Res.* 60: 297-301.
- Collin, G.B., et al. 2005. ALMS1-disrupted mice recapitulate human Alström syndrome. *Hum. Mol. Genet.* 14: 2323-2333.
- Farooqi, I.S. 2005. Genetic and hereditary aspects of childhood obesity. *Best Pract. Res. Clin. Endocrinol. Metab.* 19: 359-374.
- Hearn, T., et al. 2005. Subcellular localization of ALMS1 supports involvement of centrosome and basal body dysfunction in the pathogenesis of obesity, Insulin resistance, and type 2 diabetes. *Diabetes* 54: 1581-1587.
- Arsov, T., et al. 2006. Adaptive failure to high-fat diet characterizes steatohepatitis in ALMS1 mutant mice. *Biochem. Biophys. Res. Commun.* 342: 1152-1159.
- Arsov, T., et al. 2006. Fat aussie-a new Alström syndrome mouse showing a critical role for ALMS1 in obesity, diabetes, and spermatogenesis. *Mol. Endocrinol.* 20: 1610-1622.
- Patel, S., et al. 2006. Common variations in the ALMS1 gene do not contribute to susceptibility to type 2 diabetes in a large white UK population. *Diabetologia* 49: 1209-1213.
- Minton, J.A., et al. 2006. Syndromic obesity and diabetes: changes in body composition with age and mutation analysis of ALMS1 in 12 United Kingdom kindreds with Alström syndrome. *J. Clin. Endocrinol. Metab.* 91: 3110-3116.
- Li, G., et al. 2007. A role for Alström syndrome protein, ALMS1, in kidney ciliogenesis and cellular quiescence. *PLoS Genet.* 3: e8.

## CHROMOSOMAL LOCATION

Genetic locus: ALMS1 (human) mapping to 2p13.1.

## SOURCE

ALMS1 (K-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of ALMS1 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.

Blocking peptide available for competition studies, sc-54506 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

ALMS1 (K-20) is recommended for detection of ALMS1 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).

ALMS1 (K-20) is also recommended for detection of ALMS1 in additional species, including equine.

Suitable for use as control antibody for ALMS1 siRNA (h): sc-72345, ALMS1 shRNA Plasmid (h): sc-72345-SH and ALMS1 shRNA (h) Lentiviral Particles: sc-72345-V.

Molecular Weight of ALMS1: 461 kDa.

## RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (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 donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (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.