## SANTA CRUZ BIOTECHNOLOGY, INC.

# ALMS1 (V-17): sc-54507



#### 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 4121-4167. 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., Lin, S.Y. and Sheu, W.H. 2003. Metabolic effects of growth hormone therapy in an Alström syndrome patient. Horm. Res. 60: 297-301.
- Collin, G.B., Cyr, E., Bronson, R., Marshall, J.D., Gifford, E.J., Hicks, W., Murray, S.A., Zheng, Q.Y., Smith, R.S., Nishina, P.M. and Naggert, J.K. 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., Spalluto, C., Phillips, V.J., Renforth, G.L., Copin, N., Hanley, N.A. and Wilson, D.I. 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., Larter, C.Z., Nolan, C.J., Petrovsky, N., Goodnow, C.C., Teoh, N.C., Yeh, M.M. and Farrell, G.C. 2006. Adaptive failure to high-fat diet characterizes steatohepatitis in ALMS1 mutant mice. Biochem. Biophys. Res. Commun. 342: 1152-1159.
- Arsov, T., Silva, D.G., O'Bryan, M.K., Sainsbury, A., Lee, N.J., Kennedy, C., Manji, S.S., Nelms, K., Liu, C., Vinuesa, C.G., de Kretser, D.M., Goodnow, C.C. and Petrovsky, N. 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., Minton, J.A., Weedon, M.N., Frayling, T.M., Ricketts, C., Hitman, G.A., McCarthy, M.I., Hattersley, A.T., Walker, M. and Barrett, T.G. 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., Owen, K.R., Ricketts, C.J., Crabtree, N., Shaikh, G., Ehtisham, S., Porter, J.R., Carey, C., Hodge, D., Paisey, R., Walker, M. and Barrett, T.G. 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., Vega, R., Nelms, K., Gekakis, N., Goodnow, C., McNamara, P., Wu, H., Hong, N.A. and Glynne, R. 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; Alms1 (mouse) mapping to 6 C3.

## SOURCE

ALMS1 (V-17) 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  $\mu g$  lgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-54507 P, (100  $\mu$ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

### **APPLICATIONS**

ALMS1 (V-17) is recommended for detection of ALMS1 of mouse, rat and 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 (V-17) is also recommended for detection of ALMS1 in additional species, including bovine and porcine.

Suitable for use as control antibody for ALMS1 siRNA (h): sc-72345, ALMS1 siRNA (m): sc-72346, ALMS1 shRNA Plasmid (h): sc-72345-SH, ALMS1 shRNA Plasmid (m): sc-72346-SH, ALMS1 shRNA (h) Lentiviral Particles: sc-72345-V and ALMS1 shRNA (m) Lentiviral Particles: sc-72346-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) Immunofluo-rescence: 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 or our catalog for detailed protocols and support products.