

# AASS (H-300): sc-134661

## BACKGROUND

$\alpha$ -amino adipic semialdehyde synthase (AASS), also designated lysine ketoglutarate reductase (LKR) or saccharopine dehydrogenase (SDH), is a 926 amino acid protein that exists as a homodimer in the mitochondria. AASS acts as a bifunctional enzyme containing the lysine  $\alpha$ -ketoglutarate reductase (LKR) and saccharopine dehydrogenase activities that catalyzes the first two steps in lysine degradation. It is widely expressed with highest expression in liver and transcription of the AASS gene is induced upon starvation. Mutations in the gene encoding AASS result in various forms familial hyperlysinemias (FH), autosomal recessive disorders characterized by hyperlysinemia, lysinuria, and variable saccharopinuria. However, no adverse mental or physical effects have been found in patients with hyperlysinemia.

## REFERENCES

- Dancis, J., et al. 1976. Multiple enzyme defects in familial hyperlysinemia. *Pediatr. Res.* 10: 686-691.
- Markovitz, P.J., et al. 1984. Familial hyperlysinemias. Purification and characterization of the bifunctional amino adipic semialdehyde synthase with lysine-ketoglutarate reductase and saccharopine dehydrogenase activities. *J. Biol. Chem.* 259: 11643-11646.
- Oyanagi, K. and Nagao, M. 1998. Familial hyperlysinemia ( $\alpha$ -amino adipic semialdehyde synthase defect). *Ryoikibetsu Shokogun Shirizu*: 188-190.
- Sacksteder, K.A., et al. 2000. Identification of the  $\alpha$ -amino adipic semialdehyde synthase gene, which is defective in familial hyperlysinemia. *Am. J. Hum. Genet.* 66: 1736-1743.
- Praphanphoj, V., et al. 2001. Identification of the  $\alpha$ -amino adipic semialdehyde dehydrogenase-phosphopantetheinyl transferase gene, the human ortholog of the yeast Lys5 gene. *Mol. Genet. Metab.* 72: 336-342.
- Cleveland, B.M., et al. 2008.  $\alpha$ -amino adipate  $\delta$ -semialdehyde synthase mRNA knockdown reduces the lysine requirement of a mouse hepatic cell line. *J. Nutr.* 138: 2143-2147.
- Tonin, A.M., et al. 2009. Inhibition of creatine kinase activity by lysine in rat cerebral cortex. *Metab. Brain Dis.* 24: 349-360.

## CHROMOSOMAL LOCATION

Genetic locus: AASS (human) mapping to 7q31.32; Aass (mouse) mapping to 6 A3.1.

## SOURCE

AASS (H-300) is a rabbit polyclonal antibody raised against amino acids 627-926 mapping at the C-terminus of AASS of human origin.

## PRODUCT

Each vial contains 200  $\mu$ g IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

## STORAGE

Store at 4° C, **\*\*DO NOT FREEZE\*\***. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

## APPLICATIONS

AASS (H-300) is recommended for detection of AASS of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2  $\mu$ g per 100-500  $\mu$ g of total protein (1 ml of cell lysate)], 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).

AASS (H-300) is also recommended for detection of AASS in additional species, including equine, bovine and porcine.

Suitable for use as control antibody for AASS siRNA (h): sc-89857, AASS siRNA (m): sc-140738, AASS shRNA Plasmid (h): sc-89857-SH, AASS shRNA Plasmid (m): sc-140738-SH, AASS shRNA (h) Lentiviral Particles: sc-89857-V and AASS shRNA (m) Lentiviral Particles: sc-140738-V.

Molecular Weight (predicted) of AASS: 102 kDa.

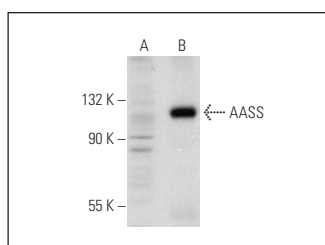
Molecular Weight (observed) of AASS: 116-128 kDa.

Positive Controls: AASS (m): 293T Lysate: sc-118147, NIH/3T3 whole cell lysate: sc-2210 or rat liver extract: sc-2395.

## 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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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.

## DATA



AASS (H-300): sc-134661. Western blot analysis of AASS expression in non-transfected: sc-117752 (A) and mouse AASS transfected: sc-118147 (B) 293T whole cell lysates.

## 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.