ASL (h): 293 Lysate: sc-110465



The Power to Question

BACKGROUND

ASL (argininosuccinate lyase), also known as ASAL or arginosuccinase, is a member of the lyase 1 family of proteins and is predominantly expressed in the liver. Localizing to the cytoplasm and existing as a homotetramer, ASL catalyzes the hydrolytic cleavage of argininosuccinic acid (ASA) to fumarate and Arginine, an essential step of the urea cycle which is crucial for the detoxification of ammonia. This reaction is also involved in the biosynthesis of Arginine. In addition, ASL shares high sequence homology with the avian and reptilian eye lens protein, δ -crystallin. Mutations in the gene encoding ASL lead to an accumulation of ASA in body fluids and result in arginosuccinic aciduria (ASAuria), an autosomal recessive disorder that is characterized by hyperammonemia, liver enlargement, convulsions, physical and mental retardation, episodic unconsciousness, and dry and brittle hair showing trichorrhexis nodosa (weak points or nodes in the hair shaft).

REFERENCES

- 1. Turner, M.A., Simpson, A., McInnes, R.R. and Howell, P.L. 1997. Human argininosuccinate lyase: a structural basis for intragenic complementation. Proc. Natl. Acad. Sci. USA 94: 9063-9068.
- Yu, B. and Howell, P.L. 2000. Intragenic complementation and the structure and function of argininosuccinate lyase. Cell. Mol. Life Sci. 57: 1637-1651.
- Sampaleanu, L.M., Vallee, F., Thompson, G.D. and Howell, P.L. 2001. Three-dimensional structure of the argininosuccinate lyase frequently complementing allele Q286R. Biochemistry 40: 15570-15580.
- Yu, B., Thompson, G.D., Yip, P., Howell, P.L. and Davidson, A.R. 2001.
 Mechanisms for intragenic complementation at the human argininosuccinate lyase locus. Biochemistry 40: 15581-15590.
- 5. Linnebank, M., Tschiedel, E., Häberle, J., Linnebank, A., Willenbring, H., Kleijer, W.J. and Koch, H.G. 2002. Argininosuccinate lyase (ASL) deficiency: mutation analysis in 27 patients and a completed structure of the human ASL gene. Hum. Genet. 111: 350-359.
- Tanaka, T., Nagao, M., Mori, T. and Tsutsumi, H. 2002. A novel stop codon mutation (X465Y) in the argininosuccinate lyase gene in a patient with argininosuccinic aciduria. Tohoku J. Exp. Med. 198: 119-124.
- 7. Christodoulou, J., Craig, H.J., Walker, D.C., Weaving, L.S., Pearson, C.E. and McInnes, R.R. 2006. Deletion hotspot in the argininosuccinate lyase gene: association with topoisomerase II and DNA polymerase α sites. Hum. Mutat. 27: 1065-1071.
- 8. Lee, H.J., Lai, Y.H., Huang, Y.T., Huang, C.W., Chen, Y.H. and Chang, G.G. 2006. Critical role of tryptophanyl residues in the conformational stability of goose δ-crystallin. Exp. Eye Res. 83: 658-666.
- Trevisson, E., Salviati, L., Baldoin, M.C., Toldo, I., Casarin, A., Sacconi, S., Cesaro, L., Basso, G. and Burlina, A.B. 2007. Argininosuccinate lyase deficiency: mutational spectrum in Italian patients and identification of a novel ASL pseudogene. Hum. Mutat. 28: 694-702.

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.

CHROMOSOMAL LOCATION

Genetic locus: ASL (human) mapping to 7g11.21.

PRODUCT

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

APPLICATIONS

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

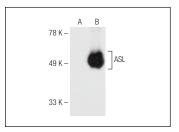
Control 293 Lysate: sc-110760 is available as a Western Blotting negative control lysate derived from non-tranfected 293 cells.

ASL (E-5): sc-374353 is recommended as a positive control antibody for Western Blot analysis of enhanced human ASL expression in ASL transfected 293 cells (starting dilution 1:100, dilution range 1:100-1:1,000).

RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-lgG κ BP-HRP: sc-516102 or m-lgG κ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz MarkerTM Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



ASL (E-5): sc-374353. Western blot analysis of ASL expression in non-transfected: sc-110760 (**A**) and human ASL transfected: sc-110465 (**B**) 293 whole cell heaters.

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.