

AGL (m): 293T Lysate: sc-178264

BACKGROUND

AGL (amylo-1, 6-glucosidase, 4- α -glucanotransferase), also known as GDE (glycogen debranching enzyme), is a 1,532 amino acid protein that exists as 3 alternatively spliced isoforms which are expressed in kidney, liver, heart and muscle in an isoform-specific manner. Exhibiting multifunctional enzyme capabilities, AGL contains two catalytic active sites, one of which acts as an 4- α -glucotransferase and the other of which acts as an amylo-1,6-glucosidase during glycogen degradation. Defects in the gene encoding AGL are the cause of glycogen storage disease type 3 (GSD3), also known as Forbes disease. GSD3 is a metabolic disorder that is characterized by the presence of abnormal glycogen due to a lack of AGL activity. Symptoms of GSD3 generally include hypoglycemia, variable myopathy, hepatomegaly and short stature.

REFERENCES

1. Ding, J.H., de Barse, T., Brown, B.I., Coleman, R.A. and Chen, Y.T. 1990. Immunoblot analyses of glycogen debranching enzyme in different subtypes of glycogen storage disease type III. *J. Pediatr.* 116: 95-100.
2. Yang, B.Z., Ding, J.H., Enghild, J.J., Bao, Y. and Chen, Y.T. 1992. Molecular cloning and nucleotide sequence of cDNA encoding human muscle glycogen debranching enzyme. *J. Biol. Chem.* 267: 9294-9299.
3. Shen, J., Bao, Y., Liu, H.M., Lee, P., Leonard, J.V. and Chen, Y.T. 1996. Mutations in exon 3 of the glycogen debranching enzyme gene are associated with glycogen storage disease type III that is differentially expressed in liver and muscle. *J. Clin. Invest.* 98: 352-357.
4. Orho, M., Bosshard, N.U., Buist, N.R., Gitzelmann, R., Aynsley-Green, A., Blümel, P., Gannon, M.C., Nuttall, F.Q. and Groop, L.C. 1998. Mutations in the liver glycogen synthase gene in children with hypoglycemia due to glycogen storage disease type 0. *J. Clin. Invest.* 102: 507-515.
5. Horinishi, A., Okubo, M., Tang, N.L., Hui, J., To, K.F., Mabuchi, T., Okada, T., Mabuchi, H. and Murase, T. 2002. Mutational and haplotype analysis of AGL in patients with glycogen storage disease type III. *J. Hum. Genet.* 47: 55-59.
6. Sakoda, H., Fujishiro, M., Fujio, J., Shojima, N., Ogihara, T., Kushiya, A., Fukushima, Y., Anai, M., Ono, H., Kikuchi, M., Horike, N., Viana, A.Y., Uchijima, Y., Kurihara, H. and Asano, T. 2005. Glycogen debranching enzyme association with β -subunit regulates AMP-activated protein kinase activity. *Am. J. Physiol. Endocrinol. Metab.* 289: E474-E481.
7. Endo, Y., Horinishi, A., Vorgerd, M., Aoyama, Y., Ebara, T., Murase, T., Odawara, M., Podskarbi, T., Shin, Y.S. and Okubo, M. 2006. Molecular analysis of the AGL gene: heterogeneity of mutations in patients with glycogen storage disease type III from Germany, Canada, Afghanistan, Iran, and Turkey. *J. Hum. Genet.* 51: 958-963.
8. Lucchiari, S., Santoro, D., Pagliarini, S. and Comi, G.P. 2007. Clinical, biochemical and genetic features of glycogen debranching enzyme deficiency. *Acta Myol.* 26: 72-74.
9. Cheng, A., Zhang, M., Gentry, M.S., Worby, C.A., Dixon, J.E. and Saltiel, A.R. 2007. A role for AGL ubiquitination in the glycogen storage disorders of Lafora and Cori's disease. *Genes Dev.* 21: 2399-2409.

CHROMOSOMAL LOCATION

Genetic locus: Agl (mouse) mapping to 3 G1.

PRODUCT

AGL (m): 293T Lysate represents a lysate of mouse AGL transfected 293T cells and is provided as 100 μ g protein in 200 μ l SDS-PAGE buffer.

APPLICATIONS

AGL (m): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive AGL antibodies. Recommended use: 10-20 μ l per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

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.

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.