



**SZABO
SCANDIC**

Part of Europa Biosite

Produktinformation



Forschungsprodukte & Biochemikalien



Zellkultur & Verbrauchsmaterial



Diagnostik & molekulare Diagnostik



Laborgeräte & Service

Weitere Information auf den folgenden Seiten!
See the following pages for more information!



Lieferung & Zahlungsart

siehe unsere [Liefer- und Versandbedingungen](#)

Zuschläge

- Mindermengenzuschlag
- Trockeneiszuschlag
- Gefahrgutzuschlag
- Expressversand

SZABO-SCANDIC HandelsgmbH

Quellenstraße 110, A-1100 Wien

T. +43(0)1 489 3961-0

F. +43(0)1 489 3961-7

mail@szabo-scandic.com

www.szabo-scandic.com

linkedin.com/company/szaboscandic



AGA (h2): 293T Lysate: sc-170753

BACKGROUND

AGA (aspartylglucosaminidase) is a 346 amino acid precursor protein that belongs to the Ntn-hydrolase family and is cleaved to produce an α chain and a β chain. Localized to the lysosome, AGA functions as a heterotetramer composed of two α and two β chains that work together to cleave the GlcNAc-Asn bond that joins oligosaccharides to target glycoproteins. Defects in the gene encoding AGA are the cause of aspartylglucosaminuria (AGU), a lysosomal storage disease that is characterized by severe mental retardation and mild connective tissue abnormalities. The gene encoding AGA maps to human chromosome 4q34.3, which encodes nearly 6% of the human genome and has the largest gene deserts (regions of the genome with no protein encoding genes) of all of the human chromosomes.

REFERENCES

- Mononen, I., Fisher, K.J., Kaartinen, V. and Aronson, N.N. 1993. Aspartyl-glycosaminuria: protein chemistry and molecular biology of the most common lysosomal storage disorder of glycoprotein degradation. *FASEB J.* 7: 1247-1256.
- Tollersrud, O.K., Heiskanen, T. and Peltonen, L. 1994. Human leucocyte glycosylasparaginase is an α/β -heterodimer of 19 kDa α subunit and 17 and 18 kDa β subunit. *Biochem. J.* 300: 541-544.
- Saarela, J., Laine, M., Oinonen, C., Schantz, C., Jalanko, A., Rouvinen, J. and Peltonen, L. 2001. Molecular pathogenesis of a disease: structural consequences of aspartylglucosaminuria mutations. *Hum. Mol. Genet.* 10: 983-995.
- Saarela, J., Oinonen, C., Jalanko, A., Rouvinen, J. and Peltonen, L. 2004. Autoproteolytic activation of human aspartylglucosaminidase. *Biochem. J.* 378: 363-371.
- Saarela, J., von Schantz, C., Peltonen, L. and Jalanko, A. 2004. A novel aspartylglucosaminuria mutation affects translocation of aspartylglucosaminidase. *Hum. Mutat.* 24: 350-351.
- Jackson, M., Clayton, P., Grunewald, S., Keir, G., Mills, K., Mills, P., Winchester, B., Worthington, V. and Young, E. 2005. Elevation of plasma aspartylglucosaminidase is a useful marker for the congenital disorders of glycosylation type I (CDG I). *J. Inherit. Metab. Dis.* 28: 1197-1198.
- Saito, S., Ohno, K., Sugawara, K., Suzuki, T., Togawa, T. and Sakuraba, H. 2008. Structural basis of aspartylglucosaminuria. *Biochem. Biophys. Res. Commun.* 377: 1168-1172.
- Michelakakis, H., Moraitou, M., Mavridou, I. and Dimitriou, E. 2009. Plasma lysosomal enzyme activities in congenital disorders of glycosylation, galactosemia and fructosemia. *Clin. Chim. Acta* 401: 81-83.
- Online Mendelian Inheritance in Man, OMIM™. 2009. Johns Hopkins University, Baltimore, MD. MIM Number: 208400. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>

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: AGA (human) mapping to 4q34.3.

PRODUCT

AGA (h2): 293T Lysate represents a lysate of human AGA transfected 293T cells and is provided as 100 μ g protein in 200 μ l SDS-PAGE buffer.

APPLICATIONS

AGA (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive AGA 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.

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.