



**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



GALE (h2): 293T Lysate: sc-116373

BACKGROUND

GALE, also known as galactowaldenase, UDP-galactose-4-epimerase or SDR1E1, is a 348 amino acid protein that functions as the third enzyme in the Leloir pathway of galactose metabolism. A member of the sugar epimerase family, GALE exists as a homodimer, binds FAD as a cofactor and catalyzes the epimerization of UDP-N-acetylglucosamine to UDP-N-acetylgalactosamine and UDP-glucose to UDP-galactose. The gene encoding GALE maps to human chromosome 1p36.11 and mutations in this gene lead to the development of complex disorder known as epimerase-deficiency galactosemia (EDG) or galactosemia type 3, which is characterized by mental retardation, liver damage, cataracts and deafness.

REFERENCES

1. Reuser, A.J., Koster, J.F., Hoogeveen, A. and Galjaard, H. 1978. Biochemical, immunological, and cell genetic studies in glycogenosis type II. Am. J. Hum. Genet. 30: 132-143.
2. Holton, J.B., Gillett, M.G., MacFaul, R. and Young, R. 1981. Galactosaemia: a new severe variant due to uridine diphosphate galactose-4-epimerase deficiency. Arch. Dis. Child. 56: 885-887.
3. Henderson, M.J., Holton, J.B. and MacFaul, R. 1983. Further observations in a case of uridine diphosphate galactose-4-epimerase deficiency with a severe clinical presentation. J. Inherit. Metab. Dis. 6: 17-20.
4. Kingsley, D.M., Kozarsky, K.F., Hobbie, L. and Krieger, M. 1986. Reversible defects in O-linked glycosylation and LDL receptor expression in a UDP-Gal/UDP-GalNAc 4-epimerase deficient mutant. Cell 44: 749-759.
5. Alano, A., Almashanu, S., Chinsky, J.M., Costeas, P., Blitzer, M.G., Wulfsberg, E.A. and Cowan, T.M. 1998. Molecular characterization of a unique patient with epimerase-deficiency galactosaemia. J. Inherit. Metab. Dis. 21: 341-350.
6. Macratesi, P., Daude, N., Dallapiccola, B., Novelli, G., Allen, R., Okano, Y. and Reichardt, J. 1998. Human UDP-galactose 4' epimerase (GALE) gene and identification of five missense mutations in patients with epimerase-deficiency galactosemia. Mol. Genet. Metab. 63: 26-30.
7. Wohlers, T.M., Christacos, N.C., Harreman, M.T. and Fridovich-Keil, J.L. 1999. Identification and characterization of a mutation, in the human UDP-galactose-4-epimerase gene, associated with generalized epimerase-deficiency galactosemia. Am. J. Hum. Genet. 64: 462-470.
8. Thoden, J.B., Wohlers, T.M., Fridovich-Keil, J.L. and Holden, H.M. 2001. Human UDP-galactose 4-epimerase. Accommodation of UDP-N-acetylglucosamine within the active site. J. Biol. Chem. 276: 15131-15136.
9. Schulz, J.M., Watson, A.L., Sanders, R., Ross, K.L., Thoden, J.B., Holden, H.M. and Fridovich-Keil, J.L. 2004. Determinants of function and substrate specificity in human UDP-galactose 4'-epimerase. J. Biol. Chem. 279: 32796-32803.

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: GALE (human) mapping to 1p36.11.

PRODUCT

GALE (h2): 293T Lysate represents a lysate of human GALE transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

GALE (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive GALE antibodies. Recommended use: 10-20 µl 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.