



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](https://www.linkedin.com/company/szaboscandic) 

Dysbindin 1 (h2): 293T Lysate: sc-177154

BACKGROUND

Hermansky-Pudlak syndrome (HPS) is a rare, genetically heterogeneous, autosomal recessive disorder. It is characterized by oculocutaneous albinism, lysosomal storage defects and prolonged bleeding due to platelet storage pool deficiency. HPS is a result of defects in various cytoplasmic organelles such as melanosomes, platelet dense granules and lysosomes. The HPS proteins, including HPS-1–6 and Dysbindin (also designated HPS-7), all interact within three distinct, ubiquitously expressed protein complexes or biogenesis of lysosome-related organelle complexes. Defects in the genes encoding for these proteins are the cause of HPS. Dysbindin binds to dystrobrevins in the dystrophin-associated protein complex (DPC) complex. Dysbindin is a cytoplasmic protein. Isoforms 1 and 2 are the result of alternative splicing.

REFERENCES

- Schosser, A. and Aschauer, H.N. 2004. In search of susceptibility genes for schizophrenia. *Wien. Klin. Wochenschr.* 116: 827-833.
- Numakawa, T., Yagasaki, Y., Ishimoto, T., Okada, T., Suzuki, T., Iwata, N., Ozaki, N., Taguchi, T., Tatsumi, M., Kamijima, K., Straub, R.E., Weinberger, D.R., Kunugi, H. and Hashimoto, R. 2004. Evidence of novel neuronal functions of Dysbindin, a susceptibility gene for schizophrenia. *Hum. Mol. Genet.* 13: 2699-2708.
- Kendler, K.S. 2004. Schizophrenia genetics and Dysbindin: a corner turned? *Am. J. Psychiatry* 161: 1533-1536.
- Benson, M.A., Sillitoe, R.V. and Blake, D.J. 2004. Schizophrenia genetics: Dysbindin under the microscope. *Trends Neurosci.* 27: 516-519.
- Zill, P., Baghai, T.C., Engel, R., Zwanzger, P., Schule, C., Eser, D., Behrens, S., Rupprecht, R., Moller, H.J., Ackenheil, M. and Bondy, B. 2004. The Dysbindin gene in major depression: an association study. *Am. J. Med. Genet. B Neuropsychiatr. Genet.* 129: 55-58.
- Bray, N.J., Preece, A., Williams, N.M., Moskvina, V., Buckland, P.R., Owen, M.J. and O'Donovan, M.C. 2005. Haplotypes at the Dystrobrevin binding protein 1 (DTNBP1) gene locus mediate risk for schizophrenia through reduced DTNBP1 expression. *Hum. Mol. Genet.* 14: 1947-1954.
- Raybould, R., Green, E.K., MacGregor, S., Gordon-Smith, K., Heron, J., Hyde, S., Caesar, S., Nikolov, I., Williams, N., Jones, L., O'Donovan, M.C., Owen, M.J., Jones, I., Kirov, G. and Craddock, N. 2005. Bipolar disorder and polymorphisms in the Dysbindin gene (DTNBP1). *Biol. Psychiatry* 57: 696-701.
- Arnold, S.E., Talbot, K. and Hahn, C.G. 2005. Neurodevelopment, neuroplasticity, and new genes for schizophrenia. *Prog. Brain Res.* 147: 319-345.

CHROMOSOMAL LOCATION

Genetic locus: DTNBP1 (human) mapping to 6p22.3.

PRODUCT

Dysbindin 1 (h2): 293T Lysate represents a lysate of human Dysbindin 1 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

Dysbindin 1 (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive Dysbindin 1 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.

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.