



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) 

FKRP (h): 293 Lysate: sc-159806

BACKGROUND

Fukutin-related protein (FKRP) is ubiquitously expressed, with highest expression in heart, skeletal muscle and placenta, and weakest expression in lung, liver, brain, kidney and pancreas. FKRP localizes to the medial Golgi apparatus through its N-terminal and transmembrane domains. It is a predicted glycosyltransferase protein that plays a role in α -dystroglycan glycosylation. Mutations in FKRP cause various diseases including congenital muscular dystrophy 1C (MDC1C), limb-girdle muscular dystrophy type 2I (LGMD2I) and congenital muscular dystrophies (CMDs) with brain malformations and mental retardation. FKRP mutations may also cause muscle-eye-brain disease (MEB) and Walker-Warburg syndrome (WWS), disorders characterized by disruption of brain and eye structure in addition to muscular dystrophy. Mislocalization of FKRP from the Golgi apparatus is a potential result of mutations in FKRP.

REFERENCES

1. Brockington, M., Blake, D.J., Prandini, P., Brown, S.C., Torelli, S., Benson, M.A., Ponting, C.P., Estourmet, B., Romero, N.B., Mercuri, E., Voit, T., Sewry, C.A., Guicheney, P. and Muntoni, F. 2001. Mutations in the Fukutin-related protein gene muscular dystrophy with secondary Laminin α 2 deficiency and abnormal glycosylation of α -dystroglycan. *Am. J. Hum. Genet.* 69: 1198-1209.
2. Online Mendelian Inheritance in Man, OMIM™. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 606596. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
3. Esapa, C.T., McIlhinney, R.A. and Blake, D.J. 2004. Fukutin-related protein mutations that cause congenital muscular dystrophy result in ER-retention of the mutant protein in cultured cells. *Hum. Mol. Genet.* 14: 295-305.
4. Müller, T., Krasnianski, M., Witthaut, R., Deschauer, M. and Zierz, S. 2005. Dilated cardiomyopathy may be an early sign of the C826A Fukutin-related protein mutation. *Neuromuscul. Disord.* 15: 372-376.
5. Dolatshad, N.F., Brockington, M., Torelli, S., Skordis, L., Wever, U., Wells, D.J., Muntoni, F. and Brown, S.C. 2005. Mutated Fukutin-related protein (FKRP) localises as wildtype in differentiated muscle cells. *Exp. Cell Res.* 309: 370-378.
6. Boito, C.A., Melacini, P., Vianello, A., Prandini, P., Gavassini, B.F., Bagattin, A., Siciliano, G., Angelini, C. and Pegoraro, E. 2005. Clinical and molecular characterization of patients with limb-girdle muscular dystrophy type 2I. *Arch. Neurol.* 62: 1894-1899.
7. Vajsar, J. and Schachter, H. 2006. Walker-Warburg syndrome. *Orphanet J. Rare Dis.* 1: 29.

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.

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.

CHROMOSOMAL LOCATION

Genetic locus: FKRP (human) mapping to 19q13.32.

PRODUCT

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

APPLICATIONS

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

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

RESEARCH USE

For research use only, not for use in diagnostic procedures.