



**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



FAM90A1 (h): 293T Lysate: sc-115644

BACKGROUND

Encoding over 1,100 genes within 132 million bases, chromosome 12 makes up about 4.5% of the human genome. A number of skeletal deformities are linked to chromosome 12 including hypochondrogenesis, achondrogenesis and Kniest dysplasia. Noonan syndrome, which includes heart and facial developmental defects among the primary symptoms, is caused by a mutant form of PTPN11 gene product, SH-PTP2. Chromosome 12 is also home to a homeobox gene cluster which encodes crucial transcription factors for morphogenesis, and the natural killer complex gene cluster encoding C-type lectin proteins which mediate the NK cell response to MHC I interaction. Trisomy 12p leads to facial development defects, seizure disorders and a host of other symptoms varying in severity depending on the extent of mosaicism and is most severe in cases of complete trisomy. The FAM90A1 gene product has been provisionally designated FAM90A1 pending further characterization.

REFERENCES

- Allen, T.L., Brothman, A.R., Carey, J.C. and Chance, P.F. 1996. Cytogenetic and molecular analysis in trisomy 12p. Am. J. Med. Genet. 63: 250-256.
- Yang, W. and Cole, W.G. 1998. Low basal transcripts of the COL2A1 collagen gene from lymphoblasts show alternative splicing of exon 12 in the Kniest form of spondyloepiphyseal dysplasia. Hum. Mutat. 1: S1-S2.
- Trowsdale, J., Barten, R., Haude, A., Stewart, C.A., Beck, S., et al. 2001. The genomic context of natural killer receptor extended gene families. Immunol. Rev. 181: 20-38.
- Zumkeller, W., Völleth, M., Muschke, P., Tönnies, H., Heller, A., Liehr, T., Wieacker, P. and Stumm, M. 2004. Genotype/phenotype analysis in a patient with pure and complete trisomy 12p. Am. J. Med. Genet. A 129A: 261-264.
- Kelley, J., Walter, L. and Trowsdale, J. 2005. Comparative genomics of natural killer cell receptor gene clusters. PLoS Genet. 1: e27.
- Nishimura, G., Haga, N., Kitoh, H., Tanaka, Y., Sonoda, T., Kitamura, M., Shirahama, S., Itoh, T., Nakashima, E., Ohashi, H. and Ikegawa, S. 2005. The phenotypic spectrum of COL2A1 mutations. Hum. Mutat. 26: 36-43.
- Segel, R., Peter, I., Demmer, L.A., Cowan, J.M., Hoffman, J.D. and Bianchi, D.W. 2006. The natural history of trisomy 12p. Am. J. Med. Genet. A 140A: 695-703.
- Stein, R. 2007. Genetics of Noonan syndrome—a new gene, and the search is still on. Clin. Genet. 72: 402-404.
- van der Burgt, I. 2007. Noonan syndrome. Orphanet J. Rare Dis. 2: 4.

CHROMOSOMAL LOCATION

Genetic locus: FAM90A1 (human) mapping to 12p13.31.

PRODUCT

FAM90A1 (h): 293T Lysate represents a lysate of human FAM90A1 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

FAM90A1 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive FAM90A1 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.

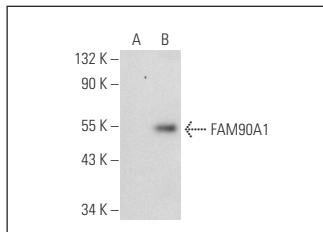
FAM90A (A-8): sc-393598 is recommended as a positive control antibody for Western Blot analysis of enhanced human FAM90A1 expression in FAM90A1 transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended:

1) Western Blotting: use m-IgG_κ BP-HRP: sc-516102 or m-IgG_κ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



FAM90A (A-8): sc-393598. Western blot analysis of FAM90A1 expression in non-transfected: sc-117752 (A) and human FAM90A1 transfected: sc-115644 (B) 293T whole cell lysates.

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.