



# 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](mailto:mail@szabo-scandic.com)

[www.szabo-scandic.com](http://www.szabo-scandic.com)

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

# FAM111A siRNA (m): sc-145017

## BACKGROUND

FAM111A, also known as KCS2 or GCLEB, is a 611 amino acid protein containing one PIP-box motif. Mutations in FAM111A are linked to Kenny-Caffey syndrome type 2, a rare dysmorphic syndrome characterized by proportionate short stature, cortical thickening and medullary stenosis of tubular bones, delayed closure of anterior fontanelle, eye abnormalities, and hypoparathyroidism. FAM111A mutations are also linked to gracile bone dysplasia (GCLEB), also known as osteocraniostenosis, a perinatally lethal condition characterized by narrowing of the medullary cavity of the skull and long bones, gracile bones with thin diaphyses, premature closure of basal cranial sutures and microphthalmia. These disorders suggest FAM111A is a key molecule for normal bone development, height gain and parathyroid hormone function. FAM111A colocalizes with PCNA in the nucleus at replication sites, and promotes S-phase entry and DNA synthesis. FAM111A also interacts with SV40 large T antigen and functions as a host range restriction factor.

## REFERENCES

1. Nagase, T., Kikuno, R. and Ohara, O. 2001. Prediction of the coding sequences of unidentified human genes. XXI. The complete sequences of 60 new cDNA clones from brain which code for large proteins. *DNA Res.* 8: 179-187.
2. Kimura, K., Wakamatsu, A., Suzuki, Y., Ota, T., Nishikawa, T., Yamashita, R., Yamamoto, J., Sekine, M., Tsuritani, K. and Wakaguri, H. 2006. Diversification of transcriptional modulation: large-scale identification and characterization of putative alternative promoters of human genes. *Genome Res.* 16: 55-65.
3. Fine, D.A., Rozenblatt-Rosen, O., Padi, M., Korkhin, A., James, R.L., Adelmant, G., Yoon, R., Guo, L., Berrios, C. and Zhang, Y. 2012. Identification of FAM111A as an SV40 host range restriction and adenovirus helper factor. *PLoS Pathog.* 8: e1002949.
4. Unger, S., Góna, M.W., Le Bèchec, A., Do Vale-Pereira, S., Bedeschi, M.F., Geiberger, S., Grigelioniene, G., Horemuzova, E., Lalatta, F. and Lausch, E. 2013. FAM111A mutations result in hypoparathyroidism and impaired skeletal development. *Am. J. Hum. Genet.* 92: 990-995.
5. Nikkel, S.M., Ahmed, A., Smith, A., Marcadier, J., Bulman, D.E. and Boycott, K.M. 2014. Mother-to-daughter transmission of Kenny-Caffey syndrome associated with the recurrent, dominant FAM111A mutation p.Arg569His. *Clin. Genet.* 86: 394-395.
6. Isojima, T., Doi, K., Mitsui, J., Oda, Y., Tokuhira, E., Yasoda, A., Yorifuji, T., Horikawa, R., Yoshimura, J. and Ishiura, H. 2014. A recurrent *de novo* FAM111A mutation causes Kenny-Caffey syndrome type 2. *J. Bone Miner. Res.* 29: 992-998.
7. Alabert, C., Bukowski-Wills, J.C., Lee, S.B., Kustatscher, G., Nakamura, K., de Lima Alves, F., Menard, P., Mejlvang, J., Rappsilber, J. and Groth, A. 2014. Nascent chromatin capture proteomics determines chromatin dynamics during DNA replication and identifies unknown fork components. *Nat. Cell Biol.* 16: 281-293.

## CHROMOSOMAL LOCATION

Genetic locus: Fam111a (mouse) mapping to 19 A.

## PRODUCT

FAM111A siRNA (m) is a pool of 3 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10  $\mu$ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see FAM111A shRNA Plasmid (m): sc-145017-SH and FAM111A shRNA (m) Lentiviral Particles: sc-145017-V as alternate gene silencing products.

For independent verification of FAM111A (m) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-145017A, sc-145017B and sc-145017C.

## STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNases and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330  $\mu$ l of the RNase-free water provided. Resuspension of the siRNA duplex in 330  $\mu$ l of RNase-free water makes a 10  $\mu$ M solution in a 10  $\mu$ M Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

## APPLICATIONS

FAM111A siRNA (m) is recommended for the inhibition of FAM111A expression in mouse cells.

## SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10  $\mu$ M in 66  $\mu$ l. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

## RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor FAM111A gene expression knockdown using RT-PCR Primer: FAM111A (m)-PR: sc-145017-PR (20  $\mu$ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

## RESEARCH USE

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