

Pcdh19 siRNA (m): sc-152059

BACKGROUND

PCDH19 (protocadherin-19) is a 1,148 amino acid single-pass type I membrane protein that contains six cadherin domains and exists as two alternatively spliced isoforms. As a potential calcium-dependent cell adhesion protein, PCDH19 is moderately expressed in ovary and most regions of the brain. PCDH19 is expressed at lower levels in primary skin fibroblasts and cerebellum. Defects in PCDH19 are the cause of epileptic encephalopathy early infantile type 9, also known as epilepsy female-restricted with mental retardation, which is a condition characterized by early onset seizures, cognitive impairment and delayed development of variable severity. Additional features include autistic signs and psychosis. The disorder is sex-limited, with the phenotype being restricted to females. The gene that encodes PCDH19 consists of approximately 118,630 bases and maps to human chromosome Xq22.1.

REFERENCES

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4. Dibbens, L.M., et al. 2008. X-linked protocadherin 19 mutations cause female-limited epilepsy and cognitive impairment. *Nat. Genet.* 40: 776-781.
5. Depienne, C., et al. 2009. Sporadic infantile epileptic encephalopathy caused by mutations in PCDH19 resembles Dravet syndrome but mainly affects females. *PLoS Genet.* 5: e1000381.
6. Jamal, S.M., et al. 2010. Novel *de novo* PCDH19 mutations in three unrelated females with epilepsy female restricted mental retardation syndrome. *Am. J. Med. Genet. A* 152A: 2475-2481.
7. Hynes, K., et al. 2010. Epilepsy and mental retardation limited to females with PCDH19 mutations can present *de novo* or in single generation families. *J. Med. Genet.* 47: 211-216.
8. Marini, C., et al. 2010. Protocadherin 19 mutations in girls with infantile-onset epilepsy. *Neurology* 75: 646-653.
9. Depienne, C., et al. 2011. Mutations and deletions in PCDH19 account for various familial or isolated epilepsies in females. *Hum. Mutat.* 32: E1959-E1975.

CHROMOSOMAL LOCATION

Genetic locus: Pcdh19 (mouse) mapping to X E3.

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.

PRODUCT

Pcdh19 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 μ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see Pcdh19 shRNA Plasmid (m): sc-152059-SH and Pcdh19 shRNA (m) Lentiviral Particles: sc-152059-V as alternate gene silencing products.

For independent verification of Pcdh19 (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-152059A, sc-152059B and sc-152059C.

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

APPLICATIONS

Pcdh19 siRNA (m) is recommended for the inhibition of Pcdh19 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 μ M in 66 μ 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 Pcdh19 gene expression knockdown using RT-PCR Primer: Pcdh19 (m)-PR: sc-152059-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.