SANTA CRUZ BIOTECHNOLOGY, INC.

MKS3 siRNA (h): sc-77580

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The Power to Question

BACKGROUND

MKS3 (Meckel syndrome type 3), also known as TMEM67 (transmembrane protein 67), JBTS6 or Meckelin, is a 995 amino acid multi-pass membrane protein that localizes to the primary cilium and to the plasma membrane. Existing as multiple alternatively spliced isoforms that are expressed in a variety of adult and fetal tissues, such as the spinal cord, MKS3 plays a role in centriole migration to the apical membrane and may be involved in the formation of the primary cilium. Defects in the gene encoding MKS3 are the cause of Meckel syndrome type 3 (MKS3) and Joubert syndrome type 6 (JBTS6), both of which are autosomal recessive disorders. MKS3 is characterized by renal cysts and anomalies of the central nervous system, while JBTS6 is characterized by cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities, thickened and reoriented superior cerebellar peduncles and an abnormally large interpeduncular fossa.

REFERENCES

1. Morgan, N.V., et al. 2002. A novel locus for Meckel-Gruber syndrome, MKS3, maps to chromosome 8q24. Hum. Genet. 111: 456-461.
2. Smith, U.M., et al. 2006. The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome and the wpk rat. Nat. Genet. 38: 191-196.
3. Baala, L., et al. 2007. The Meckel-Gruber syndrome gene, MKS3, is mutated in Joubert syndrome. Am. J. Hum. Genet. 80: 186-194.
4. Dawe, H.R., et al. 2007. The Meckel-Gruber syndrome proteins MKS1 and meckelin interact and are required for primary cilium formation. Hum. Mol. Genet. 16: 173-186.
5. Consugar, M.B., et al. 2007. Molecular diagnostics of Meckel-Gruber syndrome highlights phenotypic differences between MKS1 and MKS3. Hum. Genet. 121: 591-599.
6. Khaddour, R., et al. 2007. Spectrum of MKS1 and MKS3 mutations in Meckel syndrome: a genotype-phenotype correlation. Mutation in brief #960. Online. Hum. Mutat. 28: 523-524.
7. Brancati, F., et al. 2009. MKS3/TMEM67 mutations are a major cause of COACH syndrome, a Joubert syndrome related disorder with liver involvement. Hum. Mutat. 30: E432-E442.

CHROMOSOMAL LOCATION

Genetic locus: TMEM67 (human) mapping to 8q22.1.

PRODUCT

MKS3 siRNA (h) 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 µM solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see MKS3 shRNA Plasmid (h): sc-77580-SH and MKS3 shRNA (h) Lentiviral Particles: sc-77580-V as alternate gene silencing products.

For independent verification of MKS3 (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-77580A, sc-77580B and sc-77580C.

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 µl of the RNAse-free water provided. Resuspension of the siRNA duplex in 330 µl of RNAse-free water makes a 10 µM solution in a 10 µM Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

MKS3 siRNA (h) is recommended for the inhibition of MKS3 expression in human 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 µM in 66 µl. 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 MKS3 gene expression knockdown using RT-PCR Primer: MKS3 (h)-PR: sc-77580-PR (20 µl). 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.

PROTOCOLS

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