SANTA CRUZ BIOTECHNOLOGY, INC.

MKS3 (P-14): sc-87298

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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

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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.
8. Online Mendelian Inheritance in Man, OMIM™. 2009. Johns Hopkins University, Baltimore, MD. MIM Number: 609884. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/

CHROMOSOMAL LOCATION

Genetic locus: TMEM67 (human) mapping to 8q22.1; Tmem67 (mouse) mapping to 4 A1.

SOURCE

MKS3 (P-14) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of MKS3 of human origin.

RESEARCH USE

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

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-87298 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

MKS3 (P-14) is recommended for detection of MKS3 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000); non cross-reactive with MKS1.

MKS3 (P-14) is also recommended for detection of MKS3 in additional species, including equine, canine and porcine.

Suitable for use as control antibody for MKS3 siRNA (h): sc-77580, MKS3 siRNA (m): sc-149460, MKS3 shRNA Plasmid (h): sc-77580-SH, MKS3 shRNA Plasmid (m): sc-149460-SH, MKS3 shRNA (h) Lentiviral Particles: sc-77580-V and MKS3 shRNA (m) Lentiviral Particles: sc-149460-V.

Molecular Weight of MKS3: 108 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

PROTOCOLS

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