

SMCR8 (N-16): sc-137778

BACKGROUND

Smith-Magenis syndrome (SMS) is a rare disorder that is characterized by multiple congenital anomalies and mental retardation, with associated sleep disturbance and behavioral abnormalities. Autistic-like behaviors and symptoms begin to develop at about 18 months of age. Although there is no cure for SMS, treatment focuses on the management of its symptoms such as treating sleep disturbance, management of behaviors, speech and occupational therapies, as well as minor medical interventions. The genetic locus of 17p11.2 is deleted in patients affected with SMS. Many studies have linked the disorder to the haploinsufficiency of the retinoic acid-induced 1 (RAI1) gene that maps within the Smith-Magenis chromosome region. SMCR8 (Smith-Magenis syndrome chromosomal region candidate gene 8) is a 937 amino acid ubiquitously expressed protein that is encoded by a gene that also maps within the critical region of deletion in SMS. There are two isoforms of SMCR8 that are produced as a result of alternative splicing events.

REFERENCES

1. Moncla, A., et al. 1991. Smith-Magenis syndrome: a new contiguous gene syndrome. Report of three new cases. *J. Med. Genet.* 28: 627-632.
2. Fischer, H., et al. 1993. Constitutional interstitial deletion of 17(p11.2) (Smith-Magenis syndrome): a clinically recognizable microdeletion syndrome. Report of two cases and review of the literature. *Klin. Padiatr.* 205: 162-166.
3. Bi, W., et al. 2002. Genes in a refined Smith-Magenis syndrome critical deletion interval on chromosome 17p11.2 and the syntenic region of the mouse. *Genome Res.* 12: 713-728.
4. Gropman, A.L., et al. 2006. Neurologic and developmental features of the Smith-Magenis syndrome (del 17p11.2). *Pediatr. Neurol.* 34: 337-350.
5. Gropman, A.L., et al. 2007. New developments in Smith-Magenis syndrome (del 17p11.2). *Curr. Opin. Neurol.* 20: 125-134.
6. Elesa, S.H., et al. 2008. Smith-Magenis syndrome. *Eur. J. Hum. Genet.* 16: 412-421.
7. Online Mendelian Inheritance in Man, OMIM[™]. 2008. Johns Hopkins University, Baltimore, MD. MIM Number: 182290. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
8. Laje, G., et al. 2010. Autism spectrum features in Smith-Magenis syndrome. *Am. J. Med. Genet. C Semin. Med. Genet.* 154C: 456-462.

CHROMOSOMAL LOCATION

Genetic locus: SMCR8 (human) mapping to 17p11.2; Smcr8 (mouse) mapping to 11 B2.

SOURCE

SMCR8 (N-16) is an affinity purified rabbit polyclonal antibody raised against a peptide mapping at the N-terminus of SMCR8 of human origin.

STORAGE

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

PRODUCT

Each vial contains 100 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-137778 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

SMCR8 (N-16) is recommended for detection of SMCR8 of mouse, rat and human origin by Western Blotting (starting dilution 1:100, dilution range 1:50-1:500), immunofluorescence (starting dilution 1:25, dilution range 1:25-1:250) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000); non cross-reactive with SMCR5, SMCR7 or SMCR7L.

SMCR8 (N-16) is also recommended for detection of SMCR8 in additional species, including equine, canine and porcine.

Suitable for use as control antibody for SMCR8 siRNA (h): sc-94057, SMCR8 siRNA (m): sc-153624, SMCR8 shRNA Plasmid (h): sc-94057-SH, SMCR8 shRNA Plasmid (m): sc-153624-SH, SMCR8 shRNA (h) Lentiviral Particles: sc-94057-V and SMCR8 shRNA (m) Lentiviral Particles: sc-153624-V.

Molecular Weight of SMCR8 isoform 1: 105 kDa.

Molecular Weight of SMCR8 isoform 2: 88 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker[™] compatible goat anti-rabbit IgG-HRP: sc-2030 (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 goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (dilution range: 1:100-1:400) with UltraCruz[™] Mounting Medium: sc-24941.

RESEARCH USE

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

PROTOCOLS

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