SANTA CRUZ BIOTECHNOLOGY, INC.

SMCR7 (C-15): sc-165504

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. SMCR7 (Smith-Magenis syndrome chromosomal region candidate gene 7) is a 454 amino acid single-pass membrane protein that is encoded by a gene that also maps within the critical region of deletion in SMS. SMCR7 is expressed in all tissues with highest expression in skeletal muscle and heart. There are two isoforms of SMCR7 that are produced as a result of alternative splicing events.

REFERENCES

1. Moncla, A., Livet, M.O., Auger, M., Mattei, J.F., Mattei, M.G. and Giraud, F. 1991. Smith-Magenis syndrome: a new contiguous gene syndrome. Report of three new cases. J. Med. Genet. 28: 627-632.
2. Fischer, H., Oswald, H.P., Duba, H.C., Doczy, L., Simma, B., Utermann, G. and Haas, O.A. 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., Yan, J., Stankiewicz, P., Park, S.S., Walz, K., Boerkoel, C.F., Potocki, L., Shaffer, L.G., Devriendt, K., Nowaczyk, M.J., Inoue, K. and Lupski, J.R. 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., Duncan, W.C. and Smith, A.C. 2006. Neurologic and developmental features of the Smith-Magenis syndrome (del 17p11.2). Pediatr. Neurol. 34: 337-350.
5. Gropman, A.L., Elsea, S., Duncan, W.C. and Smith, A.C. 2007. New developments in Smith-Magenis syndrome (del 17p11.2). Curr. Opin. Neurol. 20: 125-134.
6. Elsea, S.H. and Girirajan, S. 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., Morse, R., Richter, W., Ball, J., Pao, M. and Smith, A.C. 2010. Autism spectrum features in Smith-Magenis syndrome. Am. J. Med. Genet. C Semin. Med. Genet. 154C: 456-462.

RESEARCH USE

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

CHROMOSOMAL LOCATION

Genetic locus: SMCR7 (human) mapping to 17p11.2; Smcr7 (mouse) mapping to 11 B2.

SOURCE

SMCR7 (C-15) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the C-terminus of SMCR7 of human origin.

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-165504 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

SMCR7 (C-15) is recommended for detection of SMCR7 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 other SMCR family members.

Suitable for use as control antibody for SMCR7 siRNA (h): sc-93868, SMCR7 siRNA (m): sc-153622, SMCR7 shRNA Plasmid (h): sc-93868-SH, SMCR7 shRNA Plasmid (m): sc-153622-SH, SMCR7 shRNA (h) Lentiviral Particles: sc-93868-V and SMCR7 shRNA (m) Lentiviral Particles: sc-153622-V.

Molecular Weight of SMCR7 isoforms: 49/21 kDa.

Positive Controls: human fetal heart tissue.

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.