

SMCR7 (E-1): sc-515800

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

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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.
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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/>
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CHROMOSOMAL LOCATION

Genetic locus: Mief2 (mouse) mapping to 11 B2.

SOURCE

SMCR7 (E-1) is a mouse monoclonal antibody raised against amino acids 85-244 mapping within an internal region of SMCR7 of mouse origin.

RESEARCH USE

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

PRODUCT

Each vial contains 200 µg IgG_{2a} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

SMCR7 (E-1) is recommended for detection of SMCR7 of mouse and rat origin by Western Blotting (starting dilution 1:100, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], 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).

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

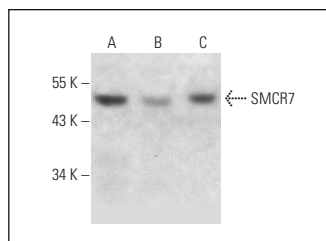
Molecular Weight of SMCR7 isoform 1/2: 49/21 kDa.

Positive Controls: NIH/3T3 whole cell lysate: sc-2210, C3H/10T1/2 cell lysate: sc-3801 or ES-D3 whole cell lysate: sc-364776.

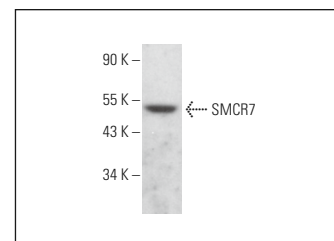
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use m-IgGκ BP-FITC: sc-516140 or m-IgGκ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

DATA



SMCR7 (E-1): sc-515800. Western blot analysis of SMCR7 expression in NIH/3T3 (A), C3H/10T1/2 (B) and ES-D3 (C) whole cell lysates.



SMCR7 (E-1): sc-515800. Western blot analysis of SMCR7 expression in BC3H1 whole cell lysate.

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 for detailed protocols and support products.