

ASPM (H-141): sc-98903

BACKGROUND

Microcephaly is a genetic disorder in which the affected individual has a head circumference less than three standard deviations below the sex- and age-related mean. The reason for the reduced head circumference is due to the formation of a small brain of normal proportions; all affected individuals are mentally retarded. ASPM (for abnormal spindle homolog, microcephaly associated), also designated microcephaly, primary autosomal recessive 5 (MCPH5), is caused by mutation in the ASPM gene. In a comprehensive mutation screen of the ASPM gene, 19 mutations were identified in a cohort of 23 consanguineous families. The mutations occur throughout the gene and are all assumed to be protein truncating. Research demonstrates that phenotypic variation in 51 affected individuals occurs in the degree of microcephaly (five to 11 SDs below normal) and of mental retardation (mild to severe), but appeared to be independent of mutation position in the gene.

REFERENCES

1. Perez-Castillo, A., et al. 1984. Is a gene for microcephaly located on chromosome 1? *Hum. Genet.* 67: 230-232.
2. Jamieson, C.R., et al. 2000. Primary autosomal recessive microcephaly: MCPH5 maps to 1q25-q32. *Am. J. Hum. Genet.* 67: 1575-1577.
3. Bond, J., et al. 2002. ASPM is a major determinant of cerebral cortical size. *Nat. Genet.* 32: 316-320.
4. Online Mendelian Inheritance in Man, OMIM™. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 608716. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>
5. Bond, J., et al. 2003. Protein-truncating mutations in ASPM cause variable reduction in brain size. *Am. J. Hum. Genet.* 73: 1170-1177.
6. Wallerman, O., et al. 2003. Evidence for a second gene for primary microcephaly at MCPH5 on chromosome 1. *Hereditas* 139: 64-67.
7. Kumar, A., et al. 2004. Genetic analysis of primary microcephaly in Indian families: novel ASPM mutations. *Clin. Genet.* 66: 341-348.
8. Kouprina, N., et al. 2005. The microcephaly ASPM gene is expressed in proliferating tissues and encodes for a mitotic spindle protein. *Hum. Mol. Genet.* 14: 2155-2165.
9. Mochida, G.H. 2005. Cortical malformation and pediatric epilepsy: a molecular genetic approach. *J. Child. Neurol.* 20: 300-303.

CHROMOSOMAL LOCATION

Genetic locus: ASPM (human) mapping to 1q31.3; Aspm (mouse) mapping to 1 F.

SOURCE

ASPM (H-141) is a rabbit polyclonal antibody raised against amino acids 790-1071 mapping within an internal region of ASPM 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 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Available as TransCruz reagent for Gel Supershift and ChIP applications, sc-98903 X, 200 µg/0.1 ml.

APPLICATIONS

ASPM (H-141) is recommended for detection of ASPM of mouse, rat and human origin by Western Blotting (starting dilution 1:200, 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).

ASPM (H-141) is also recommended for detection of ASPM in additional species, including equine, canine, bovine, porcine and avian.

Suitable for use as control antibody for ASPM siRNA (h): sc-61006, ASPM siRNA (m): sc-61007, ASPM shRNA Plasmid (h): sc-61006-SH, ASPM shRNA Plasmid (m): sc-61007-SH, ASPM shRNA (h) Lentiviral Particles: sc-61006-V and ASPM shRNA (m) Lentiviral Particles: sc-61007-V.

ASPM (H-141) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

Molecular Weight of ASPM: 410 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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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.