



ASPM siRNA (h): sc-61006

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 (5 to 11 SDs below normal) and of mental retardation (mild to severe), but appeared to be independent of mutation position in the gene.

REFERENCES

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3. Bond, J., et al. 2002. ASPM is a major determinant of cerebral cortical size. *Nat. Genet.* 32: 316-320.
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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.
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CHROMOSOMAL LOCATION

Genetic locus: ASPM (human) mapping to 1q31.3.

RESEARCH USE

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

PROTOCOLS

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

PRODUCT

ASPM siRNA (h) is a pool of 3 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10 μ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see ASPM shRNA Plasmid (h): sc-61006-SH and ASPM shRNA (h) Lentiviral Particles: sc-61006-V as alternate gene silencing products.

For independent verification of ASPM (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-61006A, sc-61006B and sc-61006C.

STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNases and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330 μ l of the RNase-free water provided. Resuspension of the siRNA duplex in 330 μ l of RNase-free water makes a 10 μ M solution in a 10 μ M Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

ASPM siRNA (h) is recommended for the inhibition of ASPM expression in human cells.

SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10 μ M in 66 μ l. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor ASPM gene expression knockdown using RT-PCR Primer: ASPM (h)-PR: sc-61006-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.