

AHI1 siRNA (h): sc-72465

BACKGROUND

AHI1 (Abelson helper integration site 1), also known as ORF1, AHI-1, JBTS3 or Joubertin, is an 1,196 amino acid signaling protein that is expressed in the brain, specifically in neurons that give rise to the crossing axons of the corticospinal tract and superior cerebellar peduncles. AHI1 contains seven WD repeats, an SH3 domain and several SH3-binding sites, and is critical for both cerebellar and cortical development. Mutations of AHI1 is associated with Joubert syndrome (JS), an autosomal recessive disorder characterized by hypotonia, ataxia, mental retardation, altered respiratory pattern, abnormal eye movements, and a brain malformation. Considered a novel oncogene, AHI1 is highly deregulated in chronic myeloid leukemia (CML). Three isoforms exist due to alternative splicing events.

REFERENCES

1. Dixon-Salazar, T., et al. 2004. Mutations in the AHI1 gene, encoding joubertin, cause Joubert syndrome with cortical polymicrogyria. *Am. J. Hum. Genet.* 75: 979-987.
2. Close, J., et al. 2004. Genome annotation of a 1.5 Mb region of human chromosome 6q23 encompassing a quantitative trait locus for fetal hemoglobin expression in adults. *BMC Genomics* 5: 33.
3. Parisi, M.A., et al. 2006. AHI1 mutations cause both retinal dystrophy and renal cystic disease in Joubert syndrome. *J. Med. Genet.* 43: 334-339.
4. Parisi, M.A., et al. 2007. Joubert syndrome (and related disorders) (OMIM 213300). *Eur. J. Hum. Genet.* 15: 511-521.
5. Kroes, H.Y., et al. 2008. DNA analysis of AHI1, NPHP1 and cyclin D1 in Joubert syndrome patients from the Netherlands. *Eur. J. Med. Genet.* 51: 24-34.

CHROMOSOMAL LOCATION

Genetic locus: AHI1 (human) mapping to 6q23.3.

PRODUCT

AHI1 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 AHI1 shRNA Plasmid (h): sc-72465-SH and AHI1 shRNA (h) Lentiviral Particles: sc-72465-V as alternate gene silencing products.

For independent verification of AHI1 (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-72465A, sc-72465B and sc-72465C.

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

AHI1 siRNA (h) is recommended for the inhibition of AHI1 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.

GENE EXPRESSION MONITORING

AHI1 (B-2): sc-515382 is recommended as a control antibody for monitoring of AHI1 gene expression knockdown by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000) or immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500).

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgG κ BP-HRP: sc-516102 or m-IgG κ 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) Immunofluorescence: use m-IgG κ BP-FITC: sc-516140 or m-IgG κ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor AHI1 gene expression knockdown using RT-PCR Primer: AHI1 (h)-PR: sc-72465-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

SELECT PRODUCT CITATIONS

1. Bozzuto, G. and Molinari, A. 2017. The product of the human AHI1 (Abelson helper integration site) gene: experimental *in vitro* data point to its involvement in tumor cell invasion. *Ann. Ist. Super. Sanita* 53: 17-24.

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.