SANTA CRUZ BIOTECHNOLOGY, INC.

BCS1L siRNA (h): sc-72637

BACKGROUND

Hepatic involvement is a common feature in childhood mitochondrial hepatopathies, particularly in the neonatal period. Respiratory chain disorders may present as neonatal acute liver failure, hepatic steatohepatitis, cholestasis, or cirrhosis with chronic liver failure of insidious onset. GRACILE (growth retardation, aminoaciduria, cholestasis, iron overload, lactacidosis, and early death) syndrome is a recessively inherited lethal disease characterized by fetal growth retardation, lactic acidosis, aminoaciduria, cholestasis, and abnormalities in iron metabolism. GRACILE syndrome is the result of mutations in BCS1L, a mitochondrial inner-membrane protein that acts as a chaperone necessary for the assembly of mitochondrial respiratory chain complex III. Mutations in BCS1L can also result in the Björnstad syndrome, an autosomal recessive disorder associated with sensorineural hearing loss and pili torti. All mutant BCS1L proteins disrupt the assembly of complex III, reduce the activity of the mitochondrial electron-transport chain and increase the production of reactive oxygen species. Clinical expression of the mutations is correlated with the production of reactive oxygen species.

REFERENCES

1. Visapää, I., et al. 2002. GRACILE syndrome, a lethal metabolic disorder with iron overload, is caused by a point mutation in BCS1L. Am. J. Hum. Genet. 71: 863-876.
2. Fellman, V. 2002. The GRACILE syndrome, a neonatal lethal metabolic disorder with iron overload. Blood Cells Mol. Dis. 29: 444-450.
3. Kotarsky, H., et al. 2007. BCS1L is expressed in critical regions for neural development during ontogenesis in mice. Gene Expr. Patterns 7: 266-273.
4. Lee, W.S., et al. 2007. Mitochondrial hepatopathies: advances in genetics and pathogenesis. Hepatology 45: 1555-1565.
5. Fernandez-Vizarra, E., et al. 2007. Impaired complex III assembly associated with BCS1L gene mutations in isolated mitochondrial encephalopathy. Hum. Mol. Genet. 16: 1241-1252.

CHROMOSOMAL LOCATION

Genetic locus: BCS1L (human) mapping to 2q35.

PRODUCT

BCS1L 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 µM solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see BCS1L shRNA Plasmid (h): sc-72637-SH and BCS1L shRNA (h) Lentiviral Particles: sc-72637-V as alternate gene silencing products.

For independent verification of BCS1L (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-72637A, sc-72637B and sc-72637C.

PROTOCOLS

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

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 µl of the RNAse-free water provided. Resuspension of the siRNA duplex in 330 µl of RNAse-free water makes a 10 µM solution in a 10 µM Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

BCS1L siRNA (h) is recommended for the inhibition of BCS1L 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 µM in 66 µl. 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

BCS1L (SS-M13): sc-134280 is recommended as a control antibody for monitoring of BCS1L 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-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) 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.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor BCS1L gene expression knockdown using RT-PCR Primer: BCS1L (h)-PR: sc-72637-PR (20 µl). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

RESEARCH USE

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