SANTA CRUZ BIOTECHNOLOGY, INC.

BCS1L (SS-M13): sc-134280

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. and Sokol, R.J. 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.
6. Hinson, J.T., et al. 2007. Missense mutations in the BCS1L gene as a cause of the Björnstad syndrome. N. Engl. J. Med. 356: 809-819.
7. Lee, W.S. and Sokol, R.J. 2007. Liver disease in mitochondrial disorders. Semin. Liver Dis. 27: 259-273.
8. Fellman, V., et al. 2008. Screening of BCS1L mutations in severe neonatal disorders suspicious for mitochondrial cause. J. Hum. Genet. 53: 554-558.
9. Blázquez, A., et al. 2009. Infantile mitochondrial encephalomyopathy with unusual phenotype caused by a novel BCS1L mutation in an isolated complex III-deficient patient. Neuromuscul. Disord. 19: 143-146.

CHROMOSOMAL LOCATION

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

SOURCE

BCS1L (SS-M13) is a mouse monoclonal antibody raised against recombinant BCS1L protein of human origin.

PRODUCT

Each vial contains 100 µg IgG_1 kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

BCS1L (SS-M13) is recommended for detection of BCS1L of 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)] and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for BCS1L siRNA (h): sc-72637, BCS1L shRNA Plasmid (h): sc-72637-SH and BCS1L shRNA (h) Lentiviral Particles: sc-72637-V.

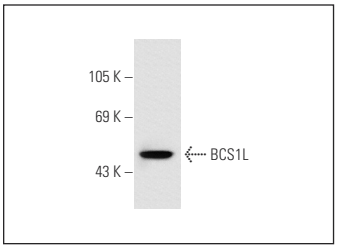
Molecular Weight of BCS1L: 48 kDa.

Positive Controls: HeLa whole cell lysate: sc-2200 or Jurkat whole cell lysate: sc-2204.

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).

DATA



BCS1L (SS-M13): sc-134280. Western blot analysis of BCS1L expression in HeLa 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.

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.