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Produktinformation

- Forschungsprodukte & Biochemikalien
- Zellkultur & Verbrauchsmaterial
- Diagnostik & molekulare Diagnostik
- Laborgeräte & Service

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See the following pages for more information!

Lieferung & Zahlungsart

siehe unsere Liefer- und Versandbedingungen

Zuschläge

- Mindermengenzuschlag
- Trockeneiszuschlag
- Gefahrgutzuschlag
- Expressversand

SZABO-SCANDIC HandelsgmbH

Quellenstraße 110, A-1100 Wien

T. +43(0)1 489 3961-0

F. +43(0)1 489 3961-7

mail@szabo-scandic.com

www.szabo-scandic.com

linkedin.com/company/szaboscandic

SANTA CRUZ BIOTECHNOLOGY, INC.

BCS1L (m): 293T Lysate: sc-118792

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.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

PROTOCOLS

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

CHROMOSOMAL LOCATION

Genetic locus: Bcs1l (mouse) mapping to 1 C3.

PRODUCT

BCS1L (m): 293T Lysate represents a lysate of mouse BCS1L transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

BCS1L (m): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive BCS1L antibodies. Recommended use: 10-20 µl per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

RESEARCH USE

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