

BSCL2 Antibody (monoclonal) (M01)

Mouse monoclonal antibody raised against a partial recombinant BSCL2. Catalog # AT1314a

Product Information

Application WB, E **Primary Accession Q96G97 Other Accession** NM 032667 Reactivity Human Host mouse Clonality monoclonal Isotype IgG1 Kappa **Clone Names** 1G4 Calculated MW 44392

Additional Information

Gene ID 26580

Other Names Seipin, Bernardinelli-Seip congenital lipodystrophy type 2 protein, BSCL2

Target/Specificity BSCL2 (NP_116056, 259 a.a. ~ 357 a.a) partial recombinant protein with GST

tag. MW of the GST tag alone is 26 KDa.

Dilution WB~~1:500~1000 E~~N/A

Format Clear, colorless solution in phosphate buffered saline, pH 7.2.

Storage Store at -20°C or lower. Aliquot to avoid repeated freezing and thawing.

Precautions BSCL2 Antibody (monoclonal) (M01) is for research use only and not for use in

diagnostic or therapeutic procedures.

Background

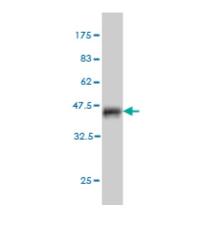
This gene encodes protein seipin, which is located in the endoplasmic reticulum and may be important for lipid droplet morphology. Mutations in this gene have been associated with congenital generalized lipodystrophy type 2 or Berardinelli-Seip syndrome, a rare autosomal recessive disease characterized by a near absence of adipose tissue and severe insulin resistance. Alternatively spliced transcript variants encoding different isoforms have been found for this gene.

References

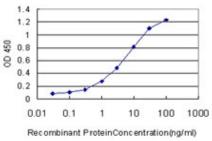
Complementary mutations in seipin gene in a patient with Berardinelli-Seip congenital lipodystrophy and dystonia: phenotype variability suggests multiple roles of seipin gene. Wu YR, et al. J Neurol Neurosurg

Psychiatry, 2009 Oct. PMID 19762912. The human lipodystrophy gene product Berardinelli-Seip congenital lipodystrophy 2/seipin plays a key role in adipocyte differentiation. Chen W, et al. Endocrinology, 2009 Oct. PMID 19574402. Two Japanese infants with congenital generalized lipodystrophy due to BSCL2 mutations. Nishiyama A, et al. Pediatr Int, 2009 Dec. PMID 19438831. A novel 16p locus associated with BSCL2 hereditary motor neuronopathy: a genetic modifier? Brusse E, et al. Neurogenetics, 2009 Oct. PMID 19396477. Clincial and pathological study of distal motor neuropathy with N88S mutation in BSCL2. Chen B, et al. Neuropathology, 2009 Oct. PMID 19323790.

Images



Antibody Reactive Against Recombinant Protein.Western Blot detection against Immunogen (36.63 KDa) .



Detection limit for recombinant GST tagged BSCL2 is approximately 0.1ng/ml as a capture antibody.

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.