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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

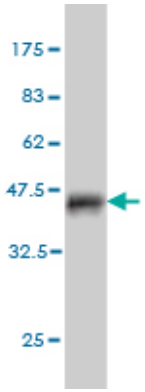
This gene encodes protein seipin, which is located in the endoplasmic reticulum and may be important for lipid droplet morphology. Mutations in this gene have been associated with congenital generalized lipodystrophy type 2 or Berardinelli-Seip syndrome, a rare autosomal recessive disease characterized 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

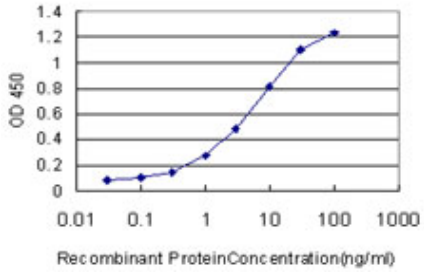
Complementary mutations in seipin gene in a patient with Berardinelli-Seip congenital lipodystrophy and dystonia: phenotype variability suggests multiple roles of seipin gene. Wu YR, et al. J Neurol Neurosurg

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.