Product name:	SACS Rabbit Polyclonal Antibody
Cat number:	ABN17571
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	Synthesized peptide derived from human protein . at AA range: 4291-4340
Reactivity:	Human,Mouse
Applications:	IHC 1:50-1:300,ICC/IF 1:50-1:200
Molecular Weight:	503kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

This gene encodes the sacsin protein, which includes a UbL domain at the N-terminus, a DnaJ domain, and a HEPN domain at the C-terminus. The gene is highly expressed in the central nervous system, also found in skin, skeletal muscles and at low levels in the pancreas. This gene includes a very large exon spanning more than 12.8 kb. Mutations in this gene result in autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS), a neurodegenerative disorder characterized by early-onset cerebellar ataxia with spasticity and peripheral neuropathy. The authors of a publication on the effects of siRNA-mediated sacsin knockdown concluded that sacsin protects against mutant ataxin-1 and suggest that "the large multi-domain sacsin protein is able to recruit Hsp70 chaperone action and has the potential to regulate the effects of other ataxia proteins" (Parfitt et al., PubMed: 19208651).disease:Defects in SACS are the cause of autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) [MIM:270550]. ARSACS is an early onset neurodegenerative disease with high prevalence in the Charlevoix-Saguenay-Lac-Saint-Jean region of Quebec. It is characterized by absent sensory-nerve conduction, reduced motor-nerve velocity and hypermyelination of retinal-nerve fibers.,function:May function in chaperone-mediated protein folding.,similarity:Contains 1 HEPN domain.,similarity:Contains 1 J domain.,tissue specificity:Highly expressed in the central nervous system. Also found in skeletal muscle and at low levels in pancreas.,