

Product datasheet for **SC304089**

Sacsin (SACS) (NM_014363) Human Untagged Clone

Product data:

Product Type:	Expression Plasmids
Product Name:	Sacsin (SACS) (NM_014363) Human Untagged Clone
Tag:	Tag Free
Symbol:	Sacsin
Synonyms:	ARSACS; DNAJC29; PPP1R138; SPAX6
Mammalian Cell Selection:	None
Vector:	<u>pCMV6-XL5</u>
E. coli Selection:	Ampicillin (100 ug/mL)

Fully Sequenced ORF: >OriGene sequence for NM_014363 edited
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3' Read Nucleotide Sequence:

>OriGene 3' read for NM_014363 unedited
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 CTCCTTACATGCCATATTGAAGAAAGGTGGGTTTTTTTTTAACTGCGCACTTTGGCCAC
 AAGATGGCTTCTGTCCACCACACTATAATTAACCTTGGGAAATGTCCACCAACCTAAGCA
 GTGTTACGTATAATGTGCTGGCAGCATTCAACCACACTGCAAGACTGCACATTGGTT
 CGTCTACTCTCACAGCCCTGACTAAGTTCAAATTTATAGTTAAGTGACTAGTGACTGAG
 ATCTCGACTTAGTCAGTATAGTATTCCATCATCCGTATCACTAATACATCTG

Restriction Sites:

Please inquire

ACCN:

NM_014363

Insert Size:

15000 bp

OTI Disclaimer: Due to the inherent nature of this plasmid, standard methods to replicate additional amounts of DNA in E. coli are highly likely to result in mutations and/or rearrangements. Therefore, OriGene does not guarantee the capability to replicate this plasmid DNA. Additional amounts of DNA can be purchased from OriGene with batch-specific, full-sequence verification at a reduced cost. Please contact our customer care team at custsupport@origene.com or by calling 301.340.3188 option 3 for pricing and delivery.

Our molecular clone sequence data has been matched to the reference identifier above as a point of reference. Note that the complete sequence of our molecular clones may differ from the sequence published for this corresponding reference, e.g., by representing an alternative RNA splicing form or single nucleotide polymorphism (SNP).

OTI Annotation: There is 7 nucleotide difference between the OriGene clone and the NCBI reference ORF. These result in the substitution of 1 aa and insertion of 1 aa.

Components: The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).

Reconstitution Method:

1. Centrifuge at 5,000xg for 5min.
2. Carefully open the tube and add 100ul of sterile water to dissolve the DNA.
3. Close the tube and incubate for 10 minutes at room temperature.
4. Briefly vortex the tube and then do a quick spin (less than 5000xg) to concentrate the liquid at the bottom.
5. Store the suspended plasmid at -20°C. The DNA is stable for at least one year from date of shipping when stored at -20°C.

RefSeq: [NM_014363.3](#), [NP_055178.2](#)

RefSeq Size: 14805 bp

RefSeq ORF: 13299 bp

Locus ID: 26278

UniProt ID: [Q9NZJ4](#)

Cytogenetics: 13q12.12

Protein Families: Druggable Genome

Gene Summary:

This gene encodes the saccin 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 saccin knockdown concluded that saccin protects against mutant ataxin-1 and suggest that "the large multi-domain saccin 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). A pseudogene associated with this gene is located on chromosome 11. Alternative splicing of this gene results in multiple transcript variants. [provided by RefSeq, May 2013]

Transcript Variant: This variant (1) represents the longer transcript and encodes the longer isoform (1). Sequence Note: This RefSeq record was created from transcript and genomic sequence data to make the sequence consistent with the reference genome assembly. The genomic coordinates used for the transcript record were based on alignment of partial transcripts and data in published reports.