

## **Product datasheet for SC310234**

# OriGene Technologies, Inc.

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### SEPN1 (SELENON) (NM\_020451) Human Untagged Clone

#### **Product data:**

**Product Type:** Expression Plasmids

Product Name: SEPN1 (SELENON) (NM\_020451) Human Untagged Clone

Symbol: SEPN1

Synonyms: CFTD; MDRS1; RSMD1; RSS; SELN; SEPN1

**Vector:** pCMV6 series

Fully Sequenced ORF: >NCBI ORF sequence for NM\_020451, the custom clone sequence may differ by one or more

nucleotides

CCCGCGCCACCGCCGCCGCCCGTTCCCTGGCGCTGCTCGGAGCCCTGCTGGCCGCC GCCGCTGCCGCCGTCCGGGTCTGCGCCCGCCACGCCGAGGCCCAGGCGGCCGCGCG CAGGAACTGGCGCTGAAGACCCTGGGGACAGATGGCCTTTTTCTCTTTTCCTCCTTGGAC ACTGACGGGGATATGTACATCAGCCCTGAGGAGTTCAAACCCATTGCTGAGAAGCTAACA GGGTCTTGTTCTGTCACCCAGACTGGAGTGCAGTGGTGCAGTCACAGCTCACTGCAGCCT CAACTTCCCTGGCTCAATTGATCCTCCTGCCTCAGCCTCCTGAGGTCAACTCCCGCGGCC AGCTGCGAGGAGGAGGAGTTGCCCCCTGACCCTAGCGAGGAGACGCTCACCATAGAAGCC CGATTCCAGCCTCTGCTCCCGGAGACCATGACCAAGAGCAAAGATGGCTTCCTAGGGGTC TCCCGCCTCGCCCTGTCCGGCCTCCGAAACTGGACAGCCGCCGCCTCACCAAGTGCAGTG TTTGCCACCCGCCACTTCCAGCCCTTCCTTCCCCCGCCAGGCCAGGAGCTGGGTGAGCCC TGGTGGATCATCCCCAGTGAGCTGAGCATGTTCACTGGCTACCTGTCCAACAACCGCTTC TATCCACCGCCGCCCAAGGGCAAGGAGGTCATCATCCACCGGCTCCTGAGCATGTTCCAC ATCAGCGACTTCTACTACACTGTGATGTTCCGGATCCATGCCGAGTTCCAGCTCAGTGAG CCGCCCGACTTCCCCTTTTGGTTCTCCCCTGCTCAGTTCACCGGCCACATCATCCTCTCC AAAGACGCCACCCACGTCCGCGACTTCCGGCTCTTCGTGCCCAACCACAGGTCTCTGAAT GTGGACATGGAGTGGCTTTACGGGGCCAGTGAAAGCAGCAACATGGAGGTGGACATCGGC TACATACCCCAGATGGAGCTGGAGGCCACGGGCCCCTCTGTGCCCTCCGTGATCCTGGAT GAGGATGGCAGCATGATCGACAGCCACCTGCCTTCAGGGGAGCCCCTGCAGTTTGTGTTT GAGGAGATCAAGTGGCAGCAGGAGCTGAGCTGGGAGGAGGCTGCCCGGCGCCTGGAGGTG GCCATGTACCCCTTCAAGAAGGTCTCCTACTTGCCGTTCACTGAGGCCTTCGACCGAGCC AAGGCTGAGAACAAGCTGGTGCACTCAATCCTGCTGTGGGGGGCCCTGGATGACCAGTCC TGCTGAGGTTCAGGGCGGACTCTCCGGGAGACTGTCCTGGAAAGTTCGCCCATCCTCACC CTGCTCAACGAGAGCTTCATCAGCACCTGGTCCCTGGTGAAGGAGCTGGAGGAACTGCAG AACAACCAGGAGAACTCGTCCCACCAGAAGCTGGCTGGCCTGCACCTGGAGAAGTACAGC TTCCCCGTGGAGATGATCATCTGCCTGCCCAATGGCACCGTGGTCCATCACATCAATGCC AACTACTTCTTGGACATCACCTCCGTGAAGCCCGAGGAAATCGAGAGCAATCTCTTCAGC TTCTCATCCACCTTTGAAGACCCGTCCACGGCCACCTACATGCAGTTCCTGAAGGAGGGA CTCCGGCGTGGCCTGCCCCTCCTCCAGCCCTAG

#### SEPN1 (SELENON) (NM\_020451) Human Untagged Clone - SC310234

**Restriction Sites:** Please inquire

**ACCN:** NM\_020451

**OTI Disclaimer:** 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). The expression of this clone is

not guaranteed due to the nature of selenoproteins.

**OTI Annotation:** This clone encodes a selenoprotein containing the rare amino acid selenocysteine (Sec). Sec is

encoded by UGA codon, which normally signals translational termination. Expression of this

clone is not guaranteed due to the nature of selenoproteins.

**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: <u>NM 020451.2</u>, <u>NP 065184.2</u>

 RefSeq Size:
 4357 bp

 Locus ID:
 57190

 UniProt ID:
 Q9NZV5

 Cytogenetics:
 1p36.11

**Protein Families:** Druggable Genome



#### **Gene Summary:**

This gene encodes a glycoprotein that is localized in the endoplasmic reticulum. It plays an important role in cell protection against oxidative stress, and in the regulation of redoxrelated calcium homeostasis. Mutations in this gene are associated with early onset muscle disorders, referred to as SEPN1-related myopathy. SEPN1-related myopathy consists of 4 autosomal recessive disorders, originally thought to be separate entities: rigid spine muscular dystrophy (RSMD1), the classical form of multiminicore disease, desmin related myopathy with Mallory-body like inclusions, and congenital fiber-type disproportion (CFTD). This protein is a selenoprotein, containing the rare amino acid selenocysteine (Sec). Sec is encoded by the UGA codon, which normally signals translation termination. The 3' UTRs of selenoprotein mRNAs contain a conserved stem-loop structure, designated the Sec insertion sequence (SECIS) element, that is necessary for the recognition of UGA as a Sec codon, rather than as a stop signal. A second stop-codon redefinition element (SRE) adjacent to the UGA codon has been identified in this gene (PMID:15791204). SRE is a phylogenetically conserved stem-loop structure that stimulates readthrough at the UGA codon, and augments the Sec insertion efficiency by SECIS. Alternatively spliced transcript variants have been found for this gene. [provided by RefSeq, Dec 2016]

Transcript Variant: This variant (2) contains an additional in-frame, Alu-derived coding exon in the 5' region compared to variant 1. The encoded isoform (2) is longer than isoform 1, containing two potential selenocysteine (Sec) residues. The first Sec found in the novel exon is not conserved, while the second Sec is highly conserved. Expression of isoform 2 was not detected in vivo or in transfection studies, leaving open the possibility that the UGA codon in the novel exon may be recognized as a stop codon, and not as a Sec codon (PMID:12700173).