

Product datasheet for SC328161

MCFD2 (NM 001171509) Human Untagged Clone

Product data:

Product Type: Expression Plasmids

Product Name: MCFD2 (NM_001171509) Human Untagged Clone

Tag: Tag Free
Symbol: MCFD2

Synonyms: F5F8D; F5F8D2; LMAN1IP; SDNSF

Mammalian Cell

Selection:

None

Vector: pCMV6-XL5

E. coli Selection: Ampicillin (100 ug/mL)

Fully Sequenced ORF: >NCBI ORF sequence for NM_001171509, the custom clone sequence may differ by one or

more nucleotides

GATGGATACATTGACTATGCTGAATTTGCAAAATCACTGCAGTAG

Restriction Sites: Please inquire **ACCN:** NM_001171509

Insert Size: 4041 bp

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).

OTI Annotation: This TrueClone is provided through our Custom Cloning Process that includes sub-cloning

into OriGene's pCMV6 vector and full sequencing to provide a non-variant match to the expected reference without frameshifts, and is delivered as lyophilized plasmid DNA.

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).



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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 001171509.1</u>, <u>NP 001164980.1</u>

RefSeq Size: 4041 bp
RefSeq ORF: 4041 bp
Locus ID: 90411
UniProt ID: Q8NI22
Cytogenetics: 2p21

Gene Summary:

This gene encodes a soluble luminal protein with two calmodulin-like EF-hand motifs at its C-terminus. This protein forms a complex with LMAN1 (lectin mannose binding protein 1; also known as ERGIC-53) that facilitates the transport of coagulation factors V (FV) and VIII (FVIII) from the endoplasmic reticulum to the Golgi apparatus via an endoplasmic reticulum Golgi intermediate compartment (ERGIC). Mutations in this gene cause combined deficiency of FV and FVIII (F5F8D); a rare autosomal recessive bleeding disorder characterized by mild to moderate bleeding and coordinate reduction in plasma FV and FVIII levels. This protein has also been shown to maintain stem cell potential in adult central nervous system and is a marker for testicular germ cell tumors. The 3' UTR of this gene contains a transposon-like human repeat element named 'THE 1'. A processed RNA pseudogene of this gene is on chromosome 6p22.1. Alternative splicing results in multiple transcript variants encoding distinct isoforms. [provided by RefSeq, Apr 2016]

Transcript Variant: This variant (5) lacks the first coding exon, compared to variant 1. This difference results in the use of an in-frame downstream start codon. The encoded protein (isoform B) has a shorter N-terminus, compared to isoform A, that is not predicted to have a signal peptide. Variants 5 and 6 encode the same isoform (B).