

## Recombinant Human MCFD2, T7 -tagged

Cat. No. MCFD2-29454TH Lot. No. (See product label)

### SPECIFICATION

**Product Overview** Recombinant full length Human MCFD2 with N terminal T7 tag; 136 amino acids, MWt 15.1 kDa.

**Species** Human

**Source** E.coli

**Description**

This gene encodes a soluble luminal protein with two calmodulin-like EF-hand motifs at its C-terminus. This protein forms a complex with LAMN1 (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.

**Conjugation** T7

**Form** Liquid

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<b>Purity</b>	>90% by SDS-PAGE
<b>Storage buffer</b>	Preservative: None Constituents: 10% Glycerol, 20mM Tris HCl, 100mM Sodium chloride, pH 7.5
<b>Storage</b>	Store at +4°C short term (1-2 weeks). Aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.
<b>Sequences of amino acids</b>	MASMTGGQQM GRGSHMEEPA ASFSQPGSMG LDKNTVHDQE HIMEHLEGVI NKPEAEMSPQ ELQLHYFKMH DYDGNLLDG LELSTAITHV HKEEGSEQAP LMSEDELINI IDGVLRDDDK NNDGYIDYAE FAKSLQ
<b>Sequence Similarities</b>	Contains 2 EF-hand domains.
<b>Full Length</b>	Full L.

## GENE INFORMATION

<b>Gene Name</b>	<a href="#">MCFD2 multiple coagulation factor deficiency 2 [ Homo sapiens ]</a>
<b>Official Symbol</b>	<a href="#">MCFD2</a>
<b>Synonyms</b>	MCFD2; multiple coagulation factor deficiency 2; multiple coagulation factor deficiency protein 2; F5F8D; LMAN1IP; SDNSF;
<b>Gene ID</b>	<a href="#">90411</a>
<b>mRNA Refseq</b>	<a href="#">NM_001171506</a>
<b>Protein Refseq</b>	<a href="#">NP_001164977</a>

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<b>MIM</b>	607788
<b>Uniprot ID</b>	Q8NI22
<b>Chromosome Location</b>	2p21
<b>Pathway</b>	Asparagine N-linked glycosylation, organism-specific biosystem; Metabolism of proteins, organism-specific biosystem; Post-translational protein modification, organism-specific biosystem; Transport to the Golgi and subsequent modification, organism-specific biosystem;
<b>Function</b>	calcium ion binding;

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