Recombinant Human MCFD2, T7 -tagged

Cat. No. MCFD2-28037TH **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant full length mature Human MCFD2 with N terminal T7 Tag; 136aa, 20.9kDa.
Species	Human
Source	E.coli
ProteinLength	120 amino acids
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

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Molecular Weight	20.900kDa inclusive of tags
Form	Liquid
Purity	>90% by SDS-PAGE
Storage buffer	pH: 7.50Constituents:0.32% Tris HCl, 0.58% Sodium chloride, 10% Glycerol
Storage	Store at +4°C short term (1-2 weeks). Aliquot and store at -20°C long term. Avoid repeated freeze / thaw cycles.
Sequences of amino acids	MASMTGGQQM GRGSHMEEPA ASFSQPGSMG LDKNTVHDQE HIMEHLEGVI NKPEAEMSPQ ELQLHYFKMH DYDGNNLLDG LELSTAITHV HKEEGSEQAP LMSEDELINI IDGVLRDDDK NNDGYIDYAE FAKSLQ
Sequence Similarities	Contains 2 EF-hand domains.

GENE INFORMATION

Gene Name	MCFD2 multiple coagulation factor deficiency 2 [Homo sapiens]
Official Symbol	MCFD2
Synonyms	MCFD2; multiple coagulation factor deficiency 2; multiple coagulation factor deficiency protein 2; F5F8D; LMAN1IP; SDNSF;
Gene ID	90411
mRNA Refseq	NM_001171506
Protein Refseq	NP_001164977

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MIM	607788
Uniprot ID	Q8NI22
Chromosome Location	2p21
Pathway	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;
Function	calcium ion binding;

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