

Product datasheet for **AR09341PU-L**

MCFD2 (27-146, T7-tag) Human Protein

Product data:

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| Product Type: | Recombinant Proteins |
| Description: | MCFD2 (27-146, T7-tag) human recombinant protein, 0.5 mg |
| Species: | Human |
| Expression Host: | E. coli |
| Expression cDNA Clone or AA Sequence: | <u>MASMTGGQOM</u> <u>GRGSHMEEPA</u> ASFSQPGSMG LDKNTVHDQE HIMEHLEGLVI NKPEAEMSPQ ELQLHYFKMH DYDGNLLDG LELSTAITHV HKEEGSEQAP LMSEDELINI IDGVLRRDDDK NNDGYIDYAE FAKSLQ |
| Tag: | T7-tag |
| Concentration: | lot specific |
| Purity: | >90% by SDS - PAGE |
| Buffer: | Presentation State: Purified State: Liquid purified protein Buffer System: 20 mM Tris-HCl buffer (pH 7.5) containing 100 mM NaCl, 10% glycerol |
| Preparation: | Liquid purified protein |
| Protein Description: | Recombinant human MCFD2, fused to T7-tag at N-terminus, was expressed in E.coli and purified by using conventional chromatography techniques. |
| Storage: | Store undiluted at 2-8°C for up to two weeks or (in aliquots) at -20°C or -70°C for longer. Avoid repeated freezing and thawing. |
| Stability: | Shelf life: one year from despatch. |
| RefSeq: | <u>NP_001164977</u> |
| Locus ID: | 90411 |
| UniProt ID: | <u>Q8NI22</u> |
| Cytogenetics: | 2p21 |
| Synonyms: | F5F8D; F5F8D2; LMAN1IP; SDNSF |



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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]

Product images: