

## MCFD2 抗原（重组蛋白）

中文名称：MCFD2 抗原（重组蛋白）

英文名称：MCFD2 Antigen (Recombinant Protein)

别名：F5F8D; SDNSF; F5F8D2; LMAN1IP

储存：冷冻（-20℃）

相关类别：抗原

概述：

Fusion protein corresponding to a region derived from 27-146 amino acids of human MCFD2

技术规格：

<b>Full name:</b>	multiple coagulation factor deficiency 2
<b>Synonyms:</b>	F5F8D; SDNSF; F5F8D2; LMAN1IP
<b>Swissprot:</b>	Q8NI22
<b>Gene Accession:</b>	BC040357
<b>Purity:</b>	>85%, as determined by Coomassie blue stained SDS-PAGE
<b>Expression system:</b>	Escherichia coli
<b>Tags:</b>	His tag C-Terminus, GST tag N-Terminus
<b>Background:</b>	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 a

utosomal 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.