Recombinant Human MCFD2

Catalog No: #GP13233

Package Size: #GP13233-1 100ug

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Description

Product Name	Recombinant Human MCFD2
Brief Description	Recombinant Protein
Immunogen Description	Fusion protein corresponding to a region derived from 27-146 amino acids of human MCFD2
Target Name	multiple coagulation factor deficiency 2
Other Names	F5F8D; SDNSF; F5F8D2; LMAN1IP
Accession No.	Swissprot:Q8NI22Gene Accession:BC040357
Uniprot	Q8NI22
GeneID	90411;
Storage	-20~-80°C, pH 7.6 PBS

Background

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.

References

Note: For in vitro research use only, not for diagnostic or therapeutic use. This product is not a medical device.

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