

## Disease

# MCFD2 associated combined factor V and VIII deficiency<sup>1, 2, 3</sup>

## Gene

### MCFD2

|                                                 |                                                                                                                                                                                                                           |
|-------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                        |
| <b>Current Treatments</b>                       | Fresh-frozen plasma (FFP), DDAVP                                                                                                                                                                                          |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                            |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                       |
| <b>Non-Molecular Test</b>                       | factor XIII and factor V activity <sup>2</sup>                                                                                                                                                                            |
| <b>Disease Frequency (per 100,000)</b>          | 0.05 <sup>2</sup>                                                                                                                                                                                                         |
| <b>Genes Included in Disease Frequency</b>      | LMAN1 and MCFD2                                                                                                                                                                                                           |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=MCFD2&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=MCFD2&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

## References

<sup>1</sup> <https://omim.org/entry/613625>

<sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/25100430/>

<sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/31558466/>