

Disease

LMAN1 associated combined factor V and VIII deficiency^{1, 2, 3}

Gene

LMAN1

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

References

¹ <https://omim.org/entry/227300>

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

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