MAYO CLINIC LABORATORIES

1-800-533-1710

F81B

Hemophilia A F8 Gene, Intron 1 Inversion Known Mutation, Whole Blood

Patient ID	Patient Name		Birth Date	Gender	Age
SA00141424	TESTINGRNV, VALIDATION		1999-09-09	M	21
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00141424	SA00141424	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		12 Jan 2021 07:00			

HA F8 Intron 1 Inversion KM, B

F81B Interpretation

[1] MCR

These results are consistent with a diagnosis of hemophilia A. This mutation has been identified in patients with severe hemophilia A. Since we have documented the presence of a mutation within the F8 gene in an affected family member, carrier testing for at risk individuals is possible.

A genetic consultation may be of benefit.

MCC Not Tested: The presence of maternal cell contamination (MCC) cannot be excluded because a peripheral blood specimen from the mother was not available for testing. If present, MCC could change the outcome of these test results.

ADDITIONAL INFORMATION

Genomic DNA is amplified by PCR with primers specific for the F8 intron 1 inversion mutation.

HA F8 Intron 1 Inversion KM, B

MCR

Positive

Reference Value
Not applicable

An intron 1 inversion was detected within the F8 gene.

HA F8 Int1 KM Reason for Referral

MCR

A F8 gene mutation was previously identified in an affected family member. Test for the presence of the familial mutation in the F8 gene.

HA F8 Int1 KM Reviewed By

MCR

BENJAMIN BAILEY

Received: 13 Jan 2021 11:33 **Reported:** 13 Jan 2021 15:26

Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292