



Hemophilia A F8 Gene, Intron 22 Inversion Known
Mutation, Whole Blood

Patient ID SA00141425	Patient Name TESTINGRNA, VALIDATION	Birth Date 1999-09-09	Gender M	Age 21
Order Number SA00141425	Client Order Number SA00141425	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 12 Jan 2021 07:00		

HA F8 Intron 22 Inversion KM, B

F822B Interpretation

1 MCR

Hemophilia A due to an intron 22 inversion within the F8 gene has previously been documented in this family.

Since the patient does NOT have the familial mutation, these results are consistent with the patient NOT being affected with Hemophilia A.

Since testing was targeted towards the familial mutation, mutations in other regions of F8 cannot be excluded. Errors in the diagnosis or pedigree provided to us, including non paternity, may lead to an erroneous interpretation of test results.

A genetic consultation may be of benefit.

MCC Negative: Using a panel of polymorphic markers to compare maternal and fetal specimens, there was no evidence of maternal cell contamination in the fetal specimen.

ADDITIONAL INFORMATION

Genomic DNA is digested with Ksp22 I restriction enzyme, ligated with T4 DNA ligase, and amplified by PCR with primers specific for the F8 intron 22 inversion mutations.

HA F8 Int22 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 Intron 22 Inversion KM, B

MCR

Negative

Reference Value
Not applicable

An intron 22 inversion was NOT detected within the F8 gene.

HA F8 Int22 KM Reviewed By

MCR

BENJAMIN BAILEY

Received: 13 Jan 2021 11:33

Reported: 13 Jan 2021 15:27

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