Hemophilia Carrier Testing Algorithm

Diagnosis of hemophilia A suspected or confirmed in a family member

Yes

F8 gene mutation previously identified in family?

Yes

If known mutation is an Intron 1 Inversion mutation, order F81B / Hemophilia A F8 Gene, Intron 1 Inversion Known Mutation, Whole Blood**

If known mutation is an Intron 22 inversion, order F822B / Hemophilia A F8 Gene, Intron 22 Inversion Known Mutation, Whole Blood**

If known mutation is a point mutation or deletion/duplication, contact a Laboratory Genetic Counselor to discuss send-out test.

No

Determine clinical severity of affected family member

Unknown

F8INV / Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood

Inversion found

Inversion not found

Moderate/Mild hemophilia: Factor VIII activity 1% to 55%

Contact a Laboratory Genetic Counselor to discuss send-out for full sequencing and possible large deletion/duplication analysis of F8 gene*

Diagnosis of hemophilia B suspected or confirmed in a family member

Yes

F9 gene mutation previously identified in family?

Yes

FIXKM / Hemophilia B, F9 Gene Known Mutation, Whole Blood**

No

Mutation not found

Contact a Laboratory Genetic Counselor to discuss send-out for large deletion/duplication analysis of F9 gene

Mutation found

FIXMS / Hemophilia B, F9 Gene Mutation Analysis, Whole Blood

* If all testing is normal, consider possibility that family member has alternate bleeding disorder

**Send copy of known familial mutation with sample for testing to be performed

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Reviewed 09/2018