Hemophilia Testing Algorithm

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

Is activity still decreased?

Hemophilia A diagnosis

Activity decreased?

F8 genetic testing has been performed on a family member and the specific mutation is known?

If the activity assays are normal, consider an alternate bleeding disorder: BDIAL / Bleeding Diathesis Profile, Limited

Severe hemophilia: activity <1%

Moderate/Mild hemophilia: activity 1% to 55%

Inversion found

Inversion not found

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

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

Vitamin K antagonist (ie, warfarin)

Child/adolescent?

Retest when >4 weeks after antagonist treatment is complete

Adjust decreased activity levels for age

Hemophilia B diagnosis

Activity decreased?

F9 genetic testing has been performed on a family member and the specific mutation is known?

Mutation not found

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

Mutation found

Severe hemophilia:

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

Inversion found

Inversion not found

Severe hemophilia: activity <1%

Moderate/Mild hemophilia: 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

*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

Reviewed 09/2018