Hemophilia Testing Algorithm

Symptomatic male

Activity decreased?

Hemophilia A diagnosis

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

YES

Severe hemophilia: activity <1%

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

Inversion found

Mutation found

Contact a Laboratory Genetic Counselor to discuss send-out testing for large deletion/duplication analysis of F8 gene

Inversion not found

Mutation not found

Moderate/mild hemophilia: activity 1% to 55%

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

F8NGS / Hemophilia A, F8 Gene, Next-Generation Sequencing, Varies

Mutation found

Mutation not found

Contact a Laboratory Genetic Counselor to discuss send-out testing for large deletion/duplication analysis of F8 gene

Activity decreased?

Hemophilia B diagnosis

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

YES

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

Activity decreased?

Vitamin K antagonist (ie, warfarin)

Child/adolescent?

YES

Retest when >4 weeks after antagonist treatment is complete

Adjust decreased activity levels for age

NO

Is activity still decreased?

YES

Hemophilia B diagnosis

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

YES

NO

Mutation found

Mutation not found

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

Mutation not found

Contact a Laboratory Genetic Counselor to discuss possibility that family member has alternate bleeding disorder