Hemophilia Testing Algorithm

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

**Known mutation must be provided for testing to be performed**

Is activity still decreased?

Activity decreased?

Hemophilia A diagnosis

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

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 the Coagulation DNA lab to discuss send-out test.

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 Coagulation DNA Lab to discuss full sequencing of F8 gene*

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Hemophilia B diagnosis

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

YES

Is activity still decreased?

NO

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

YES

Hemophilia B diagnosis

NO

Symptomatic male

Vitamin K antagonist (ie, warfarin)

Child/adolescent?

YES

Re-test when >4 weeks after antagonist treatment is complete

Adjust decreased activity levels for age

NO

Yes

No

Severe hemophilia: activity <1%

Moderate/Mild hemophilia: activity 1% to 55%

BDIAL / Bleeding Diathesis Profile, Limited

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**Known mutation must be provided for testing to be performed**