Hemophilia Testing Algorithm

Symptomatic male

Hemophilia A
FBA / Coagulation Factor VIII Activity Assay, Plasma

Activity decreased?

Hemophilia A diagnosis

YES

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

NO

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

NO

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.

Inversion found

Inversion not found

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

Severe hemophilia: activity <1%

Moderate/Mild hemophilia: activity 1% to 55%

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

Mutation not found

Mutation found

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

Severe hemophilia: activity <1%

YES

Hemophilia B diagnosis

Activity decreased?

NO

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

IF YES

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, F9 / Coagulation Factor IX Activity Assay, Plasma

 Activity decreased?

YES

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

NO

Mutation not found

Mutation found

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

Contact a Laboratory Genetic Counselor to discuss send-out for full sequencing and possible large deletion/duplication analysis of F9 gene*

Severe hemophilia: activity <1%

Moderate/Mild hemophilia: activity 1% to 55%

IF YES

Participate in Consortium/Registry

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

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

Reviewed 09/2018