Hemophilia Testing Algorithm

MAYO CLINIC LABORATORIES

Symptomatic male

Hemophilia A branch

Hemophilia A
F8A / Coagulation Factor VIII Activity Assay, Plasma

Activity decreased?

YES → **Hemophilia A diagnosis**

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

YES:
- If known variant is an Intron 1 Inversion variant, order F81B / Hemophilia A *F8* Gene, Intron 1 Inversion Known Mutation, Whole Blood*
- If known variant is an Intron 22 inversion, order F822B / Hemophilia A *F8* Gene, Intron 22 Inversion Known Mutation, Whole Blood*
- If known variant is a point variant or deletion/duplication, contact a Laboratory Genetic Counselor to discuss targeted familial variant testing*

NO:
- Severe hemophilia: activity <1%
  → F8INV / Hemophilia A *F8* Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood
  → Inversion found / Inversion not found
- Moderate/mild hemophilia: activity 1% to 55%

GNHMA / Hemophilia A, *F8* Gene, Next-Generation Sequencing, Varies

Variant found / Variant not found

Final test report from our lab will indicate options for further large deletion/duplication analysis.**

NO (Activity decreased?):
If the activity assays are normal, consider an alternate bleeding disorder: ALBLD / Bleeding Diathesis Profile, Limited, Plasma

Hemophilia B branch

Hemophilia B
F_9 / Coagulation Factor IX Activity Assay, Plasma

Activity decreased?

YES:
- Vitamin K antagonist (ie, warfarin)
- Child/adolescent?

YES:
- Retest when >4 weeks after antagonist treatment is complete
- Adjust decreased activity levels for age

Is activity still decreased?
- **NO** → If the activity assays are normal, consider an alternate bleeding disorder: ALBLD / Bleeding Diathesis Profile, Limited, Plasma
- **YES** → Hemophilia B diagnosis

NO → **Hemophilia B diagnosis**

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

YES:
Final test report from our lab will indicate options for further large deletion/duplication analysis.**

NO:
GNHMB / Hemophilia B, *F9* Gene, Next-Generation Sequencing, Varies

Variant found / Variant not found

Final test report from our lab will indicate options for further large deletion/duplication analysis.**

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*Send copy of known familial variant with sample for testing to be performed
**Contact a Laboratory Genetic Counselor for additional information or to discuss additional testing options. If all testing is normal, consider the possibility that a family member has an alternate bleeding disorder.

Clickable PDF
Click on blue tests to view test catalog information

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