Hemophilia Testing Algorithm

**Hemophilia A**
F8A / Coagulation Factor VIII Activity Assay, Plasma

**Activity decreased?**

- **Yes**
  - Hemophilia A diagnosis

- **No**
  - F8 genetic testing has been performed on a family member and the specific mutation is known?
    - **Yes**
      - If known mutation is an Intron 1 Inversion mutation, order F8INV / Hemophilia A F8 Gene, Intron 1 Inversion Mutation Analysis, Whole Blood
    - **No**
      - F8INV / Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood
        - 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

**Hemophilia B**
F9 / 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
      - **No**
        - Is activity still decreased?
          - **Yes**
            - Hemophilia B diagnosis
          - **No**
            - F9 genetic testing has been performed on a family member and the specific mutation is known?
              - **Yes**
                - FIXKM / Hemophilia B, F9 Gene Known Mutation, Whole Blood
              - **No**
                - FIXMS / Hemophilia B, F9 Gene Mutation Analysis, Whole Blood
                  - Mutation found
                  - Mutation not found
                    - Contact a Laboratory Genetic Counselor to discuss send-out for large deletion/duplication analysis of F9 gene

**Symptomatic male**

- Activity decreased?
  - **Yes**
    - Hemophilia A diagnosis
  - **No**
  - If the activity assays are normal, consider an alternate bleeding disorder: BDIAL / Bleeding Diathesis Profile, Limited

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

- **Yes**
  - If known mutation is an Intron 1 Inversion mutation, order F8INV / Hemophilia A F8 Gene, Intron 1 Inversion Known Mutation, Whole Blood
- **No**
  - If known mutation is a point mutation or deletion/duplication, contact a Laboratory Genetic Counselor to discuss send-out test.

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

- **Yes**
  - FIXMS / Hemophilia B, F9 Gene Mutation Analysis, Whole Blood
- **No**

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*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

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