Hemophilia Testing Algorithm

**Hemophilia A**
- **F8A / Coagulation Factor VIII Activity Assay, Plasma**
  - Activity decreased?
    - **YES**
      - Hemophilia A diagnosis
    - **NO**
      - Moderate/mild hemophilia: activity 1% to 55%

**Hemophilia B**
- **F9 / Coagulation Factor IX Activity Assay, Plasma**
  - Activity decreased?
    - **YES**
      - Vitamin K antagonist (ie, warfarin)
      - Child/adolescent?
        - **YES**
          - Is activity still decreased?
            - **YES**
              - Hemophilia B diagnosis
            - **NO**
              - Retest when >4 weeks after antagonist treatment is complete
              - Adjust decreased activity levels for age
        - **NO**
          - **YES**
            - Hemophilia B diagnosis
    - **NO**
      - If the activity assays are normal, consider an alternate bleeding disorder: ALBLD / Bleeding Diathesis Profile, Limited, Plasma

**Hemophilia A**
- **F8A / Coagulation Factor VIII Activity Assay, Plasma**
  - Activity decreased?
    - **YES**
      - Hemophilia A diagnosis
    - **NO**
      - Severe hemophilia: activity <1%

**F8 genetic testing has been performed on a family member and the specific mutation is known?**
- **YES**
  - F8INV / Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood
    - Inversion found
    - Inversion not found

- **NO**
  - If known mutation is an Intron 1 Inversion mutation, order F8INV / Hemophilia A F8 Gene, Intron 1 Inversion Known Mutation, Whole Blood*
  - If known mutation is a point mutation or deletion/duplication, contact a Laboratory Genetic Counselor to discuss targeted familial mutation testing
  - F8NGS / Hemophilia A, F8 Gene, Next-Generation Sequencing, Varies
    - Mutation found
    - Mutation not found

**NGSF9 / Hemophilia B, F9 Gene, Next-Generation Sequencing, Varies**
- Mutation not found
- Mutation found

**FIXKM / Hemophilia B, F9 Gene Known Mutation, Whole Blood**
- If known mutation is a point mutation or deletion/duplication, contact a Laboratory Genetic Counselor to discuss targeted familial mutation testing

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

**NGSF9 / Hemophilia B, F9 Gene, Next-Generation Sequencing, Varies**
- Mutation not found
- Mutation found

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

*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**