Diagnosis of hemophilia A suspected or confirmed in a family member

**F8 gene mutation previously identified in family?**

- **YES**
  - 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 a Laboratory Genetic Counselor to discuss targeted familial mutation testing.

- **NO**
  - Determine clinical severity of affected family member
    - Unknown
      - Severe hemophilia: Factor VIII activity <1%
        - F8INV / Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood
          - Inversion found
          - Inversion not found
      - Moderate/mild hemophilia: Factor VIII activity 1% to 55%
    - Fixation not found
      - NGSM / Hemophilia B, F9 Gene Known Mutation, Whole Blood*
      - Mutation found
      - Mutation not found

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

Diagnosis of hemophilia B suspected or confirmed in a family member

**F9 gene mutation previously identified in family?**

- **YES**
  - FIXKM / Hemophilia B, F9 Gene Known Mutation, Whole Blood*

- **NO**
  - Determine clinical severity of affected family member
    - Unknown
      - Severe hemophilia: Factor VIII activity <1%
        - F8INV / Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood
          - Inversion found
          - Inversion not found
      - Moderate/mild hemophilia: Factor VIII activity 1% to 55%
    - Fixation not found
      - NGSM / Hemophilia B, F9 Gene Known Mutation, Whole Blood*
      - Mutation found
      - Mutation not 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**