Hemophilia Carrier Testing Algorithm

1. **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 send-out test.
     - **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**
   - **NO**
     - **Mutation found**
       - FIXMS / Hemophilia B, F9 Gene Mutation Analysis, Whole Blood
     - **Mutation not found**
       - Contact a Laboratory Genetic Counselor to discuss send-out for large deletion/duplication analysis of F9 gene

2. **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**
       - Contact a Laboratory Genetic Counselor to discuss send-out for full sequencing and possible large deletion/duplication analysis of F8 gene

* 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