Hemophilia Carrier Testing Algorithm

1. **Diagnosis of hemophilia A suspected or confirmed in a family member**
   - **F8 gene variant previously identified in family?**
     - **YES**
       - If known variant is an Intron 1 Inversion variant, order F8INV / Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood*
       - If known variant is an Intron 22 inversion, order F8INV / 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**
       - Determine clinical severity of affected family member
       - **Unknown**
         - **Severe hemophilia: Factor VIII activity <1%**
         - **Moderate/mild hemophilia: Factor VIII activity 1% to 55%**
       - **F8INV / Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood**
       - **Inversion found**
       - **Inversion not found**

2. **Diagnosis of hemophilia B suspected or confirmed in a family member**
   - **F9 gene variant previously identified in family?**
     - **YES**
       - If known variant is a point variant or deletion/duplication, contact a Laboratory Genetic Counselor to discuss targeted familial variant testing.*
     - **NO**
       - NGSF9 / Hemophilia B, F9 Gene, Next-Generation Sequencing, Varies
       - **Variant found**
       - **Variant not found**

* 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.