Hemophilia Carrier Testing Algorithm

- **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
        - **F8INV / Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood**
          - Inversion found
          - Inversion not found
        - Severe hemophilia: Factor VIII activity <1%
        - Moderate/Mild hemophilia: Factor VIII activity 1% to 55%

- **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**
      - FIXMS / Hemophilia B, F9 Gene Mutation Analysis, Whole Blood

* 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

Reviewed 09/2018