Hemophilia Carrier Testing Algorithm

**Diagnosis of hemophilia A confirmed in a family member**

- **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 the Coagulation DNA lab to discuss send-out test.

- **NO**
  - Determine clinical severity of affected family member
    - Unknown
    - Severe hemophilia: Factor VIII activity <1%
    - Moderate/Mild hemophilia: Factor VIII activity 1% to 55%

  - Inversion found
    - F8INV / Hemophilia A F8 Gene, Intron 1 and 22 Inversion Mutation Analysis, Whole Blood
  - Inversion not found
    - Contact Coagulation DNA Lab to discuss full sequencing of F8 gene*

**Diagnosis of hemophilia B confirmed in a family member**

- **YES**
  - FIXKM / Hemophilia B, Factor IX Gene Known Mutation Screening**

- **NO**
  - FIXMS / Hemophilia B, Factor IX Gene Mutation Screening

* If all testing is normal, consider possibility that family member has alternate bleeding disorder
* Known mutation must be provided for testing to be performed

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