Puget Sound Blood Center
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Mutation Analysis for Bleeding Disorders

January 2015

Testing Options:

3250-05	DNA Factor VIII Inversion
3250-02	DNA Hemophilia A Mutation Evaluation
3250-10	DNA Hemophilia B Mutation Evaluation
3250-11	Genotype for known Hemophilia/VWD mutation
3250-08	DNA Von Willebrand Dis Type 2A/2B/2M
3250-09	DNA Von Willebrand Dis Type 2N

What to Order?

*For Hemophilia A patients (Factor VIII activity levels of **≤ 2%**):*

3250-05 DNA Factor VIII Inversion

We will screen for the Intron 22 inversion which accounts for 45% of severe hemophilia patients and the Intron 1 inversion (5% of the hemophilia patients have this mutation). If the patient is inversion "positive" then no further testing is necessary.

If an inversion is not found, then additional testing will be necessary.

3250-02 DNA Hemophilia A Mutation Evaluation

Note: Patient will be charged this second transaction code

For Hemophilia A patients (Factor VIII activity levels of **>2%**):
3250-02 DNA Hemophilia A Mutation Evaluation

For Hemophilia B (Factor IX):
3250-10 DNA Hemophilia B Mutation Evaluation

For carrier typing workup:

If the sample is from a mother of an affected patient whose factor VIII level is ≤2% or the severity of the Hemophilia A is unknown:

3250-05 DNA Factor VIII Inversion

We will screen for the Intron 22 inversion which accounts for 45% of severe hemophilia patients and the Intron 1 inversion (5% of the hemophilia patients have this mutation). If the potential carrier is inversion "positive" then no further testing is necessary.

The following test applies if an inversion is not found or if the factor VIII activity is >2%:

3250-02 DNA Hemophilia A Mutation Evaluation

Note: Patient will be charged this second transaction code if an inversion is not found.

For any family member that requests carrier typing for a known genotype:

3250-11 Genotype for Known Hemophilia / VWD mutation

Only indicated if the proband has been typed and the mutation is known.

Note:

Please always record the severity of the proband on the RFT. If it is not noted, we will treat the sample as a severe or "unknown" and will perform the inversion mutation which is not necessary if the patient is not severe.