Factor VIII (F8) Inversion Analysis

Versiti is pleased to offer a test for intron 22 and intron 1 Factor VIII (F8) gene inversions.

Hemophilia A is an X-linked inherited bleeding disorder caused by mutations in the F8 gene that encodes for coagulation factor VIII. The degree of plasma factor VIII deficiency correlates with both the clinical severity of disease and genetic findings. Severe hemophilia A is characterized by plasma factor VIII levels of under 1 IU/dl. Approximately 50% of severe hemophilia A cases are attributable to gene inversions in F8 introns 1 and 22. For severe hemophilia A patients, the presence or absence of F8 inversions is confirmed prior to DNA sequence analysis. For families in which the factor VIII (F8) gene inversion is present, detection of this mutation can be used for carrier testing and prenatal diagnosis for hemophilia A.

Indications for testing:

- Diagnosis of affected individuals
- Female carrier detection
- Prenatal diagnosis

Test method:

DNA based factor VIII gene analysis is carried out using PCR assays designed to detect the recombination events in intron 22 and intron 1 of the Factor VIII (F8) gene.

Assay sensitivity and limitations:

Rare inversion mutations may not be detected by these assays. Factor VIII (F8) gene mutations other than inversions are not detected by this assay. It is recommended that an affected family member be tested for the inversion mutation prior to prenatal or carrier testing.

Specimen requirements:

5 ml EDTA (lavender top) whole blood, 2 ml minimum, shipped at room temperature. A sample from an affected family member is highly recommended. Please inquire about specific requirements for the analysis of prenatal samples by calling 1-800-245-3117, ext. 6250.

SHIP

Shipping requirements:

Place the sample and completed test requisition form into plastic bags and seal. Place in an insulated container, then into a sturdy cardboard box, tape securely and ship by an overnight carrier. Ship the package in compliance with your overnight carrier guidelines. Notify the laboratory prior to shipping by calling 800-245-3117, ext. 6250.

Address package to:

Versiti Client Services
Molecular Diagnostics Laboratory
638 N. 18th Street
Milwaukee, WI 53233
800-245-3117, ext. 6250

ORDER

Required forms:

Please complete all pages of the requisition form.

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CPT Codes/Billing/Turnaround time:

Test code: 1402

CPT code: 81403

Turnaround time: 5-7 days

References:

1. Andrikovics H, Klein I, Bors A, Nemes L, Marosi A, Varadi A, Tordai A. Analysis of large structural changes of the factor VIII gene, involving intron 1 and 22, in severe hemophilia A. Haematologica. 2003;88: 778–84
2. Bagnall RD, Giannelli F, Green PM. Int22h-related inversions causing hemophilia A: a novel insight into their origin and a new more discriminant PCR test for their detection. J Thromb Haemost. 2006;4:591–8.
3. Bagnall RD, Waseem N, Green PM, Giannelli F. Recurrent inversion breaking intron 1 of the factor VIII gene is a frequent cause of severe hemophilia A. Blood. 2002;99:168–74
4. Pio, S, et al. Detection of int1h-related inversion of the factor VIII gene. Haemophilia (2010), 1–2

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