Factor V Leiden Genotyping Assay: Test Ordering Information

Test Information (v1_020112):
Factor V Leiden Genotyping Assay (CMGD
test code 4202)
CPT Codes 83891x1, 83892x2, 83900x1, 83901x5, 83834x1, 83912-Report & Interpretation
For additional information please refer to the CMGD website www.medgen.med.miami.edu.

Indications for Testing

Factor V Leiden testing is indicated and recommended if
- A patient tested positive by APC Protein Resistance. The molecular test helps to distinguish heterozygote from homozygote mutations;
- A patient has borderline APC resistance assay values. The molecular test helps to establish the genetic status;
- A patient has very low APC resistance values. The molecular test helps to differentiate heterozygote, homozygote, and pseudo-homozygote genotypes (See Pseudo-homozygous APC resistance);
- A first unprovoked VTE presented at any age (especially age is less than 50 years);
- A history of recurrent VTE is present;
- Venous thrombosis at unusual sites is present (e.g., cerebral, mesenteric, portal, and hepatic veins);
- VTE occurs during pregnancy or the puerperium;
- VTE is associated with use of oral contraceptives or hormone replacement therapy;
- A first VTE occurs in an individual with a first-degree family member with VTE before age 50 years.

Factor V Leiden testing may be considered in cases of:
- Women with unexplained fetal loss after ten weeks’ gestation;
- Women with unexplained severe preeclampsia or "HELLP" (hemolysis, elevated liver enzymes and low platelets), placental abruption, or a fetus with severe intrauterine growth restriction;
- Female smokers younger than age 50 years with a myocardial infarction or stroke;
- Individuals older than age 50 years with a first provoked VTE in the absence of malignancy or an intravascular device;
- Asymptomatic adult family members of probands with a known factor V Leiden mutation, especially those with a strong family history of VTE at a young age;
- Asymptomatic female family members of probands with known factor V Leiden thrombophilia who are pregnant or are considering oral contraceptive use or pregnancy;
- Women with recurrent unexplained first-trimester pregnancy losses with or without second- or third-trimester pregnancy losses;
- Neonates and children with non-catheter related idiopathic VTE or stroke;
- Individuals younger than age 50 years with unexplained arterial thrombosis;

Factor V Leiden testing is not recommended as:
- General population screening or routine test;
- Prenatal or newborn testing;
- Prenatal testing is not performed by the CMGD.

Specimen Requirements
2.0 ml of whole blood (minimum)
Collection Tube: Lavender Top Tube /w EDTA
Handling: Room Temp- specimen processed within 72 hours

Turn-Around Times
3 to 5 business days.

Methodology and Assay Characteristics
This test is performed using the eSensor® Technology, a solid-phase electrochemical method for determining the genotyping status of a defined panel of mutations.
This test is performed by the CMGD according to the eSensor Thrombophilia Risk product insert. The eSensor Thrombophilia Risk Test is an in vitro diagnostic for the detection and genotyping of Factor II (Prothrombin, 20210 G>A), Factor V (Leiden, 1691 G>A) and MTHFR (5, 10 methyltetrahydrofolate reductase, 677C>T and 1298 A>C) gene mutations.

Alleles Detected
This test is validated to detect the F5 gene 1691G>A R506Q mutation, also described as Factor V Leiden mutation (standard nomenclature of the mutation: c.1601G>A, p.Arg534Gln; NM_000130.4).

Analytical Sensitivity and Specificity
99 percent of analytical sensitivity and specificity.

Test Limitations
- This test is validated to detect the F5 gene 1691G>A R506Q mutation. Additional allelic variants in the F5 gene or other genes will not be detected;
- We cannot exclude the possibility of rare mutations located in close proximity to the mutation tested that
could interfere with the genotyping results and be the cause of an incorrect result;
- Results of this test should be interpreted in the context of clinical presentation and in consultation with a licensed physician and/or geneticist;
- Genetic testing using the methods applied by the CMGDL is expected to be highly accurate. However, the chance of a false positive or a false negative result, due to laboratory errors incurred during any phase of the testing, cannot be completely excluded.

Special Considerations
- Molecular diagnosis of Factor V Leiden requires analysis of non-hematopoietic tissue in a patient after hematopoietic stem cell transplantation;
- Molecular diagnosis of Factor V Leiden in liver transplant recipients requires molecular genetic testing of donor tissue;
- Molecular genetic tests are reliable in individuals on warfarin or heparin anticoagulation, and independent of thrombotic episodes.

Related Tests (visit our website at www.medgen.med.miami.edu/)
Thrombophilia Risk Genotyping Assay (CMGDL test code 4002)
Factor II G20210A and Factor V Leiden Genotyping Assay (CMGDL test code 4102)
Factor II G20210A Genotyping Assay (CMGDL test code 4302)
MTHFR Genotyping Assay (CMGDL test code 4402)

References

#Pseudo-homozygous APC resistance
Pseudohomozygotes are heterozygous for both factor V Leiden and a second mutation causing a factor V deficiency (factor V null mutation). APC resistance of Factor V Leiden pseudohomozygotes is indistinguishable from that of homozygote patients for the Factor V Leiden mutation.