



The Factor V R2 DNA Test

Factor V Leiden is the most common genetic risk factor for venous thrombosis and pulmonary embolism, present in 5% of the Caucasian population and in 20-40% of individuals with a history of venous thromboembolism. Not all individuals who are heterozygous for factor V Leiden will experience a thrombotic event. The presence of the R2 polymorphism in the factor V gene increases the likelihood of thrombosis in factor V Leiden heterozygotes. The R2 polymorphism is very common, occurring in 1 in 10 individuals. This polymorphism is part of the HR2 haplotype, a collection of polymorphisms always inherited together.

Factor V Leiden heterozygotes are at a 7-fold increased risk for venous thrombosis. Co-existence of the R2 polymorphism with factor V Leiden increases that risk by an additional 3-fold through a further increase in APC resistance. The average age of the first thrombotic event is six years younger in individuals who are heterozygous for both factor V Leiden and the R2 polymorphism than in those with factor V Leiden alone. Because of this dramatic increase in the risk for a life-threatening event, it is important to identify those factor V Leiden heterozygotes who also have the R2 polymorphism.

The R2 polymorphism is identified by PCR amplification and restriction enzyme digestion followed by gel electrophoresis. Within 24 hours of the detection of factor V Leiden, the presence of the additional risk factor, R2, can be determined without an additional blood draw, and an appropriate plan of care can then be initiated.

Indications for Testing:

- Heterozygosity for factor V Leiden demonstrated by the Factor V Leiden DNA Test (the Factor V R2 DNA test is not indicated for factor V Leiden homozygotes.)

Special Aspects of our Service:

- Rapid turnaround time
- Detailed reports with genetic interpretation, recommendations, and education
- Genetic consultation by board-certified genetic counselors and geneticists

Specimen Requirements:

DNA from a blood sample previously submitted to Kimball Genetics for factor V Leiden DNA testing

or

5 ml blood in an EDTA (lavender top) tube, room temperature

or

Cheek cell sample (please call for collection kits)

Turnaround Time:

- 1 business day

Please call Kimball Genetics for more information.

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