



<u>Procedure</u>	<u>Result</u>	<u>Units</u>	<u>Ref Interval</u>	<u>Accession</u>	<u>Collected</u>	<u>Received</u>	<u>Reported/</u> <u>Verified</u>
Factor V, R2 Mutation Detection	Comment f			17-069-110140	10-Mar-17 13:06:00	10-Mar-17 00:00:00	13-Mar-17 07:06:11
Factor V, R2 - Methodology	Comment f			17-069-110140	10-Mar-17 13:06:00	10-Mar-17 00:00:00	13-Mar-17 07:06:11
Factor V, R2 - Interpretation	Comment f			17-069-110140	10-Mar-17 13:06:00	10-Mar-17 00:00:00	13-Mar-17 07:06:11
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10-Mar-17 13:06:00 Factor V, R2 Mutation Detection:
 A-G (Normal-Mutant)
 Positive - Heterozygous for the FV R2 polymorphism.
 01 Esoterix Coagulation Lab
 8490 Upland Drive Ste 100
 Englewood, CO 80112-7116
 800-444-9111
 Adcock, Dorothy M MD

10-Mar-17 13:06:00 Factor V, R2 - Methodology:
 Patient DNA was evaluated for the factor V R2 polymorphism
 at nucleotide 4070 using polymerase chain reaction (PCR)
 and restriction fragment length polymorphism (RFLP)
 technology.
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10-Mar-17 13:06:00 Factor V, R2 - Interpretation:
 The R2 4070 polymorphism is present on one copy of the
 patient's factor V gene. Heterozygous factor V R2 alone is
 a very mild prothrombotic risk factor, with an incidence in
 the population of 5 - 12%. Heterozygous factor V R2 may be
 associated with a very mild increase in activated protein C
 (APC) resistance (for example, a decreased APCR ratio). The
 presence of the R2 polymorphism in factor V Leiden
 heterozygotes increases the risk of venous thrombosis 3 to
 4 fold further than the increased risk from factor V Leiden
 alone (7-fold).
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* Abnormal, # = Corrected, C = Critical, f = Footnote, H = High, L = Low, t = Interpretive Text, @ = Reference Lab

10-Mar-17 13:06:00 Factor V, R2 - Comment:
Simultaneous Risks: If a patient possesses two or more congenital or acquired thrombophilic risk factors, the risk of thrombosis may rise to more than the sum of the risk ratios for the individual risk factors. For instance, a combination of the factor V R2 polymorphism and the factor V Leiden mutation may confer a 16-fold increase in thrombotic risk over that conferred by the presence of an isolated heterozygous factor V Leiden mutation.

Recommendations: The factor V R2 polymorphism is an inherited characteristic. If the mutation is present, we recommend that the patient and their family consider genetic counseling to obtain additional information on inheritance and to identify other family members at risk. In the heterozygous individual married to a wild-type individual, their children have a 50% chance of inheriting this mutation. All children inherit at least one abnormal gene if the tested individual is homozygous.

Testing Characteristics: Genetic testing by PCR provides exceptionally high sensitivity and specificity. Inaccurate results using PCR are limited to rare polymorphisms in primer binding sites and to misidentification of specimens by collectors or laboratory personnel. This assay detects only the factor V R2 polymorphism at nucleotide 4070 and does not measure genetic abnormalities elsewhere in the genome.

This test was developed and its performance characteristics determined by LabCorp. It has not been cleared or approved by the Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. For inquiries or genetic consultation please call Esoterix at 1-800-444-9111.

References: Castaman et al. Haematologica 2003;88:1182.
Althenc-Gelas et al. Thromb Haemost 1999;81:193. De Visser et al. Thromb Haemost 2000;83:577. Faioni EM et al. Blood 1999;94:3062. Castoldi E et al. Blood 2004;103:4173.
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