

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: 10/30/1979
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Factor V, R2 Mutation Detection by PCR

ARUP test code 2014248

Factor V, R2 Mutation Detection

Comment

A-G (Normal-Mutant)
Positive - Heterozygous for the FV R2 polymorphism.
01 Esoterix Inc
8490 Upland Drive Ste 100
Englewood, CO 80112-7116
800-444-9111
Poirier, Brian F. MD

Factor V, R2 - Methodology

Comment

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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Factor V, R2 - Interpretation

Comment

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 APC 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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Factor V, R2 - Comment

Comment

H=High, L=Low, *=Abnormal, C=Critical

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.

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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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Factor V, R2 Mutation Detection	20-183-153207	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Factor V, R2 - Methodology	20-183-153207	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Factor V, R2 - Interpretation	20-183-153207	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Factor V, R2 - Comment	20-183-153207	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at: