

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

Physician: Doctor, Example

Patient: Patient, Example

DOB 5/26/2006 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. Performed by: 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.

Performed by: 01 Esoterix Inc 8490 Upland Drive Ste 100 Englewood, CO 80112-7116 800-444-9111

Poirier, Brian F. MD

Factor V, R2 - Interpretation

Comment

The R2 4070 polymorphism is present on one copy of the nne KZ 40/0 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 4 fold further than the increased risk from factor V Leiden alone (7-fold).
Performed by: 01 Esoterix Inc

8490 Upland Drive Ste 100 Englewood, CO 80112-7116 800-444-9111

Poirier, Brian F.

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. Performed by: 01 Esoterix Inc 8490 Upland Drive Ste 100 Englewood, CO 80112-7116 800-444-9111 Poirier, Brian F. MD

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

4848



VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
Factor V, R2 Mutation Detection	23-223-144549	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Factor V, R2 - Methodology	23-223-144549	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Factor V, R2 - Interpretation	23-223-144549	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Factor V, R2 - Comment	23-223-144549	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