Patient Name:	DUMMY	**Booking ID:**	NA
Age:	NA	**Sample Type:**	NA
Gender:	NA	**Sample Collection Date:**	DD-MM-YYYY
Referring Clinician:	NA	**Sample Receiving Date:**	DD-MM-YYYY
Test Requested:	Factor V Leiden Mutation Analysis by PCR,Sanger Sequencing	**Reporting Date:**	DD-MM-YYYY

FACTOR V LEIDEN MUTATION ANALYSIS

CLINICAL INFORMATION

NA

RESULT SUMMARY

NEGATIVE
(Not Detected)

KEY FINDINGS

Target gene mutation	Mutation detection status	Relevance
Factor V - H1299R	Not detected	None
Factor V -R506Q	Not detected	None
Factor V -Y1702C	Not detected	None

RESULT INTERPRETATION

No mutation were detected for Factor V gene variant H1299R, R506Q and Y1702C in given sample.

Result	Comment
Homozygous mutation detected	Both copies of the gene carry mutation
Heterozygous mutation detected	One copy of the gene carries mutation
Not Detected	Mutation not detected

COMMENT

- ✓ Please correlate clinically.
- ✓ For about this report, or for assistance in locating nearby genetic counseling services, please contact the Laboratory: geneticcounselors@redcliffelabs.com, or ccsupport@redcliffelabs.com.

CLINICAL INTERPRETATION

Thrombosis is the formation of a blood clots inside a blood vessel, obstructing the blood flow of the cardiovascular system. Several thrombosis associated single nucleotide polymorphisms (SNPs) have been identified and reported to significantly increase the risk of venous thrombosis. Three SNPs (R506Q, H1299R and Y1702C) in the Factor V gene are the most important genetic risk factors for inherited thrombophilia. Factor V mutation increases the relative risk of thrombosis by 5-10 fold in the heterozygous condition and by 50-100 fold in the homozygous individual. The lifetime risk for DVT is 12-20% for Heterozygote and 80% for Homozygote. Factor V mutation is a risk factor for venous as well as arterial thrombosis. It is the most common genetic risk factor for thrombosis and accounts for >90 percent of APC resistance.

TEST INFORMATION

This assay is based on DNA extracted from blood followed PCR and Sanger Sequencing. It is used as a thrombosis risk factor in patients prior to major surgery, to determine the cause of recurrent second or third trimester pregnancy loss, screening for risk of thrombosis before oral contraceptive use, estrogen replacement therapy and for presymptomatic evaluation of individuals with a family history of thrombosis or a family member identified to have FV mutations. A mutational defect in factor V causes APC (Activated Protein C) resistance which can be homozygous or heterozygous. Factor V Leiden mutation is a risk factor for venous as well as arterial thrombosis.

TEST LIMITATIONS

- ✓ Test results may vary if appropriate sample collection and transportation to lab not followed as per protocol.
- ✓ Mutations below the detection limits of the assay may not be detected. Typical detection limit for Sanger Sequencing assays is >10-20%.
- ✓ This test is laboratory developed and its performance were evaluated at National Reference Lab, Redcliffe Labs.
- ✓ PCR is a highly sensitive technique; reasons for apparently contradictory results may be due to improper quality control during sample collection, selection of inappropriate specimen and/or presence of PCR inhibitors.
- ✓ This test detects mutations only three target variants in Factor V gene and report includes only variants that meets a level of evidence threshold for cause or contribute to disease.
- ✓ Gene transcript used for clinical reporting generally represents the canonical transcript, which is usually the longest coding transcript with strong/multiple supporting evidence.

DISCLAIMER

- ❖ Test has been performed assuming that the sample received belongs to the above-named individual(s) and that any stated relationships between individuals are accepted as true.
- ❖ The results should be interpreted in the context of the patient's medical evaluation. Mutation identified in this gene does not guarantee activity of the drug in a given indication due to presence of contraindicated mutation in gene.
- ❖ The mutation information provided should only be utilized as a guide or aid and the decision to select any therapy option based on the information reported here resides solely with the discretion of the treating physician.
- ❖ This report should only be used as an aid and the treating physician should employ sound clinical judgment in arriving at any decision for patient care or treatment.

REFERENCES

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2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.

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Not all tests at all locations are under NABL scope. Availability of tests under NABL scope varies from lab to lab.