Patient Name:	Dummy	**Booking ID:**	XXX
Age:	29 Years	**Sample Type:**	EDTA Whole Blood
Gender:	Female	**Sample Collection Date:**	23-05-2024
Referring Clinician:	NA	**Sample Receiving Date:**	24-05-2024
Test Requested:	Factor II Prothrombin gene mutation analysis by PCR, Sanger Sequencing	**Reporting Date:**	28-05-2024

FACTOR II PROTHROMBIN GENE MUTATION ANALYSIS

CLINICAL INDICATION

NA

RESULT SUMMARY

NEGATIVE
(Not Detected)

KEY FINDING

Target gene mutation	Mutation detection status	Relevance
Factor II - G20210A	Not Detected	None

RESULT INTERPRETATION

No mutation was detected for the Prothrombin Factor II gene variant G20210A.

Result classification	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 SIGNIFICANCE

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 deep venous thrombosis (DVT). Prothrombin (Factor II) gene (G20210A) has been found to be associated with increased Prothrombin levels and an increase in the risk for venous thrombosis in heterozygous. Higher concentrations of Prothrombin lead to increased rates of thrombin generation, resulting in excessive growth of fibrin clots. Heterozygosity for 20210G>A is associated with a 3-fold increased risk of venous thrombosis. Affected individuals may be candidates for anti-thrombotic prophylaxis. Homozygous are rare but two copies of the mutation would increase that risk. When heterozygosity for 20210G>A is combined with heterozygosity for the Factor V Leiden mutation, the relative risk for thrombosis increases further. Combination with non-genetic risk factors such as use of oral contraceptives, also leads to substantial elevations in relative risk.

TEST INFORMATION

This assay is based on DNA extracted from blood followed by PCR and targeted mutation Sanger Sequencing. The Test may be used for evaluation of patients with early onset VTE, 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 and estrogen replacement therapy. Prothrombin G20210A mutation occurs in the noncoding region of the Factor II gene and is the second most common cause of inherited thrombophilia after FVL mutations. This test was developed and its analytical performance characteristics have been determined **by Redcliffe labs**. It has not been cleared or approved by FDA.

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 only ONE variants in Factor II gene and report includes variants that meets a level of evidence threshold for cause or contribute to disease.
- ✓ If this mutation is not found by the testing procedure, it does not mean that the risk of carrying or developing deep vein thrombosis is not present. It simply means that this specific mutation has not been found, although other mutations may be present.
- ✓ False positive or false negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, somatic or tissue-specific mosaicism.
- ✓ 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

1. Hussein et al., 2012. . Journal of Thrombosis and Thrombolysis, DOI: 10.1007/s11239-012-0731-9
2. Varga EA and Moll S. Prothrombin 20210 Mutation (Factor II Mutation). Circulation. (2004). 110:e15-e18.
3. Segers K, Dahlbäck B, Nicolaes GA. Coagulation factor V and thrombophilia: background and mechanisms. Thromb Haemost. (2007). 98(3):530-542.
4. Castoldi E, Simioni P, et al. Combinations of 4 mutations (FV R506Q, FV H1299R, FV Y1702C, PT 20210G/A) affecting the prothrombinase complex in a thrombophilic family. Blood. (2000). 96(4):1443-1448.

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Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.

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

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1. The presented findings in the Reports are intended solely for informational and interpretational purposes by the referring physician or other qualified medical professionals possessing a comprehensive understanding of reporting units, reference ranges, and technological limitations. The laboratory shall not be held liable for any interpretation or misinterpretation of the results, nor for any consequential or incidental damages arising from such interpretation.

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.

4. This report shall not be deemed valid or admissible for any medico-legal purposes.

5. The Customers assume full responsibility for apprising the Company of any factors that may impact the test finding. These factors, among others, includes dietary intake, alcohol, or medication / drug(s) consumption, or fasting. This list of factors is only representative and not exhaustive.