Genetics Uncoded:

FACTS ABOUT PROTHROMBIN-RELATED THROMBOPHILIA

AKA | Hyperprothrombinemia, Prothrombin G20210A Thrombophilia, Venous Thromboembolism, Factor II deficiency, Dysprothrombinemia

What Your Test Results Mean
Test results indicate that you have prothrombin-related thrombophilia. Risk for the current or future pregnancies is dependent on your partner’s carrier status; however, individuals with just one copy (heterozygous) of the prothrombin mutation have a 50% chance to have a child heterozygous for prothrombin-related thrombophilia. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Prothrombin-related Thrombophilia Explained
Prothrombin-related thrombophilia is the second most common inherited defect of coagulation, present in about 2% of Caucasian Americans. The severity of thrombophilia is dependent on the number of copies of the G20210A mutation as well as other factors. Individuals with one copy of G20210A and no personal history of venous thromboembolism have less than a 1% chance for venous thromboembolism during pregnancy, with a personal history, risk increases to 10%. Individuals with two copies of G20210A and no personal or family history have a 2-3% chance for venous thromboembolism during pregnancy. In the presence of family or personal history, risk for thromboembolism is greater.

Coexisting thrombophilias, and other risk factors including travel, use of oral contraceptives, use of hormone replacement therapy, use of selective estrogen receptor modulators, pregnancy status, age, and surgery also play a role in risk of venous thrombosis. Alternative methods to oral contraceptives should be considered in women prothrombin-related thrombophilia due to the increased risk of thromboembolism.

How the Genetics Work
The clinical features of prothrombin-related thrombophilia can be explained by a mutation, G20210A, in the F2 gene. All individuals have two copies of the F2 gene. Individuals with prothrombin-related thrombophilia have either one or two copies of F2 mutation. The mutation causes higher than normal levels of prothrombin, increasing the risk for venous blood clots.

Recommended Next Steps
Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?
Contact us at 1-855-776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.