



Genetics Uncoded:

# Facts about

## Factor II Prothrombin-Related Thrombophilia



### What Your Test Results Mean

**Individuals with just one copy (heterozygous) of the prothrombin variant have a 50% chance to have a child heterozygous for Factor II prothrombin-related thrombophilia.** Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

### ● Prothrombin-Related Thrombophilia Explained

Factor II 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 variant, 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 thrombophilia 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 with Factor II 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 pathogenic variant, G20210A, in the *F2* gene. All individuals have two copies of the *F2* gene. Individuals with Factor II prothrombin-related thrombophilia have either one or two copies of the *F2* variant. The variant causes higher than normal levels of prothrombin, increasing the risk for venous blood clots.

### 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.