Genetics Uncoded:
FACTS ABOUT FACTOR XI DEFICIENCY

AKA | Rosenthal Syndrome, PTA Deficiency, F11 Deficiency, Plasma Thromboplastin Antecedent Deficiency, Factor 11 Deficiency, Hemophilia C

What Your Test Results Mean
Genetic carrier screening indicates that you are a carrier of Factor XI Deficiency. Carriers typically show no symptoms of this disorder; however, carriers are at an increased risk for having a child born with Factor XI Deficiency. Risk for current or future pregnancies is dependent on your partner’s carrier status. Consulting with a genetic counselor as well as carrier testing of your partner is recommended.

Factor XI Deficiency Explained
Factor XI deficiency is an inherited form of hemophilia called Hemophilia C. Hemophilia C rarely causes spontaneous bleeds like other forms of hemophilia, but having surgery of any kind can cause excessive bleeding and the need for prophylaxis. Fresh frozen plasma is typically used for treatment. Life expectancy is considered normal if proper precautions concerning surgeries are taken.

How the Genetics Work
Factor XI is an autosomal recessive blood disorder caused by mutations in the F11 gene. In general, individuals have two copies of the F11 gene. Carriers Factor XI deficiency have a single mutation in one copy of the F11 gene while individuals with Factor XI deficiency have mutations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

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 (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.