Gene test information

PROTHROMBIN 20210A

- **Background**

  The prothrombin (coagulation factor II) 20210A mutation is a common genetic risk factor for thrombosis and is associated with elevated prothrombin levels. Higher concentrations of prothrombin lead to increased rates of thrombin generation, resulting in excessive growth of fibrin clots.

- **Prothrombin (F2 20210G>A) genotypes**

Genotype	Frequency	Commentary
F2 GG :	97%	Wild type genotype. No prothrombin 20210A variant detectable.
F2 GA :	3%	Heterozygous for prothrombin 20210A. The risk of venous thrombosis is increased approximately 2- to 4-fold compared to the wildtype..
F2 AA :	< 0.1%	Homozygous for prothrombin 20210A. The risk of venous thrombosis is increased, although the magnitude is not well defined.

- **Indications for testing**
  - a history of recurrent VTE, first VTE at younger than 50 years, or first unprovoked VTE at any age,
  - a first VTE at an unusual anatomic site, such as the cerebral, mesenteric, portal, or hepatic veins,
  - a first VTE, at any age, in a subject with a first degree family member with a VTE before the age of 50 years,
  - a first VTE related to pregnancy, the puerperium, oral contraceptive use, or hormone replacement therapy,
  - unexplained pregnancy loss during the second or third trimester.

References:

Renner W et al. Prothrombin G20210A, factor V Leiden, and factor XIII Val34Leu: common mutations of blood coagulation factors and deep vein thrombosis in Austria. Thromb Res 2000;99:35-9.

McGlennen RC et al. Clinical and laboratory management of the prothrombin G20210A mutation. Arch Pathol Lab Med. 2002 Nov;126(11):1319-25.

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