

Genes-4U

PAI-1 4G/5G

(Plasminogen-Activator-Inhibitor-1 4G / 5G Insertion / Deletion polymorphism)

Plasminogen-Activator-Inhibitor-1 is an important regulator of hemostasis. The PAI-1 gene promoter contains a 4G / 5G insertion/deletion polymorphism. The 4G variant causes enhanced transcription and is associated with enhanced plasma levels of PAI-1 protein and with a procoagulant state. Importantly, the PAI-1 gene promoter 4G variant increases the thrombotic risk when other risk factors like the factor V Leiden mutation or antiphospholipid antibodies are present. Clinical associations have been described with thrombosis (1-4), preeclampsia (5), prognosis of polytrauma and sepsis (6,7) and risk of myocardial infarction (8). The urokinase plasminogen activator-PAI system and the PAI-1 4G/5G polymorphism were also shown to play a role in the development of more aggressive colon and breast cancer (9,10).

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