Table I. Inherited thrombophilic conditions

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Risk factor** | **Prevalence** | **Inheritance** | **Age of thrombosis onset** | **Comments** |
| Protein C deficiency | Heterozygous <1%;  Homozygous rare | Autosomal dominant  http://ghr.nlm.nih.gov/condition/protein-c-deficiency | Homozygotes usually present in the newborn period with purpura fulminans. Heterozygotes can develop thrombosis in adolescence | Associated most often with venous thrombosis. Patients who require anticoagulation can develop warfarin-induced skin necrosis unless heparin is started first. |
| Protein S deficiency | Heterozygous <1%;  Homozygous rare | Autosomal dominant  http://ghr.nlm.nih.gov/condition/protein-s-deficiency | Homozygotes usually present in the newborn period with purpura fulminans. Heterozygotes can develop thrombosis in adolescence | Associated with increased venous and arterial thrombosis. Concentrations of both free and total protein S must be measured, since it is highly protein bound. Patients who require anticoagulation can develop warfarin-induced skin necrosis unless heparin is started first. |
| Antithrombin deficiency | Heterozygous <1%;  Homozygous rare | Autosomal dominant  http://ghr.nlm.nih.gov/condition/hereditary-antithrombin-deficiency | Thrombosis can occur in children as young as 10 years in heterozygotes. Homozygous deficiency usually presents with extensive thrombosis on the day of birth and most infants die within days. | Associated most often with venous thrombosis. Antithrombin is the most important inhibitor of activated clotting factors and even levels below 80% of normal can be associated with increased thrombotic risk. |
| Factor V Leiden | Heterozygous 3% to 8% of Caucasians (<1% among people of other ethnicities); Homozygous in about 1 in 5000 people | Autosomal dominant  http://ghr.nlm.nih.gov/condition/factor-v-leiden-thrombophilia | Heterozygous patients usually present as adolescents or young adults when other risk factors are present (e.g., smoking, oral contraceptive use, pregnancy, trauma, surgery) | Associated most often with venous thrombosis. Resistance to activated protein C due to a point mutation in Factor V that produces a Factor V protein resistant to cleavage by activated protein C and thus decreases the ability of protein C to inhibit thrombin formation. |
| Prothrombin thrombophilia | Heterozygous 2% of Caucasians (<1% among people of other ethnicities); | Autosomal dominant  http://ghr.nlm.nih.gov/condition/prothrombin-thrombophilia |  | Associated most often with venous thrombosis. Excess production of prothrombin leads to excess thrombin production. |