

Table III. Inherited and acquired thrombophilic disorders and incidence of venous thromboembolism

Thrombophilia	Incidence	Thrombotic risk	Incidence of first thrombosis	Incidence of recurrent thromboembolism
Antithrombin (AT) deficiency	1 in 500--5000	15-20 fold	1.8% per year (1.1 – 2.6%)	10% per year (6.1-15.4%)
Protein C (PC) activity	1 in 500 (0.14-0.5%)	15-20 fold	1.5% (1.1-2.1%)	6.0% (3.9-8.7%)
Protein S (PS) deficiency	1 in 1000 (0.03-0.13%)	15-20 fold	1.9% (1.3%-2.6%)	8.4% (5.8%-11.7%)
Factor V Leiden	Caucasian 2-10%	Heterozygotes 3.5 fold Homozygotes 18 fold	0.5% per year, heterozygotes	1.56 fold (2.8% to 7.5% per year)
Prothrombin gene G20210A mutation	Northern Europe - 1.7% (1.3-2.2%) Southern Europe – 3.0% (2.3-3.7%) US African American 0.2% Asian 0	2-3 fold	0.3% per year	1.45 fold (0-2.9% per year)
Dysfibrinogenemia	Rare 0.8% of DVT patients	Not known	Not known	Not known
Hyperhomocysteinemia	Mild homocysteinemia 5-7% or population, homocystinuria-1 in 58,000 to 1,000,000	2-3 fold for mild hyperhomocysteinemia, 50% of patients with homocystinuria suffer thromboembolism by age 30	Not known	Not known
Methylene Tetrahydrofolate Reductase C677T Polymorphism	Homozygotes 12%	1.5 fold (1.2-1.9)	Not known	Not known
Factor VIII activity	11% of controls in Leiden Thrombophilia study (LETS)	4.8 fold increased risk of initial	0.49% per year	6.7 fold increased risk
Factor IX antigen	3% in healthy controls in LETS	2.5 fold increased risk	Not known	Not known
Factor XI antigen	10% of healthy controls in LETS	2.2 fold increased risk	Not known	Not known
Antiphospholipid antibodies	3-5% of general population, up to 50% in SLE patients	4.1-16.2 fold increased risk for thrombosis with LA,		6.8 fold increase
		1-2.5 fold for		2.5 fold increase

anticardiolipin antibodies,
3.5-5.2 fold increased risk for Beta2Glycoprotein I antibodies

Abbreviations: DIC- disseminated intravascular coagulation, MTHFR- methylene tetrahydrofolate reductase , CBS- cystathionine beta synthase, SLE- systemic lupus erythematosus, LA- lupus anticoagulant, DVT

Compiled from:

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