A 19-year-old woman presents with recent-onset right leg swelling and pleuritic chest pain and is found to have iliofemoral thrombosis on Doppler ultrasound examination. Results of a ventilation-perfusion lung scan indicate a high probability of pulmonary emboli. She had been taking the oral contraceptive pill for the last three years but has now ceased. She is a non-smoker. There is no significant medical history and no known family history of venous thromboembolism.

The results of which one of the following investigations for an underlying hypercoagulable state are most likely to be affected by the presence of the extensive thrombosis?

A. Anti-phospholipid antibody screen.
B. Anti-thrombin level.
C. Assessment of prothrombin and factor V genotypes.
D. Activated partial thromboplastin time (APTT).
E. Full blood count including blood film.

The most common presentations of venous thrombosis are deep vein thrombosis (DVT) of the lower extremity and pulmonary embolism. The causes of venous thrombosis can be divided into two groups: hereditary and acquired.

Causes of venous thrombosis

Inherited thrombophilia

- Factor V Leiden mutation
- Prothrombin gene mutation
- Protein S deficiency
- Protein C deficiency
- Antithrombin (AT) deficiency
- Rare disorders
- Dysfibrinogenemia

INHERITED THROMBOPHILIA — Inherited thrombophilia is a genetic tendency to venous thromboembolism that usually presents in young patients (less than 50 years of age) and is often recurrent. The most frequent causes of the syndrome are the factor V Leiden mutation and a prothrombin gene mutation, which account for 50 to 60 percent of cases. Defects in protein S, protein C, and antithrombin (formerly known as antithrombin III) account for most of the remaining cases, while a rare cause is certain dysfibrinogenemias.

Fifty percent of thrombotic events in patients with inherited thrombophilia are associated with the additional presence of an acquired risk factor (eg, surgery, prolonged bed rest, pregnancy, oral contraceptives). Some patients have more than one form of inherited thrombophilia or more than one form of acquired thrombophilia and appear to be at even greater risk for thrombosis.

Acquired disorders

- Malignancy
- Presence of a central venous catheter
- Surgery, especially orthopedic
- Trauma
- Pregnancy
- Oral contraceptives
- Hormone replacement therapy
- Tamoxifen
- Immobilization
- Congestive failure
- Antiphospholipid antibody syndrome
- Myeloproliferative disorders
- Polycythemia vera
- Essential thrombocythemia
- Paroxysmal nocturnal hemoglobinuria
A. Anti-phospholipid antibody screen.
   Part of lupus anticoag screen – not affected by VTE - altered by use of heparin
B. Anti-thrombin level.
   If normal anti-thrombin antibody can be excluded, however if low this cannot be excluded as may be consumed by clot = answer
C. Assessment of prothrombin and factor V genotypes.
   Molecular markers will not be affected by anticoagulation or presence of VTE
D. Activated partial thromboplastin time (APTT).
   Altered by heparin use
E. Full blood count including blood film.
   No change

Wafarin will affect protein C and protein s levels