

Thrombosis and Thrombophilia

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James talks to Dr Shafqat Inam about Thrombosis and Thrombophilia and when it is appropriate to investigate for underlying causes of thromboembolism and outline initial management.

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About Dr Shafqat Inam

Shafqat Inam is currently a [Haematology](#) registrar at Royal Prince Alfred Hospital and Concord Hospital and Associate Lecturer at the [University of Sydney](#). He studied medicine at the [University of New South Wales](#) and completed his junior medical officer and basic physician training at Royal Prince Alfred Hospital. He has a particular interest in [research](#) and medical education.

Thrombosis and Thrombophilia

With Dr Shafqat Inam, Haematology Advanced Trainee at Royal Prince Alfred Hospital and Concord Hospital, New South Wales, Australia

Introduction

As a junior doctor, you are likely to encounter thromboembolic disease, both in the Emergency Department and on the wards. In the work up of any case of thrombosis the junior doctor should be aware of the possibility of an underlying thrombophilia. In this podcast, we discuss when it is appropriate to investigate for underlying causes of thromboembolism and outline initial management.

Case

You are a junior doctor working in the Emergency Department when a 35-year-old woman presents with a 3-day history of right leg swelling. Doppler ultrasound confirms a thrombus extending from the popliteal to the superficial femoral vein.



1. Once the diagnosis of DVT is made, what further history and investigations would you obtain from the patient?

- **History:**

- **Provoking Factors:** lower limb orthopaedic surgery, recent hospitalisation, long distance flight, pregnancy or the use of the oral

contraceptive pill.

- Ask for symptoms of PE such as chest pain or shortness of breath. If these symptoms are present then further testing such as CTPA may be required. Although the treatment is essentially the same for DVT and PE, it is important to know if PE is present if the patient clinically deteriorates.
- **Investigations:**
 - Doppler is sufficient for the diagnosis of DVT.
 - May consider age and sex appropriate screening tests for underlying malignancy.

2. What are thrombophilias?

- Thrombophilias are conditions which predispose an individual to thrombosis. They are relatively uncommon conditions. They include:
 - Enhanced activity of pro-coagulants.
 - Factor V Lieden mutation.
 - Prothrombin gene mutation.
 - Deficiency in natural anticoagulants.
 - Protein C
 - Protein S
 - Antithrombin III
 - Antiphospholipid Syndrome
 - Leads to both venous and arterial thrombus and associated with recurrent miscarriages.
 - Investigate for antiphospholipid syndrome by screening for the following antibodies: anticardiolipin antibody, lupus anticoagulant, Beta2 glycoprotein antibody.

3. Investigations for thrombophilias

- Thrombophilias should not be investigated routinely as the conditions are relatively uncommon and management does not change substantially.
- You may consider investigating if:
 - Site of the thrombus is unusual such as in the cerebral venous system.
 - Strong family history of thrombosis.
 - Recurrent miscarriage.

Investigating for thrombophilia

- Timing of testing is important as both the DVT itself (which can deplete certain coagulants) and the treatment of the DVT (warfarin in particular) will make testing inaccurate.
- Testing for factor V Leiden mutation and prothrombin mutation are not affected by anticoagulation therapy or presence of a DVT.
- Further screening is usually performed after the anticoagulation therapy is ceased.

4. What affects the length of anticoagulation?

- The presence of provoking factors determines the length of anticoagulation.
 - Provoked DVT - 3-6 months.
 - Unprovoked - consideration of life-long anticoagulation.

5. Management of DVT

- Acute
 - Look for evidence of vascular compromise: if present then contact the vascular team and consider catheter guided thrombolysis.
 - Anticoagulation
 - Warfarin
 - Direct thrombin inhibitor - Dabigatran
 - Factor Xa inhibitors - Rivaroxaban, Apixaban
 - These latter 3 agents do not require bridging with heparin therefore are beneficial for those presenting to the Emergency Department or GP.

NB: Direct thrombin inhibitors and Factor Xa inhibitors are contraindicated in renal failure.

- Long term
 - Compression stockings are helpful with patients with post thrombotic syndrome (presence of pain and swelling in the leg after resolution of the DVT).
 - Ensure adequate follow up so that the cessation of therapy can be considered in an appropriate time frame.

6. Below knee vs above knee DVTs

- Above knee DVTs
 - Should always be treated with anticoagulation.
- Below knee DVTs
 - Controversial. Can be treated with anticoagulation, or a repeat Doppler performed in 2 weeks, and if there is extension then treatment is commenced.

Take home messages

- Diagnosis of DVT is important as it can cause significant morbidity and mortality.
- When making the diagnosis always ask for provoking factors as this will determine the length of anticoagulation.
- Thrombophilia testing is complex and should only be performed in selected cases.

Related Podcasts

- [An approach to the management of bleeding disorders](#)

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