CASE REPORT

Congenital absence of inferior vena cava: an under recognised cause of unprovoked venous thromboembolism

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Learning points for clinicians

Congenital absence or agenesis of the inferior vena cava (AIVC) is an unfamiliar but significant cause of unprovoked deep vein thrombosis (DVT) in younger patients. If diagnosed with AIVC, management is considerably different with likely requirement of lifelong anticoagulation.

Case report

A 21-year-old male presented with a 1-week history of right thigh pain and swelling, on a background of a right partial nephrectomy as a neonate for ureterocele. He was a non-smoker, had no recent history of trauma, long-distance travel or family history of coagulopathies. Examination revealed significant right groin and thigh tenderness and swelling, palpable varicosities of the right iliac veins; and mildly tender right testicular swelling. Blood tests were unremarkable.

Ultrasound demonstrated an occlusive thrombus through the right common femoral vein extending into and throughout the external iliac vein. The inferior vena cava (IVC) was not visualized below the level of the liver and there was evidence of thrombosed pelvic varices. Testicular ultrasound was unremarkable.

Computed tomography (CT) neck to pelvis did not demonstrate any malignancy, but the vascular findings were consistent with his prior ultrasound. CT and magnetic resonance venography confirmed congenital absence of the IVC (Figure 1, arrow). He was anticoagulated with apixaban with a view to lifelong anticoagulation pending reviews from vascular and haematology teams.

Discussion

The incidence of congenital absence or agenesis of the inferior vena cava (AIVC) in the general population is 0.0005–1%, however this anomaly represents an estimated 5% of unprovoked deep vein thrombosis (DVT) in patients < 30 years of age.\textsuperscript{1–3}

The IVC is formed during embryogenesis by a complex process involving the anastomosis of the posterior cardinal, subcardinal and supracardinal veins to form a unilateral rightsided system. AIVC is one type of vascular anomaly that results from failure of these paired structures to fuse. The collateral deep venous system that develops is likely inadequate and unable to cope with the demands of increasing blood flow. This results in lower extremity chronic venous hypertension with venous stasis and predisposition to the development of thrombosis. Acute thrombotic events in AIVC patients have been reported in multiple cases to have a precipitant, most notably, strenuous muscular exercise.\textsuperscript{1}

In a case series of 10 patients with DVT and AIVC, the common presentation was proximal DVT in a young male below 40 years of age, often after major physical exertion. DVT was bilateral in 60% with no pulmonary embolism (PE). PE is an uncommon finding (9.67%), as emboli are trapped in the azygous/hemiazygous system prior to reaching the pulmonary circulation.\textsuperscript{1}
The mainstay of treatment is prolonged anticoagulation, however evidence is limited regarding duration. The concurrent use of compression elastic stockings and limb elevation has also been recommended. Patients are advised to avoid risk factors, such as excessive physical exertion, immobilisation or smoking. In some cases, surgical management including IVC reconstruction followed by oral anticoagulation has been described in Ref.4.

In conclusion, we propose AIVC is under recognised but becomes a significant cause of unprovoked DVT in the lower extremities of young patients with no known risk factors. Diagnostic work-up should include CT or magnetic resonance imaging because ultrasonography alone is unable to detect these anatomic variants. Due to the chronic venous hypertension and ongoing predisposition for thrombosis, all patients are likely to require lifelong anticoagulation. Currently, there are no standardised guidelines for this subpopulation of patients with DVT thus further research is required.

Conflict of interest: None declared.

References