Agenesis of the inferior vena cava associated with lower extremities and pelvic venous thrombosis

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The absence of the inferior vena cava is an uncommon congenital anomaly and can be misdiagnosed. We report a 14-year-old boy initially admitted as a result of a painful abdominal mass; after admission, he experienced a deep venous thrombosis in his left leg. Subsequent evaluation revealed the congenital absence of the entire inferior vena cava, with a cluster of thrombosed collateral veins in his right pelvis, corresponding to the abdominal mass. The recognition of this anomaly may be helpful in the event of differential diagnosis in retroperitoneal masses. In young patients with idiopathic deep venous thrombosis, an inferior vena cava anomaly should be considered. (J Vasc Surg 2006;44:1114-6.)

Anomalies of the inferior vena cava (IVC) are present in 0.3% to 0.5% of otherwise healthy individuals1-3 and in 0.6% to 2% of patients with other cardiovascular defects.4-7 Agenesis of the IVC (AIVC) has an incidence of 0.0005% to 1% in the general population,8 and recent reports confirm its role as a strong predisposing factor for the development of deep venous thrombosis (DVT) in young adults. Some authors estimate the prevalence of AIVC in that group of patients to be approximately 5%.4,7,9 In adults, such an anomaly can cause diagnostic problems in the paravertebral area because of their tumor-like appearance.8,10

We report a case of this anomaly in which both an abdominal mass and DVT were found. The clinical features and the diagnostic and therapeutic options are discussed.

CASE REPORT

A 14-year-old boy was admitted after a week of abdominal pain. Physical examination revealed a healthy boy with a palpable abdominal mass, but with neither hepatomegalia/splenomegalia nor portal hypertension signs. An abdominal ultrasonography scan showed a right pelvis mass suggestive of retroperitoneal lymphoma, and a computed tomography (CT) scan was performed for better evaluation and biopsy. The CT showed absence of the entire IVC, enlarged azygous and hemiazygous veins, and hypointense thrombus within a cluster of collateral veins in the pelvis that corresponded to the abdominal mass (Fig 1, A-C).

Several days after admission, an unexpected swelling in the left lower extremity was observed. An eco-Doppler ultrasound scan revealed a thrombosis of the left iliac and femoral veins, with a permeable right venous system. The patient was treated with heparin and was told to rest in bed. The edema of the leg and the abdominal mass decreased.

A magnetic resonance imaging (MRI) scan was performed, and it showed the same facts as the CT and also revealed a dilated left common iliac vein. An ascending venogram showed that the IVC was completely absent, that the azygous and hemiazygous veins collected the venous blood return from dilated paravertebral venous collaterals, and that the azygous vein drained into the superior vena cava (Fig 2). The echocardiographic examination revealed aplasia of the IVC and hepatic veins entering directly into the right atrium. No congenital defect of the heart or the abdominal visceras was found. Hypercoagulability studies were performed and did not show any defects predisposing to thrombophilia.

The patient was discharged with oral acenocoumarol therapy. Currently, after 2 years, the patient is still taking anticoagulants and using external elastic compression, and his bed is elevated at night. The swelling of the left lower extremity is almost absent, although...
1. Absence of the suprarenal IVC results from failure to coincide with clotting defects, but most of DVT. The dysgenesis of the IVC has been described in blood pressure in the veins of the legs, thus facilitating blood return through collaterals may increase the venous rise to the four segments of the adult IVC: hepatic, supra-cardinal, and supracardinal) appear in this order and give rise to the four segments of the adult IVC: hepatic, supra-renal, renal, and infrarenal (Fig 3).

AIVC is often used to describe three different entities:

1. Absence of the suprarenal IVC results from failure to form the right subcardinal vein. The hepatic segment drains directly into the right atrium, and the blood from the infrarenal IVC returns to the heart through the azygos and hemiazygos veins. There is association with other cardiac and visceral anomalies, such as dextrocardia, atrial septal defect, atrioventricular canal, situs inversus, polysplenia, or asplenia.

2. Absence of the infrarenal IVC with preservation of the suprarenal segment implies a failure of the development of the right supracaaval vein.

3. Absence of the entire IVC, as in our patient’s case, suggests that all three paired vein systems failed to develop properly, but it has no relation to the other congenital anomalies described previously.

The reasons for the developmental failure are unclear. One hypothesis is embryonic dysontogenesis, but some authors suggest that it is the result of an intrauterine or perinatal thrombosis.

AIVC could be present with DVT. An inadequate blood return through collaterals may increase the venous blood pressure in the veins of the legs, thus facilitating DVT. The dysgenesis of the IVC has been described in coincidence with clotting defects, although most of the cases are like this patient’s, in which screening for thrombophilia revealed no alteration.

Some remarkable features of AIVC malformation with DVT were found: patients with these anomalies were significantly younger, most of them had no other precipitating factors or clotting defects, the iliac veins were affected with high frequency, an excess of bilateral thrombosis was observed, and, finally, they had a higher risk of recurrent DVT. In some cases, this anomaly appears together with pulmonary embolism.

The dilated collaterals in the abdomen could be confused with enlarged pericaval lymph nodes in an abdominal CT or MRI, thus suggesting a retroperitoneal lymphoma; this was the first supposition in our patient. The thrombosed pelvic collateral veins resulting from AIVC have been described as a mimicker of acute appendicitis.

Some cases are seen incidentally in abdominal surgery or in radiologic workup. AIVC may favor chronic venous insufficiency with ulceration, but only one case with spontaneous rupture of collateral veins of the IVC has been reported. Ultrasound Doppler scanning is usually the first imaging modality in the evaluation of patients with DVT, but anomalies of the IVC may be missed. CT and MRI are the most practical methods used to visualize this pathologic process. Extensive collateral flow is observed, and the azygos and hemiazygos systems are particularly prominent; collaterals include the ascending lumbar veins, para-vertebral venous plexus, and anterior abdominal wall veins, as in this case, in which venous plexus developed on the abdominal wall.

The advantages of MRI include not only the visualization of the venous circulation without the use of contrast agents, but also the visualization of other associated congenital anomalies, and it is probably the best diagnostic tool. Phlebography has the disadvantage of being an invasive procedure, and in our case it confirmed the absolute absence of the IVC.

Although some authors recommend thrombolytic therapy, we did not use it in our patient’s case because the chronic obstructive pattern associated with AIVC could make us expect a high rate of recurrence of DVT, even if the initial thrombolysis was successful. A surgical reconstruction in a patient with large ulceration and high venous hypertension of the legs who did not respond to medical treatment has been reported, but the long-term results are unknown.

In most cases, conservative anticoagulant therapy is enough to relieve the symptoms, as in our case. Because patients with AIVC may have a higher risk for thrombotic recurrence, lifelong oral anticoagulation should be considered. Elastic stocking support and leg elevation can be used as an adjuvant treatment to prevent venous insufficiency and ulceration in such patients. Patients should also be advised to avoid additional risk factors, such as prolonged immobilization and oral contraceptive use.

Recurrent DVT is reported if the patient stops taking anticoagulants. The long-term prognosis of patients...
with AIVC is unknown, but we think that they probably do not have a normal survival rate.

In summary, our report suggests that AIVC should be considered in young patients with DVT without the concurrence of risk factors and that more investigation (CT or MRI) should be performed. Treatment is generally long-term anticoagulation because of the higher risk for thrombotic recurrence.

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REFERENCES