Congenital Absence of Inferior Vena Cava as a Rare Cause of Pulmonary Thromboembolism

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Interruption of the inferior vena cava IVC) with azygos continuation is an uncommon vascular anomaly that results from aberrant development during embryogenesis. We report a rare case of this anomaly, presenting with massive pulmonary embolism. Subsequent evaluation with abdominal CT scan revealed the congenital absence of retrohepatic IVC. The patient was successfully treated with anticoagulation. When deep venous thrombosis (DVT) develops in patients with no apparent risk factors, the presence of congenital IVC anomalies should be considered.

Key Words: Vena cava abnormalities, deep venous thrombosis, pulmonary embolism

INTRODUCTION

Congenital Absence of Inferior Vena Cava (AIVC) is an uncommon vascular anomaly and results from atresia of the retrohepatic segment of the IVC during embryogenesis.[1,2] It has been occasionally associated with a number of cardiac and abdominal anomalies.[3,4] Patients without these associated abnormalities are typically asymptomatic, making this anomaly an incidental finding. Deep venous thrombosis (DVT) has been described in patients with congenital venous mal-

formations,[4-15] and it is not until recently that this anomaly was considered as a risk factor for DVT by facilitating venous stasis.[13,16,17] We herein report a case of congenital absence of retrohepatic IVC with azygos continuation, presenting with pulmonary embolism and DVT. To our knowledge, this is only the second report on pulmonary embolism associated with IVC anomaly in the English literature. The present case emphasizes that when DVT develops, especially in patients with no apparent risk factors, congenital anomalies of the inferior vena cava (IVC) should be considered.

CASE REPORT

A 32-year-old man, without significant personal or familial medical history, was admitted to our hospital because of painful swelling of the right lower leg and dyspnea. He had been in good health until 15 days prior to admission, when he started to suffer from the above symptoms. Dyspnea was markedly aggravated enough to limit even slight physical activity 3 days before admission. He had no apparent risk factors for thromboembolic diseases, including recent trauma, surgery or family history of thrombophilia. On admission, his pulse rate was 118 beats/min and blood pressure was 120/90 mmHg. Physical examination revealed tender swelling of the right lower leg with no other pertinent finding. The tests for thrombophilia, including homocysteine, antithrombin III, protein C, protein S, antipho-

spholipid antibody and tumor markers (CEA, AFP, CA19-9), were all within normal limits. Electrocardiogram showed sinus tachycardia and chest PA showed no clear abnormality. Duplex sonography of the right leg demonstrated a thrombotic occlusion of the right popliteal vein, whereas the left leg showed no signs of abnormality. Contrast CT scan of the chest showed extensive thrombosis in the bilateral pulmonary arteries (Fig. 1, thin arrows). Worsening the situation, the running direction of the aortic arch was in the opposite orientation to the norm. The ascending aorta was lying at the left posterior side of the left main pulmonary artery, while the descending thoracic aorta toward the right side of the vertebrae (Fig. 1, Fig. 2, asterisk). Furthermore, there was an enlarged azygos vein (Fig. 1, Fig. 2, thick arrow) to the right of the aorta, emptying into the superior vena cava (Fig. 2, thin arrow). Additional malformations of IVC were identified on CT scan of the abdomen. The hepatic veins drained into suprahepatic IVC (Fig. 3, thin arrow), ventral to the aorta. To the right-posterior side of the aorta, superior to the renal veins, venous drainage formed an enlarged azygos vein (Fig. 3, thick arrow), the diameter of which was as large as that of the aorta. The hepatic segment of the IVC was absent (Fig. 4). At the level of renal veins,
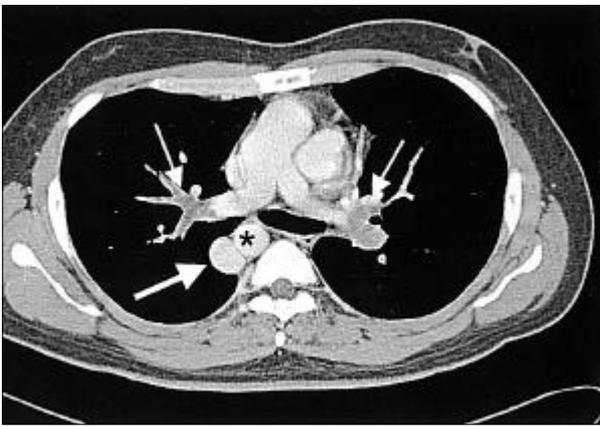

Fig. 1. Contrast CT scan of the chest showing extensive thrombosis in bilateral pulmonary arteries (thin arrows). The descending thoracic aorta (asterisk) and an enlarged azygos vein (thick arrow) are also shown.
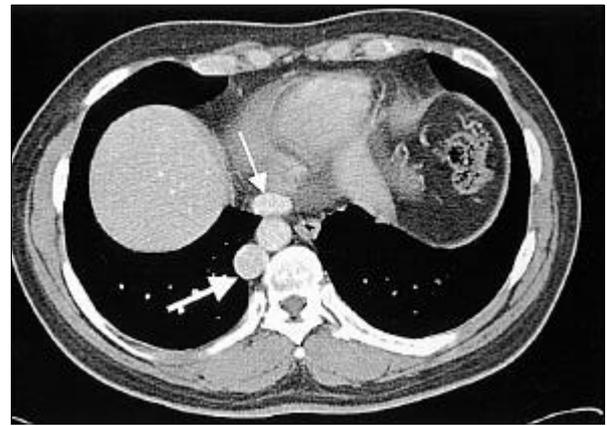

Fig. 2. Descending thoracic aorta lies to the right of the vertebrae (asterisk). The azygos vein (thick arrow) is draining into the superior vena cava (thin arrow).

Fig. 3. CT scan of the abdomen demonstrates suprahepatic IVC (thin arrow), ventral to the aorta. The azygos vein (thick arrow) is enlarged and its diameter is as large as that of the aorta.
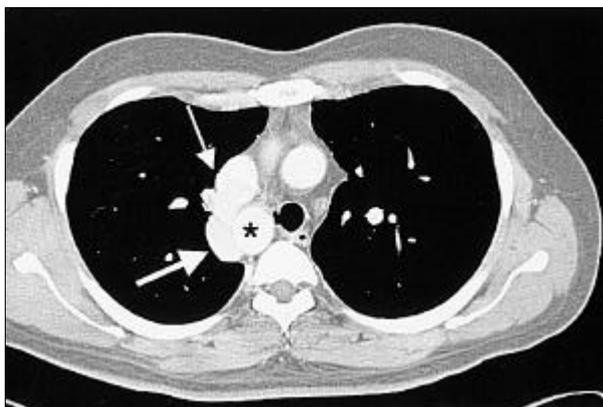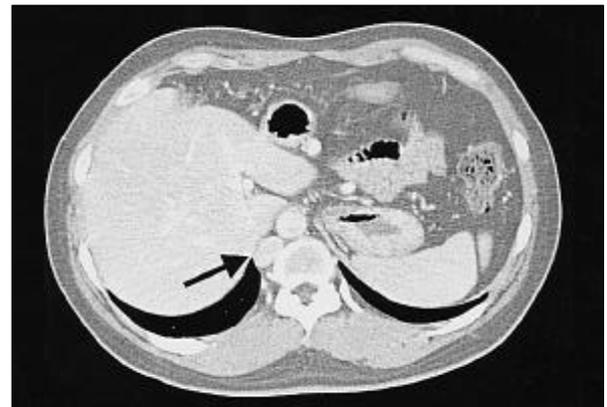

Fig. 4. CT scan through the abdomen below the diaphragm demonstrates the absence of inferior vena cava and large azygos vein (arrow) to the right of the abdominal aorta.
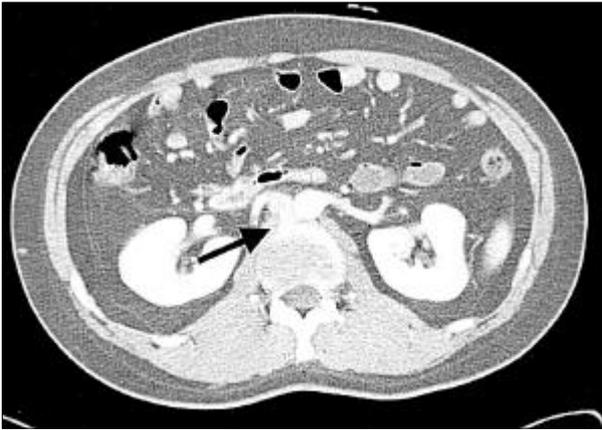
Fig. 5. At the level of the renal veins, IVC shows atrophic change and venous flow drains into the azygos vein (arrow).

venous blood returned from both renal veins and the lower half of the body was drained into the azygos vein (Fig. 4, Fig. 5, arrow). This may partially explain the very purpose of the enlarged azygos vein: the enlargement seems to have served as a compensatory mechanism against hemodynamic changes resulting from IVC malformation. The distal part of the IVC appeared completely normal. Considering the absence of obstruction of the IVC or other conditions caused by acquired enlargement of the azygos veins, a diagnosis of retrohepatic IVC interruption with azygos continuation was made. To evaluate combined congenital cardiac and visceral anomalies, such as situs inversus or polysplenia, echocardiographic examination and spleen scan were performed. Echocardiographic examination demonstrated no anomaly and spleen scan showed no evidence of polysplenia. Anticoagulation with intravenous unfractionated heparin and oral warfarin was started. Swelling of the right leg and dyspnea were gradually relieved within 3 days. Therefore, he was discharged and was followed for 15 months without recurrence of symptoms. Plans were made to put this patient on a life-long oral anticoagulation treatment.

DISCUSSION

The IVC is formed by a complex embryological process between the sixth and tenth weeks of gestation.[1,2] Three pairs of primitive veins (postcardinal, subcardinal, supracardinal veins) appear in orders, develop extensive anastomosis among themselves and undergo atrophy, resulting in subsequent development of the normal IVC. The normal IVC is converted to a unilateral, right-sided system consisting, from cauda to cranium, of the postrenal, renal, prerenal, and hepatic segments. Improper completion of the process may result in several types of IVC anomalies. Such anomalies occur in fewer than 1% of patients,[18] but the incidence is higher in patients with congenital heart diseases.[11] Fifteen types of IVC anomalies have been reported, many of which are minor variations.[1,12] The most common IVC anomalies are retroaortic and circumaortic left renal veins. IVC anomalies usually have been considered of little clinical significance. However, four types of IVC anomaly, with an incidence of 0.2 - 3%, are clinically important: duplication, transposition, retroaortic and circumaortic left renal vein.[12] Interruption of the IVC with azygos/ hemiazygos continuation is one IVC anomaly derived from the subcardinal veins.[2,4-6] When the right subcardinal vein has failed to make its connection with the hepatic sinusoids, the blood from the lower half of the body will return by the azygos veins, as in our case. In such case, the azygos veins undergo compensatory enlargement.[11] Interruption of the IVC with azygos/ hemiazygos continuation is associated with congenital cardiac or visceral malformations.[11,18] Common cardiac defects include dextrocardia, atrial septal defects, atrioventricular canal, pulmonary stenosis or any combination of these. There is a frequent association with transposed abdominal viscera, dysgenesis of lung and polysplenia. These associated anomalies were not found in our patient. Anomalies become apparent in infants when combined with heart or visceral malformations. However, the patients without these associated malformations remain asymptomatic because the deep venous collateral system is sufficiently developed and drains the venous blood from the lower extremities to the heart.[8-12] In adults, IVC anomalies are often reported as a fortuitous finding during radiologic work-up or laparotomy. The azygos continuation can masquerade as an aortic dissection or a mediastinal

mass.[11] Moreover, the enlarged azygos arch can be mistaken for a right paratracheal adenopathy on the chest radiograph.[19] We were able to make the correct diagnosis by means of CT scan, which has been accepted as a valuable modality for demonstrating IVC anomalies.[14] Although interruption of the IVC with azygos continuation is usually an asymptomatic malformation, a dozen cases of DVT have been causally linked to IVC anomaly in the English literature.[4-15] Theoretically, this anomaly may predispose to venous thrombosis because an inadequate blood return through the collaterals may increase the venous blood pressure in the veins of the leg, thereby favoring venous stasis.[4,6,11,17] Venous thrombosis is a serious medical problem causing considerable morbidity and mortality. The incidence of DVT in Western populations is estimated at one case per 1000 patient-years.[20] A multifactorial etiology involving both acquired and genetic factors interacts dynamically in the pathogenesis of DVT. Acquired risk factors are surgery, trauma, pregnancy, tumor and oral contraceptive. Genetic risk factors include deficiencies of protein C, protein S, and antithrombin III, although these were found only in about 5 to 10% of cases.[20,21] In more than 80% of patients presenting with DVT, a risk factor can be identified. Whereas the majority of previously described congenital conditions predispose to DVT by inducing a hypercoagulable state, IVC anomaly represents an unusual example of a congenital condition that predisposes to DVT by inducing venous stasis.[13] Two recent reports by Chee et al.[13] and Ruggeri et al.[16] confirmed the role of congenital IVC anomaly as a strong predisposing factor for the development of DVT in young adults. According to their reports, IVC anomalies were present in about 5% of cases of DVT in young patients. Furthermore, their data showed that bilateral DVT comprised a significant proportion of cases associated with IVC anomaly. Previously, underlying malignancy was the main condition associated with bilateral DVT, but it is now apparent that anomaly of IVC may underlie this clinical presentation and this should be born in mind when investigating these patients. In the case of IVC interruption, surgical intervention is seldom indicated. Recently, a case has been reported of a patient who did not respond suf-

ficiently to conservative treatment. A prosthetic bypass from the external iliac to the intrathoracic azygos vein was performed with complete symptomatic relief.[22] In our patient, anticoagulant-therapy alone was enough to relieve the symptoms. Considering that this anatomical anomaly is surgically uncorrectable, we decided to continue life-long, oral anticoagulation. Because patients with both vascular anomaly and thrombosis may be at higher risk for thrombotic recurrence, they should be advised to avoid additional risk factors, such as unusual physical exertion, prolonged immobilization, and oral contraceptive use.[17] In summary, the present case emphasizes that in patients with DVT apparently not associated with classic predisposing factors, complete diagnostic evaluation should be performed in order to verify any developmental anomalies of the IVC.

ACKNOWLEDGEMENTS

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