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Inferior Vena Cava Agenesia and its Association with Venous Thromboembolism: A Case Report

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Abstract

The agenesis of the Inferior Vena Cava (IAVC) is a rare vascular abnormality whose prevalence is 0.03-0.05% in the general population. It is found often in young patients (under 30 years) with proximal, typically bilateral, deep venous thrombosis (DVT) of the lower limbs, often in the absence of apparent risk factors. This cardiovascular defect is probably underestimated and the best therapeutic strategy as well as the optimal duration of anticoagulant treatment of DVT is still unclear. We described a case of recurrent iliac-femoro-popliteal DVT, not complicated by pulmonary embolism (PE), apparently of an idiopathic nature, in a healthy young man with AIVC, referred to our Department.

Keywords: Pulmonary embolism; Deep venous thrombosis; Thrombophilia

Introduction

Deep Venous Thrombosis (DVT) in young adults is considered idiopathic if a common cause of venous thromboembolism (VTE) such as recent surgery, pregnancy, immobilization, recent hormone therapy, thrombophilia or cancer is not identified quickly [1].

The congenital defects of the inferior vena cava, such as the absence or dysgenesis can be considered among the venous malformations responsible for cases of idiopathic VTE [2].

These anatomical defects, in particular the agenesia (IVCA) and hypoplasia of this vessel, may induce anomalies of the drainage of the inferior portion of the sub-diaphragmatic venous system. They may be due to an embryogenic alteration that occurs during the sixth-tenth week of embryonic life [2].

In the presence of IVCA the pathophysiological components of the "Virchow's triad" (flow stasis, parietal lesions and hypercoagulability for any cause) would act consistently as contributing factors to the development of DVT in these subjects, also in absence of traditional conditions or risk factors associated to DVT.

The knowledge of these vascular malformations turns out to be more and more fundamental in order to avoid errors in decision making not only from a diagnostic point of view, but also for a correct approach to the duration of anticoagulant treatment.

The IVCA is rare (0.3-0.5% of the general population, and 0.6-2% of patients presenting with congenital cardiovascular defects), while in clinical practice these defects associated to DVT are not so rare [3-4]. Usually they present at our outpatients clinics as idiopathic proximal DVT events involving the iliac, femoral, and popliteal veins. However, in general, these patients do not perform a routine haemodynamic evaluation of the caval district and for this reason the anatomical defect is often underdiagnosed.

Patient and Methods

We therefore present a case of recurrent iliac-femoro-popliteal

DVT, not complicated by pulmonary embolism (PE), apparently of an idiopathic nature, in a healthy young man with AIVC, referred to our Department (Angilogy Unit, Avellino, Southern Italy).

Case Report

A young 15-year-old male patient presented to our observation for swelling of the left lower limb which arose suddenly for about two days, in the absence of recent trauma. He reported occasional consumption of cannabis and beer, but not frequent use of cigarette smoking. He did not refer recent hypomobility or surgery, not known oncological diseases. Arterial blood pressure was normal (125/80 mmHg), heart rate was 82 bpm, he was eupnoic with SpO2: 98%. The body weight was 102 kg, height: 180 cm and the BMI was 31.5 kg/m².

Laboratory evaluation showed increased inflammation markers and D-dimer levels [erythrocyte sedimentation rate 50 mm/1 h, C-reactive protein 1 mg/dl (normal values <0.5 mg/dl), D-Dimer: 5 mcg/ml (normal values less than 0.5 mcg\dl), while hemoglobin, platelets, creatinine levels and liver tests were normal.

A color-Doppler ultrasonography (US) of the lower limbs was performed and revealed the presence of proximal DVT involving the external iliac vein and the common and superficial ipsilateral femoral vein. Anti-thrombotic therapy was started with low molecular weight heparin (LMWH) at therapeutic doses (enoxaparin 100 UI/kg body weight twice daily).

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Considering the patient's young age, a screening for inherited thrombophilia was also carried out, which showed the presence of heterozygosis of the H1299R mutation of Factor V.

The abdominal US examination showed the presence of hypo echoic nodular formations, of uncertain significance, in the para-aortic area and along the iliac-femoral axis. The total body CT scan showed the presence of diffuse abdominal and inguinal lymphoglandular tumefactions. A subsequent PET-CT with FDG clarifies the framework by deploying for reactive lymphadenopathy.

Oral anticoagulant therapy with vitamin K antagonists (VKA) and compression stockings (CS) were prescribed. Instrumental and hematological follow-up was planned. Subsequent echo-color-Doppler examinations showed poor recanalization of the proximal DVT and therefore anticoagulation with VKA was prolonged.

Three years later, following a voluntary suspension of VKA, the patient was hospitalized for contralateral thrombotic recurrence. The physical examination showed the presence of swelling of the right lower limb and the presence of Caput Medusae at the abdominal level (Figure 1). The echo-color-Doppler examination of the lower limbs confirmed the right proximal DVT with the presence of several collateral veins at the pelvic level. At the US examination of the abdominal vessels there was no evidence of inferior Vena Cava, presence of collateral veins in its seat and in the perirenal site, at the level of the abdominal wall and of the pelvis. A CT-scan of the whole body was performed by which it was possible to diagnose Agenesia of the whole inferior vena cava up to the sub-hepatic tract, with the presence of vicious venous circles through a network of small venous paraxial vessels, through the azygos and emiazygos and opening of collateral circuits of the abdominal wall (Figure 2). To date, after almost ten years since the first episode, the clinical and instrumental picture, despite anticoagulant and compression therapy, has not been improved, with evidence of a postthrombotic syndrome (PTS).



Figure 1: The presence of swelling of the right lower limb and the presence of Caput Medusae at the abdominal level.



Figure 2: CT-scan show the Agenesia of the whole inferior vena cava up to the sub-hepatic tract, with the presence of vicious venous circles through a network of small venous paraxial vessels, through the azygos and emiazygos and opening of collateral circuits of the abdominal wall.

Results

In consideration of our experience and literature data, it was possible to trace an identikit of the patient presenting DVT associated with IVCA, outlining its main characteristics.

First of all we could observe that usually DVT occurs as proxymal DVT in young males, in the absence of risk factors. The clinical context in which IVCA with DVT may occur shows many delays in diagnostic and therapeutic approach; frequently the suspicion of a neoplastic pathology could be posed, given the presence of vicarious vascular structures, that may simulate a lymphoma.

The thrombotic event occurs recurrently and, despite anticoagulant therapy, there are few signs of ultrasound vascular recanalization, with vicarious collateral circulation and a severe PTS. Moreover, these episodes, although extensive and massive, rarely cause PE, and this is another aspect that may induce the suspect of IVCA [5].

Discussion

The aim of this work is not only to confirm the literature data, but to focus on the diagnostic accuracy for an early recognition and evaluation of this pathology. In our experience we have been able to consider the characteristics of suspicion that can help us to discriminate, among the patients suffering from DVT, those who can have IVCA and in this way submit them to diagnostic investigations aimed at the search for this malformation.

In addition to their young age and male gender, these patients present with the objective Caput Medusae examination (following the activation of collateral circulation). They are generally sturdy and have a well-represented fat component, especially on the thighs. They are patients in whom the thrombotic event often does not manifest itself in a striking way, but mimicking other pathologies especially in the abdominal area. Citation: Cannavacciuolo F, Allegorico E, Mangiacapra S, Vivolo S, Di Micco P, et al. (2018) Inferior Vena Cava Agenesia and its Association with Venous Thromboembolism: A Case Report. J Blood Lymph S1: 003. doi:10.4172/2165-7831.S1-003

The diagnostic suspicion must be further insinuated when the patient has a history of recurrent episodes of DVT in the lower limbs and shows poor venous recanalization despite anticoagulant therapy, and not associated to classic pro-thrombotic conditions, including thrombophilia. However for such cases the role of inherited molecular thrombophilia is still unclear [6].

The association between IVCA and Factor V Leiden mutation (FVL) or other inherited thrombophilic conditions certainly increases the risk of DVT, so the possibility that the pro-thrombotic inherited condition may lead to caval thrombosis cannot be ruled out. In literature, the FVL mutation is reported in 29/161 patients (18%) [7,8].

Another key point is the intensity of anticoagulant therapy and its duration. In the literature there are no specific evidence concerning anticoagulant therapy in cases of Vena Cava malformations complicated by DVT, because of the rarity of this condition and the absence of clinical data indicating the rate of recurrence with or without an extended treatment. There are also discordant opinions about the use of more invasive procedures, such as thrombolysis or mechanical thrombolysis or thrombectomy, to be performed early, compared to standard drug therapy [9].

Furthermore, little is known about the role of all reported treatments and the development of PTS that is more serious and disabling for patients with IVCA and lower limb DVT, because of the delay in the diagnosis and in the therapeutic approach.

Conclusion

Although not considered among the main causes of DVT, vascular malformations of the inferior vena cava should always be sought, especially in the absence of other risk factors. In this context, vascular diagnostics is increasingly one of the main perspectives for the definition of VTE; in particular, it guarantees, if performed quickly and early, not only an accurate diagnosis, but above all a therapeutic efficacy. Future studies are necessary to define: the role of inherited thrombophilia in IVCA, the optimal duration and type of anticoagulation (warfarin or direct oral anticoagulants-DOACs), the possibility of specific treatments such as thrombolysis, guided catheter thrombectomy and the right approach to PTS.

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