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Inferior Vena Cava Agenesis associated with Deep Venous Thrombosis: Case Report

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ABSTRACT

Inferior vena cava agenesis is a rare congenital venous malformation, with a prevalence ranging from 0.0005 to 1% of the world's population. The present study aims to describe a case of agenesis of the inferior vena cava associated with deep vein thrombosis (DVT) in a young patient and the difficulty in maintaining adequate anticoagulation. We report the case of a 22-year-old woman who had her first thrombotic event as a child, in the renal vein, and was anticoagulated. At age 20, she had a thrombotic event in the iliac vein and went back on anticoagulation, but with adequate control of coagulation, the patient decided when to stop and return to anticoagulation without medical advice. The diagnosis of agenesis was confirmed by tomography and phlebography. Currently, she was maintained on anticoagulation and warned about the risks of the disease and inadequate anticoagulation.

Keywords: Agenesis, anticoagulation, deep venous thrombosis, diagnosis, inferior vena cava.

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I. Introduction

Inferior vena cava agenesis is a rare congenital venous malformation, with a prevalence ranging from 0.0005 to 1% of the world's population [1]. Most of the time it is asymptomatic, being an incidental finding of imaging tests or surgical procedures [2]. Patients with this condition remain asymptomatic as long as venous drainage via collateral circulation is functional, but symptoms may begin to develop when there is associated prothrombotic risk factors [3].

It is estimated that this condition is found in about 5% of patients under 30 years of age who have idiopathic deep vein thrombosis (DVT) [4]. However, this prevalence is possibly underestimated, since inferior vena cava agenesis cannot be diagnosed in imaging tests that are commonly performed in patients with DVT, such as venous Doppler ultrasonography [1], [4].

DVT is a multifactorial disease, characterized by the acute formation of thrombi in deep veins, associated with factors that promote hypercoagulability, blood stasis and endothelial injury [5]. There is evidence demonstrating that patients with agenesis of the inferior vena cava are more likely to develop DVT in the lower limbs earlier, possibly due to the tendency towards venous stasis in the lower limbs [2], [6] in which venous return depends on a system formed by collateral. Thus, the present study aims to describe a case of agenesis of the inferior vena cava associated with DVT in a young patient and the difficulty in maintaining adequate anticoagulation.

II. Case Report

A 22-year-old female patient reports having discovered a diagnosis of agenesis of the inferior vena cava in 2020, after an episode of deep venous thrombosis in the external iliac vein on the right. When questioning his mother about this condition, he reports having discovered that, as a child, he had an episode of renal vein thrombosis (he does not know exactly when), having used Marevan (sodium warfarin) until he was 6 years old, at a dose of 2.5 mg/day.

In September 2020, the patient had an episode of right external iliac vein thrombosis, resuming regular use of sodium warfarin at a dose of 5 mg/day, interrupting it when pregnancy was discovered in September 2021. During pregnancy, she underwent prenatal care in the service and denies any intercurrence. She used enoxaparin 120 mg/day from September 2021 to May 2022 (up to one month after

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cesarean delivery). In May 2022, then, she returned with the regular use of Marevan at a dose of 5 mg/day, discontinuing the medication in August 2022 on her own, remaining without the use of oral anticoagulation for almost 3 months. In November 2022, she returned to the medication at a dosage of 5 mg/day and has been presenting for 2 weeks with sudden swelling of the right lower limb, associated with pain in the inguinal region and paresthesia. She also mentions darkened lesions on the limb, which improved spontaneously and episodes of sudden dyspnea, the last of which occurred 1 week ago. He denies fever, denies cough, denies recent travel, denies local trauma and other symptoms.

On physical examination, vesicular murmurs were present bilaterally, without adventitious sounds, 99% saturated in room air, 2 rhythmic and normophonetic sounds, without audible murmurs, with a heart rate of 67 bpm. Edema of the right lower limb 1+/4+ up to the supra genicular region, with a negative Locker sign. Shows positive Homans sign, flag sign and Moses sign with new thrombotic event. The diagnosis of agenesis was performed by tomography and phlebography, as shown in images, Fig. 1 to 4.

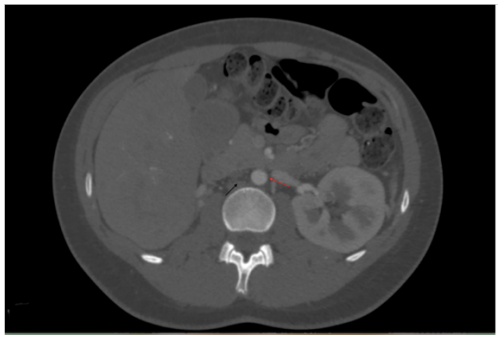

Fig. 1. Axial tomography section showing absence of inferior vena cava, indicated by the arrow the site where the vein should be located. The absence of the right kidney is also observed.

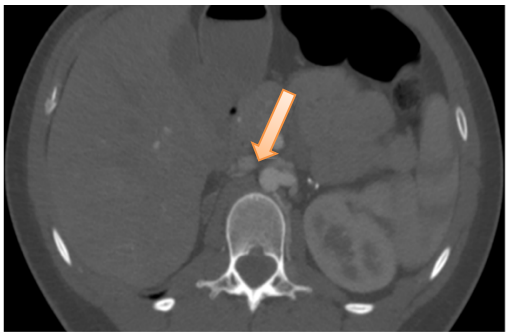

Fig. 2. Tomographic sections in the armpit indicating the renal vein left (arrow) originating from the suprahepatic vein.

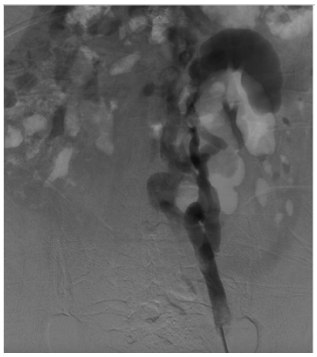

Fig. 3. Phlebography showing contrast being injected through the left iliac vein contrasting the plexus of lumbar veins without contrast inferior vena cava.

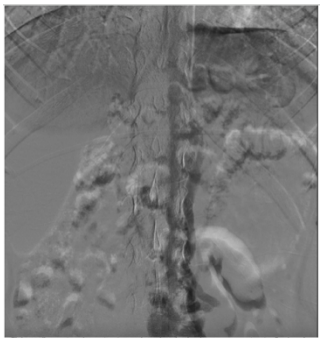

Fig. 4. Phlebography showing contrast in the plexus of lumbar veins without contrast. Inferior vena cava.

III. Discussion

The present study reports a case of agenesis of the inferior vena cava that had renal vein thrombosis as a child and thrombotic events in the lower limbs over time. The literature reports that in most cases the diagnosis occurred as an incidental finding of imaging tests or surgical procedures [2]. In the present case, it was not in the first thrombotic event of the renal vein, as a child, that she had the diagnosis of agenesis of the inferior vena cava.

What brings as an alert in this case is the lack of adequate information to the patient over these years and the lack of control in the way he used warfarin. She decided to stop and resume medication without medical advice, increasing the risk of a new thrombotic event. She had an uneventful pregnancy but was monitored by the gynecologist during this period and used adequate anticoagulants. The recurrence of thrombotic events suggests perennial maintenance of anticoagulation where the use of warfarin requires constant control of the coagulogram. The use of new oral anticoagulants would be an option for this patient, but the economic aspect limited this possibility of conduct.

Another aspect that brings a warning is regarding the investigation of congenital thrombophilias in these patients and, in their absence, the alert to investigate venous agenesis and other thrombogenic causes such as anti-cardiolipin and anti-phospholipid antibodies and neoplasms [5]. Thrombotic events should always draw attention to a specific cause of the thrombotic event. The main complication of DVT is pulmonary embolism and the chronic one is post-thrombotic syndrome. The main objective of the present study is to raise awareness regarding the diagnostic and therapeutic orientation of these patients where the control of anticoagulation is up to the physician.

IV. Conclusion

The agenesis of the inferior vena cava in this patient shows the difficulty of accurate diagnosis and adequate anticoagulation, suggesting that these patients have adequate awareness regarding their diagnosis, treatment, and intercurrences.

Conflict of Interest

Authors declare that they do not have any conflict of interest.

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