

ESPECTRO DE APRESENTAÇÃO CLÍNICA DE UMA MALFORMAÇÃO VENOSA RARA: AGENESIA DA VEIA CAVA INFERIOR

DIFFERENT CLINICAL PRESENTATIONS OF A RARE VENOUS MALFORMATION-INFERIOR VENA CAVA AGENESIS

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RESUMO

INTRODUÇÃO: A agenesia da veia cava inferior é uma das anomalias mais raras deste vaso, com uma prevalência estimada de 0,0005–1% na população geral. Porém, cerca de 5% dos pacientes com menos de 30 anos com uma trombose venosa profunda apresentam esta anomalia.

MÉTODOS E MATERIAL: Relato de dois casos clínicos de agenesia da veia cava inferior com diferentes apresentações clínicas.

CASO CLÍNICO 1: Um homem de 40 anos recorreu à urgência com queixas de edema e dor no membro inferior esquerdo, progredindo gradualmente para incapacidade de marcha. Ao exame físico foi encontrado edema e dor a palpação da perna e coxa, bem como veias colaterais abdominais superficiais proeminentes. O ecodoppler venoso mostrou trombose venosa profunda esquerda estendendo-se da veia poplítea até a veia íliaca comum. O angio-TC mostrou agenesia da veia cava infrarrenal e veias renais a drenar para os sistemas ázigos e hemiázigos. O paciente teve alta com rivaroxabano e meias elásticas. Aos 2 meses mantinha-se hipocoagulado e estava assintomático.

CASO CLÍNICO 2: Uma mulher de 35 anos, com história prévia de cirurgia de varizes de membros inferiores recorrentes e úlcera do maléolo interno esquerdo, apresentou-se na consulta com recorrência da úlcera. O angio-TC revelou ausência de veia cava infra-hepática, veias ázigos e hemiázigos dilatadas com colaterais retroperitoneais dilatadas. A paciente iniciou cuidados de penso com óxido de zinco e rivaroxabano oral.

DISCUSSÃO / CONCLUSÃO: A maioria dos pacientes com agenesia da veia cava inferior permanece assintomática. No entanto, quando sintomática, a maioria apresenta-se com uma trombose venosa profunda proximal envolvendo as veias íliaca e femoral. A TC ou RM devem ser os métodos imagiológicos usados para diagnosticar esta anomalia. Nenhum consenso claro foi alcançado sobre a estratégia terapêutica, além da anticoagulação de longo prazo e uso de meias elásticas.

Palavras-chave

Agnesia da Veia Cava Inferior; Trombose venosa profunda; Anomalias vasculares

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ABSTRACT

INTRODUCTION: *Inferior vena cava agenesis is one of the most uncommon anomalies of this vessel, with an estimated prevalence of 0.0005–1% in the general population. However, around 5% of the patients younger than 30 years with a diagnosis of deep vein thrombosis have this anomaly.*

METHODS AND MATERIAL: *Report of two clinical cases of inferior vena cava agenesis with different clinical presentations.*

CLINICAL CASE 1: *A 40-year-old man was admitted with a 3 days history of unilateral lower limb swelling and pain, gradually progressing to the inability to walk. On physical examination he had swelling, bruising and tenderness involving leg and thigh and prominent engorged superficial abdominal collateral veins.*

Venous Doppler Ultrasound showed left deep venous thrombosis extending from popliteal vein to common iliac vein. A computed tomography angiogram showed agenesis of the infrarenal vena cava and patent renal veins draining in to the azygous system and hemiazygous systems.

The patient was discharged with rivaroxaban and compression stockings. At 2 months follow up he was asymptomatic and still anticoagulated.

CLINICAL CASE 2: *A 35 year- old woman, with a previous history of recurrent lower limb varicose veins surgery and left internal malleolar ulcer at 30 years presented at medical department 3 years later with complains of ulcer recurrence. A computed tomography angiogram revealed an absent infrahepatic vena cava, prominently dilated azygos and hemiazygos veins with enlarged retroperitoneal collaterals.*

The patient initiated dressing care with oxide zinc and oral rivaroxaban.

DISCUSSION/CONCLUSION: *The majority of cases remain asymptomatic. However, when symptomatic, the majority present as proximal DVT involving the iliac and femoral veins. CT or MRI should be the imagiological methods used to diagnose this anomaly. No clear consensus has been reached on therapeutic strategy, other than long-term anticoagulation and elastic stockings.*

Keywords

Inferior Vena Cava Agenesis; Deep Vein Thrombosis; Vascular anomalies

INTRODUCTION

Inferior vena cava (IVC) formation is a complex process that occurs during embryogenesis (6–8 weeks of gestation) by development, regression, and anastomosis of three sets of paired veins: the posterior cardinal, subcardinal, and supracardinal veins. Usually, the IVC is then converted to a unilateral right-sided system consisting of the infra-renal, renal, supra-renal and hepatic segments⁽¹⁾.

Inferior vena cava agenesis (IVCA) is one of the most uncommon anomalies of this vessel, with an estimated prevalence of 0.0005–1% in the general population⁽²⁾. IVCA can be congenital, occurring in utero due to abnormal formation of the paired veins or prenatal thrombosis, or it can occur due to a postnatal thrombosis in early life^(3,4). IVCA may be found as two different variants, complete or segmental agenesis⁽³⁾. The most common agenesis is the suprarenal segment, while agenesis of the hepatic and infrarenal segment constitutes only 6% of all cases⁽²⁾.

Congenital risk factors are rare as cause of DVTs, but IVCA is emerging as a significant risk factor that may contribute to DVT in healthy, young adults⁽⁵⁾.

Around 5% of the patients younger than 30 years with a diagnosis of deep vein thrombosis (DVT) have an IVCA⁽²⁾ and so in young patients with idiopathic deep venous thrombosis, an inferior vena cava anomaly should be considered⁽⁶⁾. Physical exertion has been identified as a risk factor for DVT in patients with IVCA and as a precipitating event of the clinical presentation⁽⁷⁾, because the collaterals developed before DVT are unable to assure the increased blood flow due to major physical exertion, thereby generating venous stasis and clotting⁽⁴⁾.

METHODS AND MATERIAL

Report of two clinical cases of vena cava agenesis with different clinical presentations.

CLINICAL CASE 1

A 40-year-old man was admitted with a 3 days history of unilateral lower limb swelling and pain, gradually progressing to the inability to walk. The patient had never had these symptoms before and had no family history of thrombosis. On questioning he referred a history of extreme physical activity the previous days. He had a negative medical or surgical history and was not taking any medication. On physical examination, was found swelling, bruising and tenderness involving leg and thigh. Acute prominent engorged abdominal superficial collateral veins were also seen. Venous Doppler Ultrasound (VDU) showed left DVT extending from popliteal vein to common iliac vein. A CT showed agenesis of the infrarenal segment of the IVC and patent renal veins draining in to the azygous system on the right side and hemiazygous on the left side (FIG. 1 AND 2).

The patient was discharged with rivaroxaban and compression stockings. At 2 month follow up the patient was still anticoagulated and asymptomatic.

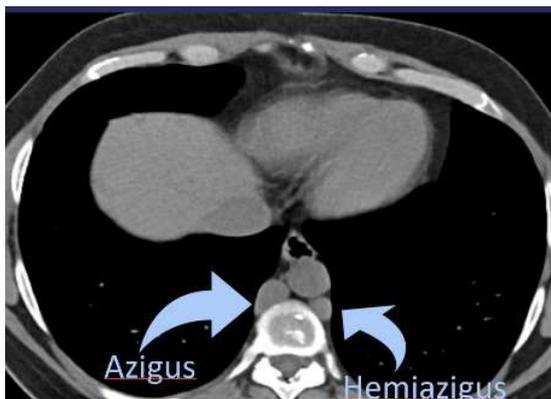


Figure 1 IVC not visible at suprarenal level; Ectasia of azygos and hemiazygos system



Figure 2 IVC not visible at renal level

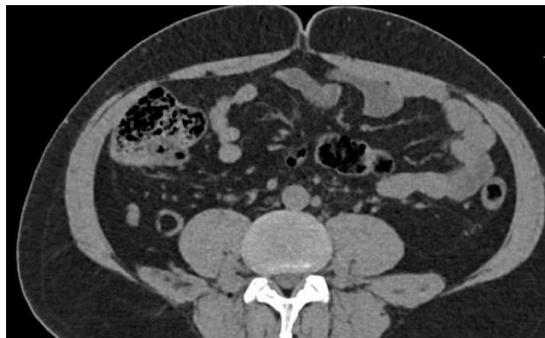


Figure 3 IVC not visible at infra renal level

CLINICAL CASE 2

A 35 years old woman, with a previous history of recurrent lower limb varicose veins surgery and left internal malleolar ulcer presented at medical department with complains of ulcer recurrence. She had no other medical or surgical history and was not taking any medication. On physical examination she had a left internal malleolar ulcer without varicose veins. A CT was order and revealed infra-renal, renal and suprarenal, IVC agenesis with preservation of the intrahepatic segment and exuberant collateralization (lumbar venous plexus draining into azygous and hemiazygous systems; epigastric and internal mammary veins of right predominance). (FIGURE 3, 4 AND 5)

The patient initiated dressing care with oxide zinc and oral rivaroxaban. During the follow-up she developed a right popliteal DVT, after inadvertent anticoagulation discontinuation, having restarted hipocoagulation.

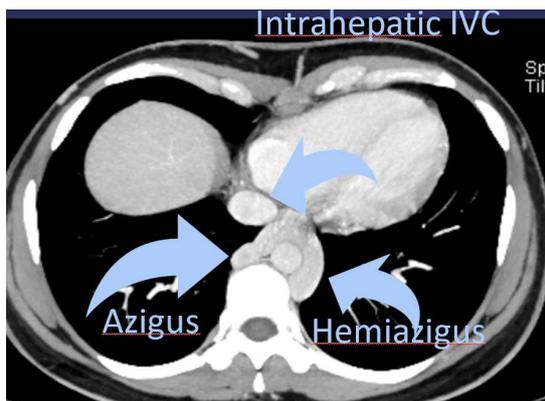


Figure 4 Ectasia of azygos and hemiazygos systems

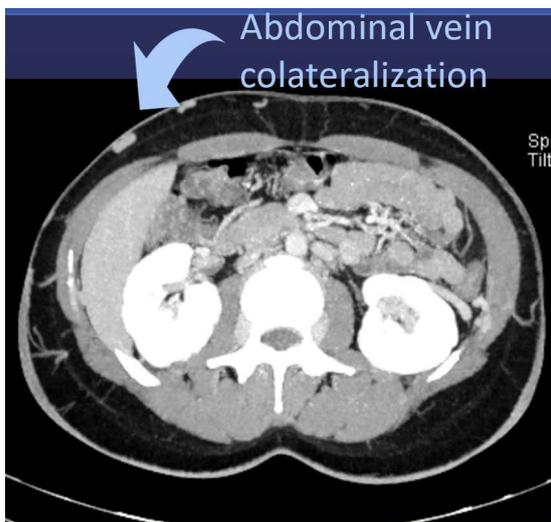


Figure 5 Extensive superficial abdominal veins collateralization (blue arrow)

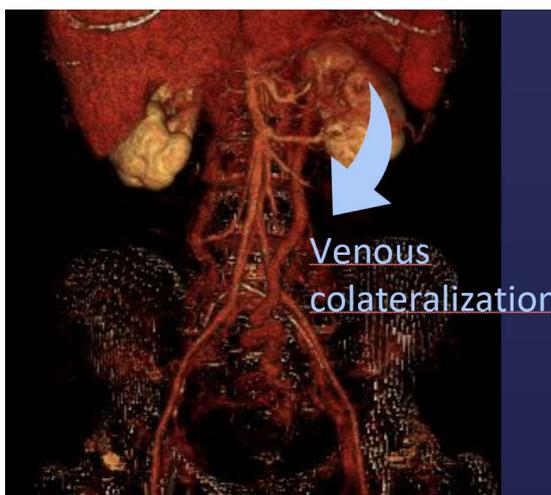


Figure 6 Extensive venous collateralization (blue arrow)

DISCUSSION

Some common features of patients with IVCA are young age (mean age of 30 years) and male gender⁽²⁾. The majority of patients with IVCA remain asymptomatic because of the extent of collateral venous network in the abdomen and lower extremity (azygous, hemiazygous, lumbar, para-vertebral, and abdominal wall veins)⁽²⁾. However, as we observed in clinical case 1, there will always be venous stasis compared to the general population⁽²⁾ and so, when symptomatic, the majority of cases present clinically as proximal DVT⁽⁷⁾. The thrombosis preferentially involves the iliac and femoral veins, often bilaterally, and usually occurs

after exposure to a precipitating factor (physical trauma or injury, prolonged immobility, use of birth control pills)⁽⁹⁾. Pulmonary embolism is a rare complication of DVT in IVCA, probably due to the small diameter of the vessels leading to the pulmonary circulation in the absence of the IVC⁽⁶⁾. However there are some reports of pulmonary embolus associated with IVCA, the possible explanation being that enlarged azygous or hemiazygous veins could act as conduits for emboli to the pulmonary circulation. IVCA may also favor chronic venous insufficiency with development of ulceration as we observed in the clinical case 2⁽³⁾.

Ultrasonographic doppler scanning is usually the first imaging modality in the evaluation of patients with DVT, but anomalies of the IVC may be missed⁽⁶⁾. All young patients presenting with idiopathic DVT should be investigated for inferior vena cava anomalies with CTA or MRI if ultrasound does not visualize the inferior vena cava⁽⁷⁾. Imaging criteria for IVCA diagnosis were the absence of the IVC lumen, associated with evidence of connection of the existent caval segment to the azygos system, and identification of venous collaterals (paravertebral venous system and its communications with the ascending lumbar veins and azygous–hemiazygous systems; gonadal, periureteral and other retroperitoneal veins; abdominal wall veins; hemorrhoidal venous plexus; and the portal venous system). It is possible to differentiate between an old thrombosis and IVCA: venous collaterals can sometimes be found in both entities, so the caval lumen should always carefully be sought (missing caval lumen in IVCA, while the lumen of an old thrombosis is usually narrowed, resembling a hypotrophic vessel)^(4,8). Venography is the gold standard for imaging diagnosis, but it is also highly invasive⁽⁹⁾.

Additional congenital anomalies have been associated with IVCA and involved aplasia/hypoplasia of the right kidney, situs inversus, congenital heart diseases, polysplenia and asplenia⁽⁷⁾.

Optimal treatment of DVTs associated with IVCA is unclear. The rarity of IVCA precludes performing clinical trials to determine the best treatment strategy and case series are likely the most informative⁽⁷⁾.

Once diagnosed, aggressive treatment must be started because of the high risk of post-thrombotic syndrome, but the length of treatment with vitamin K antagonist and the use novel oral anticoagulants are a matter of debate. A 2010 case series, the largest to date, compiled a list of 72 patients with IVCA-associated DVT. Nearly all of these 72 patients were treated with long-term warfarin but the length of

anticoagulation varied. Considering the irreversible risk for future thrombosis, lifelong anticoagulation may be necessary^(10,4) Elastic stockings support and leg elevation should be also suggested, as well as avoidance of additional risk factors such as prolonged immobilization and oral contraceptive use⁽⁹⁾.

In some cases, venous recanalization by thrombolysis can be a good alternative particularly in the younger patients because of their higher risk of long lasting disability.

If medical treatment fails venous bypass graft/stenting should be considered. Venous bypass graft from the common iliac vein to the azygous vein or proximal cava segment is considered to prevent DVT or to heal the ulcers if conservative treatments fails⁽¹⁰⁾.

CONCLUSION

IVCA is a rare congenital abnormality that should be suspected when confronted with a typical clinical pattern, particularly a young patient [$< 30/40$ years old], especially males, with proximal DVT after intense physical exertion. Ultrasonography is not useful for IVCA diagnosis but CT scans and/or MRA can provide the diagnosis. Prolonged, perhaps even life-long anticoagulation, is usually prescribed along with elastic stockings.

REFERENCES

1. Man L.; Hendricks N.; Maitlan H. IVC agenesis: a rare cause of deep vein thrombosis. *J Thromb Thrombolysis* 2015 Oct.
2. Gil R., Pérez A., Arias J., Pascual F., and Romero E. Agenesis of the inferior vena cava associated with lower extremities and pelvic venous thrombosis. *J Vasc Surg* 2006; 44:1114-6.
3. Pomeranz C., Cullen D., Bellah R. Deep venous thrombosis in a child with inferior vena cava and renal anomalies: KILT syndrome. *Pediatric Radiology*; 11 January 2018
4. Lambert M., Marboeuf P., Midulla M., Trillot N., Beregi J., Mounier-Vehier C., Hatron P. and Jude B. Inferior vena cava agenesis and deep vein thrombosis: 10 patients and review of the literature. *Vascular medicine* 15(6) 451-459, 2010.
5. Singh K., Poliquin J., Syversten G., Kohler D. A rare cause of venous thrombosis: Congenital absence (agenesis) of the inferior vena cava. *int J angiolog* 2010;19(3):e110-e112.
6. Aranda J., Cerda C., Mussali S., Flores J. Inferior Vena Cava Agenesis: An Unusual Cause of Deep Vein Thrombosis and Pulmonary Embolism in Young Adult Patients. *EJVES Short Reports*, 2018 39 12e15.
7. O'Connor D., O'Brien N., Khani T., Sheehan S. Superficial and Deep Vein Thrombosis Associated With Congenital Absence of the Infrahepatic Inferior Vena Cava in a Young Male Patient. *Dublin, Ireland Ann Vasc Surg* 2011; 25: 697.e1-697.e4
8. Yang C, Trad HS, Mendonça SM, Trad CS. Congenital inferior vena cava anomalies: a review of findings at multidetector computed tomography and magnetic resonance imaging. *Radiol Bras.* 2013 Jul/Ago;46(4):227-233
9. Protti G, Elia F, Bosco F, Aprà F. An eminent absence: agenesis of inferior vena cava underlying bilateral iliac vein thrombosis. *EJCRIM* 2020
10. Sitwala P, Ladia V., Brahmhatt P., Jain V., Bajaj K. Inferior Vena Cava Anomaly: A Risk for Deep Vein Thrombosis. *North American Journal of Medical Sciences.* Nov 2014, Volume 6, Issue 11.