

A rare case of inferior vena cava agenesis associated with deep venous thrombosis

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ABSTRACT

Inferior vena cava (IVC) agenesis is an extremely rare malformation, most times diagnosed randomly, representing an important risk factor in the development of spontaneous deep venous thrombosis (DVT) in children and young adults.

We present the case of an 8-year-old male child admitted in our clinic for the following complains: right lumbar pain with paravertebral irradiation. The personal pathological history revealed a recent appendectomy. At the time of admission, an abdominal ultrasound was performed which revealed right nephromegaly, and also raised the suspicion of IVC agenesis. We performed a thoraco-abdominal angio-CT exam with contrast enhancement which confirmed the diagnosis of IVC agenesis and right common iliac vein, venous thrombosis of the right external iliac vein, right common femoral vein and right superficial femoral vein. We excluded a potential antiphospholipid syndrome or a thrombophilia, and the genetic tests ruled out the possibility of a family predisposition. We initiated anticoagulant therapy with close monitoring of the coagulation parameters, the evolution being favorable.

Despite the very low frequency of this malformation, the early and correct diagnosis is essential for these patients' management and prognosis.

Keywords: deep venous thrombosis, inferior vena cava agenesis, child

Abbreviations

IVC – inferior vena cava

DVT – deep venous thrombosis

PE – pulmonary embolism

INR – international normalized ratio

INTRODUCTION

The anomalies of inferior vena cava (IVC), such as agenesis of the infrahepatic segment of IVC or hypoplasia have a very low incidence, affecting approximately 0.5% of general population (1,2). IVC agenesis is an extremely rare malformation, most times discovered randomly, being frequently associated with other congenital malformations such as situs inversus, cardiac malformations, polysplenia or asplenia (1). The main causes that can lead to anomalies of the IVC consist in the perturbation of the embryonic development process of the venous

system or the perinatal venous thrombosis with the alteration of the venous system development (3).

IVC anomalies represent an important risk factor in the development of spontaneous deep venous thrombosis (DVT) in children and young adults (4).

The incidence of venous thromboembolism defined as DVT and/or pulmonary embolism (PE) increased significantly during the recent years being diagnosed in approximately 1/200 among the hospitalized pediatric patients (5,6).

DVT represent a rare pathology in pediatrics, its diagnosis representing a challenge for pediatricians

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due to the lack of specific clinical symptoms (7). Most of the children that develop this pathology present multiple risk factors due to the multifactorial etiology of DVT (8). The incidence of this condition increased significantly due to the survival of children with formerly known fatal pathologies, and due to the extremely advanced technologies that allow this fact. In children below the age of 15 years, the incidence is of approximately 5 in 100,000 cases (9). In the USA, this pathology owns a prevalence between 34 and 58 cases in 10,000 admissions (6,10).

Recent studies sustain the fact that ultrasonography by venous compression represents the first-choice diagnostic tool in case of DVT, offering the advantage of an easy accessibility, utility safety and reduced costs. Despite these facts, the gold standard in the diagnosis of DVT remains the angio-CT (9,11).

The drug therapy in case of DVT includes fondaparinux and direct inhibitors of thrombin, but low-molecular weight heparin or non-fractionated heparin are most frequently used (12).

We present a case of DVT associated with IVC agenesis in an 8-year-old male child with the aim of underlining the rarity of this pathology and the importance of an early, prompt and correct diagnosis.

CASE REPORT

Reasons of admission

We present the case of an 8-year-old male patient admitted in our clinic for the following complaints: right lumbar pain with paravertebral irradiation. The personal history revealed that approximately 7 days before the admission in our clinic, the patient underwent an appendectomy, with slowly favorable post-operative evolution due to the symptoms described above, raising the suspicion of a pyelonephritis. The family history was not relevant.

Clinical examination

At the time of admission, the clinical exam pointed out the following pathological elements: influenced general status, pallor, post-appendectomy scar in the right iliac fossa, painful abdomen at superficial and deep palpation in the right hypochondrium and right lateral side of the abdomen, painful right renal area.

Diagnostic assessment

The initial abdominal ultrasound pointed out right nephromegaly, the sizes of the right kidney of

approximately 118/49 mm, increased echogenicity of the cortical with the disappearance cortico-medullar differentiation, without the visualization of the IVC. Therefore, we raised the suspicion of IVC agenesis.

We performed a thoraco-abdominal angio-CT with contrast enhancement that confirmed the results of the abdominal ultrasound and completed it as it follows: inhomogeneous liver due to the presence in the 8 segment of a triangular area with perfusion disorders because of the thrombosis of the portal branch, thrombus in the portal vein close to the spleno-mesenteric confluent with the diameter of 7/4 mm, the right kidney increased in volume with mildly inhomogeneous nephrogram possibly due to the venous drainage that cannot be identified, absent inferior vena cava or possibly with thrombosis, the drainage of the left kidney through the left spermatic vein, and afterwards in the paravertebral veins, azygos and hemiazygos veins (Fig. 1, 2, 3).



FIGURE 1. Thrombosis of right iliac vein



FIGURE 2. Thrombosis of right renal vein, agenesis of inferior vena cava

The venous Doppler ultrasound pointed out the lack of the suprahepatic veins, inferior vena cava and right common iliac vein, the thrombosis of the right external iliac vein, uncompressible with hy-

perechoic images in the lumen, thrombosis of the right common femoral vein, uncompressible with hyperechoic image in the lumen, and thrombosis of the right superficial femoral vein.



FIGURE 3. Renal loading asymmetry with the persistence of the right kidney cortical retention of contrast

In order to rule out the diagnosis of antiphospholipid syndrome, we performed the following laboratory tests that came back normal: anti-beta 2 glycoprotein IgM antibodies (1,3U/ml), anticardiolipin IgM antibodies (1,2 MPL-U/ml), absent lupus anticoagulant. The genetic tests did not show any mutation of the factor V Leiden (A506G), or factor II (prothrombin gene G20210A). Also, in order to exclude a potential thrombophilia, we performed antithrombin III, von Willebrand factor, C and S proteins, and homocysteine, which were all within the normal ranges.

Therapeutic assessment

After establishing the diagnosis of IVC agenesis and right common iliac vein, venous thrombosis of the right external iliac vein, right common femoral vein and right superficial vein, we initiated anticoagulant therapy with high molecular weight heparin and close monitoring of the coagulation parameters. Afterwards, we initiated therapy with low-molecular heparin, with favorable evolution. The patient was discharged presenting a good general status, with the recommendation of anticoagulant therapy with acenocoumarin, close monitoring of INR, and prophylaxis with elastic socks.

DISCUSSIONS

IVC presents a complex and well-documented embryogenesis. Anatomically, it is formed of 4 segments: hepatic, suprarenal, renal and infrarenal (13, 14). The malformations of IVC can be represented by: malrotation and its position on the left side, the

drainage of IVC into the intrathoracic azygos vein, circumaoctic left renal vein, the duplication of IVC with normal intrathoracic position or the duplication of IVC with drainage into the azygos and hemiazygos veins and the absence of the infrarenal segment of the IVC or its complete absence (15). Certain authors sustain the fact that the absence of IVC can be a result of perinatal or intrauterine thrombosis that leads to obliteration, and afterwards its resorption (13,14). If the infrarenal segment of IVC does not present a normal development, the iliofemoral veins will drain into the azygos and hemiazygos veins through paravertebral anterior collateral veins. Due to the much inferior diameter of these collateral veins in comparison to that of the IVC, it is obvious the fact that this collateral pathway can lead to chronic venous stasis and afterwards to the thrombosis of the inferior limbs (13). The previously mentioned mechanism is also sustained in the case of our patient.

During embryogenesis, the right metanephrons drain into the IVC. Therefore, the agenesis of IVC can impair the development of the right kidney, with renal hypoplasia or even agenesis. Certain authors define this anomaly as KILT syndrome, which associates the anomaly of IVC, of the right kidney, and the thrombosis of the inferior limbs (1,11,13). In the case presented above, the patient presented nephromegaly, the diagnosis of KILT being unsustainable.

Venous thromboembolism represents a severe pathology among children with a major clinical importance due to its increased rate of morbidity and mortality. The correct, prompt and precise diagnosis of DVT owns a critical importance also due to the risks associated with the therapy (7,11,17,18). Among the risk factors that lead to the development of DVT, we mention: genetic factors, trauma, surgical interventions, neoplasms, central venous catheters, venous anomalies, chronic inflammation and sepsis (8,11,17,18). In the case of our patient, even though the genetic risk is absent, we underline the fact that before the admission in the Pediatrics Clinic, he underwent an appendectomy that required prolonged bed immobilization, and also, the patients presented a rare vascular anomaly, agenesis of IVC and right common iliac vein. Thus, the accumulation of all these factors led to the development of DVT.

It is well known the fact that congenital anomalies of IVC represent a risk factor for the development of DVT in case of young adults, but the prevalence of these anomalies in pediatric patients with

spontaneous DVT is not very precise. Recent studies point out the fact that DVT tends to have a more increased predominance in males, with an increased incidence at the age of young adults (4,11,15,18). This fact is sustained also by our case report that revealed the presence of DVT in an 8-year-old male child.

In case of patients with DVT due to the agenesis of IVC there is not a standard treatment. DVT is a pathology rarely encountered in children, and therefore, a clear therapeutic protocol is not defined for this condition. Nevertheless, low-molecular heparin is preferred for children that present DVT. Certain authors recommend at least 6 months of anticoagulant therapy, but most of the times the anticoagulant therapy is required lifelong, even despite the exclusion of a thrombophilia in order to rule out the recurrences (11). Similarly, in the case of our patient, the treatment was guided by the clinical evolution and the results of the repeated ultrasounds, the anticoagulant therapy being required

lifelong due to the associated vascular anomaly. Most likely, further studies will contribute to the establishment of the long-term prognosis in case of patients with DVT, and to the elaboration of an optimal therapeutic protocol for DVT secondary to vascular malformations.

CONCLUSIONS

Congenital anomalies of IVC represent an important risk factor for the development of inferior limbs DVT in case of children and teenagers. In the lack of significant family history, of a thrombophilia or a possible genetic mutations, the case presented above cumulated procoagulant factors, such as: chronic venous stasis, impaired venous returns due to post-appendectomy prolonged immobilization and venous hypertension due to the absence of main venous trunks leading imminent to the development of inferior limbs deep venous thrombosis.

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