ISSN- O: 2458 - 868X, ISSN–P: 2458 – 8687
National Library of Medicine - ID: 101731606

International Journal of Medical Science and Innovative Research (IJMSIR)
IJMSIR : A Medical Publication Hub
Available Online at: www.ijmsir.com
Volume – 9, Issue – 4, August – 2024 , Page No. : 21 – 26

A Case Report on KILT Syndrome (Kidney and Inferior Vena Cava Abnormalities with Legthrombosis) in A Young Male

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Citation this Article: Dr. Ranjit Prasad, Dr. G. Rakesh, Dr. Sreepad Yellu, Dr. Neil Nunes Aurelius, "A Case Report on KILT Syndrome (Kidney and Inferior Vena Cava Abnormalities with Legthrombosis) in A Young Male", IJMSIR - August - 2024, Vol – 9, Issue - 4, P. No. 21 – 26.

Type of Publication: Case Report

Conflicts of Interest: Nil

Abstract

First discovered in 2002, KILT syndrome is an extremely unusual trio consisting of kidney abnormalities, inferior vena cava (IVC) malformations, and deep vein thrombosis (DVT). This disorder, which is more common in younger people, is linked to an increased risk of recurrent thrombotic episodes and typically manifests itself accidentally during imaging. Additionally, it's important to closely observe any signs of possible renal problems.

We present the case of a 20-year-old male patient who complained of right flank pain and right lower limb discomfort that had persisted for a period of 10 days. His right loin and lower abdomen pain has been becoming worse. There was nothing noteworthy in his medical background, risk factors, or family tree. Upon inspection, it was found that the right lower limb had edema of variable diameter. Vascular thrombosis was seen in several veins on CT scans, including the common femoral, internal iliac, and right common iliac veins. The inferior vena cava (IVC) below the liver was very small, measuring just 0.8 cm in diameter, and the IVC segment located inside the liver was completely missing. There were enlarged venous collaterals in the lumbar area, retro peritoneum, and pelvis. Additionally, there was enlargement of the azygos and hemiazygos veins. While the left kidney was hypertrophied, the right kidney was hypoplastic. The right renal vein was visible but not filled with blood.

No emboli were seen in the pulmonary arterial system. There were no anomalies found in the urinalysis results, and the renal function tests came out normal, with a creatinine level of 92 µmol/L. Nothing came up during the thrombophilia screening. Oral anticoagulation with rivaroxaban was administered after low molecular weight heparin was administered as part of the treatment. Hydration and pain control were part of the supportive care. The patient reported less pain and marked reduction in leg edema upon discharge.

Because of its link to recurrent deep vein thrombosis (DVT) and renal problems, KILT syndrome warrants investigation despite its rarity. Inadequate venous

drainage increases the risk of deep vein thrombosis (DVT) in intravenous malformations, such as hypoplasia and azygos continuation. Patients presenting with limb symptoms that do not have an obvious cause should be evaluated for KILT syndrome. A multidisciplinary treatment strategy and lifelong anticoagulation are necessary for effective management. Improving treatment techniques and understanding long-term consequences require more research.

Keywords: KILT syndrome, blood clots, anatomical features, inferior vena cava.

Introduction

KILT syndrome, a rare condition linking kidney and inferior vena cava (IVC) abnormalities to limb thrombosis, was first described in 2002. Typically identified incidentally through imaging, this condition carries a significant risk of recurring blood clots, especially in young individuals. It also necessitates monitoring for potential kidney problems. We present a case of a 20-year-old experiencing flank pain and lower limb discomfort, subsequently diagnosed with KILT syndrome. Advanced imaging techniques vividly illustrated the unusual anatomical features.

Case Report

A 20-year-old boy arrived at the hospital complaining of right loin ache and discomfort in his right lower limb when walking, for past 10days. Before being admitted, he experienced escalating discomfort in the right loin and lower abdomen. He had no notable medical background, pertinent risk factors, or familial medical history. The physical examination showed edema in the right lower limb with a variation in circumference.

CT scan revealed a significant presence of deep venous thrombosis (DVT) in the right common iliac, internal iliac, external iliac, and common femoral veins (Figure 1). The portion of the inferior vena cava (IVC) below the liver was seen to be underdeveloped, with a diameter of 0.8 cm, whilst the portion of the IVC within the liver was not present. The presence of enlarged venous collaterals was observed in the pelvis, retroperitoneum, and along the lumbar epidural and paravertebral venous plexuses. The azygos and hemiazygos veins exhibited enlargement as seen in Figure 3. The right kidney exhibited hypoplasia, measuring 4.6 cm in bipolar length. In contrast, the left kidney had compensatory hypertrophy, measuring 12.0 cm in bipolar length. The right renal vein was found to be patent, but it was also seen to be hypoplastic in terms of its diameter.

The examination of the pulmonary arterial system revealed no indication of pulmonary embolism. The patient exhibited normal renal function, as shown by a creatinine level of 92 μmol/L. The urinalysis results indicated the absence of microscopic blood in the urine or abnormal levels of protein. The thorough investigation for thrombophilia did not reveal any significant findings. The treatment began with the administration of low molecular weight heparin, which was then followed by oral anticoagulation with rivaroxaban at a daily dosage of 20 mg. Additional supporting interventions were sufficient water and pain relief. After being released from the hospital, the discomfort decreased and there was significant improvement in the edema of the right lower leg. A comprehensive and interdisciplinary approach to care was organized, which included specialists in hematology, nephrology, pediatric surgery, physiotherapy, and occupational therapy.

The comprehensive, ongoing care approach involved the use of anticoagulant medication for life, frequent outpatient monitoring of blood pressure, urine protein, and renal function, and guidance to refrain from participating in contact sports or engaging in intense workouts.

Periodic monitoring with CT scans showed that the deep vein thrombosis (DVT) has improved, although there is still some remaining blood clot in the right common, external, and internal iliac veins. There was a gradual enlargement of the superficial veins in the abdomen and groin area on the right side. The patient reported no substantial discomfort when walking and had just little remaining edema in the leg.

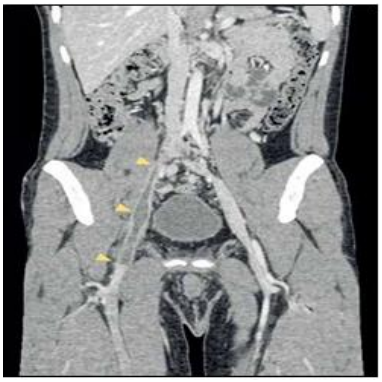

Figure 1: CT-Plain abdomen and pelvis shows deep venous thrombosis of the right common iliac, external iliac, and common femoral veins (arrowheads).

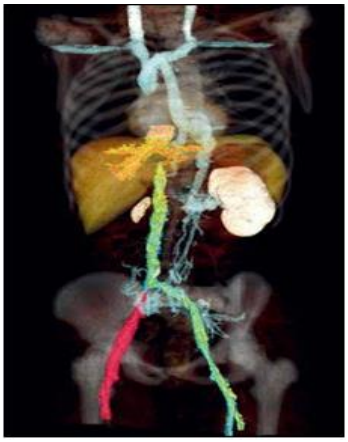

Figure 2: Volume rendering demonstrating extensive deep venous thrombosis within the right common iliac, external iliac, internal iliac, and common femoral veins (red). Focal interruption of the inferior vena cava (green) seen at intrahepatic segment with azygos continuation draining into the superior vena cava (blue). Note the hypoplastic right kidney with compensatory enlargement of the left kidney.

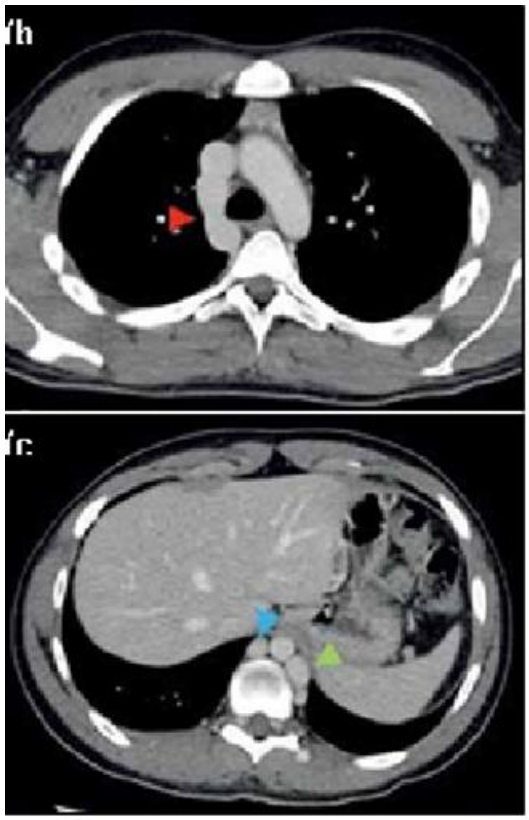

Figure 3: CT-Chest plain, axial section showing corresponding dilated azygos arch (red arrowhead) draining into the superior vena cava.

Axial CT scan at the level of diaphragm showing absence of intrahepatic inferior vena cava with distended azygos (blue arrowhead) and hemiazygos (green arrowhead) veins.

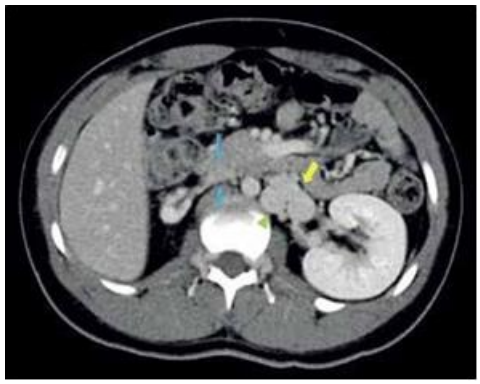

Figure 4: Axial CT- Abdomen, post-contrast image at the level of kidneys showing marked right renal hypoplasia with hypoplastic right renal vein draining into the profoundly atretic inferior vena cava (0.7 cm) [thin blue

arrows]. Compensatory hypertrophy of the left kidney with dilated left renal vein (yellow arrow) seen.

Discussion

Due to its rare occurrence, the level of clinical suspicion may not be elevated even when there are symptoms of limb swelling and discomfort.

IVC abnormalities are well recognized as separate risk factors for DVT, with a prevalence of 0.3% to 0.5% in the general population and 0.6% to 2% in individuals with pre-existing cardiovascular problems.[4] The process of IVC creation during embryonic development is intricate and takes place from the fourth week to the eighth week of embryonic life. The structure is composed of three pairs of paired veins, namely supracardinal, posterior carinal, and subcarinal veins.[5] Within the literature, a range of 15 to 60 distinct abnormalities of the inferior vena cava (IVC) have been documented. The most prevalent variants are IVC duplication (most prevalent, occurring in 2%-3% of cases), left-sided IVC, left retroaortic or circumaortic renal vein, and agenesis of the IVC.[4] IVC hypoplasia/agenesis leads to the development of a large number of collateral veins, such as the azygos, hemiazygos, and lumbar veins, as seen in our instance. In the context of the existing literature, our instance would be classified as "azygos continuation of the inferior vena cava." These collateral blood vessels have insufficient venous drainage, even if they have enlarged structurally due to increasing pressure and stagnant blood flow. This increases the likelihood of recurrent deep vein thrombosis (DVT).[10] The clinical presentation of deep vein thrombosis (DVT) can vary, ranging from swelling and discomfort in the lower leg to more unusual symptoms such as pain in the loin or lower back, as observed in our case. Nevertheless, these symptoms may not inevitably indicate an underlying KILT condition.[11]

There is a connection between kidney abnormalities and abnormalities in the inferior vena cava (IVC) as well as deep vein thrombosis (DVT) in the leg among a certain set of individuals. The trio was initially documented by Van Veen et al1 in 2002 and subsequently designated as KILT syndrome.[9]

According to research conducted by Sagban et al[12], it was discovered that 6% of instances of IVC agenesis had right renal hypoplasia, whereas 2.7% had left renal hypoplasia. Similarly, the right kidney is more frequently impacted, as shown in our case. From an embryological perspective, it seems logical that the venous blood from the right metanephros flows directly into the inferior vena cava (IVC), while the venous drainage of the left metanephros occurs through the gonadal vein and lumbar perforators.

Pulmonary embolism is infrequently observed in individuals with KILT syndrome or underlying IVC defects, most likely due to the requirement for the clot to be pushed by the comparatively narrow azygos and hemiazygos veins instead of the IVC.[13]

Ultrasound is effective in diagnosing lower extremities deep vein thrombosis (DVT), however it is not suitable for identifying irregularities in the inferior vena cava (IVC). A chest radiograph may reveal an increase in the size of the azygos shadow, which may be observed as the expansion of the right paratracheal stripe.

Contrast CT scan or magnetic resonance angiography are the primary imaging techniques for diagnosing KILT syndrome.

Currently, there is no definitive agreement on how to effectively manage KILT syndrome. The majority of case studies recommend the use of long-term anticoagulation because of the intrinsic lifetime risk profile associated with IVC abnormalities. It is suggested to provide comprehensive and interdisciplinary treatment for young

individuals with hypertension and renal function issues caused by renal hypoplasia. This care should include pain management, physical rehabilitation, and regular monitoring.[11] It is advisable to avoid physical effort since it might potentially raise the risk of deep vein thrombosis (DVT) in individuals with underlying inferior vena cava (IVC) abnormalities[13.] In our situation, the potential long-term result and forecast have not been established yet. Additional longitudinal follow-up investigations are necessary.

Conclusion

This instance effectively demonstrates a distinctive cause and risk factor for deep vein thrombosis (DVT), which is more prevalent among young adults. The frequent occurrence of kidney and IVC abnormalities provides valuable information on the early development of KILT syndrome throughout pregnancy and embryogenesis. In a young patient who comes with idiopathic deep vein thrombosis (DVT) with no known genetic or acquired blood clotting disorders or obvious risk factors, more investigation is warranted.

It is advisable to utilize CT or magnetic resonance angiography to examine for any pelvic/central venous malformation and renal abnormalities.

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