
CASE REPORT

Kidney and Inferior Vena Cava Abnormalities with Leg Thrombosis (KILT Syndrome) in a Young Healthy Male: a Case Report

LHQ Chin, M Cheung, GHT Ng, WWM Lam

Department of Radiology, Queen Mary Hospital, Hong Kong

INTRODUCTION

Kidney and inferior vena cava (IVC) abnormalities with leg thrombosis (KILT syndrome) was first described in the literature in 2002.¹ To date, several case reports have described this rare phenomenon, often with incidental detection of the triad of anomalies on cross-sectional imaging. This radiological diagnosis is clinically important because of the lifelong risk of recurrent venous thrombosis, especially in young individuals, and the need to screen for co-existing renal abnormalities. We present the case of a 17-year-old male with newly diagnosed KILT syndrome who presented with loin pain and lower limb discomfort. Post-processing with volume and cinematic rendering techniques were also incorporated to illustrate the unique radiological findings.

CASE REPORT

A 17-year-old male presented to the hospital with a 2-week history of right loin pain and right lower limb discomfort upon ambulation. Prior to admission, he had developed increasing right loin and lower abdominal pain. He had no significant medical history, relevant risk factors or family history. Physical examination

revealed right lower limb swelling with circumferential difference.

Computed tomography (CT) showed extensive deep venous thrombosis (DVT) involving the right common iliac, internal iliac, external iliac, and common femoral veins (Figure 1). The infrahepatic segment of the IVC appeared hypoplastic (0.8 cm calibre), whilst the intrahepatic IVC was absent. Multiple dilated venous collaterals were seen in the pelvis, retroperitoneum, and along the lumbar epidural and paravertebral venous plexuses. The azygos and hemiazygos veins were also dilated (Figures 2 and 3). The right kidney was hypoplastic (4.8 cm bipolar length) with compensatory hypertrophy of the left kidney (13.0 cm bipolar length). The right renal vein was patent but also hypoplastic in calibre. The distended left renal vein was seen predominantly draining into the distended hemiazygos vein (Figure 4). Screening of the pulmonary arterial system showed no evidence of pulmonary embolism.

Radionuclide scan using technetium-99m combined with dimercaptosuccinic acid showed small faint homogenous

Correspondence: Dr LHQ Chin, Department of Radiology, Queen Mary Hospital, Hong Kong
Email: chin.leanne@gmail.com

Submitted: 11 Aug 2020; Accepted: 7 Oct 2020

Contributors: All authors designed the study. LHQC acquired and analysed the data, and drafted the manuscript. MC, GHTN and WWML critically revised the manuscript for important intellectual content. All authors had full access to the data, contributed to the study, approved the final version for publication, and take responsibility for its accuracy and integrity.

Conflicts of Interest: All authors have disclosed no conflicts of interest.

Funding/Support: This study received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

Data Availability: All data generated or analysed during the present study are available from the corresponding author on reasonable request.

Ethics Approval: The patient was treated in accordance with the tenets of the Declaration of Helsinki. The patient provided written informed consent for all treatments and procedures, and for publication of this case report.



Figure 1. Coronal curved planar reformation demonstrating deep venous thrombosis of the right common iliac, external iliac, and common femoral veins (arrowheads).

tracer uptake in the right kidney with differential renal function of 99% and 1% for the left and right kidneys, respectively (Figure 5).

The patient's renal function was normal with creatinine of 95 $\mu\text{mol/L}$. Urinalysis showed no evidence of microscopic haematuria or proteinuria. Extensive thrombophilia workup was unremarkable.

Treatment was commenced with low molecular weight heparin followed by oral anticoagulation (rivaroxaban 20 mg daily). Other supportive measures comprised adequate hydration and analgesia. Upon discharge, pain subsided with much improvement of right lower limb swelling. Multidisciplinary follow-up care was arranged, involving paediatricians, haematologists, renal physicians, paediatric surgeons, physiotherapists and,

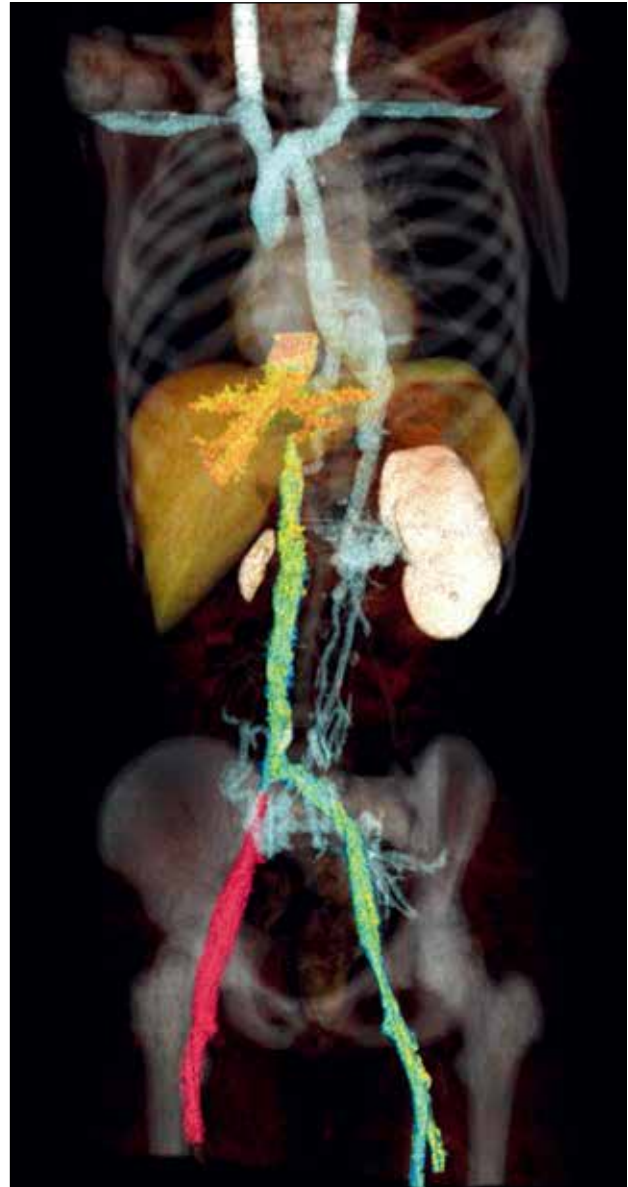


Figure 2. Volume rendering demonstrating extensive deep venous thrombosis within the right common iliac, external iliac, internal iliac, and common femoral veins (red). Focal interruption of the inferior vena cava (green) seen at intrahepatic segment with azygos continuation draining into the superior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

occupational therapists. His joint long-term management plan included lifelong anticoagulation medication, regular outpatient monitoring of blood pressure, urine protein and renal function, and advice to avoid contact sports or strenuous exercises.

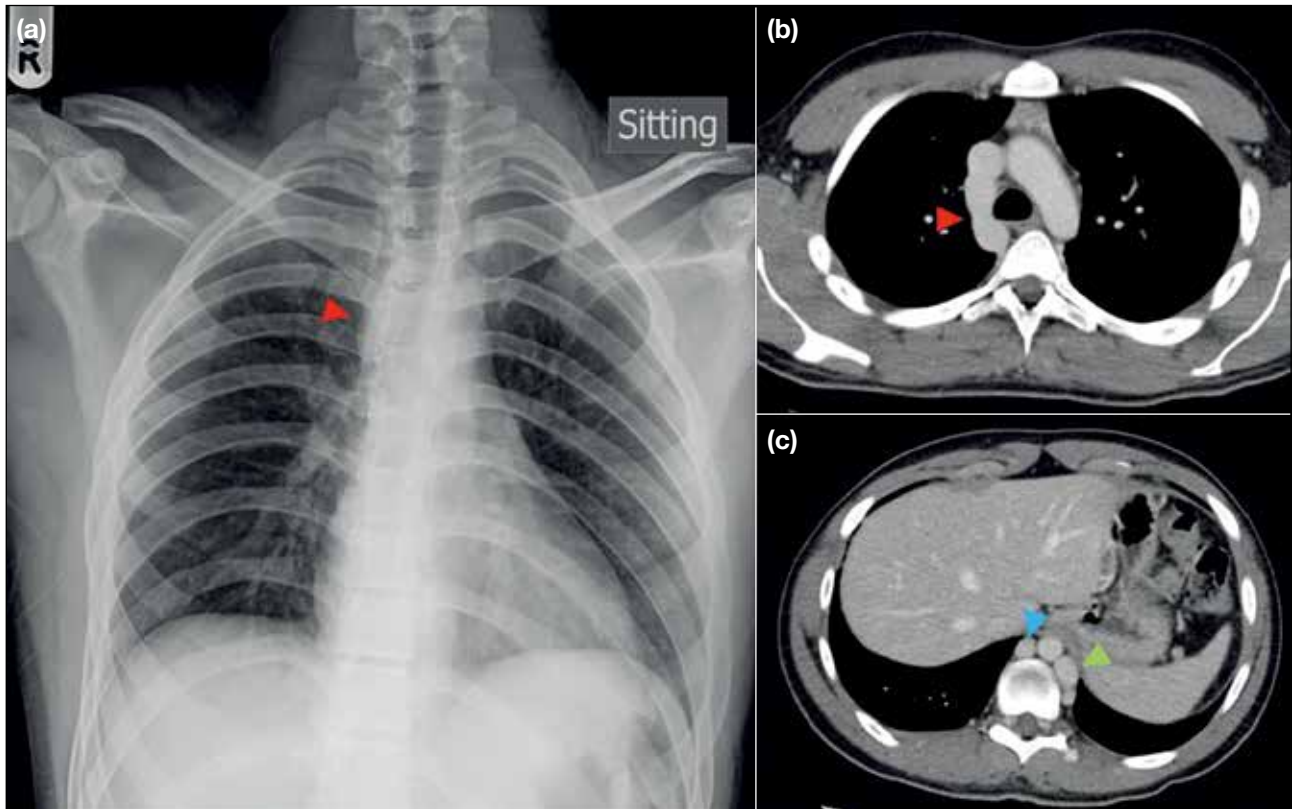


Figure 3. Chest radiograph (a) showing prominent azygos shadow (red arrowhead) seen as widening of the right paratracheal stripe. Axial computed tomography (CT) scan of the thorax (b) showing corresponding dilated azygos arch (red arrowhead) draining into the superior vena cava. Axial CT scan at the level of diaphragm (c) showing absence of intrahepatic inferior vena cava with distended azygos (blue arrowhead) and hemiazygos (green arrowhead) veins.



Figure 4. Axial post-contrast computed tomography 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 calibre) [thin blue arrows]. Compensatory hypertrophy of the left kidney with dilated left renal vein (yellow arrow) seen predominantly draining into the dilated hemiazygos vein (green arrowhead).

Interval follow-up CT scans revealed improvement of the DVT with residual laminar thrombus seen in the right common, external, and internal iliac veins. There was interval development of dilated superficial abdominal veins along the right lower abdominal wall and inguinal region (Figure 6). The patient did not complain of significant pain upon ambulation and had minimal residual leg swelling.

DISCUSSION

In the paediatric population (aged 0-18 years), DVT is rare, occurring in about 0.7 to 2.1 cases per 100 000 children compared with 100 to 150 per 100 000 adults.² Given its low incidence in young patients, the index of clinical suspicion may not be high despite a clinical presentation of limb swelling and pain. As in our case with spontaneous DVT, there is a quoted higher incidence of underlying IVC anomalies (5%) compared with the general population (0.5%).³

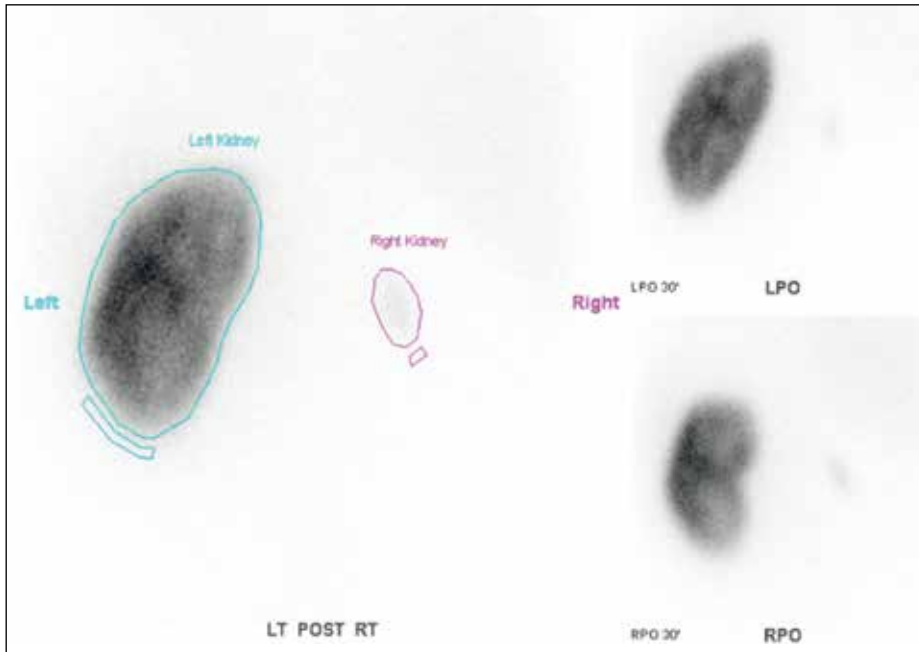


Figure 5. Dimercaptosuccinic acid scan showing marked right renal hypoplasia with trace amount homogenous uptake.



Figure 6. Cinematic rendering technique demonstrating dilated superficial epifascial veins over the right lower abdominal wall and inguinal region.

population, and in 0.6% to 2% of those with underlying cardiovascular defects.⁴ The embryonic process of IVC formation is complex, occurring between the fourth week and eighth week of embryonic life. It is formed from three sets of paired veins (supracardinal, posterior carinal, and subcarinal veins).⁵ In the literature, 15 to 60 different IVC anomalies have been described. The more common types include IVC duplication (most common, 2%-3%), left-sided IVC, left retroaortic or circumaortic renal vein and agenesis of the IVC.⁴ IVC hypoplasia/agenesis results in extensive collateral formation, most commonly including the azygos, hemiazygos and lumbar veins,⁶⁻⁸ as similarly illustrated in our case. Relative to the literature, our case would be referred to as “azygos continuation of the IVC”.⁹ These collaterals have inadequate venous drainage despite structural enlargement with increased venous pressure and stasis leading to increased risk for recurrent DVT.¹⁰

Interestingly, clinical presentation of DVT can be variable ranging from lower limb swelling and pain to more atypical symptoms of loin or low back pain, as seen in our case. However, these symptoms do not necessarily suggest underlying KILT syndrome.¹¹

Renal anomalies have been found to be associated with IVC anomalies and leg DVT in a certain group of patients. This triad was first described by Van Veen et al¹ in 2002 and later named KILT syndrome.⁹ A study by

IVC anomalies are well-known independent risk factors for DVT, occurring in up to 0.3% to 0.5% of the general

Sagban et al¹² found that right and left renal hypoplasia were identified in 6% and 2.7% of IVC agenesis cases, respectively. Likewise, as in our patient, the right kidney is more commonly affected. This makes embryological sense, as the venous return from the right metanephros goes directly to the IVC whilst venous drainage of the left metanephros is through the gonadal vein and lumbar perforators.

Pulmonary embolism has been rarely reported in patients with KILT syndrome or underlying IVC anomalies, likely because the clot would need to be propelled via the relatively small azygos and hemiazygos veins instead of the IVC.¹³

Although ultrasound may diagnose lower extremity DVT, it is not useful for detecting IVC anomalies. Chest radiograph may show enlargement of the azygos shadow, evidenced as widening of the right paratracheal stripe. Contrast CT scan or magnetic resonance angiography are preferred imaging modalities to diagnose KILT syndrome. Dimercaptosuccinic acid renal scan will provide information on differential renal function.

There is no clear consensus on the management of KILT syndrome to date. Most case reports advocate long-term anticoagulation due to the inherent lifelong risk profile associated with IVC anomalies. Holistic multidisciplinary care including analgesia, physical rehabilitation, and active surveillance for young-onset hypertension and renal function due to renal hypoplasia are recommended.¹¹ Advice against physical exertion is recommended since it may increase the risk for DVT with underlying IVC anomalies.¹³ As in our case, the prospective long-term outcome and prognosis are yet to be determined. More longitudinal follow-up studies are required.

CONCLUSION

This case nicely illustrates a unique cause and risk factor for DVT, occurring more commonly in paediatric and young adult populations. The common co-existence of kidney and IVC abnormalities also offers insight into the

early in-utero and embryogenesis of KILT syndrome. In a young patient who presents with idiopathic DVT and no thrombophilia or apparent risk factors, further imaging with CT or magnetic resonance angiography is recommended to look for underlying pelvic/central venous malformation and renal abnormalities.

REFERENCES

1. Van Veen J, Hampton KK, Makris M. Kilt syndrome? *Br J Haematol.* 2002;118:1199-200.
2. Radulescu VC. Management of venous thrombosis in the pediatric patient. *Pediatric Health Med Ther.* 2015;6:111-9.
3. Chee YL, Culligan DJ, Watson HG. Inferior vena cava malformation as a risk factor for deep venous thrombosis in the young. *Br J Haematol.* 2001;114:878-80.
4. Lauener S, Bütikofer A, Eigenheer S, Escher R. Thrombophlebitis hiding under a KILT — case report on 40 years long-term follow-up of neonatal renal vein thrombosis. *BMC Pediatr.* 2019;19:183.
5. Spentzouris G, Zandian A, Cesmebasi A, Kinsella CR, Muhleman M, Mirzayan N, et al. The clinical anatomy of the inferior vena cava: a review of common congenital anomalies and considerations for clinicians. *Clin Anat.* 2014;27:1234-43.
6. Bami S, Vazquez Y, Chorny V, Goldfisher R, Amodio J. Deep venous thrombosis of the leg, associated with agenesis of the infrarenal inferior vena cava and hypoplastic left kidney (KILT syndrome) in a 14-year-old child. *Case Rep Pediatr.* 2015;2015:864047.
7. Muntean C, Bucur G, Simu I, Tripon F, Marginean O. Deep venous thrombosis associated with inferior vena cava abnormalities and hypoplastic kidney in siblings. *Acta Med Marisiensis.* 2016;62:266-8.
8. Pomeranz CB, Cullen DL, Bellah RD. Deep venous thrombosis in a child with inferior vena cava and renal anomalies: KILT syndrome. *Pediatr Radiol.* 2018;48:1521-5.
9. Bass JE, Redwine MD, Kramer LA, Huynh PT, Harris JH Jr. Spectrum of congenital anomalies of the inferior vena cava: cross-sectional imaging findings. *Radiographics.* 2000;20:639-52.
10. Ruggeri M, Tosetto A, Castaman G, Rodeghiero F. Congenital absence of the inferior vena cava: a rare risk factor for idiopathic deep-vein thrombosis. *Lancet.* 2001;357:441.
11. Fung JK, Yeung VH, Chu SK, Man CW. KILT (kidney and IVC abnormalities with leg thrombosis) syndrome in a 41-years-old man with loin pain and fever. *Urol Case Rep.* 2017;12:6-8.
12. Sagban TA, Scharf RE, Wagenhäuser MU, Oberhuber A, Schelzig H, Grabitz K, et al. Elevated risk of thrombophilia in agenesis of the vena cava as a factor for deep vein thrombosis. *Orphanet J Rare Dis.* 2015;10:3.
13. Lambert M, Marboeuf P, Midulla M, Trillot N, Beregi JP, Mounier-Vehier C, et al. Inferior vena cava agenesis and deep vein thrombosis: 10 patients and review of the literature. *Vasc Med.* 2010;15:451-9.