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SWOLLEN LEGS AND FALSE NEPHROTIC SYNDROME: A CASE OF PRIMARY LYMPHATIC DYSPLASIA WITH INTERMITTENT CHYLURIA

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ABSTRACT

Chyluria, defined as the presence of chyle in urine, is a rare condition caused by an abnormal communication between the lymphatic system and the urinary tract. It can present with massive proteinuria and characteristic milky urine, mimicking nephrotic syndrome. We report the case of a 23-year-old woman with a history of seronegative ocular myasthenia gravis who developed progressive bilateral lower extremity oedema and intermittent nephrotic-range proteinuria despite normal serum albumin and lipid profiles. A renal biopsy, performed to investigate proteinuria, was complicated by a subcapsular hematoma. Shortly thereafter, the patient developed dysuria, urinary retention, and milky urine, leading to the diagnosis of chyluria. Imaging revealed thrombosis of the left iliac vein, associated with hereditary thrombophilia (heterozygosity for factor V Leiden and homozygous *G20210A* prothrombin mutation), prompting anticoagulation therapy. Lymphoscintigraphy suggested a lympho-vesical fistula and bilateral inguinal lymph node dysplasia. Conservative management, including urinary catheterization, fluid optimization, and dietary modifications with a low-fat, medium-chain triglyceride-enriched regimen, resulted in resolution of chyluria. At 6-month follow-up, there was no recurrence of symptoms, proteinuria improved, and anticoagulation was continued. This case highlights the importance of a multidisciplinary approach in managing rare presentations of chyluria, nephrotic proteinuria, and venous thrombosis.

KEYWORDS

Intermittent chyluria, nephrotic syndrome, lymphatic fistula, venous thrombosis

LEARNING POINTS

- Chyluria should be considered in patients presenting with milky urine and proteinuria, especially after excluding glomerular pathology.
- Non-parasitic causes, such as trauma or iatrogenic procedures like renal biopsy, are increasingly recognized in non-endemic regions.
- Conservative management, particularly dietary modifications with medium-chain triglycerides can lead to significant clinical improvement without invasive interventions.

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INTRODUCTION

The lymphatic system operates as a low-pressure network that begins in lymphatic capillaries, where interstitial fluid is absorbed. Skeletal muscle contractions aid lymph propulsion, while smooth muscle and unidirectional valves ensure forward flow. Superficial lymphatics drain the skin and subcutaneous tissues into deep lymphatics, passing through regional lymph nodes for immune response activation. Lymph eventually enters the circulatory system via the thoracic duct at the left venous angle, while lymph from the right upper body drains into the right lymphatic duct. Chyluria, a rare condition, occurs when lymphatic fluid leaks into the urinary tract, giving urine a characteristic milky appearance. This is caused by a lymphatic-urinary fistula that allows chyle—composed of lymph, chylomicrons, triglycerides, and proteins—to pass into the renal pelvis or bladder. In endemic regions like Southeast Asia and Africa, parasitic infections such as *Wuchereria bancrofti* are the leading cause. In non-endemic areas, chyluria is typically linked to non-parasitic factors, including trauma, lymphatic obstruction, retroperitoneal fibrosis, malignancies, or congenital anomalies *(Table 1)*. Symptoms include milky urine, dysuria, proteinuria, and occasionally urinary retention due to chylous clots. The case we describe is unique due to its association with post-renal biopsy trauma, concurrent iliac vein thrombosis, and an underlying thrombophilic state, all of which complicated the diagnostic and therapeutic course.

CASE DESCTRIPTION

The patient, a 23-year-old woman, had a known history of seronegative ocular myasthenia gravis and bronchial asthma. In August 2023, the patient developed peripheral oedema, predominantly in the lower limbs, prompting the initiation of nephrology follow-up. Despite normal serum albumin and lipid profiles, initial laboratory investigations revealed intermittent proteinuria, reaching the nephrotic range (9.6 g/day). Proteinuria persisted over the subsequent months, and by February 2024, urinary protein levels increased, necessitating further evaluation. A diagnostic hospitalization was scheduled for May 2024 to investigate the persistent nephrotic proteinuria. A renal biopsy was performed, revealing no significant glomerular pathology; however, the procedure was complicated by the development of a 1.5 cm subcapsular hematoma. Within days of the biopsy, the patient developed worsening dysuria, urinary retention, and milky urine. Urinary analysis confirmed the presence of elevated triglycerides, supporting a diagnosis of chyluria *(Table 2)*. Concurrently, the patient experienced increasing abdominal pain. A computed tomography (CT) scan of the abdomen and pelvis revealed thrombosis of the left iliac vein, likely exacerbated by her underlying thrombophilic state. Therapeutic anticoagulation with low-molecular-weight heparin (LMWH) was promptly initiated. To explore the cause of chyluria, multiple investigations were performed. Cystoscopy showed no evidence of fistulous tracts. However, lymphoscintigraphy revealed activity in a lymph node in the right inguinal-crural region and curvilinear activity originating from the right obturator region, extending cranially and medially into the bladder. These findings raised suspicion for a possible right lympho-vesical fistula *(Fig. 1)*. Abdominal magnetic resonance imaging (MRI) demonstrated resolution of the left perirenal hematoma and peritoneal effusion but revealed a minimal residual subcapsular fluid collection (3 mm) at the lower pole of the left kidney. No masses, collections, or lymphadenopathy were detected. A thrombus persisted in the left iliac vein and inferior vena cava. The patient was switched from LMWH to a direct oral anticoagulant (DOAC) and managed conservatively. Urinary catheterization was performed to relieve retention, and fluid optimization

Category	Cause	Description
Parasitic	Lymphatic filariasis	Infection by *Wuchereria bancrofti*, leading to lymphatic obstruction and abnormal urinary connections
	Other parasites	Rarely caused by other parasites affecting the lymphatic system
Non-parasitic	Congenital malformations	Abnormal communication between lymphatic and urinary systems presents from birth
	Trauma	Blunt or penetrating injuries causing lymphatic vessel damage
	Surgical complications	Retroperitoneal surgeries causing lymphatic-urinary fistula formation
	Neoplasm	Neoplasms compressing or invading lymphatic vessels, such as lymphomas or retroperitoneal malignancies
	Infections	Non-parasitic infections like tuberculosis causing lymphatic vessels inflammation
	Pregnancy	Increases intra-abdominal pressure during pregnancy causing lymphatic leakage into the urinary tract
Idiopathic	Unknown causes	Occurs without any identifiable underlying pathology

Table 1. Classification of the causes of chyluria.

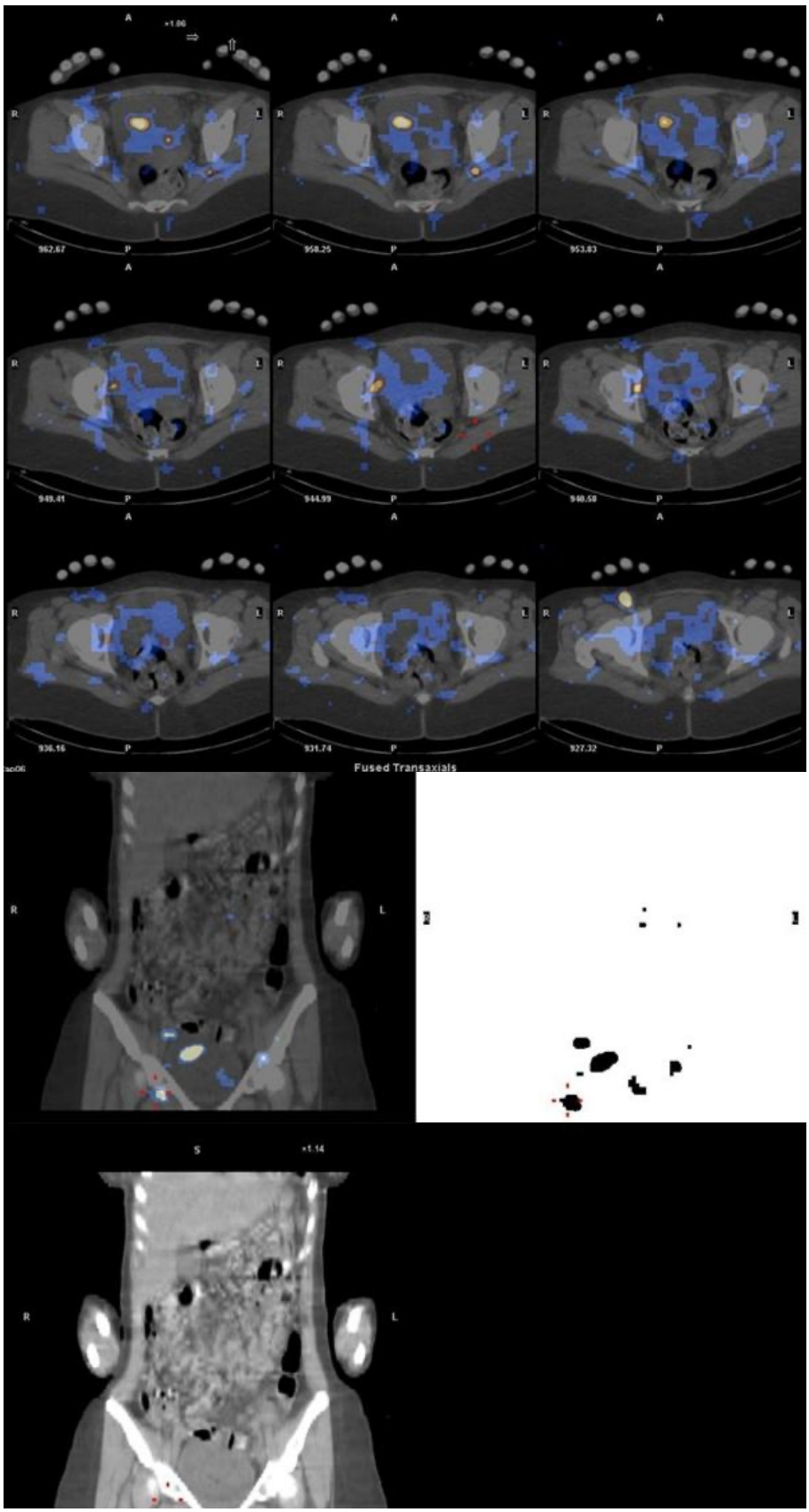


Figure 1. Lymphoscintigraphy showing curvilinear activity originating from the right obturator region, extending medially and cranially into the bladder, suggesting a right lympho-vesical fistula.

measures were implemented. Gradual improvement was noted over the following days, with resolution of milky urine and normalization of urinary output. The catheter was successfully removed without further retention or chyluria. After collegial discussion of the case, it was decided to perform an elective lymphoscintigraphy of the superficial and deep lymphatic system, due to suspected primitive dysplasia, with evaluation at an ultra-specialized centre. The conclusion was bilateral inguinal lymph node dysplasia from primary lymphedema of the lower limbs, associated with chylous dysplasia in the splanchnic region. A dietary regimen was recommended, consisting of a hypocaloric, low-fat diet

Exam	Value	Reference value
Urine proteins (g/24h)	9.6	< 0.15
Urine triglycerides (mg/dl)	542	1-10
Serum protein (g/l)	65	57-80
Serum albumin (g/l)	34.1	29-47
Serum triglycerides (mg/dl)	192	< 160
Serum vitamin D (nmol/l)	8.6	47.7-144

Table 2. Laboratory tests.

with medium-chain triglycerides (MCTs) and MCT oil. At the 6-month internal medicine follow-up, after assessing the thrombotic and bleeding risk balance, secondary venous thromboembolism prevention with reduced-dose DOAC was recommended.

DISCUSSION

Chyluria, characterized by the presence of chyle in the urine, is a rare condition resulting from an abnormal communication between the lymphatic system and the urinary tract. While parasitic infections, particularly *Wuchereria bancrofti* in endemic areas, are the most common cause, non-parasitic aetiologies, such as trauma, surgical procedures, malignancy, tuberculosis, and congenital anomalies, are increasingly recognized in non-endemic regions. In this case the renal biopsy likely acted as a traumatic trigger for the development of a recurrent chyluria due to an underlying lymphatic dysplasia. The pathophysiology of chyluria involves obstruction, stasis, or trauma of the lymphatic vessels, causing retrograde lymphatic flow into the renal pedicle lymphatics. The subsequent dilatation, proliferation, and rupture of these lymphatics into the renal pelvis or bladder result in lympho-pelvicalyceal fistula formation. This condition is further classified into mild, moderate, and severe grades based on the degree of chyle passage, weight loss, and radiographic findings, with our case exhibiting severe chyluria[1]. The diagnostic workup for chyluria aims to identify the underlying cause and localize the site of lymphatic leakage. While cystoscopy may confirm milky urine or blood clots, lymphoscintigraphy remains critical in identifying abnormal lymphatic activity and fistulous tracts. In this case, lymphoscintigraphy demonstrated lymphatic activity originating in the obturator region and extending into the bladder, consistent with a right lympho-vesical fistula. Advanced imaging techniques, including non-contrast MRI, provide additional structural information; however, sensitivity remains limited. Direct cannulation of the lymphatic system with contrast lymphangiography (CLL) offers superior diagnostic accuracy and, when combined with cone-beam CT (CBCT), can identify subtle fistulous connections. Furthermore, CLL has a therapeutic role, as the inflammatory reaction induced by lipiodol leakage can aid in sealing the fistula[2,3]. A unique aspect of this case was the concurrent development of iliac vein thrombosis, likely exacerbated by the patient's underlying thrombophilic state, characterized by heterozygosity for factor V Leiden and homozygosity for the *G20210A* prothrombin mutation. Prompt anticoagulation therapy was critical. The management of chyluria ranges from conservative to invasive approaches: conservative measures include bed rest, high fluid intake, and dietary modifications with a low-fat diet enriched with medium-chain triglycerides (MCTs). MCTs bypass the intestinal lymphatics, reducing chyle production and facilitating resolution; minimally invasive treatments such as retrograde pelvic instillation sclerotherapy use agents like silver nitrate or povidone-iodine to induce an inflammatory reaction, fibrosis, and closure of the lymphatic-pelvic communication. However, complications like renal scarring and hydronephrosis can occur. Surgical interventions, including lymphatic disconnection or ligation, are reserved for refractory cases. In our case, conservative management, including dietary optimization and fluid management, resulted in spontaneous resolution of chyluria[4]. This case highlights several key points: 1) chyluria, although rare, should be considered in the differential diagnosis of patients with milky urine and proteinuria, particularly after excluding glomerular pathology; 2) non-parasitic causes such as trauma, including iatrogenic triggers like renal biopsy, are increasingly recognized in non-endemic regions; underlying thrombophilic states can complicate the clinical course, necessitating vigilance and prompt anticoagulation therapy; 3) lymphoscintigraphy and advanced imaging play a critical role in diagnosing and localizing lymphatic abnormalities, guiding therapeutic decisions; 4) conservative management, particularly dietary modifications with MCT-enriched regimens, remains a cornerstone of therapy and can achieve significant clinical improvement without invasive interventions.

CONCLUSION

This case underscores the importance of a systematic diagnostic approach and a multidisciplinary management strategy in addressing chyluria, particularly when associated with nephrotic proteinuria and venous thrombosis. Conservative management can be highly effective and should be prioritized before considering invasive interventions.

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