Precision Embolization of a Rare Intra-articular Genicular Artery Pseudoaneurysm in Loeys-Dietz Syndrome

Khadiga Al Rasbi[1] and Alok Kumar Mittal[1,2*]

[1] Radiology Resident, Oman Medical Specialty Board, Muscat, Oman

[2]Department of Radiology and Molecular Imaging[1]. Sultan Qaboos University Hospital, Muscat, Oman

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*****Corresponding author: dralokaiims@gmail.com

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Abstract

Loeys-Dietz syndrome (LDS) is a rare autosomal dominant connective tissue disorder associated with aggressive vascular pathology, predominantly affecting large vessels. Peripheral vascular complications, particularly in small, intra-articular are exceedingly rare and poorly characterized. A 31-year-old male with genetically confirmed LDS presented with a 10-day history of right knee swelling and pain, without preceding trauma. Imaging revealed a large intra-articular pseudoaneurysm of the right inferolateral genicular artery, accompanied by hemarthrosis. Given the patient's vascular fragility, endovascular embolization was performed using coils and glue, supplemented by lipiodol embolization of adjacent tortuous genicular arteries. The patient exhibited significant clinical improvement post-procedure and was discharged in stable condition. This case represents the first reported occurrence of a spontaneous intra-articular pseudoaneurysm in a patient with LDS. It underscores the importance of maintaining a high index of suspicion for vascular anomalies in connective tissue disorders and demonstrates the efficacy of minimally invasive embolization in managing such complex presentations.

Keywords: Loeys-Dietz syndrome, Genicular artery, Pseudoaneurysm, Embolization, Connective tissue disorder.

Introduction

Pseudoaneurysms of the genicular arteries are uncommon vascular abnormalities, typically arising from traumatic or iatrogenic causes.[1,2] Rarely, they may develop spontaneously in the context of underlying connective tissue disorders such as Marfan syndrome and Ehlers-Danlos syndrome.[3,4] Loeys-Dietz syndrome (LDS), a rare autosomal dominant multisystem connective tissue disorder first described in 2005, predisposes affected individuals to extensive vascular complications due to inherent arterial wall fragility caused by heterozygous mutations in genes encoding transforming growth factor-beta (TGF-β) receptors (TGFBR1 and TGFBR2). The dysregulation of the TGF-β signaling pathway leads to defective extracellular matrix remodeling and reduced structural integrity of the arterial wall. These mutations result in fragmentation of elastic fibers, smooth muscle cell disarray, and abnormal collagen deposition. Consequently, even minor hemodynamic stress or physiological trauma can precipitate to weakened arterial walls, increasing their risk of aneurysm formation and arterial dissection in LDS patients.[5] Although up to 75% of cases result from de novo mutations, its prevalence remains uncertain, and its full clinical spectrum continues to evolve.[6] The latest classification of LDS has six types based on genetic mutations, all linked with aortic aneurysms and arterial tortuosity but varying in features like craniofacial

abnormalities, skeletal, and cardiac involvement. Types differ by gene (TGFBR1, TGFBR2, SMAD3, TGFBR3, SMAD2) and the presence of additional findings such as osteoarthritis or mitral valve prolapse.[7]

LDS is characterized by a triad of arterial aneurysms, vascular tortuosity, and craniofacial abnormalities. Compared to similar connective tissue disorders such as Marfan syndrome, there is an increased risk of aortic dissection and rupture at smaller diameters and younger ages.[6]

While the vascular complications of large arteries in LDS are well-documented, much less is known about the involvement of smaller, peripheral arteries. The pathophysiological mechanisms that increase susceptibility to aneurysmal changes in these vessels remain poorly understood.[5]

Pseudoaneurysms are a known but rare complication in connective tissue disorders like Marfan syndrome and Ehlers-Danlos syndrome.[3] They are most observed in larger, high-pressure arteries such as the aorta and pulmonary vessels . In contrast, pseudoaneurysms in smaller arteries, particularly in more peripheral or intra-articular locations, are exceedingly rare in the context of connective tissue diseases.[8] Such pseudoaneurysms pose a significant risk of rupture, bleeding, and functional impairment, particularly when located intra-articularly, where they may present with joint swelling, pain, or hemarthrosis. To the best of our knowledge, there have been no reported cases of spontaneous intra-articular artery pseudoaneurysms in individuals with LDS. This makes the present case especially unique and deserving of further investigation.

Management of pseudoaneurysms in LDS presents unique challenges due to the underlying vascular fragility, which complicates surgical interventions.[6] Minimally invasive endovascular embolization has emerged as a preferred strategy, offering targeted therapy with reduced procedural morbidity.

Herein, we present the first documented case of a spontaneous intra-articular pseudoaneurysm of the inferolateral genicular artery in a patient with genetically confirmed LDS, successfully managed with transarterial embolization. This report aims to highlight a novel vascular manifestation of LDS and underscores the importance of considering spontaneous pseudoaneurysms in uncommon anatomical locations in patients with connective tissue.

Case Report

A 31-year-old male with a known diagnosis of Loeys-Dietz syndrome presented with a 10-day history of right knee swelling and pain. There was no history of preceding trauma. On physical examination, the patient exhibited marked swelling and tenderness in the right knee. Laboratory investigations revealed a hemoglobin level of 8.8 g/dL, a white blood cell count of 8.5 × 10^3/uL, and an elevated C-reactive protein (CRP) level of 67 mg/L.

Ultrasound of the right knee demonstrated a significant joint effusion and a well-circumscribed, hypoechoic mass on the lateral aspect of the joint, measuring approximately 2.8 × 4.6 cm. Color Doppler imaging revealed a characteristic swirling flow pattern, known as the "yin-yang sign," consistent with a pseudoaneurysm. Subsequent CT angiography of the lower limbs confirmed the presence of a large knee effusion, indicative of hemarthrosis, and identified an intra-articular pseudoaneurysm within the lateral compartment of the knee joint, measuring 5 × 3 cm [Figure 1].

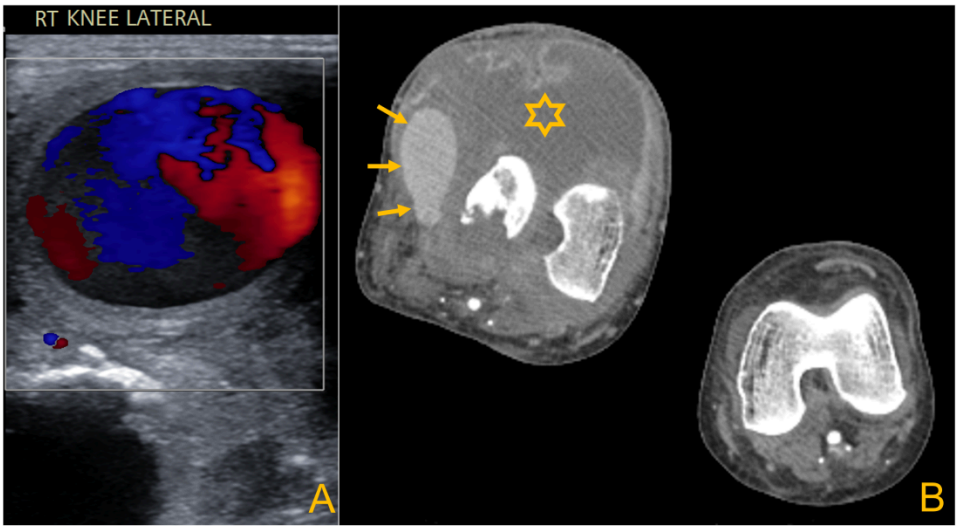

Figure 1: Pre-embolization imaging of the right knee joint. (a) Ultrasound Doppler along the lateral aspect of the right knee demonstrates a large anechoic intra-articular cystic lesion with internal color flow exhibiting the characteristic Yin-Yang sign, suggestive of a pseudoaneurysm. (b) Axial section from CT angiography of both lower limbs at the knee level reveals a large, well-defined, lobulated, contrast-filled outpouching along the lateral aspect of the right knee joint (arrows), consistent with a pseudoaneurysm. A significant hyperdense knee joint effusion is also noted (star).

The patient was referred to interventional radiology for transarterial embolization. A diagnostic lower limb angiogram identified a large pseudoaneurysm originating from the right inferolateral genicular artery. Embolization was performed using two 3 × 3 mm metallic coils (Cook Medical) and a 30% mixture of N-butyl cyanoacrylate and lipiodol in a 1:3 ratio [Figure 2]. Additionally, the other genicular arteries appeared notably enlarged and tortuous, accompanied by a diffuse soft tissue blush surrounding the knee joint. These vessels were embolized with plain lipiodol.

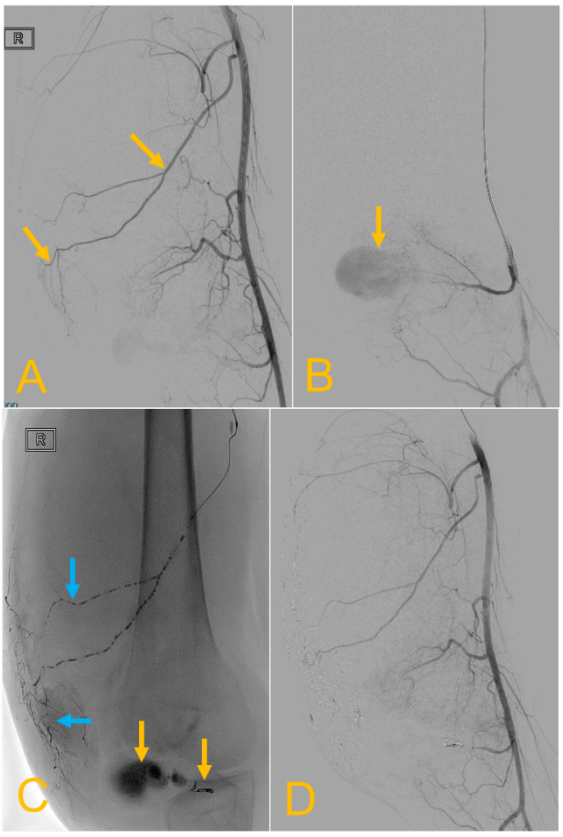

Figure 2: Digital subtraction angiography (DSA) of the right lower limb. (a) Diagnostic lower limb catheter angiogram demonstrates a hypertrophied, tortuous descending genicular artery (DGA) (yellow arrow) with significant soft tissue blush (blue arrow). (b) Super-selective angiogram of the inferior lateral genicular artery (ILGA) reveals a large pseudoaneurysm (arrow) with associated soft tissue blush. (c) Post metallic coil and N-butyl cyanoacrylate–lipiodol mixture (1:3 ratio) embolization of the ILGA (yellow arrows) and plain lipiodol embolization of the DGA (blue arrows). (d) Completion angiogram shows complete exclusion of the pseudoaneurysm and resolution of abnormal soft tissue blush.

Lipiodol was selected over other embolic agents such as polyvinyl alcohol (PVA) particles or gelfoam for the management of the perivascular blush due to its radiopacity and transient embolic effect, offering precise control of diffuse vascular leakage in the context of fragile arterial walls.

Following the procedure, the patient experienced significant improvement in knee pain and swelling and was discharged in stable condition a few days later.

Discussion

Pseudoaneurysms involving smaller arteries, particularly in peripheral or intra-articular locations, are exceedingly rare in the context of connective tissue disorders. Pseudoaneurysms of the genicular arteries are uncommon vascular lesions, with a limited number of cases documented in the literature. The majority of reported genicular artery pseudoaneurysms have been iatrogenic, occurring postoperatively — most frequently following total knee arthroplasty or arthroscopic knee procedures.[1] Trauma-related cases have also been described.[2] To date, this appears to be the first reported case of a genicular artery pseudoaneurysm associated with an underlying connective tissue disorder, specifically Loeys-Dietz syndrome (LDS).

LDS is a rare, autosomal dominant, multisystem connective tissue disorder first characterized in 2005 by mutations in genes encoding transforming growth factor-beta (TGF-β) receptors (TGFBR1 and TGFBR2), which are serine/threonine kinase receptors.[5] Numerous studies have shown that the TGF-β signaling pathway regulates various critical cellular processes such as cell proliferation, angiogenesis, and matrix transformation; therefore, mutations in genes involved in this pathway are the major cause for the pathogenesis of LDS, including aggressive vascular pathology, aneurysms, and arterial tortuosity.[9]

Its clinical hallmarks typically include aortic aneurysms and dissections, arterial tortuosity, hypertelorism, high-arched palate, and an abnormal uvula.[6] Compared with other inherited connective tissue diseases such as Marfan syndrome and Ehlers-Danlos syndrome, LDS tends to follow a more aggressive course, with earlier onset of vascular complications and a propensity to involve relatively smaller arteries. Furthermore, arterial involvement in LDS is often extensive, affecting multiple vascular territories simultaneously.[8] Although the genetic mutations responsible for LDS have been well-characterized, there remains a significant gap in the literature regarding its peripheral vascular manifestations. Only a few published cases have described true aneurysms in the lower limb arteries of LDS patients.[10,11] In addition, Beaulieu et al.[6] analyzed peripheral vascular manifestations in LDS patients, noting that these patients often harbor additional aneurysms and dissections throughout their vasculature beyond the central aortic disease. This pattern is consistent with the diffuse arteriopathy seen in other connective tissue disorders such as Marfan and Ehlers-Danlos syndromes, where peripheral aneurysms and arterial tortuosity may also occur due to inherent vascular fragility, requiring vigilant surveillance and timely intervention. However, to our knowledge, no prior reports have documented the occurrence of false aneurysms (pseudoaneurysms) in peripheral arteries in this population. The absence of significant trauma in our patient further supports the likelihood that this "spontaneous" pseudoaneurysm arose as a direct consequence of the underlying connective tissue disorder.

Spontaneous peripheral pseudoaneurysms in connective tissue diseases are exceedingly rare but have been sporadically reported. For example, a spontaneous pseudoaneurysm of the common peroneal artery has been documented in a patient with Ehlers-Danlos syndrome,[3] and a spontaneous popliteal artery pseudoaneurysm in a patient with polyarteritis nodosa.[4] These cases, along with the present report, emphasize the importance of considering underlying systemic disorders when evaluating atypical vascular lesions.

The vascular supply to the knee is derived from five genicular arteries: the descending genicular artery (DGA) from the superficial femoral artery (SFA), and the superior medial (SMGA), superior lateral (SLGA), middle (MGA), inferior medial (IMGA), and inferior lateral genicular arteries (ILGA), all branches of the popliteal artery . Of these, the inferomedial genicular artery is most commonly affected by pseudoaneurysm formation, while the inferior lateral genicular artery — the source of the pseudoaneurysm in our case — is the least frequently involved.[12]

Genicular artery embolization is a well-established, minimally invasive technique, historically employed in the management of recurrent knee hemarthrosis and, more recently, for symptomatic knee osteoarthritis.[13] Although surgical resection with or without embolization has traditionally been the most frequently reported treatment for genicular artery pseudoaneurysms, management in LDS presents distinct challenges due to inherent vascular fragility. This increases the risk of complications such as arterial dissection during invasive procedures — as encountered in our patient during femoral artery access. Given these concerns, endovascular embolization offers a safer and more controlled therapeutic option in this high-risk population. In Addition, genicular artery embolization utilizes selective embolization to address pathologic neovascularity while minimizing non-target ischemia. MicroCoils provide controlled, precise occlusion at the pseudoaneurysm neck, while adjunctive use of lipiodol addresses small distal branches or persistent filling, ensuring complete exclusion of the pseudoaneurysm sac. This approach allows targeted treatment with reduced risk of ischemic complications in the periarticular structures, aligning with best practices in embolization of geniculate branches.[13]

At present, no specific guidelines exist for the management of peripheral arterial complications in LDS, with most of the available literature focusing on aortic root and arch repairs.[6] As such, case reports like ours are critical to expanding the clinical understanding of LDS and will be instrumental in informing future recommendations for the identification and management of rare, yet potentially serious, peripheral vascular complications in this population.

Conclusion

This case highlights the rare occurrence of spontaneous intra-articular pseudoaneurysms in LDS and demonstrates the safety and effectiveness of endovascular embolization as a first-line intervention in such complex vascular presentations.

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