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Case Report

Vascular Anomalies in Connective Tissue Disorders and Chronic Kidney Disease: A Case Report on Intra/Extra-Articular Knee Pseudoaneurysm

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Abstract

We present the case of a 31-year-old male with End-Stage Renal Disease (ESRD) and ultimately was found to have Loeys-Dietz Syndrome (LDS), a rare connective tissue disorder, who developed a pseudoaneurysm in the genicular artery leading to recurrent hemarthrosis of the knee and distal thigh. The case highlights the importance of a multidisciplinary approach, timely genetic diagnosis, and comprehensive vascular management in rare syndromic presentations. This report also provides a review of the literature on vascular complications in LDS and ESRD.

Case Report

A 31-year-old male with no known medical or surgical history initially presented at the age of 17 with symptoms of vertigo, nausea, and fainting. A focused review of his medical history and physical examination revealed features consistent with a syndromic diagnosis, including marfanoid habitus, hypogonadism, skeletal abnormalities, gynecomastia, and tall stature, which raised suspicion for Klinefelter syndrome, though karyotyping showed a normal chromosomal pattern. During his initial presentation, he was found to have renal insufficiency and hypertension. Investigations, including imaging studies such as Ultrasound (US) and a renogram, revealed the presence of a solitary small left kidney. Over a 10-year period, his renal function gradually worsened, and he was ultimately diagnosed with End-Stage Renal Disease (ESRD) and initiated on hemodialysis through an Arteriovenous Fistula (AVF). At the age of 27, he experienced his first right knee monoarthritic attack, which was aspirated and improved without the need for hospitalization. Later that year, he presented again to the emergency department with swelling in his right knee, which was tender, warm, and extended from the knee to mid-thigh. Ultrasound suggested inflammatory arthritis, likely gout, and synovial fluid analysis showed no growth. Partial improvement of the swelling was noted after short-term intra-articular steroid therapy. Further evaluation also confirmed aortic root dilation on echocardiogram, raising the need for additional genetic testing to rule out Connective Tissue Disorders (CTDs). The patient continued to suffer from recurrent right knee swelling without any event of trauma or fever, despite multiple aspirations and arthroscopies over a period of 4 years. He required frequent hospitalizations under orthopedic surgery for the same complaints of a swollen, warm, tender knee with a restricted range of motion. Subsequent ultrasound studies revealed a complex effusion with multiple echogenic areas and mild subcutaneous edema, while MRI indicated a massive fluid collection extending from the femur to the knee joint, with no signs of osteomyelitis. CT Angiogram findings: A dense, massive effusion is noted in the right knee, with enhancing synovium suggesting hemarthrosis of varying blood age and chronic synovitis. A 5 x 3 cm pseudoaneurysm is identified arising from the peripheral branches of the right popliteal artery, located laterally to the lateral femoral condyle and tibia. This pseudoaneurysm is intra-articular, however, extended to the distal mid-thigh extra-articular component.

The patient was admitted for an interventional radiology procedure, during which antegrade angiography of the right lower limb was performed through the right common femoral artery, extending to the knee joint under local anesthesia. During the procedure, a dissection of the right superficial femoral artery was noted following the puncture, though there was no significant narrowing to limit blood flow. Super-selective angiography was then carried out on all four genicular arteries, revealing tortuosity and a large intra-articular pseudoaneurysm in the right inferior-lateral genicular artery. To address this, super-selective angioembolization was performed using metallic coils and lipiodol, followed by embolization of the remaining genicular arteries with plain lipiodol. The procedure proceeded without any immediate complications. During the same hospital admission, his knee gradually became tense, with swelling that was harder than before, and remained warm and tender. A decision was made to perform a lateral knee incision and surgical dissection of the popliteal artery branch pseudoaneurysm. The patient underwent pseudoaneurysm excision using lateral parapatellar knee approach (Figure 1). The patient did well post operatively, however, he required another two sessions of washout for hematoma collection. As sequence patient kept on long-term tranexamic acid. Genetic exome sequencing finally confirmed a diagnosis of autosomal dominant Loeys-Dietz syndrome.

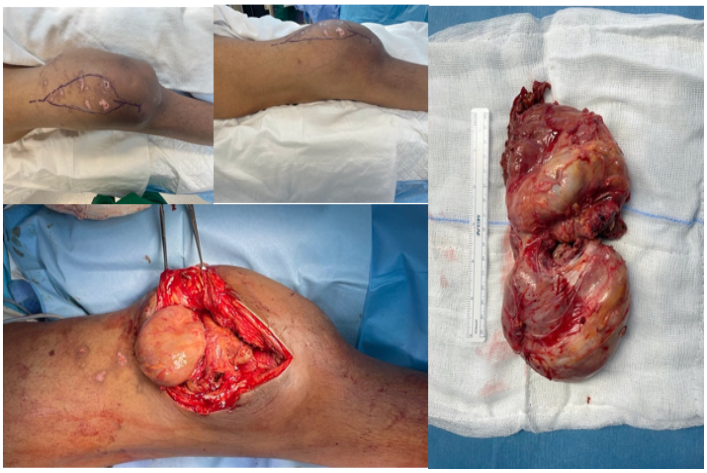

Figure 1: Intraoperative picture of swollen knee and distal thigh, and excisional capsular pseudoaneurysm that measures about 17cm long.

Discussion

End-Stage Renal Disease (ESRD) are associated with multiple vascular complications due to vascular calcification, and systemic endothelial dysfunction. Vascular complications often affect both large and small vessels and can involve any vascular territory, including peripheral arteries [1]. Vascular calcification is a hallmark of ESRD, affecting both the intimal and medial layers of the arterial wall. This process leads to stiffening of the arteries and can increase the risk of arterial rupture, aneurysms, and pseudoaneurysms [2]. Medial calcification, also known as Mönckeberg's sclerosis, can also be observed in the peripheral arteries in ESRD patient [3]. Although pseudoaneurysms are most commonly associated with dialysis access sites they can also occur in non-access sites as ESRD are more commonly associated with underlying vascular fragility, atherosclerosis, and calcium deposition in the arterial walls [4]. This is particularly relevant in patients who have undergone vascular interventions (e.g., angioplasty, stenting) or who are on medications such as anticoagulants or antiplatelet agents [5,6]. Connective Tissue Disorders (CTDs) are often associated with vascular anomalies, including aneurysms, pseudoaneurysms, and other vascular complications that vary in severity and clinical manifestation [7]. Loeys-Dietz Syndrome (LDS), a rare connective tissue disorder, is primarily caused by mutations in the TGF-β Receptor Genes (TGFBR1, TGFBR2) [8]. This disorder is characterized by vascular aneurysms, arterial tortuosity, and an increased risk of dissection and rupture. Vascular complications, including pseudoaneurysms, arise due to arterial fragility caused by abnormal collagen and elastin in the arterial walls [9-11]. The underlying pathophysiology of LDS involves disruption of the TGF-β signaling pathway, which plays a critical role in maintaining vascular integrity [8,12]. Intra-articular pseudoaneurysms are rare vascular anomalies where blood collects within a joint space, forming a pseudoaneurysm that leads to blood being confined in the surrounding tissues by a fibrous capsule. These types of pseudoaneurysms are extremely rare and can occur in various joints [13]. Causes other than CTDs and ESRD, includes trauma, infections such as septic arthritis, and Iatrogenic causes such as joint injections or arthroscopy [14]. The encapsulated hematoma, over time, can form a pseudoaneurysm which differentiates it from true aneurysms that involve the vessel wall. The most common presenting symptom, as blood accumulation in the joint space leads to significant pain and swelling in the affected joint. Restricted Joint mobility, bruising, hemarthrosis, and the development of a pulsatile mass in some cases [15,16]. Advanced imaging such as Computed Tomography (CT) Angiography, Arteriography, and Magnetic Resonance Imaging (MRI) can provide a more comprehensive view of the vascular anatomy and help identify the source of the pseudoaneurysm [17-19]. In larger or symptomatic pseudoaneurysms, surgical exploration and repair of the underlying affected vessel may be necessary.The pseudoaneurysm can be excised, and a vascular repair may be performed to restore the integrity of the vessel [20]. In cases where joint integrity is compromised, reconstruction may also be necessary. In some cases, particularly with pseudoaneurysms in accessible areas such as the knee or elbow, endovascular techniques such as coil embolization or stent grafting may be used as minimally invasive

options to treat the pseudoaneurysm [21,22].

Conclusion

Intra/Extra-articular pseudoaneurysms are rare vascular complications that can occur in patients following trauma, surgery, or as a result of underlying vascular or connective tissue disorders. Early diagnosis through imaging techniques such as ultrasound, MRI, and CT angiography is essential for effective management. Treatment may range from conservative care to surgical or endovascular intervention depending on the size, symptoms, and underlying causes**.**

Clinical Message

Recurrent and difficult-to-treat joint effusions, particularly in patients with a complex medical history or unusual clinical features, should raise clinical suspicion for underlying vascular causes, such as pseudoaneurysms or vascular malformations. These conditions can present as joint swelling or effusion and may be mistaken for more common causes like inflammatory arthritis or trauma. Early recognition of vascular etiologies is crucial, as delayed diagnosis or mismanagement can lead to significant morbidity, including joint damage, hemorrhage, or embolic events. Early intervention, including endovascular embolization or surgical management, can help prevent further joint damage and improve patient outcomes. Therefore, when faced with recurrent joint effusions that do not respond to conventional treatments, clinicians should consider the possibility of a vascular cause and initiate appropriate diagnostic work-up and management.

Learning Point of the Article: The need for high clinical suspicion and advanced imaging to confirm vascular complications such as intra-articular pseudoaneurysms in patients presenting with recurrent large joint effusions.

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