Bilateral popliteal artery aneurysms in a young man with Loeys-Dietz syndrome

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Loeys-Dietz syndrome is a recently described genetic connective tissue disorder. The syndrome is associated with multiple nonvascular phenotypic anomalies but also aggressive arteriopathy, which has so far principally been shown to cause aortic root dilatation with subsequent dissection and rupture. We report the first ever case of a young man diagnosed with Loeys-Dietz syndrome with asymptomatic large bilateral popliteal artery aneurysms. We have successfully resected these aneurysms and revascularized with synthetic graft. (J Vasc Surg 2012;56:486-8.)

Loeys-Dietz syndrome (LDS) is a recently described autosomal dominant syndrome characterized by the triad of arterial tortuosity and aneurysms, hypertelorism, and a bifid uvula or cleft palate. Most of the arteriopathy manifests in the thoracic aorta, although aneurysmal degeneration has been noted rarely in other locations within the thorax, abdomen, and pelvis. It is caused by heterozygous mutations in the genes encoding transforming growth factor β receptors 1 and 2 (TGFBR1 and TGFBR2, respectively). LDS has more recently been subcategorized into types I and II based on the presence or absence (respectively) of craniofacial abnormalities. Affected patients have a higher risk of aortic dissection or rupture at an early age compared with other connective tissue disorders. However, surgical intervention in LDS is generally successful with lower in-hospital mortality than in those with vascular Ehlers-Danlos or Marfan syndrome.

CASE REPORT

A 28-year-old male was referred to our unit by the cardiothoracic department of our hospital. The patient had undergone aortic root and aortic valve replacement in 2007 after experiencing an acute Stanford A aortic dissection. A connective tissue disorder was suspected clinically at that time and Ehlers-Danlos syndrome was ruled out, but confirmation of TGFBR1 mutation resulting in LDS type II has only recently been achieved through genetic testing. There was no relevant family history. The patient had noticed a swelling behind his right knee and could sense a palpable mass, and was referred by his general practitioner for further investigations. On examination, he had good femoral and distal pulses, and the popliteal aneurysms could easily be palpated as a pulsatile mass just above the knee crease on the right side and at the level of the popliteal fossa on the left. He underwent duplex arterial scanning and computed tomography angiography (Fig 1).

The patient’s case was discussed at our vascular multidisciplinary meeting, and it was decided that open repair of the larger aneurysm (right) should proceed first. Endovascular treatment was rejected due to the severe tortuosity of the proximal neck. Also, while little is known about the natural history of peripheral arterial aneurysms in LDS, extrapolating from other connective tissue diseases such as Marfan syndrome, there was also concern about disease progression in the contiguous artery. This would risk endoleaks, migration, and the potential need for further re-intervention.

PROCEDURE

Through the medial approach, the distal superficial femoral artery was dissected and controlled with a sling (Fig 2). The below-knee popliteal artery was then exposed and clamped to avoid peripheral embolization while manipulating the sac. The entire aneurysm was dissected out, the inflow, outflow, and all tributaries were ligated, and the aneurysm was excised. Revascularization was achieved with an 8-mm polytetrafluoroethylene (PTFE) heparin bonded Propanet graft (W. L. Gore and Associates, Flagstaff, Ariz.). The proximal anastomosis was fashioned end-to-end using a 6-0 polypropylene suture, which was then covered with a PTFE Teflon tube to mitigate against future anastomotic dilatation. The below-knee popliteal artery was ligated and the distal anastomosis performed in an end-to-side manner for convenience. He was recommenced on anticoagulant therapy immediately postoperatively due to his replaced heart valve. Aside from a thigh hematoma that required evacuation, he recovered well and was discharged on postoperative day 7. As a separate staged procedure, we proceeded to repair the left popliteal aneurysm in an identical way (Fig 3) approximately 2 months later.

DISCUSSION

The median reported survival of LDS patients is only 37 years. The mean age of death is 26 years, secondary to thoracic aortic dissection as the leading cause (67%), followed by abdominal aortic dissection (22%) and cerebral bleeding (7%).
Among the general population, popliteal artery aneurysms are the most common peripheral aneurysms; they occur mostly in men and have an incidence of 0.1% to 2.8%.2,3 Twenty-five to 80 percentage points of popliteal artery aneurysms are asymptomatic at the time of operation, but when clinical symptoms occur, they include intermittent claudication, rest pain due to aneurysm thrombosis, and distal embolization.4 While their etiology is debated, the most common association is atherosclerosis; however, less frequently they may be posttraumatic, congenital, mycotic, inflammatory, arise due to popliteal artery entrapment syndrome, or be part of a connective tissue disorder, especially at younger ages.3

To our knowledge, we have presented the first case of a young adult with LDS presenting with popliteal artery aneurysms. The case is particularly challenging because of the young age of the patient and the uncertain natural history of the disease, especially in someone who has already survived thoracic dissection. In our case, he was asymptomatic despite these large aneurysms, although this may relate to the fact that he was fully anticoagulated on warfarin because of his aortic valve replacement. Open surgery has traditionally been the gold standard, with the
medial or posterior approach being favored among most surgeons.\textsuperscript{2,3} Recently endovascular techniques have been introduced as a potential treatment alternative with promising results.\textsuperscript{5,6} The success of flexible covered stents is related to the anatomy of the popliteal artery aneurysm (degree of tortuosity, diameter and length of proximal and distal landing zones, and relation to the knee joint). Although recent prospective studies show similar results compared with open surgery,\textsuperscript{5,6} there is a lack of data to support the long-term patency of covered stents, especially in patients with connective tissue disorders.

Specific surgical technical considerations in this case included: (1) The proximal neck angulation of 90 degrees and the patent genicular arteries in close proximity were considered to be technical contraindications for endovascular repair. (2) Even if the aneurysm was technically suitable, open surgery was chosen due to the reported good postoperative outcomes from the aortic arch aneurysm repairs for such patients, and, as mentioned previously, the unknown natural history of the arterial disease.\textsuperscript{7} (3) The proximal anastomosis was reinforced with the use of a PTFE tube partially covering the native arterial part of the anastomosis for better mechanical support, preventing further aneurysmal dilatation, although we recognize that there is no firm evidence for this. (4) Synthetic ePTFE graft was used as the conduit in favor of long saphenous vein partially because of its good reported patency in popliteal aneurysms\textsuperscript{8} but also because of at least the theoretic risk of aneurysmal degeneration of vein grafts due to the connective tissue disease.

CONCLUSIONS

The vascular surgeon should be aware of LDS as a connective tissue disorder which manifests as aggressive arteriopathy of not only the aortic arch but also of peripheral vessels.\textsuperscript{9}

Recent data indicate that early surgical intervention is the treatment of choice for LDS patients suffering from arterial aneurysms with good operative outcome.\textsuperscript{1} Although endovascular techniques represent a new appealing alternative for these patients, bearing in mind the overall short survival rate,\textsuperscript{10} progressive enlargement and tortuosity of the vessel could be a potential source of treatment failure, especially in patients who have already survived the leading cause of death in LDS, thoracic dissection.\textsuperscript{11} We will keep him under close surveillance for metachronous aneurysms in the years to come.

REFERENCES


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