Endovascular repair of tortuous recurrent femoral-popliteal aneurysm in a patient with Loeys-Dietz syndrome

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ABSTRACT

Loeys-Dietz syndrome is a rare connective tissue disorder with widespread arterial tortuosity and aneurysms. This syndrome is most notable for its aortic disease, including aortic root dilation and aortic dissection or rupture. Although not as well studied, peripheral artery aneurysms are a prevalent concurrent manifestation and have previously been repaired with both open and endovascular approaches. There are minimal data about the durability and technical considerations of endovascular repair in this disease. We report a case of a patient who developed an extremely tortuous recurrent femoral-popliteal artery aneurysm secondary to aneurysmal degeneration around previously placed stents that was treated with an endovascular approach. (J Vasc Surg Cases and Innovative Techniques 2018;4:156-9.)

Loeys-Dietz syndrome (LDS) is an autosomal dominant connective tissue disorder characterized by arterial tortuosity and aneurysms, hypertelorism, and bifid uvula or cleft palate. Additional skeletal, craniofacial, and cutaneous manifestations have been described since its discovery in 2005, which has led to the classification of multiple subtypes. Vascular manifestations, including widespread tortuosity, aneurysms, and dissections, are the most consistent features. The spectrum of LDS disorders is caused by mutations in the transforming growth factor β pathway. The resulting overexpression of transforming growth factor β leads to medial degeneration, thereby causing vessel dilation and laxity.

The aggressive vasculopathy of LDS can lead to life-threatening dissections and aneurysm ruptures. Patients with LDS are at high risk of aortic dissection and rupture at earlier ages and at smaller aortic diameters than in the general population. Although aortic disease is the most studied vascular manifestation, tortuosity and aneurysmal degeneration can occur anywhere in the vascular tree: most patients (80%) have peripheral artery manifestations, most commonly in the carotid and subclavian arteries. Less frequently, LDS patients may also develop superficial femoral artery (SFA) and popliteal artery aneurysms. Herein, we describe a patient with LDS who presented with a recurrent femoral-popliteal artery aneurysm around a prior endovascular repair. The patient consented to presentation of this case.

CASE REPORT

A 66-year-old man with LDS presented with enlarging asymptomatic SFA and popliteal artery aneurysms around previously placed stents that were found on routine surveillance computed tomography angiography (CTA).

The patient was first diagnosed with LDS by TGFBR1/2 genetic testing in 2005. He had previously carried a diagnosis of Marfan syndrome (MFS) based on a family history of early aortic dissections. In 2006, he was referred to vascular surgery for management of bilateral popliteal aneurysms. Given the aggressive vasculopathy of LDS, he was started on close surveillance with annual three-dimensional CTA of the head, neck, chest, and abdomen and popliteal duplex ultrasound examination for monitoring of new and existing aneurysms.

His past medical history was significant for hypertension, atrial fibrillation, and factor V Leiden; past surgical history was remarkable for 11 cardiac and vascular operations for LDS manifestations. Cardiac operations included aortic root replacement and two redo operations for coronary artery dehiscence and pseudoaneurysm. Vascular operations included open repair of abdominal aortic aneurysm and iliac aneurysms, left subclavian aneurysm repair, resection of an acquired arteriovenous fistula, and multiple interventions for bilateral popliteal aneurysms. His 4.4-cm right popliteal aneurysm was first repaired in 2006 through an open posterior approach, which later required right SFA stenting and ultimately an SFA to below-knee popliteal artery bypass graft. His left popliteal aneurysm grew to 4.6 cm in 2011, at which point he underwent placement of four overlapping stents.

On our evaluation, he was noted to have significant aneurysmal degeneration proximal and distal to the popliteal stents on the left. Three-dimensional CTA was obtained, demonstrating a 4.8- x 4.4-cm aneurysm (measured at 2.2 cm 1 year earlier) in the mid-SFA proximal to the stent and new aneurysmal degeneration distal to the stents in the below-knee...
popliteal artery (Fig 1). The stented segment of the artery was markedly tortuous.

Given the rapid growth of the aneurysm and risk of thromboembolism, surgical repair was indicated. The patient preferred an endovascular approach because of wound healing complications with previous open repair. Informed consent was obtained.

Access was obtained through an open left femoral artery exposure for direct visualization of the ectatic vessel. Antegrade access with a 14F sheath was obtained to avoid prior aortic reconstructions (Fig 2). After confirmation of location under angiography, a 13-mm × 10-cm Gore Viabahn (W. L. Gore & Associates, Flagstaff, Ariz) stent was deployed in the below-knee popliteal aneurysm. A second 13-mm × 10-cm Gore Viabahn stent was deployed in the left SFA aneurysm. Both stents were placed with substantial overlap with the old stents. Completion angiography demonstrated no evidence of endoleak and good distal runoff into tibial vessels (Fig 2). Distal pulses were easily palpable after repair. He was discharged home in good condition on postoperative day 1. He was seen in follow-up 1 month after repair, with duplex ultrasound demonstrating aneurysm exclusion and no evidence of endoleak, and will resume his annual surveillance imaging protocol.

DISCUSSION

In this case report of the rare disease LDS, we describe a recurrent left femoral-popliteal aneurysm secondary to aneurysmal degeneration around previous stents, which we successfully managed with an endovascular approach.

Our institutional experience with LDS has demonstrated that the average age at diagnosis is 28 years, with peripheral aneurysms seen in the majority (80%) of patients.9 Our patient was initially misdiagnosed as having MFS, but on diagnosis of LDS, his vascular disease on imaging included bilateral popliteal aneurysms in addition to iliac, abdominal aortic, and superior
mesenteric aneurysms. Vessel tortuosity throughout the vascular tree is unique to LDS in comparison to other connective tissue disorders like MFS. Vertebral artery tortuosity and mean tortuosity score have been used to differentiate LDS from MFS. Vascular tortuosity, along with a lack of lens dislocation, was critical in establishing this patient’s diagnosis.

Peripheral aneurysms can be repaired through an open or endovascular approach. Endovascular management of tortuous vessels is technically challenging because of the risk of stent migration and kinking. A tortuous popliteal aneurysm, as in this case, would typically favor open repair through a medial or posterior approach. Furthermore, there is uncertainty about the durability of endovascular repairs in LDS patients secondary to the concern that disease progression could lead to aneurysmal degeneration of contiguous vessels.

To mitigate the risks posed by vessel tortuosity, our patient’s previous endovascular repair of the left popliteal aneurysm was performed with 50% stent overlap, as previously described. Although aneurysmal degeneration did occur around these stents, this original repair proved to be durable, lasting 6 years without migration or kinking. The current reintervention was not indicated for technical failure of the previous intervention but rather for progression of underlying disease. Technical success of endovascular repair in LDS patients has been demonstrated in the literature at short follow-up intervals of 6 months to 2 years. This case demonstrates the natural history of disease during a longer interval of follow-up of 6 years and underscores the importance of regular surveillance imaging for aneurysmal degeneration around previous repairs.

Small series and case reports in LDS patients provide promising evidence of the safety and efficacy of endovascular repair. One case report of bilateral iliac aneurysms demonstrated successful endovascular repair with sac regression and no evidence of endoleak on short-term follow-up. Another presented a case of endovascular coil embolization of an internal mammary aneurysm. Our institutional experience with management of peripheral vascular disease in LDS patients showed no perioperative complications after endovascular repair vs 11.8% perioperative morbidity for open repair. Given the rarity of this disease, there is still a need for long-term follow-up of endovascular repair in larger cohorts, but these data nonetheless support the endovascular option in repair of distal peripheral aneurysms in selected patients with LDS.

CONCLUSIONS

LDS is a disease that leads to aneurysmal degeneration and vessel tortuosity throughout the vascular tree. Given the natural history of the disease, patients require multiple vascular operations and reinterventions during their lifetime. The case presented herein supports active surveillance with CTA to evaluate for new, enlarging aneurysms and degenerative changes around previous repairs. Tortuosity of long diseased vessel segments can make endovascular repair challenging, but we demonstrate that an endovascular approach is feasible despite unfavorable anatomy. Given that LDS is a relatively rare
disease, continued prospective evaluation is needed to determine optimal management strategies for peripheral artery aneurysms in affected patients.

REFERENCES
1. Loeys BL, Dietz HC. Loeys-Dietz syndrome. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJ, Stephens K et al., editors. GeneReviews. Seattle, Wash: University of Washington; 2017.
2. MacCarrick G, Black JH 3rd, Bowdin S, El-Hamamsy I, Frischmeyer-Guerrerio PA, Guerrero AL, et al. Loeys-Dietz syndrome: a primer for diagnosis and management. Genet Med 2014;16:576-87.
3. Loeys BL, Chen J, Neptune ER, Judge DP, Podowski M, Holm T, et al. A syndrome of altered cardiovascular, craniofacial, neurocognitive and skeletal development caused by mutations in TGFBR1 or TGFBR2. Nat Genet 2005;37:275-81.
4. Loeys BL, Schwarze U, Holm T, Callewaert BL, Thomas GH, Pannu H, et al. Aneurysm syndromes caused by mutations in the TGF-β receptor. N Engl J Med 2006;355:788-98.
5. Fukui D, Miyagawa S, Soeda J, Tanaka K, Urayama H, Kawasaki S. Overexpression of transforming growth factor β1 in smooth muscle cells of human abdominal aortic aneurysm. Eur J Vasc Endovasc Surg 2003;25:540-5.
6. Maliszewski JJ, Miller DV, Lu J, Dietz HC, Halushka MK. Histopathologic findings in ascending aortas from individuals with Loeys-Dietz syndrome (LDS). Am J Surg Pathol 2009;33:194-201.
7. Lee RS, Fazel S, Schwarze U, Fleischmann D, Berry GJ, Liang D, et al. Rapid aneurysmal degeneration of a Stanford type B aortic dissection in a patient with Loeys-Dietz syndrome. J Thorac Cardiovasc Surg 2007;134:242-3.e1.
8. Augoustides JG, Plappert T, Bavaria JE. Aortic decision-making in the Loeys-Dietz syndrome: aortic root aneurysm and a normal-caliber ascending aorta and aortic arch. J Thorac Cardiovasc Surg 2009;138:502-3.
9. Beaulieu RJ, Lue J, Ehler BA, Grimm JC, Hicks CW, Black JH 3rd. Surgical management of peripheral vascular manifestations of Loeys-Dietz syndrome. Ann Vasc Surg 2017;38:10-6.
10. Kono AK, Higashi M, Morisaki H, Morisaki T, Tsutsumi Y, Akutsu K, et al. High prevalence of vertebral artery tortuosity of Loeys-Dietz syndrome in comparison with Marfan syndrome. Jpn J Radiol 2010;28:273-7.
11. Mohan IV, Bray PJ, Harris JP, May J, Stephen MS, Bray AE, et al. Endovascular popliteal aneurysm repair: are the results comparable to open surgery? Eur J Vasc Endovasc Surg 2006;32:149-54.
12. Stephenson MA, Vlachakis I, Valenti D. Bilateral popliteal artery aneurysms in a young man with Loeys-Dietz syndrome. J Vasc Surg 2012;56:486-8.
13. Geisbusch P, Kotelis D, von Tengg-Kobligk H, Hychlik-Durr A, Allenberg JR, Bockler D. Thoracic aortic endografting in patients with connective tissue diseases. J Endovasc Ther 2008;15:144-9.
14. Ohman JW, Charlton-Ouw KM, Azizzadeh A. Endovascular repair of an internal mammary artery aneurysm in a patient with Loeys-Dietz syndrome. J Vasc Surg 2012;55:837-40.
15. Casey K, Zayed M, Greenberg JI, Dalman RL, Lee JT. Endovascular repair of bilateral iliac artery aneurysms in a patient with Loeys-Dietz syndrome. Ann Vasc Surg 2012;26:107.e5-10.

Submitted Dec 19, 2017; accepted Mar 2, 2018.