Late Peripheral Thoracic Aneurysms following Aortic Root Surgery in Patients with Loeys–Dietz Syndrome

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ABSTRACT

The Loeys–Dietz syndrome is a multisystem connective tissue disorder characterized by aortopathy, arterial tortuosity, peripheral aneurysms, and skeletal features. The peripheral arteriopathy is an important cause of morbidity and potential mortality. This report presents 2 cases: the first demonstrating a 5-cm pseudoaneurysm of the right internal mammary artery and the second demonstrating a 2.3-cm aneurysm of the left internal mammary artery, each following aortic root surgery. These were successfully treated with percutaneous techniques. No complications were seen at follow-up as long as 2 years. Patients with Loeys–Dietzs syndrome require comprehensive long-term vascular follow-up and are likely to require percutaneous vascular interventions.

ABBREVIATIONS

EDS = Ehlers–Danlos syndrome, LDS = Loeys–Dietz syndrome, LIMA = left internal mammary artery, RIMA = right internal mammary artery, TGF-ßR = transforming growth factor-ß receptor

The Loeys–Dietz syndrome (LDS) is a multisystem connective tissue disorder with autosomal-dominant inheritance and is associated with mutations in the genes for transforming growth factor-ß receptor (TGF-ßR) 1 and 2 (1). The mortality and morbidity associated with various forms of aortopathy is often caused by aortic dissection (1). Increasing aortic size is associated with increased risk of dissection and rupture. Management strategies have largely been guided by serial follow-up of aortic size to determine the timing of prophylactic intervention (2). Patients with LDS appear to have a more malignant course than those with other aortopathies such as Marfan syndrome, and indeed recent guidelines have a lower threshold for recommendation of elective aortic root replacement in these patients (2). In addition, these patients also have more extensive peripheral arteriopathy (1,3). The present report includes two illustrative cases to demonstrate that this is an important source of ongoing morbidity and that these patients require more comprehensive follow-up than needed for more common forms of aortopathy. The manuscript was exempt from the requirement for institutional review board approval.

CASE REPORTS

Case 1

A 38-year-old woman with no medical history of note presented in December 2008 with severe chest pain. Computed tomographic (CT) angiography demonstrated type A aortic dissection extending from the aortic root to the bifurcation involving the innominate artery and right common carotid artery. Emergency aortic valve, root, and ascending aorta replacement was performed with the use of a 25-mm Carbomedics composite graft (Sulzer Carbomedics, Austin, Texas). The postoperative course was complicated, and the chest was reopened twice because of ongoing bleeding. She was discharged after 6 weeks.

At clinic review 1 month after discharge, no acute problems were noted. Routine follow-up CT demonstrated successful surgical repair with residual dissection

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None of the authors have identified a conflict of interest.

J Vasc Interv Radiol 2015; 26:1539–1543

http://dx.doi.org/10.1016/j.jvir.2015.04.014

flap but no aneurysmal areas. The patient was well at annual clinical follow-up in 2010. In view of the presentation with type A aortic dissection at a young age, genetic testing for inherited aortopathy was performed and confirmed LDS.

The patient presented to the hospital in April 2011 with a brief episode of chest pain that resolved spontaneously. She was discharged with arrangement for outpatient cardiologic follow-up. She remained well, and a routine magnetic resonance (MR) imaging scan was performed in July 2011. This demonstrated a 5-cm pseudoaneurysm of the right internal mammary artery (RIMA), which was confirmed on CT (Fig 1). The patient was admitted, and coil embolization was performed via the right brachial artery (Figs 2, 3) with the use of a 5-F vertebral catheter (Merit Medical, South Jordan, Utah). Two 5-mm Nester coils (Cook, Bloomington, Indiana) were successfully deployed. Follow-up scanning, initially with CT and most recently with MR imaging in August 2014, have demonstrated that the pseudoaneurysm remains occluded and the aneurysm sac has resolved.

Case 2

A 55-year-old man presented in April 2008 with acute ascending aortic dissection and underwent emergency aortic valve and root replacement. The postoperative course was complicated by acute compartment syndrome of the left lower leg necessitating an above-knee amputation. Subsequent imaging showed that, in addition to the presenting ascending aorta dissection, an additional femoral artery dissection was present and the ischemic injury was likely to have been caused by cannulation of the false lumen during cardiopulmonary bypass.

The family history was suggestive of an inherited aortopathy, and genetic testing identified a TGF-ßR1 mutation. Following identification of the mutation, screening for peripheral arteriopathy was performed. MR angiography of the head and neck vessels identified a 17-mm cavernous sinus aneurysm. This has been asymptomatic and has remained stable on annual imaging follow-up.

In 2009, clinical assessment showed a pulsatile swelling in the right groin. CT confirmed a 5-cm right iliac aneurysm, and the patient underwent successful surgical repair with an aortobifemoral graft.

In April 2013, the patient was admitted with chest pain. CT demonstrated a 23-mm aneurysm of the left internal mammary artery (LIMA) that had not been present on previous imaging (Fig 4). This was amenable to percutaneous closure and was initially approached from the right femoral route. The narrow aortic true lumen was selected and the left subclavian artery was catheterized. A complex LIMA aneurysm was demonstrated, but the origin was small and displaced by the aneurysm sac, making catheterization difficult. This approach was eventually abandoned, and a left brachial approach was adopted to catheterize the LIMA with the

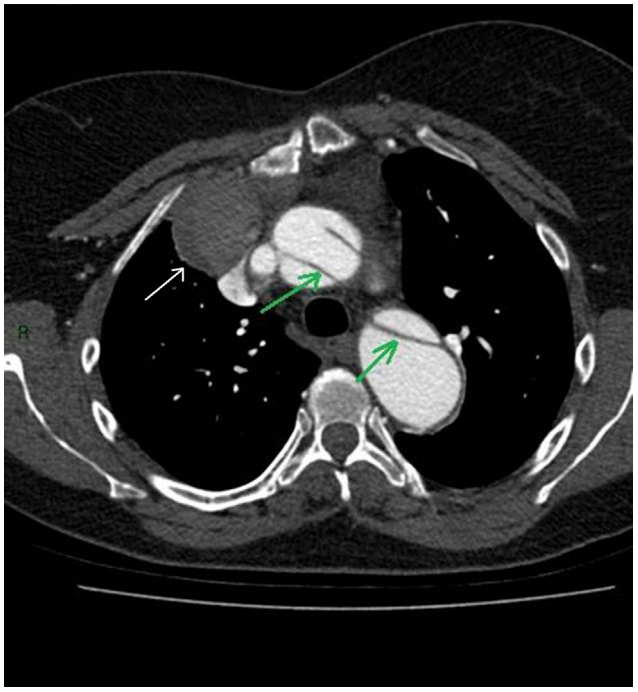

Figure 1. Transverse image from CT angiogram. The 5-cm RIMA pseudoaneurysm is seen anterior and to the right of the ascending aorta (white arrow). The residual dissection flap is seen within the ascending and descending aorta (green arrows).

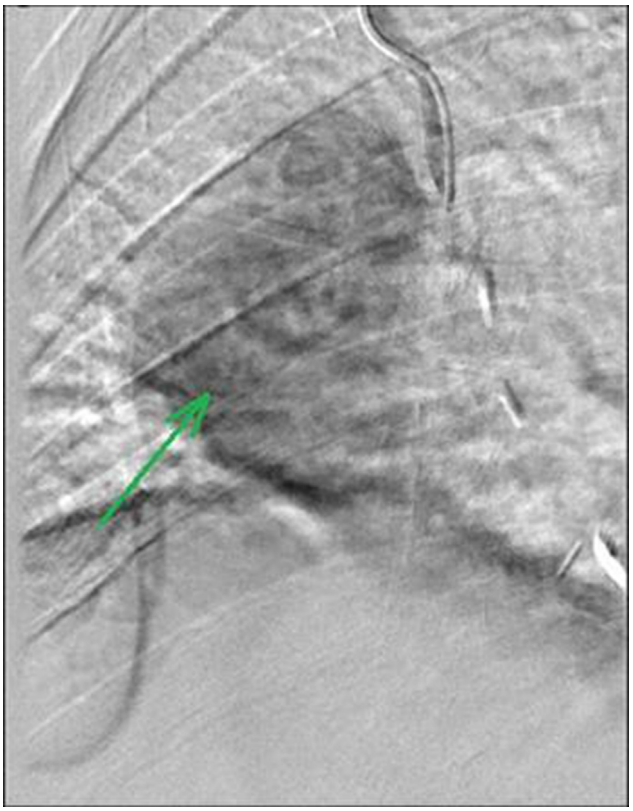

Figure 2. Angiogram demonstrates a pseudoaneurysm (arrow) with contrast material appearing after injection from the RIMA.