CASE REPORT

A rare cause of recurrent aortic dissection

Yashwant Agrawal [a,*], Vishal Gupta [b]

[a] Department of Internal Medicine/Pediatrics, Western Michigan University Homer Stryker School of Medicine, Kalamazoo, MI; [b] Department of Cardiology, Michigan State University/Borgess Medical Center, Kalamazoo, MI

[a,b] USA

We report the case of a 19-year-old man with a history of Loeys–Dietz syndrome (LDS), which was diagnosed when he had a Stanford type A aortic dissection. He also had multiple aneurysms including ones in the innominate, right common carotid, and right internal mammary arteries. He had had multiple procedures including Bentall's procedure, repeat sternotomy with complete arch and valve replacement, and coil embolization of internal mammary artery aneurysm in the past. His LDS was characterized by gene mutation for transforming growth factor-β receptor 1. He presented to our facility with sudden onset of back pain, radiating to the right shoulder and chest. He was diagnosed with Stanford type B aortic dissection and underwent thoracic aorta endovascular repair for his aortic dissection. This case represents the broad spectrum of pathology associated with LDS where even with regular surveillance and aggressive medical management the patient developed Stanford B aortic dissection.

Keywords: Aortic aneurysm, Aortic dissection, Aortic root replacement, Loeys–Dietz syndrome

Introduction

Loeys–Dietz Syndrome is an inherited connective tissue disorder of the vascular system. With no specific diagnostic criteria to recognize the syndrome, genetic testing is mandatory to identify transforming growth factor mutations. Most patients present with aortic aneurysmal rupture and/or dissection. Frequent monitoring with imaging is required to monitor such patients.

Case report

The patient was a 19-year-old man who in October 2013 had a Stanford type A aortic dissection with contained rupture and aortic root aneurysm (Fig. 1). He had Bentall's procedure with No. 25 St. Jude valved conduit and ascending aortic replacement with No. 24 Vaskutek branched graft. Genetic testing had revealed involvement of transforming growth factor β receptor 1 (TGFBR1) gene consistent with LDS. Imaging studies including computed tomography angiogram (CTA) and magnetic resonance angiogram of the head and

Disclosure: Authors have nothing to disclose with regard to commercial support.
Received 1 October 2015; revised 14 October 2015; accepted 12 November 2015.
Available online 21 November 2015

* Corresponding author at: Department of Cardiology, Borgess Medical Center, 1521 Shaffer Road, Kalamazoo, MI 49048, USA.
E-mail address: yashwantagrawal.agrawal@gmail.com (Y. Agrawal).

P.O. Box 2925 Riyadh – 11461KSA
Tel: +966 1 2520088 ext 40151
Fax: +966 1 2520718
Email: sha@sha.org.sa
URL: www.sha.org.sa

Peer review under responsibility of King Saud University.
URL: www.ksu.edu.sa
http://dx.doi.org/10.1016/j.jsha.2015.11.005

Production and hosting by Elsevier

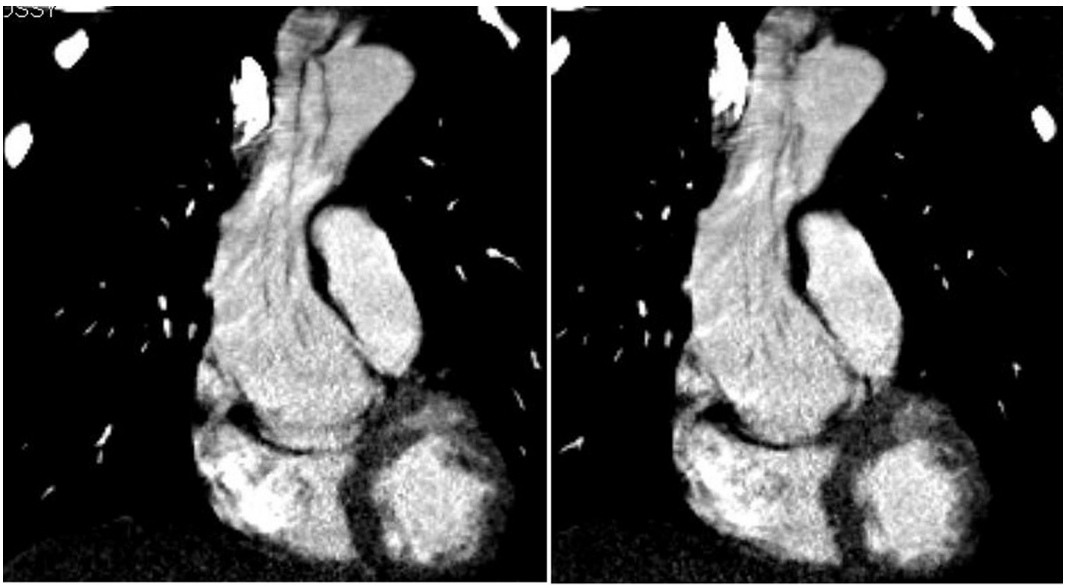

Figure 1. Computed tomography of chest, abdomen, and pelvis revealing Stanford type A aortic dissection with contained rupture and aortic root dissection extending into the right common carotid artery.

Figure 2. Computed tomography angiogram of the neck, chest and abdomen showing right common carotid artery and innominate artery aneurysm.

neck revealed multiple tortuous intracranial and cervical arteries consistent with LDS. He also had a 13 mm irregular right common carotid aneurysm.

In November 2013, when the patient had chest pain, concerns for new dissection arose and a

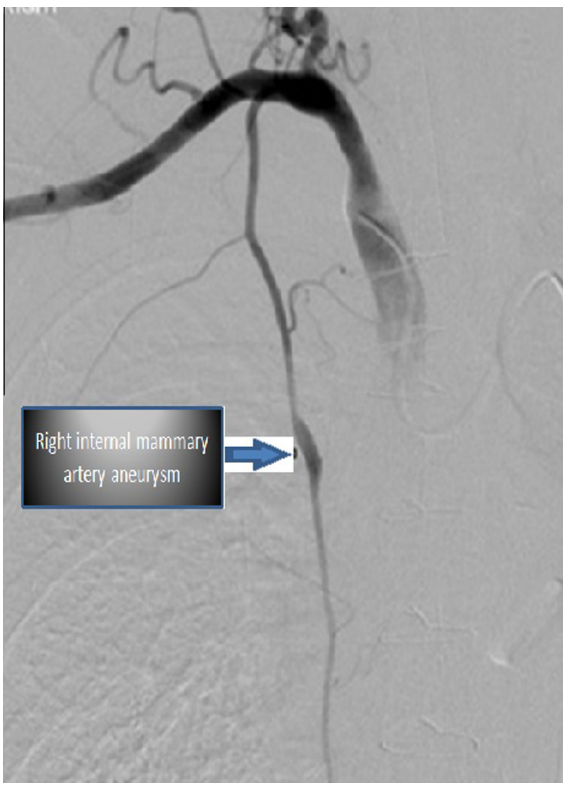

Figure 3. Fluoroscopy imaging showing right internal mammary artery aneurysm.

CTA of the neck, chest, and abdomen was performed, revealing an aortic arch dissection flap extending from the distal aspect of the graft to the mid-descending thoracic aorta. It also extended into the innominate, right subclavian, right common carotid, origins of the left subclavian and left vertebral arteries. Increasing size of innominate artery aneurysm and complete occlusion of the innominate artery just prior to its bifurcation were also found (Fig. 2).

In March 2014, diagnostic neurological arteriography to define the patient's vasculature was performed. It revealed vertebrobasilar steal syndrome with occlusion of the innominate artery and retrograde flow into the right vertebral artery filling the right subclavian artery. Follow-up testing revealed rapid enlargement of the aortic arch and innominate aneurysms related to chronic aortic dissection. For this pathology, surgery was indicated.

In June 2014, repeat sternotomy with complete aortic arch replacement with an elephant trunk No. 24 vascutek branched graft was performed. A bypass to the innominate artery bifurcation including separate bypasses to the right carotid and subclavian arteries was also performed. The right common carotid aneurysm was also repaired. Biopsy of the aorta showed mural myxoid degeneration with medial elastin fiber disorganization consistent with history of LDS.

In December 2014, coil embolization of right internal mammary artery aneurysm was performed (Fig. 3). A steady increase in the size of the aneurysm from 2 mm to 8 mm, noted on follow-up computed tomography scans between April 2014 and August 2014, prompted this intervention.

He presented in February 2015 to the emergency department at our facility with sudden onset of sharp back pain, 10/10 in intensity radiating to

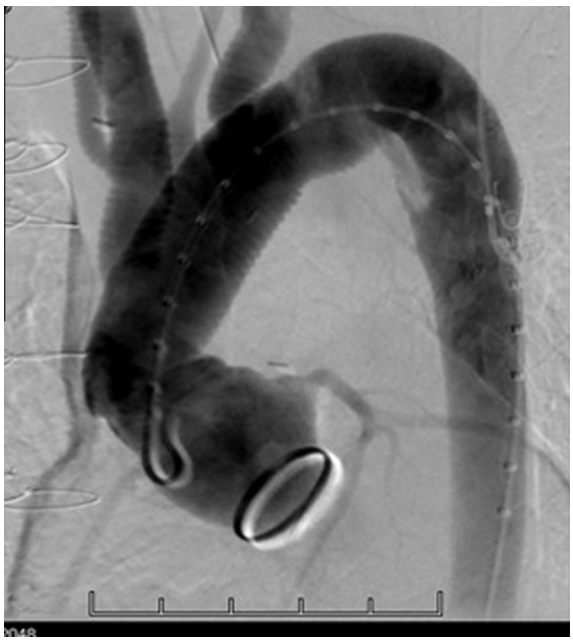

Figure 5. Thoracic aortogram during the right internal mammary artery aneurysm coil embolization.

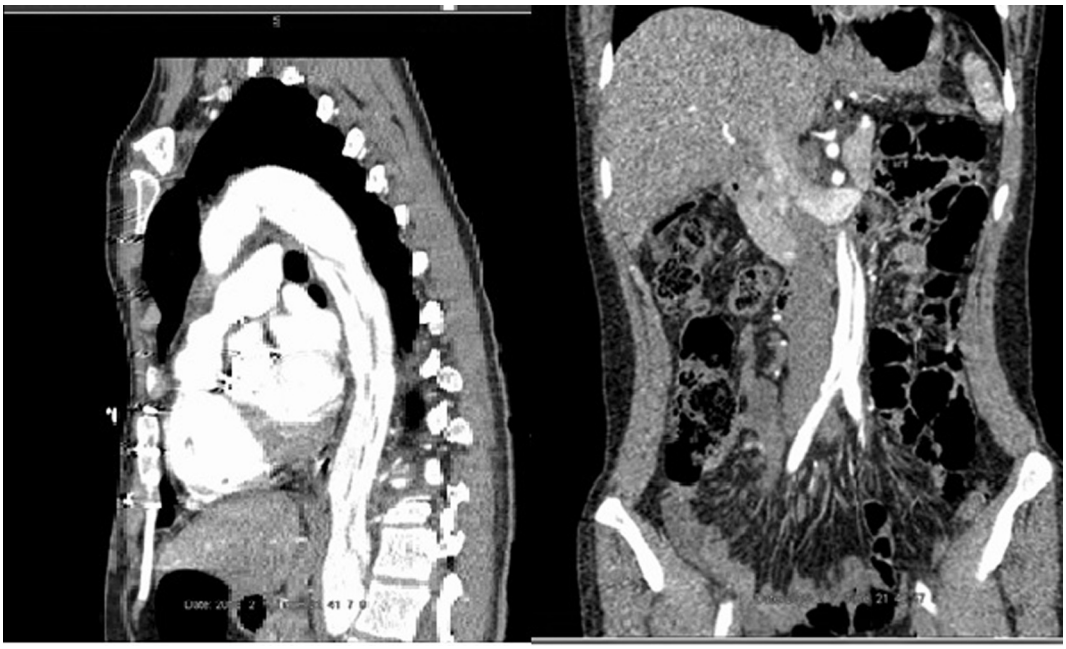

Figure 4. Computed tomography of chest, abdomen, and pelvis revealing Stanford Type B aortic dissection extending from the left subclavian artery to the proximal right common iliac artery.

the right shoulder and chest. Vitals including blood pressure were in the normal range. Physical examination was pertinent for a mechanical click consistent with aortic valve replacement surgery, which the patient had had more than a year previously. No craniofacial abnormalities were noteworthy, except for the patient being myopic. Family history was significant, with his father being tested for LDS. Laboratory data were unremarkable.

With a history of LDS predisposed to aneurysms and dissections, a CTA of the chest, abdomen, and pelvis was performed. This revealed a Stanford type B aortic dissection extending from the left subclavian artery to the proximal right common iliac artery (Fig. 4). All major aortic branches were patent. The patient underwent thoracic aorta endovascular repair with multiple overlapping Gore C-TAG devices, including three proximal extensions and primary repair of the right common femoral artery. The postoperative course was uncomplicated and the patient was discharged in 6 days. The patient has since been asymptomatic and is doing well at his regular out-patient follow-up appointments.

Discussion

LDS is a connective tissue disorder, first described in 2005 [1], with an autosomal dominant mode of inheritance. The syndrome is associated with genetic mutations in TGFBR1 genes leading to an increase in TGF-β activity [1]. The causality of this process is disorganized elastin fibers and collagen overproduction, leading to vessel wall instability, instigating aneurysms, dilatations, and dissections. Primarily, two types of LDS have been described, Type 1 with and Type 2 without craniofacial abnormalities [2]. Our patient presented with no obvious craniofacial anomalies as seen in Type 1 LDS.

Loeys and Dietz [1,2] described LDS as a fatal genetic disease with a mean survival age of 26 years. The likely cause of mortality is aortic aneurysms, dissections and/or rupture. Moreover, it is associated with multiple aneurysms and arterial tortuosity throughout the body [1,2].

Yearly screening with echocardiogram for ascending aorta anatomy has been proposed [3] for LDS patients. After obtaining a basal magnetic resonance angiogram and CTA of head to pelvis, at least a biennial visualization of each vasculature structure is proposed in these patients [4]. These recommendations are flexible: patients might require more frequent imaging as per their phenotype. A thoracic aortogram performed in December 2014, when the patient underwent coil embolization of internal mammary artery, did not reveal new pathology (Fig. 5). LDS is a largely an unknown entity with this case highlighting its unpredictable and aggressive nature, where frequent surveillance can also leave more surveillance desired.

References

[1] 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.

[2] Loeys BL, Schwarze U, Holm T, Callewaert BL, Thomas GH, Pannu H, et al.. Aneurysm syndromes caused by mutations in the TGF-beta receptor. N Engl J Med 2006;355:788–98.

[3] Arslan-Kirchner M, Epplen JT, Faivre L, Jondeau G, Schmidtke J, De Paepe A, et al.. Clinical utility gene card for: Loeys–Dietz syndrome (TGFBR1/2) and related phenotypes. Eur J Hum Genet 2011;19. http://dx.doi.org/10.1038/ejhg.2011.68.

[4] MacCarrick G, Black 3rd JH, Bowdin S, El-Hamamsy I, Frischmeyer-Guerrerio PA, Guerrerio AL, et al.. Loeys–Dietz syndrome: a primer for diagnosis and management. Genet Med 2014;16:576–87.