Concomitant coronary artery disease in a patient with Loeys-Dietz Syndrome and multiple aortic aneurysms

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Introduction

Loeys-Dietz syndrome (LDS) is a recently described connective tissue disorder characterized by early and aggressive onset of aneurysm formation and dissection of the aorta.[1] The lesions in these patients present early, often in childhood, and at much smaller aortic diameters.[2] Mean age of survival for patients diagnosed with LDS is 37 years.[3] These patients also have a typical phenotype: including a bifid uvula, hypertelorism, cleft palate, and vascular tortuosity derived from genetic alterations in TGFβ.[1, 3] Specifically, management of thoracic vascular abnormalities drives the treatment of these patients. We describe a patient with LDS who presented with multiple aortic aneurysms, cerebral aneurysms and surprisingly, concomitant coronary artery disease.

Case

A 52-year-old man presented to the emergency room with lower back pain. He had strained his back a few months previously and the pain never resolved. He eventually underwent an MRI and was found to have an eight centimeter abdominal aortic aneurysm. He was referred for aortic consultation and after further questioning, was found to have a significant family history. His brother had previously undergone successful valve sparing aortic root replacement and was diagnosed with LDS. Our patient had refused both genetic testing and imaging at the time of his brother’s diagnosis. After genetic testing, he was found to have the TGFBR1 mutation, which was the same mutation as his brother, and the diagnosis of LDS was made. He underwent further imaging studies and was found to have a 5 cm aortic root aneurysm, 8 cm infra-renal aortic aneurysm, 7mm left cavernous internal carotid artery aneurysm and a 7 mm basilar artery aneurysm [Figure 1]. Because of size and symptoms, plans were made for initial abdominal aortic aneurysm repair, followed by subsequent aortic root replacement. He underwent successful open aorto-bi-iliac infra-renal aneurysm repair.

In preparation for his aortic root surgery, a cardiac catheterization was performed and a 75% stenosis of his proximal left anterior descending artery was found [Figure 2]. Echocardiogram confirmed the dilated aortic root and revealed a tricuspid aortic valve with mild aortic insufficiency.

After recovering from his abdominal aortic aneurysm repair, he underwent a valve sparing aortic root replacement with coronary artery bypass grafting. The aortic root was replaced with a Valsalva graft, the aortic valve spared and re-implanted in the fashion of a David IV procedure. The distal aortic (proximal to the innominate artery) and coronary button anastomoses were

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patients with dissections of the coronary arteries,[4, 5] but never
with a stenotic lesion requiring intervention. The prevalence of
coronary artery disease is unknown in patients with LDS, likely
because most patients with this syndrome do not progress to an age
when coronary angiography would be indicated. When evaluating
surgical vascular disease in these complex connective tissue
patients, concomitant coronary disease cannot be dismissed, and
should be investigated in appropriate patients based on established
clinical guidelines.

Conclusion

We present a patient with LDS who had concomitant coronary
artery disease, highlighting the value of a thorough workup
in patients with complicated connective tissue disorders. This
emphasizes the need to consider routine cardiac diagnoses and
appropriate workup for patients with such vascular diseases.

Disclaimers

The views expressed in this article are those of the authors and
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This work was not been previously presented in any format.

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Discussion

LDS is a connective tissue disorder first described by Loeys et al
in 2005.[1] Similar to Marfan's Syndrome, this disease is autosomal
dominant and patients have a severe defect in elastogenesis as
a result of a mutation in the TGFBR gene. It is well known for
its aggressive vascular involvement, especially aortic dissections
and aneurysms. Because of the high risk of aortic dissections and
aneurysmal disease, patients with LDS undergo intervention
sooner than the average population.[2, 3] When one vascular
defect is found, it is recommended that complete body imaging
be done in order to evaluate for the presence of other aneurysmal
disease. The life expectancy of these patients is decreased when
compared with the general population. Typical of this disease, our
patient was found to have multiple vascular aneurysms. Though
not the oldest patient described with LDS, our patient is much
older than average life expectancy and because of his relatively
advanced age, we elected to perform a coronary angiogram. To our
knowledge, this is the first described case of coronary artery disease
in a patient with LDS. There have been previous descriptions of

Figure 2: LHC showing ostial stenosis of the left anterior
descending coronary artery. (Arrow denotes stenotic lesion.)

reinforced with Teflon felt. This was performed with standard
cardiopulmonary bypass, distal aortic cross clamp and antegrade
myocardial protection. The left anterior descending coronary artery
was grafted using the left internal mammary artery, harvested in a
pedicle fashion. The procedures and recovery were unremarkable.
His one month follow-up echocardiogram showed no aortic
insufficiency and his computed tomography scan showed intact
aneurysm repairs.