THE SPECTRUM OF AORTIC DISEASE IN LOEYS-DIETZ SYNDROME

ACC Poster Contributions
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Background: Loeys-Dietz syndrome (LDS), due to mutations in TGFBR1 and 2, is characterized by aortic aneurysms and dissections at younger ages than those typically seen with Marfan syndrome. We describe the cardiovascular manifestations of LDS in a single tertiary center.

Methods: Records were reviewed from 26 patients with LDS evaluating cardiovascular features and aortic disease.

Results: Patients ranged from age 9-55 years, with 10 males and 16 females. 19/26 underwent aortic surgery at ages 14-43 years. 4 adults suffered aortic dissection: 3 as the initial presentation. Aortic root size was 3.9 cm at the time of dissection in one patient. One patient died from an aortic arch dissection months after prophylactic root replacement and at the time of dissection the arch measured 3.5 cm. Patients with aortic dissection required multiple subsequent aortic operations. 4 patients underwent total aortic replacement. Branch vessel involvement requiring repair included aneurysms of the brachiocephalic and subclavian arteries. 11 patients underwent valve-sparing (VS) aortic root replacement and 8 patients underwent composite aortic valve and root replacement (CVG). Of the 11 patients with VS root replacement, one required reoperation due to failed Ross procedure in a patient previously diagnosed as bicuspid aortic valve disease alone. Two others who underwent a Yacoub-type “remodeling” VS procedure developed early splaying of the remaining aortic sinus tissue with progressive enlargement of the aorta. Patients who have had a David “reimplantation” VS procedure or CVG have had stable repairs. Other cardiac procedures have included repair of an atrial septal defect, patent ductus arteriosus and replacement of the mitral valve.

Conclusions: Patients with LDS have aggressive aortic and arterial disease. They differ from Marfan patients by the characteristic triad of hypertelorism, bifid uvula and arterial tortuosity. LDS patients are at risk of aortic catastrophes at relatively small aortic size and often require multiple aortic or vascular procedures. Prophylactic aortic root replacement with David VS technique and CVG appear effective. Serial imaging of the entire arterial tree is necessary.