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*Brief Report*

# Valve-Sparing Replacement of the Aortic Root for a 2-year-old Child with Loeys-Dietz Syndrome

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**Abstract** Loeys-Dietz syndrome is a newly recognized constellation that presents with aortic aneurysm or dissection similar to Marfan's syndrome. We describe successful surgical treatment in a 2-year-old with the syndrome in whom we performed a valve-sparing replacement of the aortic root because of significant dilation of the aortic root and the ascending aorta.

Keywords: Genetic syndrome; aortic root dilation; ascending aortic aneurysm

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Loeys-Dietz syndrome,[1] an autosomal dominant genetic syndrome, demonstrates many clinical characteristics similar to Marfan's syndrome, including aortic aneurysm, aortic tortuosity, and aortic dissection. Additional classic physical characteristics include orbital hypertelorism, cleft palate, and bifid uvula. We describe successful treatment by means of valve-sparing replacement of the aortic root because of significant dilation of the aortic root and ascending aorta in a 2-year-old with the syndrome.

## Clinical Summary

A male infant was delivered at 40 weeks gestation by Caesarean section. He was brought to the neonatal intensive care unit due to his respiratory distress. An echocardiogram revealed patency of the arterial duct. The duct was surgically ligated when he was 2 months of age without any complications. Bilateral inguinal hernias were repaired when he was aged 5 months. At this time, multiple broken ribs at different stages of healing were noted on the chest X-ray. An extensive workup, including genetic testing, led to the diagnosis of Loeys-Dietz syndrome. An echocardiogram at 2 months of age had revealed Z-score of 1.6 in the ascending aorta, but subsequent serial echocardiograms documented rapid dilation, the root reaching 35 mm in diameter, equivalent to a Z-score of 11.5, when he was 22 months of age. By this time, he had developed aortic regurgitation. The patient was referred to Rainbow Babies and Children's Hospital Multidisciplinary Connective Tissue Program for further management. He weighed 12.2 kg, representing the 50th centile, and his height was 91.5 cm, at the 97th centile. An early diastolic murmur, graded at 2 from 6, and medium to high-pitched, was audible at the left lower sternal border, but there were no systolic murmurs. Echocardiography confirmed moderate aortic regurgitation as well as mild to moderate left ventricular dilation, with systolic and diastolic dysfunction. The aortic valve was dilated, being measured at 21.4 mm in diameter. Computed tomography of the chest showed dilation of the aortic root and proximal ascending aorta, with tortuosity of the distal transverse and descending aortic segments (Fig. 1).

At 24 months of age, the patient underwent replacement of the aortic root, but with sparing of the aortic valve. Total cardiopulmonary bypass was established between the distal ascending aorta and both caval veins. Myocardial protection was

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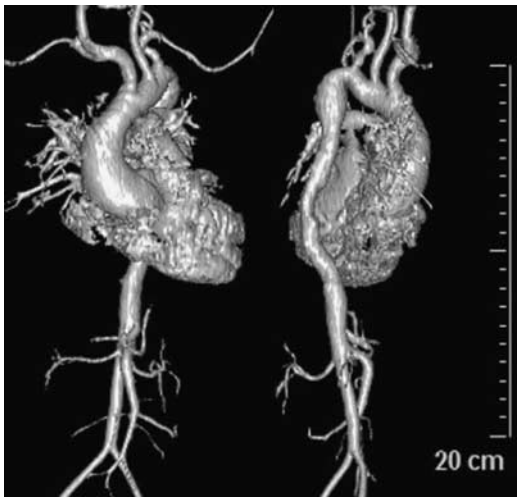


**Figure 1.**
*Computed tomography emphasizes the remarkable proximal aortic dilation and distal tortuosity, with dilation of the aortic branches, including the superior mesenteric artery.*

achieved using moderate systemic hypothermia at 32°C, and antegrade cold blood cardioplegia, followed by retrograde cardioplegia, repeated every 20 minutes. The aortic root was replaced, with re-implantation of the valve, according to the technique described by David and associates.[2] A 20 mm Gelweave Valsalva prosthesis (Terumo Cardiovascular Systems Corp., Ann Arbor, Michigan) was used for the reconstruction. The aortic cross clamp was taken off and the cardiopulmonary bypass was safely discontinued. Intraoperative echocardiography after termination of the cardiopulmonary bypass showed mild aortic regurgitation from the central area. The postoperative clinical course was uneventful, and the child was discharged home on the 6th postoperative day. Follow-up echocardiography one month after the surgery showed normal left ventricular size, with significantly improved systolic and diastolic function.

## Discussion

Loeys-Dietz syndrome is classified in 2 types, the first with craniofacial features such as hypertelorism and bifid uvula, and the second type without these features. Patients falling into the first type tend to demonstrate more severe cardiovascular anomalies than those placed in the second group. Our patient would have been categorized in the first type, and had undergone dramatic progression of both aortic dilation and regurgitation prior to his referral. Significant dilation of the aorta is seen even in small children with the syndrome.[3–6] Current indications[3,7] include surgery for those with an aortic root having Z-scores greater than 3.0 for those falling in to the first type, and Z-scores greater than 4.0 for those with the second

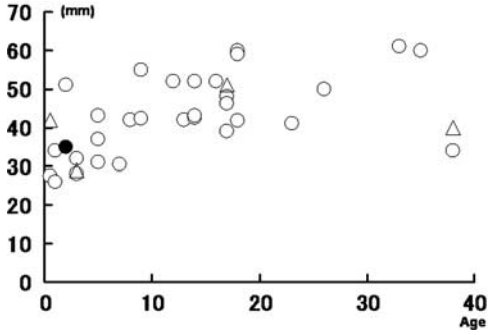


**Figure 2.**
*Aortic diameter in patients with Loeys-Dietz syndrome as assembled by searching the literature. White circles indicate surgical cases. White triangles indicate deceased cases. The black circle indicates our surgical case.*

type, or those with rapidly changing dimensions in the root (Fig. 2). Recognition of this syndrome with diligent serial follow-up of the cardiovascular system, even in small children as indicated by our report, is needed to prevent sudden death or aortic dissection.[4,5]

Replacement of the root whilst sparing the valve is becoming a standard approach for patients having Marfan's syndrome and aneurysmal aortic roots.[1] We successfully accomplished the valve-sparing replacement using a 20 mm graft without any sign of significant valvar dysfunction in our 2-year-old. Ours is one of the youngest patients with Loeys-Dietz syndrome who has undergone successful surgical treatment using the valve-sparing approach for significant dilation of the aortic root and proximal ascending aorta. Replacement of the valve with an allograft is an alternative approach in a small child, albeit that degeneration of such allografts is well recognized in children.[8] It would also be possible to replace both the aortic valve and root using a mechanical valved composite conduit as in the Bentall procedure. The risk of lifelong anticoagulation in young patients, however, is substantial, and may contribute to decreased survival.[9] Even in small children such as our patient, therefore, a valve-sparing approach might be the preferred approach. Continued follow-up will be needed to assess the long-term ramifications of this approach for children with Loeys-Dietz syndrome.

Recently, use of angiotensin II type 1 receptor blockers has demonstrated effectiveness in the prevention of aortic dilation in patients with Marfan's syndrome.[10] Their effectiveness in patients with Loeys-Dietz syndrome is unproven, albeit that an inhibitor of angiotensin-converting enzyme was shown to prevent further dilation of the ascending aorta.[4] Although our patient was treated postoperatively with losartan, a blocker of angiotensin II type 1 receptors, further serial follow-up of his cardiovascular system will be essential.

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