



Type a Aortic Dissection and Impaired Systolic Function in a Child with Loeys-Dietz Syndrome

Yan Li*, Qian Chang, Xiangyang Qian, Xiaogang Sun and Mingyao Luo

Department of Cardiovascular Surgery, National Center for Cardiovascular Diseases and Fuwai Hospital, China

Abstract

Loeys-Dietz syndrome (LDS) is a recently recognized genetic aortic aneurysm syndrome characterized by the triad of hypertelorism, bifid uvula or cleft palate, and generalized arterial tortuosity with aneurysms and dissections throughout the arterial tree with a high risk of early-onset aortic dissection but with no known myocardial involvement [1]. We report a child who was diagnosed as acute type A aortic dissection, a novel transforming growth factor beta (TGF β) receptor type I (TGFBR1) mutation, and impaired systolic function after urgent surgical repair.

Keywords: LDS: Loeys-dietz Syndrome; TGF β : Transforming Growth Factor β ; TGFBR1: TGF Beta-receptor Type I; LVEF: Left Ventricular Ejection Fraction

Case Presentation

A 7-year-old boy visited the emergency room with a complaint of sudden-onset chest pain and dyspnea. At the age of 6, this boy was diagnosed as aortic root aneurysm with mild aortic regurgitation. At the age of 7 years he showed height 140 cm and arm span 136 cm. A thorough physical examination demonstrated dolichocephaly, hypertelorism, hypoplastic alae nasi, high-arched palate, microretrognathia, joint hypermobility, which suggested LDS. Echocardiography demonstrated a large aortic root aneurysm with a dissection membrane, left ventricle diameter 50 mm, and severe aortic regurgitation. Therefore, a CT imaging was made rapidly, which clarified acute type a aortic dissection without involvement of the coronary arteries and aortic root diameter 75 mm (Figure 1). Suspicion for LDS was confirmed by genetic testing, which identified a novel heterogeneous non-frame shift deletion c.G678_680del AGA in exon 4 of TGBFR1 in the highly conserved kinase domain of the gene, which could subsequently result in the heterogeneous p.Glu228del variation (Figure 2). An urgent operation was performed by a median sternotomy and total cardiopulmonary bypass (CPB). Aortic root replacement was done using a modified Bentall technique. The postoperative course was uneventful, and the patient was discharged 10 days postoperatively without any complications. The postoperative CT imaging and echocardiogram revealed patent epicardial coronary arteries, global systolic dysfunction (left ventricular ejection fraction, LVEF, 45%), and dilated left ventricle (left ventricle diameter 65 mm). He was treated with diuretics, digoxin, and an angiotensin-converting enzyme inhibitor. He developed fatigue on

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*Correspondence:

Yan Li, Department of Cardiovascular Surgery, National Center for Cardiovascular Diseases and Fuwai Hospital, Chinese Academy of Medical Sciences and Peking Union Medical College, 167 Beilishi Rd, Beijing, 100037, China, Tel: 86-10-88398269; Fax: 86-10-68332500; E-mail: fuwai8359@126.com

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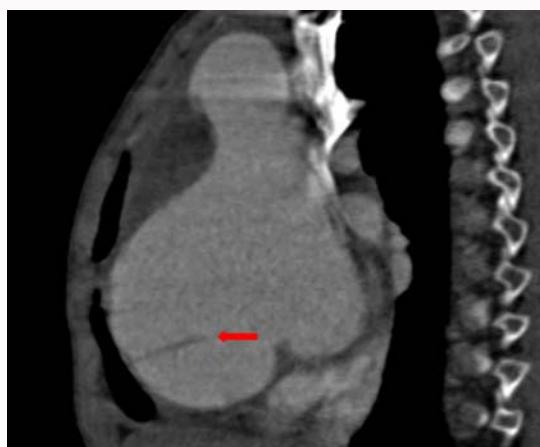


Figure 1: Sagittal reconstruction shows dilation of Valsalva sinuses with a dissection membrane (red arrow).

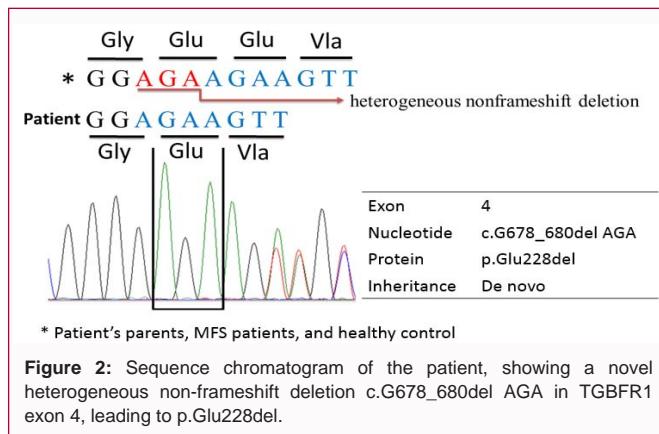


Figure 2: Sequence chromatogram of the patient, showing a novel heterogeneous non-frame shift deletion c.G678_680del AGA in TGBFR1 exon 4, leading to p.Glu228del.

postoperative day 45, and echocardiogram revealed global systolic dysfunction again (LVEF 34%). His fatigue and left ventricular systolic function improved to 42% with medical therapy on postoperative day 90. Cardiomyopathy was suspected and a strict surveillance was recommended.

Discussion

The most important finding of LDS is the aggressive and rapid progression of aortic pathology even though the patients are young

in age. Notably, what makes these TGF β -associated cardiovascular diseases more challenging is that they affect many cardiac tissues in the same patients [3,4]. Only one case of cardiomyopathy due to microvascular dysplasia a LDS adult has been described [4], however, cardiomyopathy in LDS children has not previously been described to the best of our knowledge. Although it is possible that LDS and cardiomyopathy are unrelated, it seems plausible that the TGFBR1 mutation in LDS could cause microvascular dysplasia and subsequent myocardial dysfunction. Further study may provide an opportunity to clarify the significance of this finding.

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