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CASE REPORT

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Thoracoabdominal aortic replacement in a 6-year-old boy with Loeys-Dietz syndrome

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Abstract

Connective tissue disorders such as Marfan- and Loeys-Dietz syndrome (LDS) can lead to aortic aneurysms and aortic dissections in children. Patients with LDS often necessitating multiple aortic surgeries throughout their lives to extend their lifespan. A boy with LDS underwent Bentall procedure at the age of three for aortic aneurysm. At the age of six, this boy was referred to the hospital again due to severe abdominal pain. Computed tomographic angiography (CTA)indicates aortic dissection (DeBakey Type III, Stanford Type B). After a multidisciplinary team discussion, a successful thoracoabdominal aortic replacement was performed.

Keywords Loeys-Dietz syndrome, Thoracoabdominal aortic replacement, Aortic dissection, Children

Background

Loeys-Dietz syndrome (LDS) is an autosomal dominant genetic disorder that causes connective tissue abnormalities in multiple systems of the body. LDS is characterized by traits of arterial tortuosity and aneurysms, hypertelorism, and bifid uvula or cleft palate [1, 2]. We describe a case involving a 6-year-old boy with LDS who successfully underwent thoracoabdominal aortic replacement. The purpose of reporting this case is its educational significance for cardiothoracic surgeons and pediatricians.

Case presentation

A 6-years-old boy with abdominal pain was admitted to our hospital. Physical Examination after Admission: Heart rate 140 beats per minute, respiratory rate 20 breaths per minute, blood pressure 116/65 mmHg, weight 20 kg, arterial saturation 98%. The patient is tall and thin. There is bilateral exotropia and pectus carinatum. There are deformities in the joints of the limbs: contracture of the interphalangeal joints of the fingers, bilateral clubfoot, and deformity of the toe joints with flexion.

The boy was previously seen in our hospital in 2019, and echocardiography examination revealed ascending aortic aneurysm (Aortic Root Z-Scores:11.7), congenital aortic insufficiency (severe) and patent ductus arteriosus. Genetic testing revealed a heterozygous mutation in the TGFBR2 gene (C.1411G>T, p.D471Y), leading to the diagnosis of LDS type 2. Emergency aortic valve and ascending aorta replacement surgery (Bentall procedure, Mechanical valve 25, Graft 28 mm), aortic arch resection with artificial graft replacement (D: 24*10 mm, L: 50 cm), ductus arteriosus ligation was performed. After 1-year post-Benatll procedure, the computed tomographic angiography (CTA) showed the maximum aortic diameter of 12 mm. Besides the cardiac surgery in 2019, the patient also underwent craniotomy for the resection of a prepontine arachnoid cyst at our hospital in 2020, and hand surgery for polydactyly in an external hospital in 2023.

CTA was performed during admission. CTA indicates aortic dissection (DeBakey Type III, Stanford Type B), involving the aortic arch to the abdominal aorta (at the mid-level of the left kidney). The intimal tear is located

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at the T5 level. A significant hematoma surrounds the aorta at the level of the diaphragm(Fig. 1.A). Electrocardiogram is normal. Echocardiography shows left ventricular hypertrophy, mechanical valve function is normal, and the descending aorta exhibits an intimal flap. On the first day of admission, the boy developed dyspnea, with arterial saturation dropping to 93%. Endotracheal intubation was performed for assisted ventilation. The boy was not anticoagulated with warfarin or low molecular heparin at time of dissection. International normalized ratio remained between 2.0 and 2.8. Following multidisciplinary discussions, thoracoabdominal aortic replacement surgery was performed.

The patient was placed in the right lateral position, exposing the left inguinal region. The chest cavity was opened at the left fifth intercostal space. The aorta revealed an aneurysmal dilatation. The maximum diameter of the dissecting aortic aneurysm was approximately 5.5 cm. At the level of the diaphragm, the aneurysm formed a large surrounding hematoma. Cannulation was performed through the inferior vena cava and the abdominal aorta to establish extracorporeal circulation. Proximal cross-clamping was applied to the left subclavian artery, and cross-clamping of the aorta was performed at the level above the celiac trunk. The aneurysm was incised, and the thrombus was extracted. Trimming of the aorta was performed, with proximal aorta and artificial blood vessel (MAQUET, polyester, D:10 mm) subjected to an end-to-end anastomosis. The abdominal aorta, mesenteric artery, and bilateral renal arteries were cut at an angle, and an end-to-side anastomosis was performed with the artificial graft. The T10-T11 intercostal artery was anastomosed to the side of the artificial blood vessel with an island flap, and the remaining intercostal arteries were sutured closed. The overall extracorporeal circulation time was 276 min. The patient was discharged on the 11th day postoperatively. A team consisting of pediatric and cardiac surgeons conducted follow-up on the patient. The girl had mild chest pain postoperatively, with no neurological complications observed. CTA scans were planned at the first and third months postoperatively.

The postoperative CTA of the patient at one (Fig. 2.D) and three months (Fig. 2.E) revealed that the artificial blood vessel was unobstructed and there was no stenosis at the anastomosis sites. However, compared to the CT scans taken one month postoperatively, the false lumen of the descending aortic arch in the third month postoperative CTA appeared enlarged, forming a localized bulge (Fig. 2.F). The Adamkiewicz artery is visualized. We plan

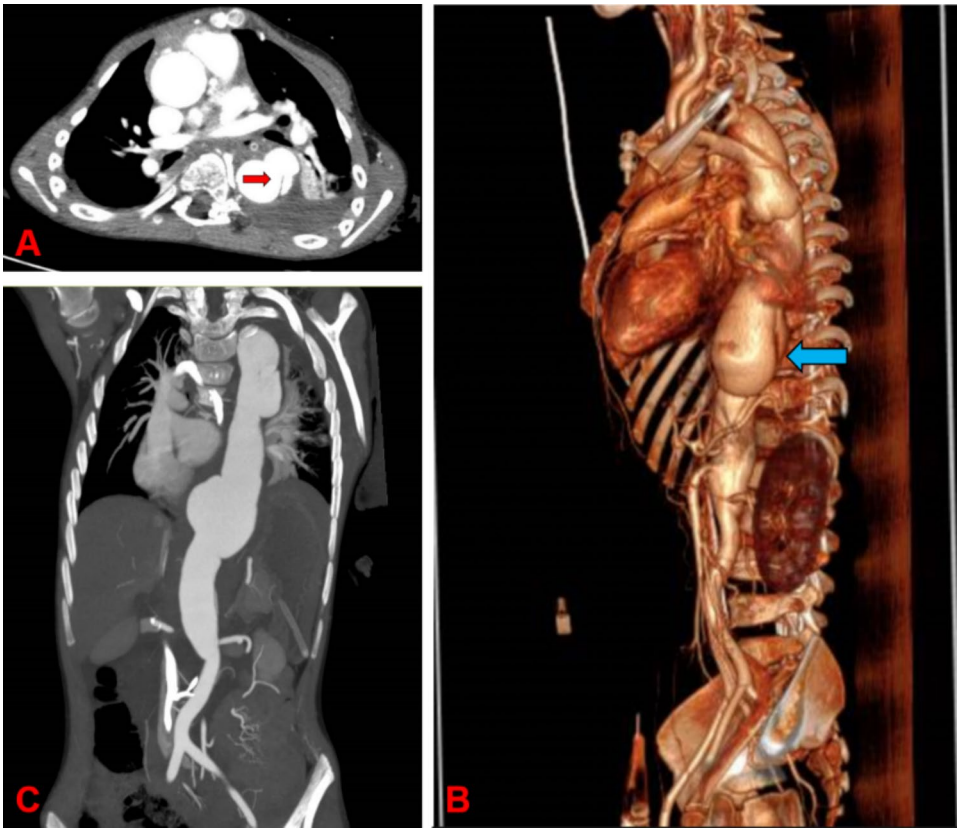

Fig. 1 **A**) a large intimal fissure (red arrow). **B**) Preoperative 3-dimensional computed tomographic image showing the aortic dissection near the diaphragm (blue arrow) forming a huge hematoma around it. **C**) Marked dilatation of the thoracoabdominal aorta and formation of a huge aneurysm

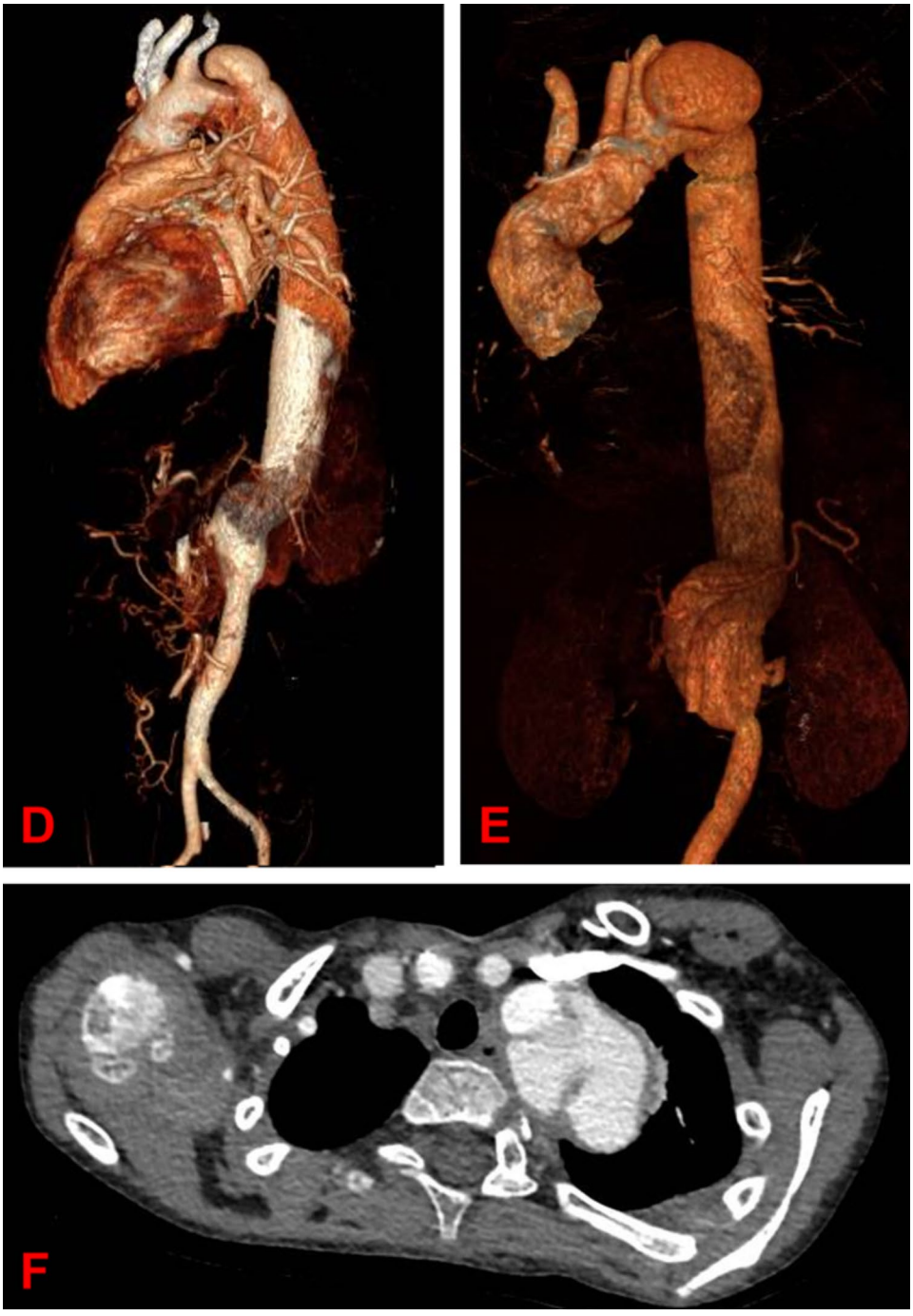

Fig. 2 **D**) Postoperative CTA three-dimensional reconstruction in the first month. **E**) Postoperative CTA three-dimensional reconstruction in the third month. **F**) CT scan in the third month after surgery showed an enlargement of the false lumen in the descending thoracic aorta

to perform aortic arch replacement on the boy. However, for the boy's better recovery and overall health improvement, we aim to maximize the interval between the second and third interventions.

Discussion and conclusions

Patients with LDS syndrome may experience aortic dissection and aneurysm at a very young age, and compared to the disease presentation in adults, the onset of aortic dissection and aneurysm is more abrupt and progresses rapidly in children [3]. The indications for prophylactic treatment of the aorta in patients with LDS include: adult patients with an aortic diameter exceeding 4 cm;

aortic root diameter growth > 0.5 cm/year; pediatric LDS syndrome patients with an aortic diameter exceeding the 99th percentile for the average age of children [4]. For pediatric and adolescent patients, the diameter of the artificial graft selected for surgery should be slightly larger than the measured aortic diameter obtained from CT scans, allowing the graft to meet the growing needs during the developmental process [3]. It is not recommended to use endovascular stent intervention therapy in patients with genetic aortic syndromes (LDS syndrome and Marfan syndrome). Open repair of the descending aorta and thoracoabdominal aorta is the preferred approach, as stent intervention therapy may lead to late stent graft leakage due to continuous vessel expansion or persistent perfusion of the false lumen [4]. After surgery, patients should undergo an annual comprehensive CT examination of the aorta to assess the compatibility of artificial graft with the body size. The occurrence of claudication and reduced exercise tolerance may suggest that the size of the graft is insufficient to meet the blood supply to the lower extremities, indicating the potential need for repeat surgery to replace them with larger diameter interposition graft [5]. Multidisciplinary teams contribute to the treatment and follow-up of patients with LDS. Family education of such conditions at pre-discharge and outpatient setup might be life-saving for underaged minors.

Abbreviations

LDS Loeys Dietz syndrome
CTA Computed Tomographic Angiography

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Not applicable.

Author contributions
Zhen was a major contributor in writing the manuscript. Yu was the main participant in the surgery. All authors read and approved the final manuscript.

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Data availability
No datasets were generated or analysed during the current study.

Declarations

Ethics approval and consent to participate
The ethics committee of Guangdong Provincial People's Hospital, China, approved the study and waived the requirement for written informed consent.

Consent for publication
We have obtained consent from the parents to publish the case.

Competing interests
The authors declare no competing interests.

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