

Ruptured abdominal aortic aneurysm repair in pediatric Marfan syndrome patient

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ABSTRACT

Marfan syndrome is a well-described autosomal dominant connective tissue disorder with a constellation of anatomic characteristics including aortic degeneration as a result of the spontaneous mutation of the fibrillin gene, *FBNI*. Whereas life-threatening dissection and ascending aneurysmal rupture have been thoroughly documented in the literature, aneurysms of the abdominal aorta and those present in the pediatric population have only rarely been reported. In this case report, we describe presentation, successful open surgical repair, and recovery of a pediatric Marfan syndrome patient with a ruptured abdominal aortic aneurysm. (*J Vasc Surg Cases and Innovative Techniques* 2018;4:20-3.)

Marfan syndrome (MFS) has been well studied across many specialties.¹⁻⁴ In vascular surgery, only a few papers have described ruptured abdominal aortic aneurysm (AAA) undergoing surgical repair.^{5,6} Within the literature, pediatric cases of ruptured aortic aneurysms have not been well described compared with the adult population as the presentation is rare, and most descriptions were post mortem.⁷⁻¹⁸ There have been a few documented cases of pediatric aortic repair in asymptomatic and unruptured AAA.¹⁹ What follows is a description of the clinical course of a pediatric MFS patient presenting with ruptured AAA. Informed consent was obtained in person from the patient and his mother to report the case.

CASE REPORT

The patient is a 15-year-old boy previously diagnosed with MFS. His genetic mutation is a 3-nucleotide deletion with 23-nucleotide insertion in exon 40 on the *FBNI* gene, resulting in frameshift. He is the only known member of his family to be diagnosed with MFS and has previously undergone operations for repair of his mitral valve and pectus excavatum. He has been observed by specialists at three other outside medical institutions for his condition, but he was not previously known to have an AAA. The only imaging of his abdominal aorta was a magnetic resonance imaging scan performed 11 months before presentation demonstrating focal infrarenal ectasia with a diameter of 29 mm. The patient is an active young man who enjoyed

sports such as golf, and he had already been counseled to modify his activities and to engage in only mild physical exercise. He has no history of any tobacco use. His home medications included losartan 100 mg and aspirin 81 mg daily.

The patient presented acutely to the emergency department with symptoms of lower abdominal pain (7/10) radiating to his back. Physical examination revealed a tall young man of thin habitus with arachnodactyly and a soft, pulsatile mass in his upper abdomen; he was without any neurologic deficits. A bedside ultrasound examination visualized a 64-mm-diameter AAA. At that time, the patient was hemodynamically stable; he was sent for a computed tomography angiography (CTA) scan, which showed no evidence of aortic dissection but demonstrated an infrarenal AAA measured to be 73 mm in transverse diameter. There appeared to be a left posterior rupture with active extravasation of contrast material and a large retroperitoneal hematoma extending from the diaphragm into the pelvis and displacing solid organs and bowel (*Fig 1*). After the scan, the patient became acutely hypotensive, had an episode of emesis, and began to experience respiratory distress. The patient was rapidly intubated for imminent respiratory failure and volume resuscitated with 1 L of normal saline and 2 units of uncrossmatched O-negative packed red blood cells. Shortly thereafter, the patient was sent to the operating room for emergent AAA repair. Given the emergent nature of the patient's presentation and condition, formal written consent was not able to be obtained; however, the surgeon was able to discuss all anticipated risks and benefits with the patient's family before the procedure, and they verbally agreed to proceed.

The patient was emergently repaired for his AAA through a transabdominal approach, and it was noted that he had immediate bulging of his bowels (*Fig 2, A*) and a large retroperitoneal hematoma (*Fig 2, B*) that appeared to have had a tamponade effect, and no active bleeding was observed. This allowed time for adequate vascular control to be obtained before exploration of the aneurysm sac. The aneurysm sac was estimated to be 10 cm in diameter, and the rupture was identified to be on the left lateral aspect and 2 cm in length (*Fig 2, C*). Clamping of the aorta was performed using Fogarty HydraGrip clamps at the level of the fourth portion of the duodenum. The aorta was able to be successfully repaired with a 14-mm Dacron

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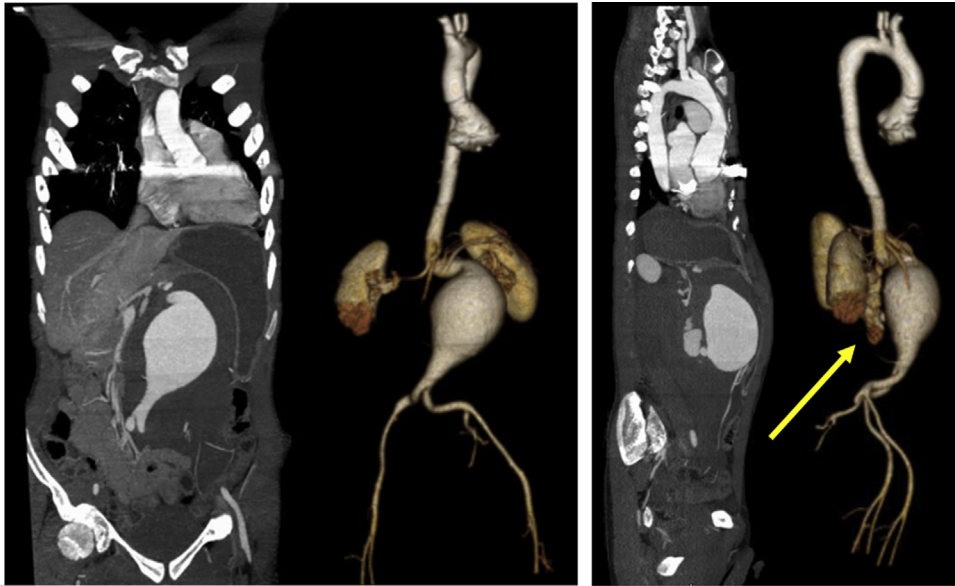


Fig 1. Two views of a multilevel composite coronal view and three-dimensional reconstructed computed tomography angiography (CTA) image of the patient's 73-mm abdominal aortic aneurysm (AAA). Posterior area of rupture is indicated by the arrow.

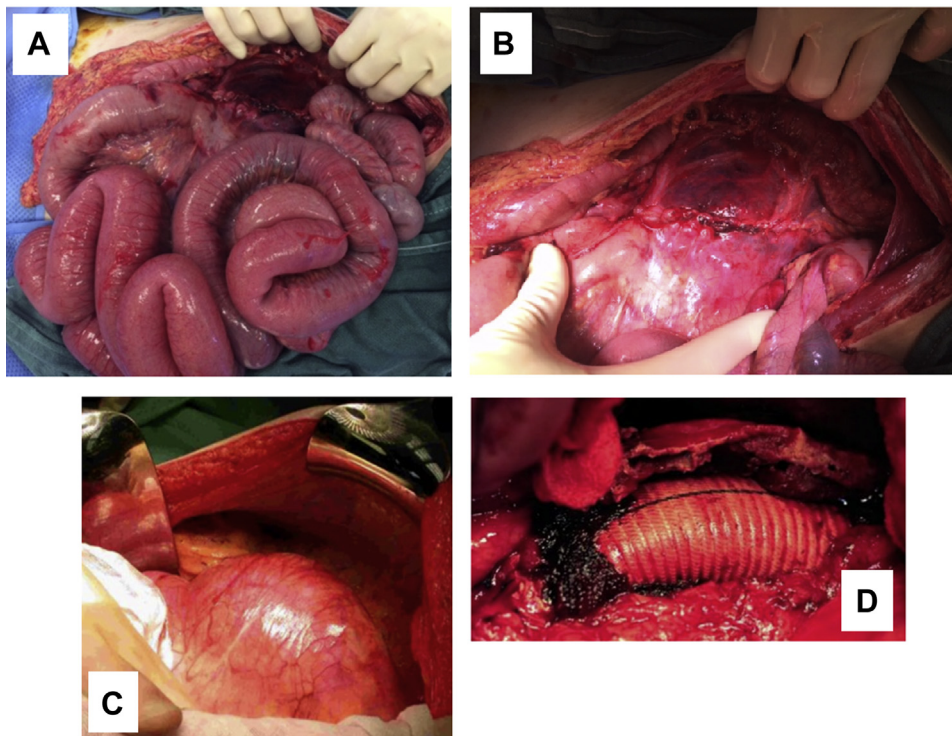


Fig 2. Intraoperative photographs. **A**, Bulging bowels encountered on entry to the peritoneum. **B**, Retroperitoneal hematoma. **C**, Abdominal aortic aneurysm (AAA), intraoperatively estimated to be 10 cm in diameter. **D**, Incised aneurysm sac with completed open AAA repair using Dacron tube graft, distal anastomosis with hemostatic agent Surgicel applied.

tube graft, with the anastomoses made in standard fashion without felt buttresses to healthy-appearing, nonaneurysmal aortic tissue (Fig 2, D). Because of observed intestinal edema, the patient's abdomen was unable to be closed primarily, and

an ABThera open abdomen negative pressure dressing (Acelity, San Antonio, Tex) was applied at the end of the case.

The patient's postoperative course was 2 weeks in length, and he was initially transferred to the pediatric intensive care unit for

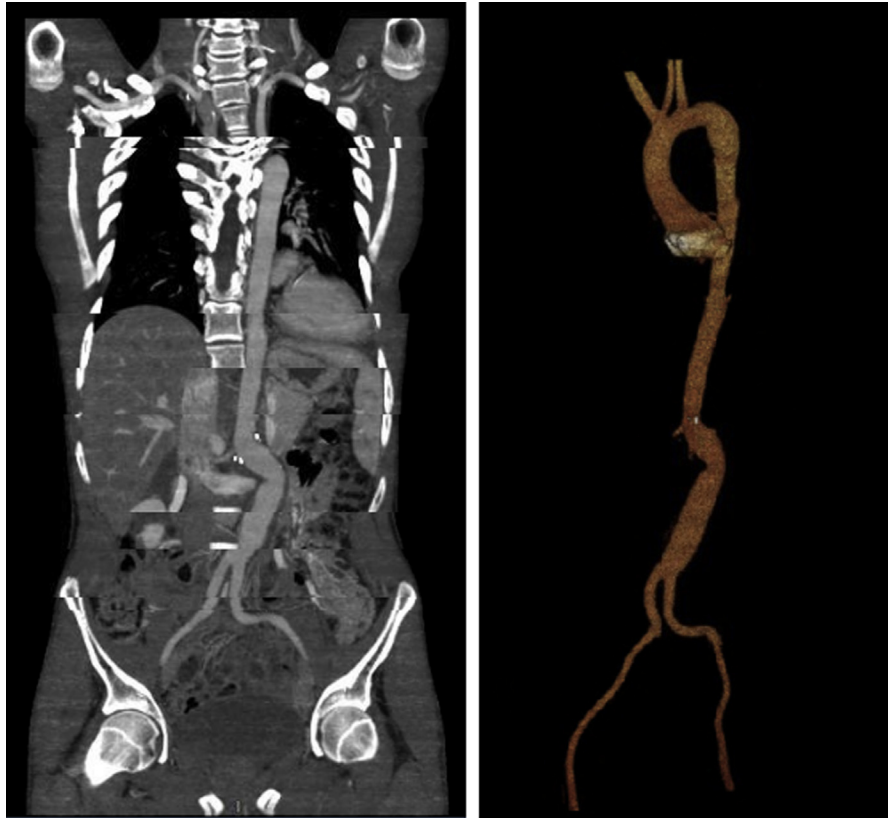


Fig 3. Postoperative computed tomography angiography (CTA) scan at 1 year, multilevel composite coronal image and three-dimensional reconstruction.

immediate postoperative care. He required pediatric intensive care unit care through postoperative day 5, after which he was able to be transferred to the general inpatient floor. During his inpatient course, the patient required application of a Wittmann patch to assist abdominal closure, which was ultimately completed on postoperative day 7 with concurrent removal of the patch. Because of concerns of nutritional deficit, total parenteral nutrition was begun before his transition back to oral intake. On postoperative day 14, the patient was discharged to home, with appropriate inpatient rehabilitation of his activities of daily living. He had no additional antihypertensive medications prescribed on discharge. At his 1-month outpatient follow-up appointment, the patient was doing well and slowly returning to his normal activities, and his surgical incision was healing appropriately. At his 1-year postoperative visit, he was noted to have been prescribed a different angiotensin receptor blocker (irbesartan 300 mg) by his genetics physicians but was fully healed and doing well. Repeated CTA imaging at 1 year noted stable postoperative changes (Fig 3).

DISCUSSION

This is an account of a pediatric MFS patient with a ruptured AAA who was successfully treated by open surgical repair. Whereas most cases of sudden death in MFS patients are a result of dissection or rupture of the ascending thoracic aorta, this case demonstrates that there may be a role for screening and monitoring of

patients for AAA in the pediatric MFS population. Although current recommendations are standard for the ascending aorta by annual echocardiography on MFS diagnosis, surveillance of the remaining aorta is less defined, with recommendations of intermittent CTA or magnetic resonance angiography beginning at young adulthood.¹ For MFS, arch imaging by echocardiography is recommended at the time of diagnosis and every 6 months thereafter until growth is stable and then annually. Growth >4.5 cm may require more frequent surveillance and possible repair.²⁰ There are no current recommendations on AAA screening in MFS patients. This patient had been observed by a surgical team regarding his MFS but had not had imaging of his abdominal aorta in 11 months, which was performed at that time in preparation for mitral valvuloplasty.

Current guidelines for non-MFS abdominal ultrasound AAA screening are for significant AAA family history in patients older than 60 years, cigarette history in men 65 to 75 years old, and clinically suspected AAA on physical examination.²¹ For non-MFS thoracic aortic aneurysm (TAA), echocardiography screening is recommended in first-degree relatives of those with TAA. In patients with known genetically associated TAA, genetic screening is recommended with subsequent imaging for those with the mutation. Screening of second-degree relatives of those with TAA is also reasonable.²⁰

This report also demonstrates that emergent open repair can be successful in the setting of this rare connective tissue disease. Once aneurysmal dilation has reached a diameter requiring repair, most reports in the adult population have described successful repair with the standard open method, with few reports of endovascular aortic repair.^{5,6} Given the natural history of connective tissue disorders such as MFS, especially in a pediatric population, open surgical repair onto tissues that have not yet become aneurysmal appears to be the safest treatment strategy.

CONCLUSIONS

Whereas aneurysms of the ascending aorta are characteristic of MFS patients, AAA may be a rare but potentially devastating complication in these patients, even in the pediatric population. In addition to surveillance of the ascending aorta, for the abdominal aorta, there may be a role for biannual noninvasive observation in these patients using abdominal ultrasound. However, size criteria indicating surgical AAA repair currently exist only for the adult population and differ with smaller size criteria for those who are noted to be young, healthy, or female.²² Given the rapidity with which this MFS patient's AAA grew in 11 months from 2.9 cm to 7.3 cm on presentation with rupture, in MFS patients, 6-month-interval imaging with repair indicated according to current adult sizing criteria of >5.0 to 5.5 cm is recommended. Further surveillance plans for this patient after repair include annual abdominal ultrasound examinations with follow-up CTA between 2 and 3 years to rule out pseudoaneurysm, infection, or proximal degeneration. This report demonstrates that in situations of emergent ruptured AAA among pediatric patients with MFS, open surgical repair is a viable treatment option.

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