Ann Thorac Cardiovasc Surg 2024; 30: 22-00044
Online May 19, 2022
doi: 10.5761/atcs.cr.22-00044

Case Report

Scattered Media Elastic Fibers from the Aortic Root to the Ascending Aorta in a 30-Year-Old Marfan Syndrome Patient

Tomohiro Nakajima,[1] Yutaka Iba,[1] Syuichi Naraoka,[1] Tsuyoshi Shibata,[1] Shintaro Sugita,[2] and Nobuyoshi Kawaharada,[1]

We present a case report of a 30-year-old Marfan syndrome patient who underwent a David procedure for severe aortic valve insufficiency and Valsalva aneurysm. Harvested aortic walls were examined by pathologists. Although the tunica media of the ascending aorta contained aligned elastic fibers, the aortic root media lacked aligned elastic fibers.

Keywords: Marfan syndrome, aortic root dilation, elastic fiber

Introduction

There are many reported cases of Marfan syndrome (MFS) patients presenting with severe aortic valve insufficiency and aortic root dilation.[1)] Although surgical techniques and outcomes have been discussed, many physicians were not interested in the pathological features of these lesions.[2)] In this study, we report the impact of a case in which a 30-year-old MFS patient lacked elastic fibers in the tunica media of the sinus of Valsalva.

Case Report

A 30-year-old man was introduced to our hospital owing to ascending aortic dilation and severe aortic valve regurgitation. The patient had a history of surgery for scoliosis at 3 years of age. His thoracic cavity was distorted and diagnosed as funnel chest. He also had a family history of vascular disease and his father died at 54 years due to aortic dissection. The patient fulfilled the revised Ghent criteria for the diagnosis of MFS:[3)] aortic root dilation with Z >2 and systemic score >7 (wrist and thumb sign, pectus carinatum deformity, scoliosis or thoracolumbar kyphosis, and facial features; **Figs. 1A** and **1B**). Severe aortic valve regurgitation was detected in this patient (**Fig. 1A**), and volume rendering computed tomography showed a 70-mm aortic root dilation (**Fig. 1B**).

We conducted valve-sparing aortic root surgery (operation time was 355 min, cardiopulmonary bypass time was 225 min, and aorta clamp time was 192 min). The aortic root to the ascending aorta was replaced with a Valsalva graft of 26 mm. The harvested aortic walls were examined by pathologists. The patient was discharged from our hospital postoperative day 15 without any events.

Figure 2A shows the aortic wall from the root to the ascending aorta, which part was shown with black arrow by Elastica van Gieson stain. The following features were recognized histologically: almost normal arterial tissue without ruptured elastic fibers in the ascending aorta, ruptured elastic fibers in the transition region to the aortic root, and arterial tissue in which elastic fibers had disappeared in all layers at the aortic root. Higher power microscopy showed scattered elastic fibers at the shifting lesion (**Fig. 2B**).

[1]*Department of Cardiovascular Surgery, Sapporo Medical University School of Medicine, Sapporo, Hokkaido, Japan*
[2]*Department of Surgical Pathology, Sapporo Medical University School of Medicine, Sapporo, Hokkaido, Japan*

Received: March 7, 2022; Accepted: April 19, 2022
Corresponding author: Tomohiro Nakajima. Department of Cardiovascular Surgery, Sapporo Medical University School of Medicine, South-1, West-16, Chuo-ku, Sapporo, Hokkaido 060-8543, Japan
Email: t.nakajima@sapmed.ac.jp

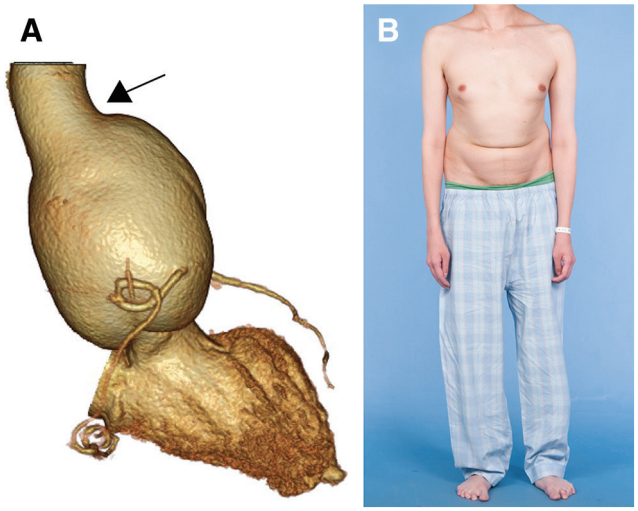

Fig. 1 (**A**) Preoperative volume rendering CT. The aortic root was dilated like a pear. The black arrow indicated the sites collected for pathological examination. (**B**) Appearance of the patient. In the appearance, humpback and lumbar flexion were recognized.

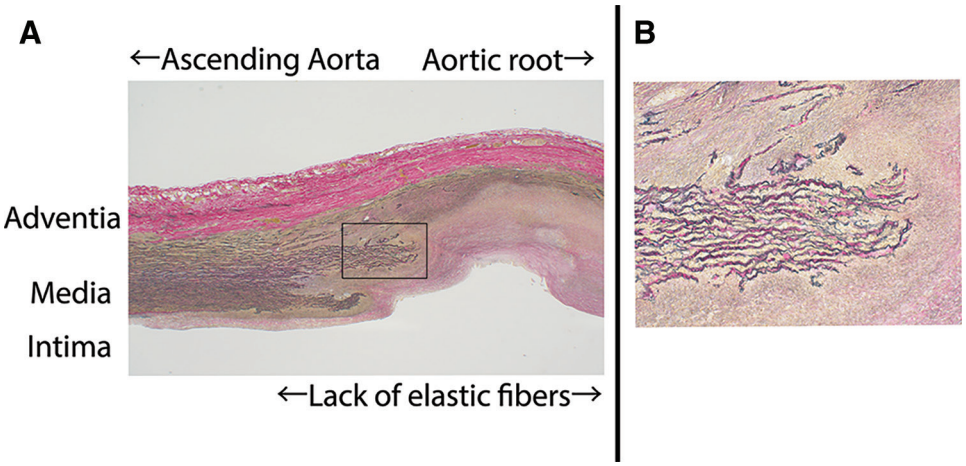

Fig. 2 Elastica van Gieson staining. (**A**) From ascending aorta to aortic root media elastic fibers disappeared gradually. (**B**) High-power field showed scattered elastic fibers.

Discussion

MFS affects a wide range of age groups, from children to adults, and is known to cause heart valve disease, aortic dissection, and sudden death. The frequency is reported to be 1 in 5000 to 10,000 people.[4] Inheritance is autosomal dominant with a 50% chance of inheritance in children. However, not all people with MFS develop heart disease or aortic dissection. The prevalence of cardiovascular disease in MFS remains unknown.

It is characterized by a combination of heart and blood vessel disorders, muscle and bone abnormalities, and eye disorders. Among these, aortic disorders are directly life-threatening. The wall of the aorta is composed of three layers: the intima, the media, and the adventitia. In MFS, the connective tissue in the media is weakened. A tear in the weakened wall of the aorta can cause acute aortic dissection with severe pain. Furthermore, because blood pressure is constantly applied to the weakened wall of the aorta, the blood vessels may gradually swell to form an aortic aneurysm. However, most patients do not develop any symptoms.[5]

The lesions are considered to be caused by the application of hemodynamic load against the

background of "fragile media." The medial lesion is presumed to be a decrease in elastic lamina and a decrease in cross-linking of fibers between elastic lamina, and the causes are hypertension and hereditary connective tissue diseases, such as MFS. Cystic medial necrosis, a previously reported medial lesion, is common in connective tissue diseases such as MFS. However, in previous reports, necrosis or elastic fiber defects of the tunica media were limited to the extreme part.[6)]

Conclusion

In this case, the elastic fibers of the tunica media were absent along the entire wall of the sinus of Valsalva aneurysm in an MFS patient. In contrast, the ascending aorta of this patient showed the same histological features as normal healthy people. We reported this case because it was an impactful image.

Acknowledgment

We thank Leah Cannon, PhD, from Edanz (https://jp.edanz.com/ac) for editing a draft of this manuscript.

Disclosure Statement

The authors report no conflicts of interest in this study.

References

1) Patel HJ, Deeb MG. Ascending and arch aorta pathology, natural history, and treatment. Circulation 2008; **118**: 188–95.
2) Takayama T, Miyata T, Nagawa H. True abdominal aortic aneurysm in Marfan syndrome. J Vasc Surg 2009; **49**: 1162–5.
3) Loeys BL, Dietz HC, Braverman AC, et al. The revised Ghent nosology for the Marfan syndrome. J Med Genet 2010; **47**: 476–85.
4) Guala A, Teixido-Tura G, Dux-Santoy L, et al. Decreased rotational flow and circumferential wall shear stress as early markers of descending aorta dilation in Marfan syndrome: a 4D flow CMR study. J Cardiovasc Magn Reson 2019; **21**: 63.
5) Guala A, Teixidó-Tura G, Rodríguez-Palomares J, et al. Proximal aorta longitudinal strain predicts aortic root dilation rate and aortic events in Marfan syndrome. Eur Heart J 2019; **40**: 2047–55.
6) Deleeuw V, de Clercq A, de Backer J, et al. An overview of investigational and experimental drug treatment strategies for Marfan syndrome. J Exp Pharmcol 2021; **13**: 755–79.