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CASE REPORT

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Marfan syndrome, an insidious disease with an unexpected course

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Abstract

Introduction: *The study presents a 45-year-old male with Marfan syndrome admitted to the cardiac surgery department with a suspected acute dissecting aneurysm of the ascending aorta involving distal thoracic and abdominal aorta.*

Case report: *Patient was qualified for the surgical intervention and the Bentall procedure with valve On-X and ascending aorta replacement with vascular prosthesis Gelweave was performed. Patient was readmitted 8 months later for reoperation due to the progress of the aortic arch aneurysm. During the redo surgery mediastinal pseudoaneurysm originating from the anastomosis between vascular prosthesis and aortic arch was identified. Aortic arch was replaced with vascular prosthesis Thoraflex. After 3 months the patient presented chronic cardiac tamponade decompressed with substernal surgical access. 1 month later patient was readmitted with pseudoaneurysm of the ascending aorta. Supracoronary section of the ascending aorta was replaced with a new Gelweave vascular prosthesis and the bleeding that originated from the left coronary artery ostium was treated accordingly. Marfan syndrome require multidisciplinary care and often multistage risk surgical treatments, including postoperative bleeding or pseudoaneurysms. There are no clear data on the choice of valve prosthesis. Each time, the surgeon must center the risk of hemorrhagic complications against the necessity for reoperation due to degeneration of the biological prosthesis.*

Keywords: Marfan syndrome; genetic disease; aortic aneurysm; thoracoabdominal; aortic dissection; thoracic surgery; cardiovascular surgical procedures

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Introduction

Marfan syndrome being an autosomal dominant inherited genetic disorder affects an average of 1/5,000 people [1]. Mutation of the fibrillin-1 gene (FBN1) is located on chromosome 15q21.1 andencodes fibrillin 1 glycoprotein, which is incorporated into the extracellular matrix (ECM). The microfibrils formed in this way allow the subsequent binding of epidermal growth factor (EGF). A fibrillin monomer is finally connected

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to other cellular elements to form a dense network that provides tissue elasticity [2]. In addition, disrupted interaction between fibrillin 1 and latent transforming growth factor binding proteins (LTBPs) causes a release of active transforming growth factor-beta (TGF-β). TGF-β by numerous enzymatic pathways consecutively weakens the fibrillar elastic matrix intensifying disease symptoms [2]. A disruption of the fibrillar network contributes to significant structural abnormalities of connective tissue within multiple systems, including the cardiovascular system [1]. Patients are burdened with aortic aneurysms and very serious complications — aortic dissections [1]. The most commonly described aortic aneurysm in Marfan syndrome is aortic root ectasia [2]. 20–25% of the patients with Marfan syndrome develop thoracic aortic aneurysm dissection [3]. Finally, the average survival for a patient with Marfan syndrome reaches 50 years [4].

Case report

A 45-year-old patient was urgently admitted to the cardiac surgery department with a suspected acute dissecting aneurysm of the ascending aorta and dissection involving the distal thoracic and abdominal aorta. The patient was qualified for the Bentall procedure. The aortic valve and root were resected, and the mechanical valve On-X 27/29 (On-X life technologies) and vascular prosthesis Gelweave 8 × 30 mm (TerumoAortic) were implanted (Fig. 1). Aortic arch was inspected, no re-entry was found. Intima and media of the aorta was reconnected with a sponge sealant patch (TachoSil). Estimated EuroSCORE was 38.68% and GERAADA Score was 84.7%. Postoperative course was uneventful.

8 months later, patient was readmitted for re-do surgery due to the progress of the aortic arch aneurysm detected on follow-up angio-CT. The dissection located within the aortic arch extending up to the left common carotid artery and left subclavian artery ending at the level of the common iliac arteries. Patient had 18.64% EuroSCORE and 60.1% GERAADA Score. Intraoperatively mediastinal pseudoaneurysm originating from the anastomosis between vascular prosthesis and aortic arch was identified. Aortic arch was replaced with aortic arch vascular prosthesis Thoraflex 30/38/12/8/10 × 10 mm (TerumoAortic) implantation (Fig. 2). The patient was admitted 3 months later due to chronic cardiac tamponade. A decompression through substernal surgical access was performed obtaining 400ml of pericardial effusion. EuroSCORE was 6.46%. One month after previous short hospital stay, patient was readmitted on an emergency basis due to a new pseudoaneurysm of the ascending aorta (Fig. 3). We calculated EuroSCORE to 36.65 % and GERAADA Score to 79.9%. During the surgery hematoma around the aortic valve was removed. The supracoronary section of the ascending aorta was again replaced with a new Gelweave 32 mm (TherumoAortic) vascular prosthesis. Bleeding

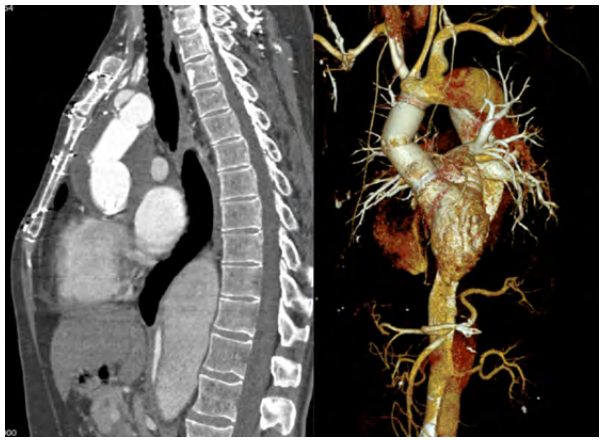

Figure 1. CT angiography showing On-X 27/29 mechanical valve and vascular prosthesis Gelweave 8 × 30 mm

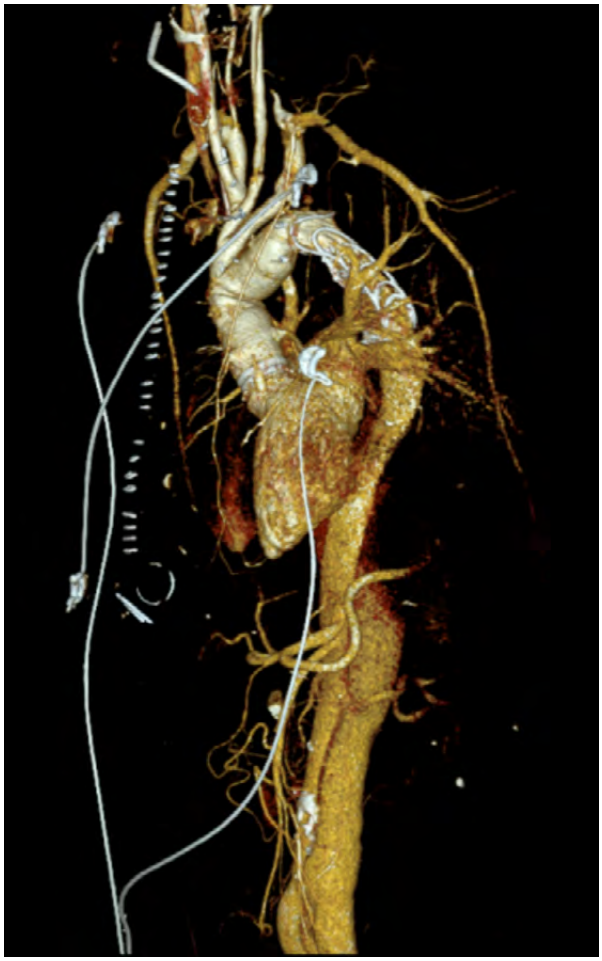

Figure 2. CT angiography 3D reconstruction showing On-X 27/29 mechanical valve and vascular prosthesis Gelweave 8 × 30 mm and aortic arch replaced with aortic arch vascular prosthesis Thoraflex 30/38/12/8/10 × 10 mm

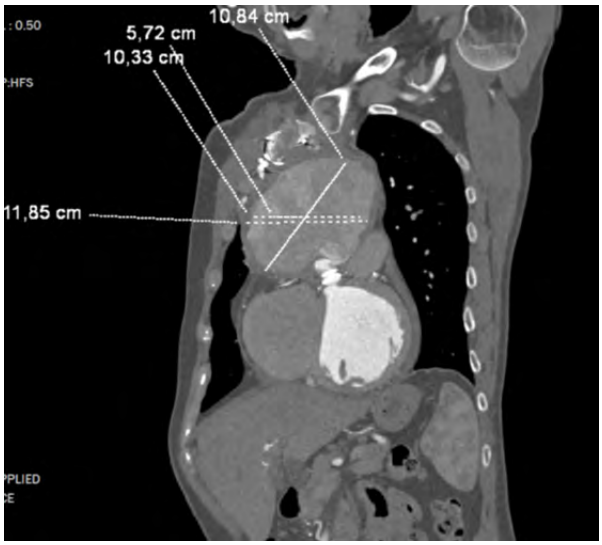

Figure 3. CT angiography showing pseudoaneurysm of the ascending aorta with its dimensions

that originated from the left coronary artery ostium was identified and treated accordingly. Extracorporeal circulation with the lowest temperature of 20 Celsius degree lasted 188 minutes. The postoperative course was uneventful. The patient was referred to the cardiology department for rehabilitation.

Discussion

50% patients with Marfan syndrome required surgical intervention of the aorta, with type A dissection being the most common cause [5].

Reports suggest that the mortality rate after aortic arch replacement surgery (using the frozen elephant trunk technique) in patients with prior the Bentall procedure is 11.5% [7]. Elective surgery has a lower frequency of reoperations number. Only 10% require third surgery interventions versus 50% admitted on an emergency basis [8].

There are no clear data on the choice of valve prosthesis. Each time, the surgeon must center the risk of hemorrhagic complications against the necessity for reoperation due to degeneration of the biological prosthesis. Survival and freedom from reoperation at 1 and 5 years are similar for both mechanical and biological valves (with a slight advantage for biological valves). At 10-year follow-up, biological valves have a reduced survival rate with a significant increase in reoperation frequency (69.4%) compared to mechanical prostheses (57.6%) [10].

Conclusions

Marfan syndrome requires multidisciplinary complex care and often multistage surgical treatment with high risks, including postoperative bleeding or pseudoaneurysms. Recently, a long-lasting biological aortic valve could be an alternative option for young patients with Marfan syndrome. This eliminates the necessity of antiplatelet therapy in the form of warfarin, significantly increasing the risk of bleeding [11]. Despite the above educating the patient about characteristic symptoms and appropriate management is an important part of prevention. In a patient with the above clinical manifestation, immediate treatment is necessary to minimize the risk of death.

Article information and declarations

Ethics statement: Ethical approval was not required.
Author contributions: Grzegorz Hirnle — concept, data gathering, writing, editing, supervision; Sebastian Krych — data gathering, writing, editing; Emilia Kupczyk — data gathering, writing, editing; Maria Kawulok — data gathering, writing, editing; Tomasz Hrapkowicz — writing, editing, supervision
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