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Clinical Case

Successful Bentall Operation in a Black African Woman with Marfan Syndrome and Type A Aortic Dissection: A Case Report

Opération de Bentall réussie chez une femme noire africaine atteinte du syndrome de Marfan et d'une dissection aortique de type A: à propos d'un cas

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ABSTRACT

Marfan syndrome is an autosomal dominant genetic disease characterized by mutations in the fibrillin-1 gene that lead to a connective tissue disorder affecting mainly the skeletal, ocular, and cardiovascular systems. Its reported prevalence in the general population is around 1/3,000–1/5,000, with no described racial predilection. Cardiovascular manifestations cause over 90% of the deaths in these patients, mainly due to aortic aneurysms and related complications. Medical therapy includes the administration of beta blockers and angiotensin II receptor inhibitors to slow the aortic growth rate, and prophylactic root surgery with either a composite root replacement (Bentall procedure) or valve-sparing procedures (reimplantation or remodeling) is the leading choice in patients with a diagnosed aneurysm. We report the case of a 26-year-old Black female with suspected Marfan syndrome and a family history of aneurysm and sudden death in first-degree relatives who underwent urgent repair of a 49.5 mm aortic root aneurysm with a composite root replacement procedure.

RÉSUMÉ

Le syndrome de Marfan est une maladie génétique autosomique dominante. Les manifestations cardiovasculaires de cette maladie causent plus de 90% des décès chez ces patients, principalement dus aux anévrismes aortiques et aux complications associées. La prise en charge comporte un traitement médical et une chirurgie prophylactique de la racine aortique (procédure de Bentall ou procédure d'épargne valvulaire). Nous rapportons le cas d'une femme noire de 26 ans avec un syndrome de Marfan suspecté et des antécédents familiaux d'anévrisme et de mort subite, qui a subi une réparation urgente d'un anévrisme de la racine aortique de 49,5 mm par une procédure de Bentall.

INTRODUCTION

Marfan syndrome (MFS) is a systemic connective tissue disorder resulting from mutations in the fibrillin-1 (FBN1) gene with an estimated prevalence of 1/3,000–1/5,000 in the general population [1]. It is characterized by inheritable autosomal transmission, with a familial history reported in approximately 75% of patients [2]. The diagnosis of MFS, based on the revised Ghent nosology, involves identifying major and minor criteria from the patient's familial history and cardiovascular, ocular, and skeletal abnormalities [3]. The clinical manifestations involve various organs, such as those of the skeletal system (pectus excatum, scoliosis) and ocular system (ectopia lentis), but cardiovascular lesions, especially with aortic aneurysm, are associated with the most feared complications [4,5]. Indeed, silent aortic disease in MFS is often revealed during acute and life-threatening events, such as dissection and rupture, which are responsible of 90% of deaths in MFS patients [6,7]. An extensive familial screening, including genetic counselling and molecular testing in suspected cases, is the key to preventing adverse outcomes by enabling the provision of medical therapy (beta blockers and angiotensin II receptor antagonists) and timely surgical intervention.

Although no particular predilection has been demonstrated in specific populations [8], the scarce published data on MFS in sub-Saharan Africa (SSA) suggest a lower prevalence of surgical treatment in the region, mainly due to limited access to cardiovascular specialists. The current paper reports the case of a young black African female with MFS who presented with type A aortic dissection and a family history of aortic aneurysm

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and sudden death who underwent successful management through composite root replacement surgery.

CASE PRESENTATION

A 26-year-old black African female with recent onset of atypical chest pain one month earlier was referred by the attending cardiologist to our cardio-surgical unit for suspicion of type A aortic dissection. A computed tomography angiography (CTA) performed before admission revealed a 49.6 cm aortic root aneurysm with a suspicion of intimal tear (Figure 1A). The family history revealed two cases of aortic aneurysm and sudden death in first-degree relatives (mother and uncle). Physical examination showed a relatively increased height (175 cm for 56 kg) and features of joint laxity, suggesting a possible MFS character. The vital signs during admission were within normal range (blood pressure: 130/70 mmHg; heartrate: 81 bpm; body temperature: 36.6 °C), and cardio-pulmonary auscultation revealed a 2/6 diastolic murmur in the left parasternal area (fifth intercostal space) with clear lungs. A 12-lead electrocardiogram showed a regular sinus rhythm with mild signs of ventricular overload. A transthoracic echocardiogram confirmed the presence of a dilated aortic root with moderate functional aortic valve insufficiency (Figure 1B). No signs of primary mitral lesions were noted, but a mild dilation of the left ventricle was present that determined a tiny functional mitral regurgitation. The left ventricular contractility was preserved (65%), and no pericardial abnormality was seen. Considering the reported history of atypical chest pain, the sudden death of two relatives with aortic aneurysm, and the suspicion of root dissection, a consensual decision was taken for an urgent aortic root surgery.

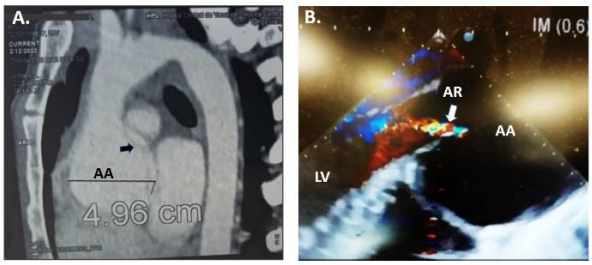

Figure 1. Preoperative Computed Tomography Angiography Scan (A) and Transesophageal Echocardiography (B) showing the aortic root aneurysm and valve regurgitation. The black arrow indicates the intimal tear. (*AA: aortic aneurysm; LV: left ventricle; AR: aortic regurgitation*).

A full median sternotomy was performed and cardiopulmonary bypass established through a distal aortic arch and right atrium cannulation. After aortic cross clamping, the aneurysm was opened and a crystalloid cardioplegic solution was administrated selectively in the coronary ostia and repeated every 30 minutes. The aneurysm was completely resected. The coronary buttons were prepared, and a morphologically tricuspid aortic valve was resected due to the presence of cusp margin elongation and mild fenestration. A St. Jude Medical Masters Series 21 mm mechanical aortic prosthesis was then sutured to a 26 mm Dacron graft and implanted using 2.0 interrupted mattress sutures reinforced with pledgets. Coronary buttons were anastomosed to the Dacron graft using 5.0 polypropylene sutures reinforced with Teflon felt. The distal anastomosis was then realized with two 4.0 polypropylene running sutures. The patient was weaned from CPB following the usual de-airing maneuvers. Postoperative transesophageal control showed good position and function of the mechanical valve and complete repair of the aortic aneurysm. Intraoperative steps are illustrated in Figure 2.

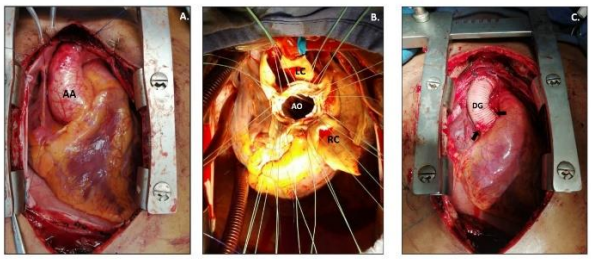

Figure 2. Intraoperative views: A) aortic root aneurysm; B) exposure of the aortic orifice and preparation of the coronary buttons; C) completed repair with a 24 mm Dacron graft. The arrows indicate the sites of coronary anastomoses. *(AA: aortic aneurysm; AO: aortic orifice; LC: left coronary; RC: right coronary; DG: Dacron graft).*

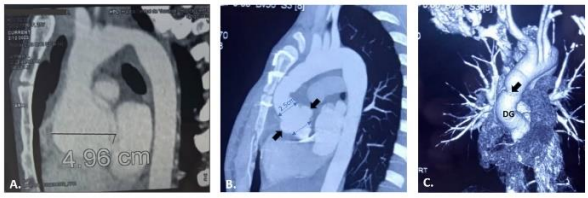

Figure 3. Comparison between preoperative (A) and post-surgical Computed Tomography Angiography scans (B and C). The black arrows indicate the sites of coronary buttons anastomoses in B, and the distal Dacron graft anastomosis in C. *(AA: aortic aneurysm; AO: aortic orifice; LC: left coronary; RC: right coronary; DG: Dacron graft).*

The postoperative course was uneventful, and the patient was transferred to the intensive care unit. One week after surgery, she was discharged in good clinical condition. A CT scan two months after discharge (Figure.3) showed a good surgical result with complete resection of the aneurysm.

DISCUSSION

Because MFS results from the effects of FBN1 mutations, the pathogenesis of the related aortopathy has traditionally been ascribed to a disordered elastic microfiber structure that leads to accelerated aortic wall dilation [9]. MFS aortic aneurysms are characterized by earlier presentation in young patients with a predilection for aortic root dilation (valsalva sinuses). Indeed, severe forms have been reported from the neonatal period and during infancy, with a higher risk of aortic dissection and rupture when compared to non-MFS aneurysms [10–12]. The

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major guidelines on aortic diseases have recommended early root replacement at lower diameters in MFS (5.0 cm in Class I) as compared to non-MFS patients (5.5 cm) [13,14]. Moreover, lower surgical thresholds (≤4.5 cm) should be considered if high risk features are present, such as a family history of dissection, a diffused aortic root dilation, or rapid aortic growth (≥0.3 cm/year). This "aggressive" repair policy has significantly improved the long-term survival of these patients (≥80% at 10 years) [15,16] despite the risk of reoperation, which remains a major concern due to progression of the residual or distal aneurysmal disease [17–9]. Indeed, the mainstay of MFS aneurysm therapy has been a radical resection of the whole dilated aortic wall with root replacement procedures, namely, Bentall and valve-sparing root replacement (VSRR) procedures, which have yielded similar survival rates in recent comparative series [20–22]. VSRR techniques (mainly by reimplantation) are suitable in cases with preserved aortic valve anatomy and provide better freedom from valve-related events by avoiding lifelong oral anticoagulation, which is important in the young population. Conversely, composite graft replacement (Bentall) is preferred in cases with primary aortic valve deformities (fenestrations, fibrosis, elongated free margins) to limit the risk of late valvular dysfunction and reoperation, especially in younger patients [23]. However, the patient's expectations and clinical characteristics in addition to the team's expertise remain key factors in the individual decision-making process.

In our case, the surgical strategy was driven mainly by the need of urgent repair, the nature of the aortic valve lesions, and the expected compliance to oral anticoagulation therapy of the patient. The history of sudden death in first-degree relatives, the suspicion of root dissection on the preoperative CTA (Fig. 1), and the atypical pain one month earlier motivated an urgent root replacement (as for type A Stanford aortic dissection). A Bentall operation with mechanical prosthesis was preferred over a VSRR technique due to the moderate primary valve regurgitation with aortic free margin elongation and mild fenestrations on the cusps. Despite the patient's young age, the choice of the mechanical prosthesis (over biological) was motivated by the patient's cultural background (university student), suggesting an acceptable compliance to long-term oral anticoagulation therapy. Moreover, we were concerned by the financial burden on the patient's family in case of repeated surgery for early valve failure as could have been the case with biological Bentall or VSRR. The patient did not present any other cardiac lesions related to MFS (mitral prolapse or distal aortic pathology) at the time of surgery. Nevertheless, we recommended life-long therapy with beta blockers and angiotensin II receptor inhibitors to slow the aortic growth rate, considering the proven susceptibility to dissection and the potential risk of distal aneurysm development after proximal repair.

Although several cases of MFS patients with aneurysm disease have been reported in SSA, they were limited to case reports, and very few were addressed surgically, as there is still very little access to cardiovascular surgery in the region. Mongo et al. [24] and others in Congo [25] present similar cases of severe TAA and dissection managed conservatively in the absence of a local surgical solution. To the best of our knowledge, this is the first report of aortic aneurysm surgery in a Marfan patient in Central Africa.

CONCLUSION

MFS and connective tissue disorders should be suspected in young patients with TAA and a family history of sudden death, aneurysm, or dissection, even in sub-Saharan communities. The current case supports the recent advances in the surgical management of complex cardiac pathologies in our region.

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