Large pulmonary artery aneurysm associated with Marfan syndrome

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Pulmonary artery aneurysms are rare, and published data on the subject are limited. The first case of Marfan syndrome associated with a large pulmonary artery aneurysm complicated by pulmonary artery thrombi and pulmonary hypertension is described. Serial echocardiograms showed progressive dilation of the aneurysm, which reached a massive

size of 90 mm. Cardiac catheterization, pulmonary angiography and computed tomography angiography were used in the preoperative evaluation. The patient died suddenly while awaiting a decision for surgical intervention, thereby underscoring the need for aggressive management.

Key Words: *Marfan syndrome; Pulmonary artery aneurysm; Pulmonary hypertension*

Pulmonary artery (PA) dilation is a minor cardiovascular criterion in the diagnosis of Marfan syndrome (1). A large PA aneurysm in Marfan syndrome has never been described, and the risk of PA dissection or rupture in such a patient is unknown. PA aneurysm is exceedingly rare and its natural history is unknown; therefore, clear guidelines for its management are lacking. We report an unusual case of Marfan syndrome complicated by PA thrombi, pulmonary hypertension and sudden death, underscoring the need for an aggressive management strategy.

CASE PRESENTATION

An asymptomatic 18-year-old man with Marfan syndrome and a PA aneurysm was admitted for investigation for surgery. He fulfilled the Ghent nosology criteria for diagnosis of Marfan syndrome, with involvement of three organ systems (ie, cardiac, musculoskeletal and skin) (1). He was previously diagnosed with idiopathic pulmonary hypertension at 11 years of age and his clinical condition remained stable over the ensuing years. Other significant medical history included moderate mitral regurgitation from bileaflet mitral valve prolapse.

Over time, he developed a massive PA aneurysm; at the time of admission, he was being considered for surgery. His chest x-ray showed an enlarged pulmonary trunk and PAs with plethoric lung fields (Figure 1A). Serial echocardiograms revealed a dilated aortic sinus with a diameter of 42 mm that remained stable over the years. In contrast, the main PA showed progressive enlargement, from 37 mm at 11 years of age, to 48 mm at 12 years of age and 74 mm at 15 years of age. His most recent echocardiogram revealed a huge PA aneurysm measuring 90 mm, with spontaneous echocardiographic contrast indicating sluggish blood flow. The pulmonary root was also dilated at 48 mm and was associated with moderately severe pulmonary insufficiency. Both the right and left PAs

Figure 1 A Chest x-ray showing a huge main pulmonary trunk and dilated right pulmonary artery with plethoric lung fields. B Transthoracic echocardiogram showing main pulmonary trunk measuring 90 mm and dilated right and left pulmonary arteries. The white arrow indicates pulmonary thrombus within the right pulmonary artery

were also dilated at 27 mm and 35 mm, respectively, and an in situ thrombus was seen in the right PA (Figure 1B). Doppler echocardiography revealed severe pulmonary hypertension with an estimated PA systolic pressure of 118 mmHg.

Computed tomography (CT) angiography with three-dimensional reconstruction confirmed the large main pulmonary trunk aneurysm (Figure 2A) and revealed thrombi in the right PA and its branches (Figure 2B). Laboratory blood investigations were negative for systemic inflammatory conditions or infections. The patient's coagulation profile was normal, with no evidence of an underlying thrombophilic state. There was no evidence of deep venous thrombosis from either his medical history or duplex ultrasound. The pulmonary thrombi were believed to be due to sluggish flow within

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the dilated pulmonary vessels. The patient was anticoagulated with warfarin while awaiting consideration for surgical repair of the PA aneurysm because its large size and the presence of pulmonary hypertension posed a significant risk for dissection and/or rupture. To determine suitability for surgery and to aid postoperative management, cardiac catheterization was performed. The PA pressure was 82/38 mmHg, with a mean of 53 mmHg. There was no significant decrease in PA pressure with oxygen, inhaled iloprost or intravenous adenosine infusion.

While awaiting the final decision, the patient was readmitted with complaints of abdominal discomfort, nausea and poor appetite. An abdominal x-ray and CT of the abdomen and pelvis were both unremarkable, and he was treated for viral gastritis and dehydration. While in the ward, he suffered sudden cardiorespiratory arrest and was hypotensive with pulseless electrical activity. Subsequent resuscitation was unsuccessful and no postmortem was performed. Sudden death was attributed to severe pulmonary hypertension and catastrophic PA rupture and/or dissection.

DISCUSSION

PA aneurysm is a rare entity, with approximately eight cases per 100,000 autopsies (2,3). Large PA aneurysm associated with Marfan syndrome has not been previously reported. Marfan syndrome is an inherited connective tissue disorder associated with defects in the fibrillin-1 gene, and is known to result in dilation of the aortic root, ascending aorta and PA (1-4). Currently, the diagnosis of Marfan syndrome is made according to the criteria of the 1996 Ghent nosology (1), which requires the presence of major criteria in two organ systems plus involvement of a third organ system. Although Marfan syndrome involves mainly the skeletal, ocular and cardiovascular systems, the respiratory system, skin and dura may be involved (1). Mitral valve prolapse, which was present in our patient, is common in Marfan syndrome, with a reported prevalence of 28% in a recent series (5). Of note, the majority of patients with mitral valve prolapse in that study had bileaflet involvement and a significantly large aortic root diameter, suggesting a more diffuse disease process (5).

Other causes of PA aneurysm include patent ductus arteriosus, ventricular and atrial septal defect, and transposition of the great arteries with ventricular septal defect. Less common causes include infections (eg, tuberculosis and mycotic aneurysms), connective tissue diseases including Ehlers-Danlos syndrome, and generalized inflammatory processes such as Behçet's syndrome or giant cell arteritis (2,3,6,7). The clinical manifestations are nonspecific, and patients are frequently asymptomatic. Cyanosis may occur because of bronchial compression by the aneurysm; chest pain and hemoptysis, when they occur, raise the concern of impending rupture (2,3,6). Multiple diagnostic modalities are commonly used, including chest x-ray, echocardiography, CT angiography, magnetic resonance angiography or pulmonary angiography, particularly before planning surgery. There are no clear guidelines for management, partly due to the rare nature of this condition.

Surgical intervention is the treatment of choice, particularly in the presence of symptoms, pulmonary hypertension,

Figure 2) **A** Computed tomography angiography with three-dimensional reconstruction showing a grossly dilated main pulmonary artery (white arrows). **B** Large main pulmonary artery trunk with thrombus in a branch of the right pulmonary artery (black arrow)

underlying connective tissue disease or progressive dilation on serial monitoring. Reported surgical procedures include aneurysmectomy with Dacron graft placement or autologous pericardial replacement, arterial aneurysmorrhaphy or plication angioplasty, and very often pulmonary valve replacement is also required in the presence of an abnormal pulmonary valve or significant pulmonary insufficiency (8-11). Lung transplantation with PA reconstruction or heart-lung transplantation has also been reported (12,13). Conservative management has been advocated in certain cases of 'low-pressure' PA aneurysms in which the risk of complication is lower (11).

Our patient presented a complex situation, with both a huge PA aneurysm and an underlying connective tissue disorder, further compounded by severe irreversible idiopathic pulmonary hypertension and PA thrombi. The rate of progression of his PA dilation and the high risk of rupture or dissection in the presence of severe pulmonary hypertension would normally require urgent surgical intervention. Certainly, if the aorta was 90 mm in diameter, there would not have been any hesitation. However, the presence of a lethal underlying condition with a guarded long-term prognosis, together with the high risk of operation, made surgery of the PA less feasible. Realistically, had he survived longer, a heart and lung transplantation may have been his only option.

CONCLUSION

Echocardiographic monitoring of the PA is important in patients with connective tissue disorders, particularly when there is concurrent pulmonary hypertension. Marfan syndrome patients have enjoyed an improved life expectancy in recent years, presumably due to advances in the management of aortic complications. Further investigation into the natural history of PA dilation in patients with Marfan syndrome is needed to better understand its clinical and prognostic significance and guide management.

DISCLOSURE: This work was performed at the National Heart Centre, Singapore, and the Mazankowski Alberta Heart Institute, Edmonton, Alberta, and has not been previously presented.

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