Aortic Dissection in Marfan’s Syndrome

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Patient: 39 years, male

Clinical History:

The patient started to experience shortness of breath. He has a history of Marfan’s syndrome and family history is significant for his mother having had aortic dissection.

Imaging Findings:

The patient presented with shortness of breath. A series of investigations were conducted. These included, in chronological order, a portable chest x-ray which showed a widened mediastinum (Fig 1). A non-gated CT angiogram of the chest, abdomen and pelvis with “smart prepping” was subsequently performed. The CTA revealed a dissection flap that originated from the dilated aortic root and measuring 8-cm. It then gave rise to the sinotubular region measuring 8.3cm, then the 9cm ascending aorta, to a 5cm diameter descending aorta that included a 3.7-cm false lumen. The abdominal aorta measured 6.3cm in diameter with the true lumen measuring at 1.4cm. The left subclavian, right renal and celiac artery arose from the true lumen. The false lumen gave rise to the right innominate, left carotid, left renal and inferior mesenteric artery (Fig 2a-g,3). No ischemic organs were observed. The dissection extended as far as the right common iliac artery which measured 3.4cm.

Carotid and extremity Doppler ultrasound, and renogram with Tc 99m Mag-3 were performed to assess presence of renal dysfunction due to rising creatinine to 313 umol/L. Doppler sonogram studies revealed grossly patent deep venous system. Renogram revealed bilateral impaired renal function but perfusion was present though diminished at 40% right and 60% left uptake.

Discussion:

Marfan syndrome is an inheritable autosomal dominant connective tissue disorder with 25% of cases arising de novo. Specifically it is a mutation of the fibrillin-1 (FBN1) gene. These patients classically present with tall stature, long slender limbs, arachnodactyly, pectus deformity, high arched palate with dental crowding and skin striae distensae [1]. In the cardiovascular system, this disease manifests as progressive degeneration and weakening of the aortic wall leading to aortic insufficiency and aneurysm of the ascending and descending aorta. The aneurysm may progress to or is complicated by dissection, as well as mitral valve prolapse and regurgitation [1].

The term acute aortic dissection is most appropriately assigned to acute dissection of the thoracic aorta, though any part of the aorta can be affected. The entry point starts with a critical initial intimal tear allowing blood to enter the aorta that leads to dissection of the medial layer. Dissection is classified according to descending (Type B) and/or ascending aorta (Type A) involvement. Acute aortic dissection has the potential to compromise oxygen supply to major organs and limbs leading to end-organ ischaemia. Thus, delay in diagnosis or misdiagnosis with inappropriate treatment can result in up to 1-3% mortality per hour during the first 48hrs [2].

The initial screening investigation usually conducted is the chest X-ray. Classic signs includes widened mediastinum...
 (>8cm) and abnormal aortic contour (Fig 1). Widened mediastinum is present in 64% of Type A and 56% of Type B dissections [3]. The sensitivity of this modality is quite low, ranging from approximately 47% for Type A to 77% for Type B [4], therefore, a normal chest x-ray appearance does not rule out aortic dissection.

CT has been used as the investigation of choice in cases of aortic dissection as it provides a non-invasive approach and excellent visualization of the aortic pathology to accurately plan operative therapy [5]. Visualization of true lumen separated from false lumen by a dissection flap confirms the diagnoses. Sensitivity in diagnosis is 93%. Runza et al [6] suggests that ECG-gated contrast-enhanced CT can allow differentiation between the three causes of acute chest pain with the highest morbidity and mortality: acute myocardial infarction, pulmonary embolus, and acute aortic dissection by decreasing the effects of cardiac motion to optimize cardiac images. Benefits of CT modality is the ability to concurrently identify associated features of aortic branch occlusion such as acute aortic valve regurgitation, acute myocardial infarction or cardiac tamponade, cerebral ischemia, spinal cord ischemia, or acute mesenteric vascular occlusion.

These patients are haemodynamically unstable and thus transthoracic and transoesophageal echocardiography can be a reasonable alternative to conduct at the bedside rather than transporting them to the CT unit. The limitation of the transoesophageal echo is its inability to visualize the abdominal aorta however. Magnetic resonance imaging can provide superb images of the cardiovascular system but its use in emergent situations is limited due to availability.

**Differential Diagnosis List:** Type A aortic dissection in Marfan's Syndrome patient.

**Final Diagnosis:** Type A aortic dissection in Marfan's Syndrome patient.

**References:**


Description: Enlarged mediastinum, marked dilatation of the thoracic ascending and descending aorta. Lungs were clear with no effusions. Origin:
Figure 2

a

Description: Dilated sinotubular region of the aorta measuring 8.3-cm. Origin:

b

Description: Dilated aortic arch with false lumen. Origin:
Description: Intimal flap separating true and false lumen in dilated 9-cm ascending and 5-cm descending aorta. Origin:

Description: The left renal artery arises from the false lumen while the right renal artery arises from true lumen. Origin:
Description: The celiac artery is supplied by the true lumen. Origin:

Description: The dissection flap continues down the 6.3-cm diameter abdominal aorta to the bifurcation. Origin:
Description: The dissection continues down into the right iliac artery branch. Origin:
Description: Tortuous and dilated thoracic and abdominal aorta. Origin: