

Marfan's syndrome presenting with abdominal aortic aneurysm: A case for vigilance

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Summary

We present the case of a 16-year old student with Marfan's syndrome and abdominal aortic aneurysm who presented with a diagnostic conundrum. He presented with a three months history of progressive painful left upper abdominal mass and back pain. It became severe in the last two weeks before presentation and was associated with constipation. This mass was thought to be of splenic origin but the initial ultrasound suggested a pancreatic pseudocyst. Review of his previous hospital record revealed that he had been treated for severe myopia which started at infancy. Another ophthalmic review at our centre revealed bilateral ectopia lentis. He had no cardiac signs and no family history of cardiovascular diseases. He is the 6th of 8 siblings, all the family members are alive and healthy except one sibling who died at 7 months. The diagnosis of abdominal aortic aneurysm was only made at laparotomy and confirmed by on-table aortogram. He had excision of the aneurysmal sac and replacement with on-lay dacron tube graft. He died on the 4th post-operative day. A diagnosis of abdominal aortic aneurysm was not made at initial presentation because of the rarity of this condition in our environment and incompetence of the ultrasonographer. Aortic aneurysm in Marfan's syndrome is commonly found in the thoracic part of the aorta, however in this case, it is abdominal. A high index of suspicion is necessary to avoid missing this pathology, therefore the need for vigilance.

Keywords: *Marfan's syndrome, Abdominal aortic aneurysm.*

Résumé

Voilà le cas d'un étudiant de 16ans, victime de syndrome de Marfan et d'anévrisme aortique abdominal qui a présenté une déviation diagnostique. Il a présenté une histoire de trois mois de douleur progressive de la masses abdominale supérieure ainsi qu'un mal de reins qui s'est fait sévère pendant les deux dernières semaines avant la présentation et qui était associé à la constipation.

On croyait que cette masse est d'origine splénique mais les ultrasons initiaux suggèrent une pseudokyste pancréatique. Les examens de ses dossiers précédant révèlent qu'il a été traité pour une myopie sévère qui a commencé depuis son enfance. Un autre examen ophtalmique dans notre Centre révèle une "ectopie lentis" bilatérale. Il n'y a pas montré des symptômes cardiaques et il n'y a pas dans la famille une histoire des maladies cardiovasculaires. Cet étudiant est le 6 ans la famille de 8 enfants. Toute la famille est vivante et en bonne santé sauf un des enfants qui est mort depuis l'âge de 7 mois. Le diagnostic d'anévrisme aortique abdominal n'était fait qu'à laparotomie et était confirmé par une aortogramme sur table. Il a eu une excision du sac anévrisimal qui a été remplacé par un tube de greffe de dacron. Il est mort le 4 jour après l'opération. Une diagnostic abdominal n'a pas été faite au début de la présentation à cause de la rareté de cette condition dans notre environnement et de

l'incompétence d'ultrasonographe.

D'habitude, anévrisme aortique dans la syndrome de Marfan se trouve dans la partie thoracique de l'aorte, néanmoins, dans ce cas c'est abdominal. Pour bien comprendre cette pathologie il faut une haute indexe de soupçon, donc la nécessité de vigilance.

Introduction

The definition of Marfan's syndrome is based on the characteristic changes that occur in three connective tissue systems-skeletal, ocular and cardiovascular system¹. It is inherited as an autosomal dominant condition and the defective gene has been mapped out to the fibrillin-1 (FBN1) gene on chromosome 15^{1,2,3,4}. Fifteen to 30% of this genetic defect may be due to new mutation. "Skipped generation" as a result of a variable expressivity is also relatively common¹. The abnormality of the connective tissue is due to defective organisation of the collagen and/or elastin fibres⁴. The structural properties of elastin and collagen are complementary. Elastin acts as the main load bearing component and provides elastic recoil while collagen provides a strong inextensible "safety net" at high loads. Therefore, both must fail if aneurysm and rupture are to occur.

The commonest features of Marfan's syndrome are asthenic build, pectus excavatum (funnel chest), pectus carinatum (pigeon chest), instability of joints, arachnodactyly (spider fingers), dolichostenomelia and kyphoscoliosis. Other features are high arched palate, mitral and aortic incompetence, severe myopia and ectopia lentis (subluxation of the lens)^{4,7}. All these features are seldom present in one person. The commonest complications of Marfan's syndrome are usually cardiovascular which include: mitral and aortic incompetence, dissecting aneurysm or spontaneous rupture of the aorta⁸. The aortic aneurysm in Marfan's syndrome is usually thoracic,^{1,3,6,9-12} while abdominal aortic aneurysms are mainly due to atherosclerosis^{5,13}. Most of the abdominal aortic aneurysms that have been reported in Marfan's syndrome were in association with thoracic aortic aneurysm^{14,17}.

Although an impression of abdominal aortic aneurysm may be made on clinical features, confirmatory investigations should include abdominal ultrasound^{5,13} and CT scan and MRI,¹⁶ if possible. Transoesophageal echocardiography has also been useful in detecting thoracic aortic aneurysm¹⁸. Aortogram is seldom used because the cavity of the aneurysm is often filled with clot and so may not demonstrate the actual outer diameter of the aneurysm¹³.

The main stay of treatment in symptomatic patients is excision and repair with tube graft prosthesis. In an asymptomatic aneurysm with a diameter of >5cm, repair should be offered because of the high risk of rupture and high rate of mortality^{5,13}. The outcome of treatment is better when it is done in specialised centres. Even then, there is a high peri-operative mortality of 40-50% in emergency surgery compared to 3-5% in elective surgery¹⁹. The commonest post-operative complications are respiratory (lower lobe consolidation,

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atelectasis and "lung shock")¹³. Other complications include haemorrhage, renal failure, embolisation, wound dehiscence, aortic-duodenal fistula^{13,20} infected graft⁹, and spinalischaemia that may lead to paraplegia^{13,14}.

We present a case of Marfan's syndrome presenting with abdominal aortic aneurysm (AAA), an unusual entity in this tropical environment.

Case report

A 16-year old male secondary school student presented with a three month history of painful left sided abdominal mass. The pain radiated to the back, chest and left lower limb. Two weeks prior to presentation the pain became severe and was associated with constipation. Physical examination revealed an asthenic male with a pulse rate of 102/minute, blood pressure of 110/60mm Hg and a palpable non-pulsatile left upper abdominal mass. A preliminary diagnosis of a splenic mass was made. His packed cell volume was 34% with a genotype of AA. An abdominal ultrasound was done which showed a huge cystic mass behind and below the stomach. A diagnosis of pancreatic pseudocyst was then made. He had an exploratory laparotomy and a fusiform infrarenal aneurysm of the abdominal aorta was found, which was confirmed by on-table aortogram using urografin.

A review of the patient's former hospital record revealed that at 11 years of age he was managed for acute glomerulonephritis and was noted to have poor vision right from infancy as a result of severe myopia. He was lost to follow-up until five years later when he presented with the abdominal mass at our centre. Ophthalmological review confirmed the presence of bilateral ectopia lentis.

At surgery, the aneurysm was very close to the left renal artery, and pushed mainly to the left. After proximal and distal control, the sac (containing clots and multiple plaques) was opened. A preclotted dacron tube graft was layed within the sac and anastomosed to the normal aorta starting with the proximal end. The renal ischaemia time was 15 minutes. The sac was closed over the graft. The patient had 5 units of autologous whole blood at operation. He made good progress in the immediate post-operative period and had return of bowel function on the 3rd day. On the 4th day he vomited a large amount of faeculent vomitus, passed loose stool and developed severe chest pain. He went into shock and died shortly thereafter.

Discussion

Marfan's syndrome is not an uncommon disease entity. The diagnosis should be suspected in a patient with asthenic built, funnel chest, arachnodactyly, joint instability upward dislocation of the lens and aortic incompetence⁶. However, the full gamut of these features are seldom present in one person. Without vigilance and a high index of suspicion, the diagnosis may be missed because of the rarity of this pathology in our environment. Most of the patients may be discovered when they present with one or two complications of this condition in addition to the typical features. This patient initially had severe myopia but later presented with an abdominal mass.

The commonest complication in Marfan's syndrome is usually cardiovascular including aortic aneurysm. Unlike the common findings of thoracic aortic aneurysm in Marfan's syndrome,^{1,3,6,9-12} this patient presented with an abdominal aortic aneurysm. Echocardiography did not show a thoracic component. Where abdominal aortic aneurysm was reported elsewhere, there was associated thoracic (thoraco-abdominal) aneurysm¹⁴⁻¹⁷. Abdominal aortic aneurysm may be asymptomatic but the

presence of abdominal and/or back pain are signs of impending rupture⁵. This patient had features of impending rupture. In addition, he had pain in the left lower limb which was attributed to nerve compression¹³.

To confirm the diagnosis and to determine the diameter of the aneurysm, abdominal duplex ultrasound is superior to aortogram, because often the aneurysmal cavity is filled with clots and cannot be demonstrated¹³. This patient had on-table aortogram which confirmed that the non-pulsatile mass was of vascular origin. Other investigations for diagnosis include CT scan and MRI. The patient had abdominal ultrasound but a diagnosis was not made. The use of Doppler ultrasound with colour flow mapping would have been helpful. The treatment of choice is an on-lay prosthetic graft.

The aneurysmal sac is usually closed over the graft in order to prevent the formation of aorto-duodenal fistula⁵. The place of non-surgical treatment is restricted to the use of beta blockers to slow down the rate of aortic dilatation^{21,22}. The sudden death of this patient in the fourth post-operative day could be as a result of pulmonary embolism or aortic graft dehiscence. Post mortem was rejected by the parents.

In view of the conundrum of diagnosis in this patient, we posit that asthenic patients with abdominal mass should be investigated for Marfan's syndrome and abdominal aortic aneurysm if we are to pick this rare entity.

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