3 Diagnosis and Management of Acute Complications Associated with Marfan Syndrome: Pitfalls in the Emergency Setting

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Introduction

The complications of Marfan syndrome (MFS), which may present as medical emergencies, are well recognised [1–3] and include aortic dissection/rupture, pneumothorax, retinal detachment, lens dislocation and joint dislocation. The optimum management of these complications has also been well described. Some of the above-mentioned complications are life-threatening, the most notorious being aortic dissection. If this condition is undiagnosed the early mortality is 1 % per each hour delay [4, 5]. The outcome is proportional to the time spent in making the diagnosis and moving the patient from the Emergency Department to the operating room. Unfortunately establishing the diagnosis of aortic dissection/rupture in the Emergency Department proves difficult, leading to avoidable mortality and morbidity. We have attempted in this chapter to highlight some of the pitfalls in the diagnosis. Our objective is that these complications will be recognised in a timely manner so that patients are moved to the appropriate facility with the least possible delay.

Chest Pain in a Young Adult with Known or Suspected MFS

If an MFS patient presents with chest pain, the first and the only diagnosis to consider at the beginning is aortic dissection or rupture. Only when this life-threatening condition is excluded is one free to consider other possibilities.

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Aortic Dissection

Acute dissection of the thoracic aorta is a life-threatening situation and carries a high mortality if definitive care is delayed. If untreated the mortality rate is 28 % within 24 h, 50 % in 48 h, 70 % within 1 week and 90 % or more within months [6, 7]. Marfan syndrome is a predisposing factor for aortic dissection. If a patient with known or suspected Marfan syndrome presents to the Emergency Department with chest pain, the diagnosis should be acute dissection of the aorta until proven otherwise. Such an attitude will make it imperative for us to consider the probability of the condition, thus avoiding the catastrophic situation where these patients are sent home with an alternative diagnosis.

It is important to recognise that 50 % of aortic dissections in women younger than 40 occur during pregnancy [8].

Presenting Symptoms and Signs in Aortic Dissection

In aortic dissection the mechanism for all the symptoms and signs are:

1. Dissection/rupture of the aorta.
2. Compression of adjoining structures by the expanding aorta.
3. Ischaemia caused by occlusion of the branches of the aorta.

The common presenting symptoms are as follows:

Chest pain. The pain is severe and sudden in onset. The maximum pain is at the onset. Patients often describe the pain as cutting, searing, ripping, or tearing. It is commonly retrosternal and interscapular and may be felt both above and below the diaphragm. A common characteristic of aortic dissection pain is midline truncal pain, but aortic chest pain may be atypical. Chest pain is the commonest symptom which could be due to cardiovascular, pulmonary, gastrointestinal, neurological or musculoskeletal causes. A high degree of suspicion in this special group of patients will lead to the diagnosis being made expeditiously.

Gastrointestinal symptoms. Abdominal pain can be the presenting complaint. This may be due to acute mesenteric ischaemia due to occlusion of the superior mesenteric vessels. One of the clues in this situation is pain, which is totally out of proportion to the abdominal findings.

Shortness of breath. Patients with acute dissection may present with acute shortness of breath, which could mimic asthma. This is due to acute pulmonary oedema from sudden onset of severe aortic insufficiency. The other causes are compression of trachea from the expanding ascending aorta. In Marfan syndrome the diagnosis to be considered with these symptoms are first aortic dissection and then spontaneous pneumothorax.

Fainting episode. About 5 % of all patients with aortic dissection present with a history of a syncopal attack [9]. Causes for this vary from cardiac tamponade to

hypovolaemia to conduction disorders of the heart. Again, in Marfan syndrome patients one should think of the worst case situation, which is aortic dissection.

Neurological complaints. About 20 % of patients with aortic dissection may present with a neurological symptom or we may elicit a neurological sign. The commonest cause of these symptoms is ischaemia of the spinal cord due to spinal artery occlusion or else cerebral vessel occlusion.

Clinical Signs in Aortic Dissection

Patients with acute aortic dissection may show a wide variety of signs. It is not uncommon, however, for there to be few clinical signs. Some clinical signs are very non-specific and some may clearly indicate the diagnosis. Possible signs include:

General signs – Anxious patient, diaphoresis, pale, raised pulse rate, raised respiratory rate, mottled extremities.

Cardiovascular – Hypotensive, normotensive, hypertensive, pulsus paradoxus, wide pulse pressure, aortic regurgitation murmur, muffled heart sounds (pericardial tamponade), asymmetrical pulses.

Head and neck – Horner's syndrome, vocal cord irritation or paralysis.

Respiratory system – Decreased breath sounds, basal crepitations, wheezing.

Central nervous system – Altered level of consciousness, hemiplegia, and paraplegia.

Investigations

Bearing in mind the high and time-dependent mortality of aortic dissection one should ask the question as to how suspicious are you of aortic dissection? If the suspicion is high then very early consultation with the local cardiothoracic surgeons is essential.

The most important adjunct to making the correct diagnosis is a high degree of suspicion. If according to the clinical information, the probability of dissection is high or moderate, the objective will be to transfer the patient to a cardiothoracic surgical centre. The other group of patients in whom the diagnosis of aortic dissection is considered to be a low probability should rapidly undergo diagnostic procedures to rule out the possibility.

ECG may mislead one through showing ischaemic changes or may be relatively normal. Chest x-ray can also be misleadingly normal. It is said that the chest x-ray will show one or more features of aortic dissection in 80 % of the cases of aortic dissection [10], but nonetheless in practice the chest x-ray may be easily passed as normal and provide dangerously false reassurance. Radiological features suggesting aortic dissection/rupture are: widened mediastinum, depression of the left main bronchus, small basal effusion, oesophagus shifted to the right (seen when a nasogastric tube is inserted before the x-ray), and a left-sided pleural cap.

Case History A supine chest x-ray of 26 year old female presenting to A & E with severe chest pain was normal. ECG showed ischaemic changes. The chest x-ray was initially used to exclude a dissection and diagnosis of acute myocardial infarct was made. Forty-five minutes later dissection was considered and confirmed by CT.

The other diagnostic tests available are aortography, computed tomography (CT), magnetic resonance imaging (MRI), transthoracic echocardiography (TTE) and transoesophageal echocardiography (TOE). It has to be recognised that excessive delays in awaiting investigation could do more harm to the patient. In this situation one has to weigh the risks to the patient and benefits carefully, in close early consultation with the local cardiothoracic surgeons, at any hour of the day or night.

Aortography is not as sensitive as believed earlier. In a study in 1989 its sensitivity and specificity were judged to be 88 % and 94 % respectively [11]. The sensitivity of CT is similar to that of aortography with values for sensitivity ranging from 83 to 90 %. Specificity ranges from 90 to 100 % [10, 12, 13]. A study in 1993 concluded that sensitivity and specificity of MRI could be 95 % and 100 % respectively [14, 15].

Even though TTE can give a rapid bedside diagnosis it has a low sensitivity of 60–80 % and low specificity of 60–90 % [10, 11, 14]. However, TOE has a high sensitivity of 95–100 % and specificity ranges from 70 to 95 % [9, 10, 13, 16].

Transfer of the Patient to the Regional Cardiothoracic Centre

It is very common for this group of patients to be transferred urgently from the initial receiving hospital to regional cardiothoracic centres.

The process of transfer has to be properly planned and then executed accurately and speedily. Transporting ill patients between hospitals is hazardous and adds to the patient morbidity and mortality. Seriously ill patients do not easily tolerate lifting, tipping, or any sudden abrupt movement. Sudden acceleration and deceleration is associated with potentiation of cardiovascular instability. It is observed that during transfer there is deterioration of oxygenation in seriously ill patients. Every hospital department which moves patients (inter hospital or intra hospital) should have a transfer checklist which has to be completed before moving the patient. This checklist is normally structured in three main parts: (1) Preparation for the transfer (administrative aspects); (2) Preparation of equipment, drugs and personnel; (3) Preparation of the patient. Prior to transfer of the patient, it is important to ensure the patient is stable and will remain stable throughout the journey. The patient should be accompanied by the most appropriate, experienced medical personnel. Specifically in this group of patients, if the patient is hypertensive, it is necessary for the blood pressure to be reduced. If the patient is hypertensive one has to bring down the pressure carefully by using a nitroprusside infusion to maintain the blood pressure around 90 mm of Hg to 100 mmHg. A beta-blocker given intravenously will reduce the velocity of the ventricular contraction thus helping to contain the dissection. All these patients should have adequate analgesia in the intravenous form.

Pneumothorax

Spontaneous pneumothorax is more common in Marfan syndrome patients than in the general population [17, 18]. This should be excluded early in the investigation of an MFS patient with chest pain or unexplained breathlessness by doing an erect PA chest x-ray. If confirmed, the management will be as for any spontaneous pneumothorax. If it is small and the patient is asymptomatic one can manage expectantly. However, if the patient is symptomatic the pneumothorax should be aspirated.

Endocarditis

Marfan syndrome patients have an increased risk of endocarditis compared to the general population [19, 20]. This applies to MFS patients in whom no cardiac lesion has been demonstrated as well as those more obviously at risk through implants or known mitral valve disease. In the emergency care setting this is relevant both in terms of diagnosis of unexplained symptoms, and during treatment, when the need to follow up-to-date guidelines for antimicrobial prophylaxis is important [21].

General Anaesthesia

It is known that general anaesthesia in Marfan syndrome patients carries an increased morbidity and mortality risk [22]. These risks include cardiac and pulmonary complications, and difficult endotracheal intubation. In addition to consideration of antimicrobial prophylaxis for Marfan syndrome patients during procedures under general anaesthetic in the emergency care setting, the need for an experienced anaesthetist is therefore emphasised.

Visual Disturbance in a Patient with Marfan Syndrome

The propensity of patients with Marfan syndrome to develop important ocular complications is quite high. These complications are best treated early in their development. Early consultation with the ophthalmologists is essential to achieve good outcomes. Wakita et al. evaluated the ocular complications associated with Marfan syndrome and listed them in order of frequency of the incidence and their findings are as shown below [23].

1. Dislocation of the lens 72.6 %.
2. Retinal detachment 36.4 %.
3. Glaucoma 15.2 %.

Lens Dislocation

Lends dislocation is the most common ocular abnormality in MFS, affecting 60–80 %. In the majority of cases it is present before 10 years of age. Ectopia lentis refers to a subluxated lens and may occur in Marfan syndrome. Once the lens has become displaced enough, it is no longer able to focus light and monocular diplopia or blurred vision occurs. Dislocation into the vitreous can occur in these conditions as well as in trauma. If increased intraocular pressure (IOP) occurs in the setting of a dislocated lens, a vitrectomy and lens removal are necessary to save the eye. Acutely, medication to lower the IOP can be used. Early referral to an ophthalmologist is important to ensure optimum outcome.

Retinal Detachment

Detachment of the retina is a well-recognised complication in MFS. The incidence of retinal detachment in aphakic eyes (16 %) is higher than in the phakic group (9 %) [23]. In order to get good results, early recognition of the condition and referral are very important.

Summary

Patients with Marfan syndrome can suddenly develop life- or vision-threatening complications. Aortic dissection is a life-threatening condition and if recognised early and referred in time the outcome remains return to normal life. Similarly, eye complications associated with Marfan syndrome should be referred to specialists early to avoid permanent visual impairment.

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