Case Report

Marfan syndrome presenting with transient renal insufficiency

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Introduction

Patients with Marfan syndrome frequently develop an aortic dissection [1]. We describe a patient who lived more than 5 years with a progressive dissection of the thoracoabdominal aorta and presented with acute renal insufficiency after a progression of the dissection beyond the origin of the renal arteries. After 6 days of complete anuria, however, serum creatinine levels normalized and diuresis started spontaneously. Chronic aortic dissection rarely causes renal insufficiency [2,3].

Case report

A 51-year-old woman was referred to our hospital because of acute renal insufficiency. Five years earlier the diagnosis of Marfan syndrome had been made, based on a very tall habitus with relatively long extremities and fingers (arachnodactyly), a lens subluxation of the left eye, and a thoracic aeurysm measuring 10 cm at its maximum and extending to the abdominal aorta. Furthermore the patient had a dilating cardiomyopathy with a severely decreased left ventricular function (ejection fraction 18%), valvular heart disease (mitral, tricuspidal, and aorta valve insufficiency), and paroxysmal ventricular tachycardias. The aortic dissection had been considered inoperable after extensive evaluation because of the cardiac condition of the patient. She was treated with frusemide, flecainide, and captopril.

At the time of admission she had been suffering from severe pain in the back and abdomen for 5 days, as she had had before during a previous progression of the dissection. She had not passed urine for the last 3 days. Further history did not reveal additional information.

On physical examination a woman with much pain and a Marfan-like appearance was seen. Her blood pressure was 90/60 mmHg (which was not unusual for her), the heart rate was 80 beats/min. Systolic and diastolic murmurs were heard, as well as crepitations at the lower segment of the lungs. On examination of the abdomen a 10-cm broad solid, not pulsating tender mass was found; pulsations of the femoral arteries were absent. Laboratory investigations revealed the following results: Hb 6.8 mmol/l, sodium 125 mmol/l, potassium 6.4 mmol/l, calcium 2.23 mmol/l, phosphate 4.11 mmol/l, serum urea 65 mmol/l, creatinine 647 μmol/l; liver function and CPK were normal. An ultrasound of the abdomen showed an increase of the aortic aneurysm dissection up to 13 cm, with the renal arteries originating in the dissected aorta; there was no hydronephrosis.

A chest X-ray showed an unchanged cardiomegaly with interstitial pulmonary oedema. The ECG showed a sinus rhythm, an intermediate axis, and broad QRS complexes (0.20 ms).

It was postulated that the acute renal failure was due to the inclusion of both renal arteries caused by progression of the aortic dissection in this patient with Marfan syndrome.

Our patient and her husband had been very well informed about the poor prognosis. At their urgent request, however, haemodialysis was started, as they had lived with a disease which had been estimated as having a very poor prognosis for 5 years already.

Dialysis sessions were uneventful. The patient had an episode of ventricular tachycardia twice, which was successfully treated with cardioversion and intravenous administration of flecainide. On the fourth day diuresis started spontaneously and serum creatinine returned to normal levels (Table 1). On the seventh day after admission, however, the patient was found dead without any warning signs.

Postmortem examination revealed a ruptured thoracic aneurysm with approximately 3 litres of blood in the thoracic cavity. The dissected aneurysm measured over 20 cm, extending below the iliac bifurcation, con-

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Table 1. Dialysis on days 1, 2, 3. Values for urea in mmol/l, for creatinine in μmol/l, before and after (B/A) dialysis

<table>
<thead>
<tr>
<th>Day</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
<th>6</th>
<th>7</th>
</tr>
</thead>
<tbody>
<tr>
<td>Urea</td>
<td>65/48</td>
<td>57/41</td>
<td>47/36</td>
<td>42</td>
<td>38</td>
<td>30</td>
<td>21</td>
</tr>
<tr>
<td>Creat</td>
<td>647/497</td>
<td>600/458</td>
<td>283/221</td>
<td>200</td>
<td>116</td>
<td>88</td>
<td>67</td>
</tr>
</tbody>
</table>

sisting mainly of a huge thrombosed false lumen with a small true lumen, the kidneys being totally absorbed in the aneurysmatic process (Figure 1). The aortic intima exhibited only minor atherosclerotic changes. The heart was hypertrophic. Advanced autolysis precluded adequate microscopic examination of the kidneys.

Discussion

Of all cases of aortic dissection 10% are caused by Marfan syndrome. The others result mainly from atherosclerosis, hypertension, infections (usually Staphylococcus aureus, Salmonella, Lues). Aortic dissection has been reported in Ehlers–Danlos syndrome, Noonan syndrome, Turner syndrome, and pregnancy [4–7]. The prognosis of a thoracic dissection is poor, with 25% of patients dying within 24 h and 90% within 1 year [8]. That this patient with Marfan syndrome lived for 5 years with a large progressive, inoperable dissecting aneurysm is very remarkable.

Marfan syndrome is an autosomal, dominant inherited disorder, although frequently skipped generations are observed due to its variable expression. In 15–30% of the cases it is caused by new mutations [9]. Marfan syndrome is a connective tissue disease, defined on characteristic findings in the skeleton, the eye and the cardiovascular system. Patients are relatively long compared to their relatives, have disproportionately long extremities and fingers (arachnodactyly), and often have chest deformities; they frequently have in their medical history an ectopia lentis and they often suffer from a mitral valve prolapses and aortic aneurysm [10].

It was remarkable that diuresis started spontaneously and the serum creatinine normalized after 6 days of complete anuria. We hypothesize that inclusion of the renal arteries in the dissection led to a reduction of blood flow, causing renal insufficiency and anuria, but that the blood flow was enough to prevent total infarction (and thus irreversible renal insufficiency). This might have been caused by compression on the renal arteries by the false lumen of the dissected aorta. Other causes of renal insufficiency are less likely; especially, the patient had not been suffering from prolonged periods of hypotension, not even during periods of ventricular tachycardia. Due to the bad condition of the patient no renograms were made.

Two things in this patient were unusual, one being the fact that she had lived with a progressive aneurysm for over 5 years, the other was that after 6 days of complete anuria the renal function improved to nearly normal creatinine and serum urea levels. Unfortunately the patient died of a fatal thoracic dissection.

Acknowledgements. We wish to thank Dr Willebrand, pathologist, for the postmortem examination of this patient.

Fig. 1a, b. Dorsal aspect of the abdominal aorta at postmortem examination, showing the dissected aorta (B), the kidneys (A), and the caudal extension of the aneurysm beyond the level of the iliac arteries (C).
References

9. Roberts WC, Honig HS. The spectrum of Cardiovascular disease in Marfan syndrome. A clinicomorphologic study of 18 necropsy patients and comparison to 151 previously reported necropsy patients. Am Heart J 1982; 104: 115–135

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