An unusual case of cardiomyopathy

Introduction

Hereditary haemochromatosis (HHC) is an autosomal recessive disorder of iron metabolism characterized by increased iron absorption and deposition in the liver, pancreas, heart, joints and pituitary gland. This article presents an unusual case where the only manifestations were cardiac and which progressed despite effective phlebotomy.

Discussion

The gene for HHC is found on the short arm of chromosome 6 and is designated HFE. The patient in this case was homozygous for C282Y, the most common mutation in HFE, present in around 80% of people with HHC (Hanson et al, 2001). However, apart from the cardiac manifestations he did not display any other clinical features of HHC. Liver biopsy was in keeping with haemochromatosis, but there was no evidence of cirrhosis.

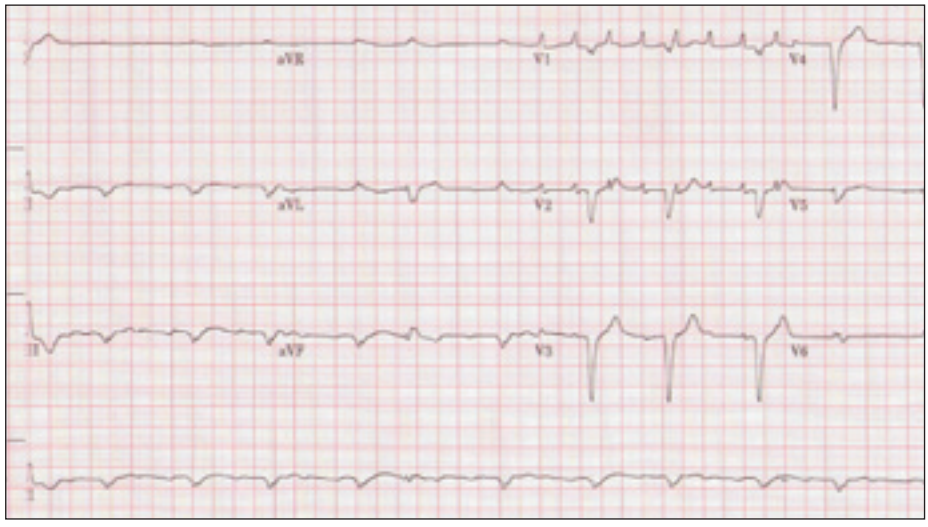

Figure 1. Patient's electrocardiogram 1 year before admission.

Cardiac manifestations of HHC are seen in one third of patients and include cardiomyopathy and arrhythmias (Niederau et al, 1996). The cardiomyopathy can be dilated or, more rarely, restrictive. The electrocardiogram in HHC often shows low-voltage and non-specific ST and T-wave changes. The most common disturbances in rhythm are atrial tachyarrhythmias (*Figure 1*), ventricular ectopics, ventricular tachyarrhythmia, sick sinus syndrome and varying degrees of atrioventricular block. The cardiac abnormalities are thought to relate to iron deposition in the myocardium and conducting system (Dabestani et al, 1988; Wang et al, 1994).

The mainstay of treatment for HHC is venesection, with good evidence that institution of phlebotomy before the onset of cirrhosis can significantly reduce mortality, potentially returning life expectancy to normal (Niederau et al, 1996). There are also case reports of severe cardiomyopathy

Case Report

A 56-year-old man presented with a 3-week history of worsening dyspnoea and lethargy. Clinical examination revealed signs of marked congestive cardiac failure and hypotension with a systolic blood pressure (BP) of 70 mmHg. Laboratory investigations revealed a urea of 29 mmol/litre and a creatinine of 310 μmol/litre with normal liver function tests, apart from a bilirubin of 40 μmol/litre. Initial treatment included oxygen, intravenous diuretics and ionotropes although his BP remained low. Subsequently, an intra-aortic balloon pump (IABP) was inserted with 1:1 augmentation and his clinical picture improved gradually. Cardiac catheterization was performed which showed normal coronary arteries and high right-sided heart pressures with a right ventricular pressure of 58/14 mmHg, pulmonary artery pressure of 58/28 mmHg, and a mean pulmonary capillary wedge pressure of 29 mmHg.

His electrocardiogram (ECG) 1 year before his current admission is shown in *Figure 1*. This revealed an atrial tachycardia with a cycle length of approximately 320 ms with upright P waves in lead V1 and inverted P waves in the inferior leads. There was a left bundle–branch block morphology with a QRS duration of around 100 ms and relatively high degree atrioventricular block with a ventricular rate of around 60–70 beats per minute.

He had initially presented 5 years earlier in congestive cardiac failure. ECG at that time showed first degree heart block with a QRS duration of 100 ms. Transthoracic echocardiography revealed a dilated left ventricle at 6.1 cm in diastole and a left ventricular ejection fraction <20%. Viral serology at the time was negative, he did not drink alcohol and did not suffer from ischaemic heart disease. His only other medical history consisted of hypothyroidism for which he was treated with thyroxine. His brother had died at the age of 44 years suffering from liver cirrhosis, diabetes and heart failure, which raised the question of hereditary haemochromatosis (HHC). The diagnosis of HHC was confirmed by HLA testing showing he was homozygous for the C282Y mutation of the responsible gene, a raised serum ferritin at 900 mg/litre and a liver biopsy in keeping with HHC (increased iron deposition in the liver cells but with normal acinar architecture and no inflammation or cirrhosis).

After stabilization with an IABP, his treatment options for cardiogenic shock included either insertion of a left ventricular assist device to improve BP and renal function or cardiac transplantation. Unfortunately the patient became septic from his IABP insertion site and so neither were viable options. He died shortly afterwards.

Dr Christopher A Miller *is Senior House Officer and* **Dr Simon G Williams** *is Consultant Cardiologist, Cardiac Transplant Unit and Department of Cardiology, Wythenshawe Hospital, Manchester M23 9LT*

Correspondence to: Dr SG Williams

secondary to HHC responding remarkably well to phlebotomy (Madani and Bormanis, 1997). However, in this case the cardiomyopathy progressed despite regular phlebotomy maintaining low ferritin levels (40 μg/litre).

Once congestive cardiac failure develops, survival is usually limited unless cardiac transplantation is considered. A review of the world literature until 2003 reported 11 patients with HHC who had undergone heart transplantation (two of which had concomitant liver transplants) as well as five patients with other causes of iron-overload cardiomyopathy (Caines et al, 2005). All continued repeated phlebotomy post-transplant. Five-year survival of the 16 patients was 81% and only one patient developed histological evidence of mild iron accumulation in the cardiac allograft. While transplantation does not halt the course of the disease, it does extend survival once symptoms are refractory to maximal medical therapy. **BJHM**

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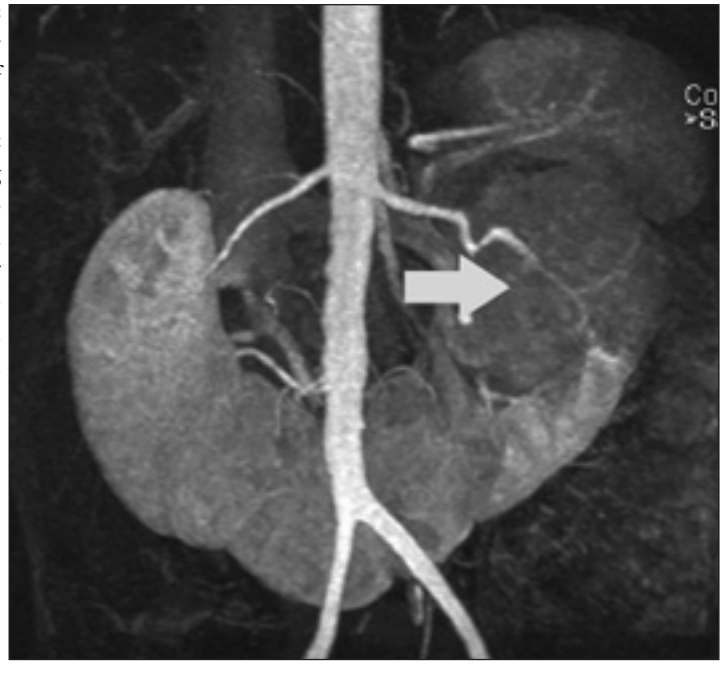
Oncocytoma in a horseshoe kidney

A 60-year-old man presented to the urological service for investigation of haematuria. As part of his work up, he underwent renal ultrasound. This identified the presence of a mass within the upper pole of the left component of a horseshoe kidney. In view of the complex nature of the abnormality, a gadolinium-enhanced magnetic resonance angiogram was performed. A maximum intensity projection (MIP) image clearly demonstrated the mass in the upper pole of the left component of the horseshoe kidney (*Figure 1*). At surgery the left component of the horseshoe kidney was removed and the mass was found to be an oncocytoma.

Horseshoe kidney is a common congenital renal abnormality. The incidence of horseshoe kidney in the population is estimated to be 1/400 to 1/600 births based on autopsy data from the 1940s and 1950s (Weizer et al, 2003). The malformation is a result of abnormal fusion of the metanephric blastemas of both kidneys in the 6th or 7th week of gestation. Fusion of the lower poles of the kidneys causes failure of the kidneys to ascend and rotate in the abdominal cavity.

The association between horseshoe kidney and tumours conditions is extremely rare with less than 200 cases reported in the literature (Stimac et al, 2004). Oncocytoma is an unusual renal tumour with 115 cases reported in the literature and only four cases reported in association with horseshoe kidney (Klimberg et al, 1986; Destito et al, 1992; Mottola et al, 1992; Stimac et al, 2004). Oncocytoma has an overall 5-year survival rate of 65% (Lewi et al, 1986). Magnetic resonance imaging is very useful to confirm the renal anomaly, tumour situation, vascularity and to assist in planning surgical intervention. **BJHM**

Figure 1. Maximum intensity projection (MIP) of a magnetic resonance angiogram demonstrates the anatomy of the horseshoe kidney and the oncocytoma in the left upper pole.



Dr Orla Buckley *is Specialist Registrar,* **Dr John Feeney** *is Specialist Registrar,* **Dr Tony Geoghegan** *is Consultant Radiologist and* **Dr William C Torreggiani** *is Consultant Radiologist, Department of Radiology, The Adelaide and Meath Hospital, Tallaght, Dublin 24, Ireland*

Correspondence to: Dr WC Torreggiani

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