CASE STUDY

Localized myopathy in a young man with abetalipoproteinaemia – myocardial infarction?

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Myocardial infarction has not previously been reported as a complication of abetalipoproteinaemia (ABL). We describe a case of probable asymptomatic myocardial infarction in a 24-year-old male with ABL and no previous cardiac history. Electrocardiographic and imaging evidence of the injury is presented.

Introduction

Abetalipoproteinaemia (ABL) described by Bassen and Kornzweig[1] is characterized by the absence of serum apolipoprotein B. The clinical features have been well described[2], but cardiac disease has not been a prominent feature. Cardiac failure, premature ventricular complexes (on ECG), and murmurs have been reported occasionally[1,3–5]. We report here the association of probable myocardial infarction with documented ABL.

Case report

W.E. is a 25-year-old white male who was first admitted to hospital at age 12 years for evaluation of diarrhoea and fat intolerance. He was the product of a non-consanguineous marriage of parents with normal plasma lipoproteins. He had failed to thrive from birth with intermittent vomiting and diarrhoea, and delayed psychomotor development. On physical examination he was below the third percentile in height and weight, and vibration sense was absent in the lower extremities. Peripheral blood smear showed numerous acanthocytes. Blood chemistry revealed an elevated skeletal creatinine phosphokinase and elevated lactic dehydrogenase. Chest X-ray and ECG were both normal. Serum cholesterol was 0.96 mmol/l, serum triglycerides were undetectable. No beta or prebeta lipoproteins were seen on lipoprotein electrophoresis and no apoliprotein B was detected by serum radial immunodiffusion.

The rapid progression of neurological symptoms at age 22 years led to reevaluation. He denied cardiac symptoms, dyspnoea or smoking history. There was no history of illicit drug abuse. There was no family history of premature vascular degenerative disease. On physical examination he was thin and alert, with a blood pressure of 130/80 mmHg and pulse rate of 70 beats/min. Cardiac and fundoscopic examinations were normal. Carotid and extremity pulses were normal and without bruits. Neurological examination revealed marked spinocerebellar degeneration.

A peripheral blood smear demonstrated acanthocytes and a normal haemoglobin. Total serum triglyceride was <0.2 mmol/l, total serum cholesterol <1.2 mml/l. Blood vitamin A levels were undetectable. Vitamin E levels were 0.8 μg/ml) (range 5 to 20 μg/ml) on 400 U/d oral supplementation. There was a complete absence of serum apoliprotein B on immunodiffusion. Electrocardiography revealed Q waves in II, III and V5 and V6, and inverted T waves in V3–V6 consistent with an inferolateral myocardial infarction (Figure 1). Bicycle exercise testing revealed a mildly reduced exercise tolerance with a peak work load of 600 kpm/min and peak oxygen consumption of 28.3 ml/min per kg. There was no change in the ECG during exercise. Thallium scanning both at rest and during exercise revealed a constant apical perfusion defect. A gated blood pool scan using ^{99}Tc (Figure 1) showed that during diastole (image No. 1), the left ventricle demonstrated an apical bulge. During systole (image No. 10), the apex was akinetic but the rest of the wall motion was normal; global left ventricular function was well preserved with an ejection

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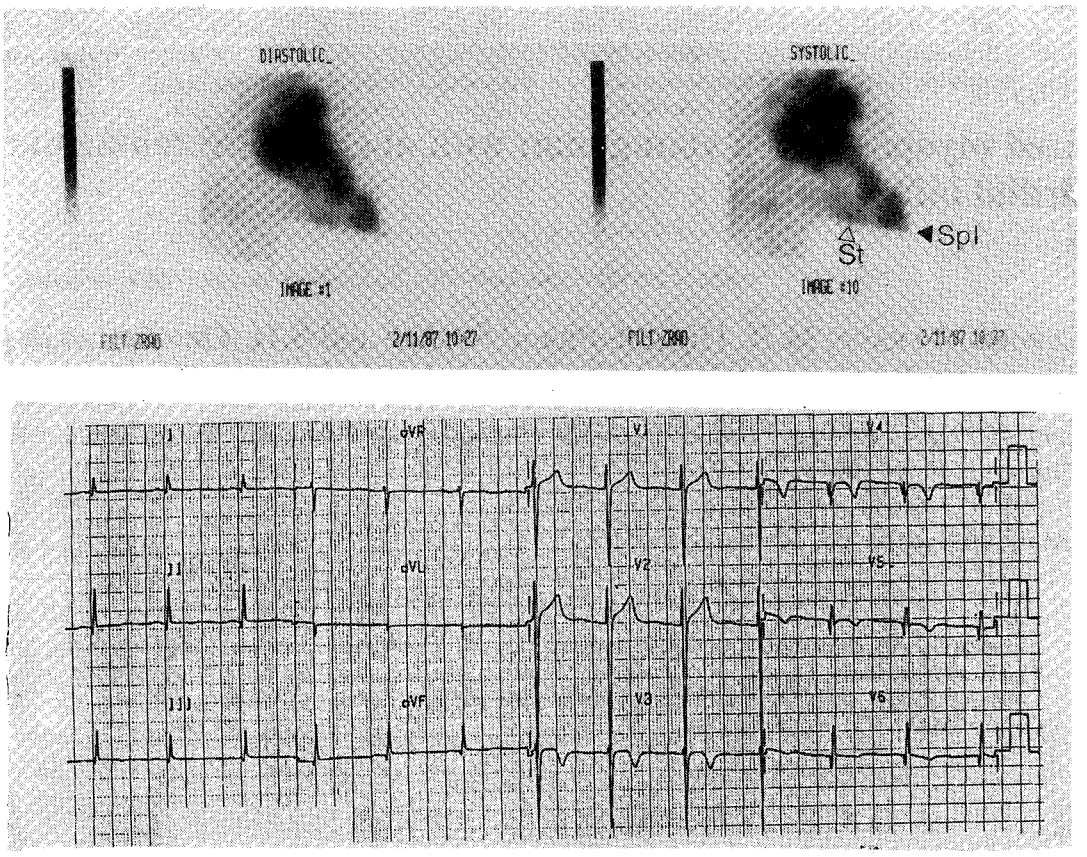

Fig.1. Resting 12 lead ECG and gated blood pool scan (^{99}Tc) showing diastolic and systolic frames. Arrows indicate spleen (Spl) and stomach (St).

fraction of 50%. Right ventricular function was normal. Normal coronary vessels were seen on coronary angiography.

Comments

An ECG pattern consistent with a past inferolateral myocardial infarction, and evidence of myocardial destruction, was present in this 22-year-old male with ABL. Myocardial infarction had not previously been reported as a feature of ABL. Dische et al[3] reported a case of ABL in a 10-year-old male who had palpitations, multifocal PVC on ECG and biventricular enlargement on chest X-ray. At post-mortem, he had an enlarged and dilated heart with fibrotic myocardium and endocardium but grossly normal coronary circulation. Histologically, the myocardium had extensive interstitial fibrosis. The coronary arteries had marked intimal fibrous thickening and some fragmentation of internal elastic lamellae.

Sobrevilla et al[4] reported a 36-year-old female with ABL, cardiac failure and premature ventricular complexes (PVC) on ECG. Post-mortem examination revealed an organizing mural thrombus in the left ventricle, but no histological evidence of myocarditis or myocardial infarction. The coronary vessels were thin, transparent and patent.

Although it is possible that our patient had a localized cardiomyopathy, the gated blood pool scan was not consistent with an apical hypertrophic cardiomyopathy. The electrocardiographic and imaging studies made it much more likely that he sustained a myocardial infarction as a result of a perfusion abnormality. The absence of beta lipoproteins suggested the absence of coronary atherosclerosis but the unusual nature of the remaining lipoproteins[2] made lipoprotein risk factor definition insecure. Neither of the cases with detailed post-mortem cardiac evaluations[3,4] revealed coronary atherosclerosis or localized cardiomyopathy. In our patient, coronary angiography revealed a normal coronary circulation without evidence of embolization. The aetiology of the myocardial infarction may have been vasospastic, but embolism could not be excluded. Cocaine abuse can cause coronary vasospasm, but use of illicit drugs was denied[5].

A skeletal myopathy has been described in ABL[6] and was very similar in features to the skeletal myopathy occurring in vitamin E deficiency[7–9]. Neither a smooth muscle disorder nor cardiac disease have been reported in association with vitamin E deficiency. There are neurologic and cardiac similarities between ABL and Friedreich's ataxia. Myocardial infarction and ECG abnormalities have been reported in young individuals with Friedreich's ataxia. These were associated with proven occlusive coronary artery disease of unknown aetiology[10,11]. The angiographically normal coronary arteries in this patient indicate that, despite clinical similarities between Friedreich's ataxia and ABL, the myocardial necrosis differed in pathogenesis.

This case, and the previous pathologic descriptions[3,4] of the heart in ABL, make it possible that these patients have a perfusion abnormality resulting in diffuse or localized myocardial necrosis.

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摘要

血漿β脂蛋白缺乏症并發心肌梗塞一例報告

血漿β脂蛋白缺乏症并發心肌梗塞尚未見報道。本文報道一例以前無心臟病史的２４歲男性血漿β脂蛋白缺乏症患者發生無症狀性心肌梗塞。心電圖及影像學檢查證實心肌損傷的存在。