Unusual presentation of more common disease/injury

Kidney infarction in Friedreich's ataxia with dilated cardiomyopathy

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Summary

A 37-year-old man with advanced Friedreich's ataxia was referred to our emergency department with acute exacerbated abdominal pain of unclear aetiology. Laboratory tests showed slightly increased inflammatory parameters, elevated troponin and B-type natriuretic peptide, as well as minimal proteinuria. Transthoracic echocardiography revealed a pre-existing dilated cardiomyopathy. Abdominal sonography showed no pathological alterations. Owing to persistent pain under analgesia, a contrast-enhanced CT-abdomen was performed, which revealed a non-homogeneous perfusion deficit of the right kidney, although neither abdominal vascular alteration, cardiac thrombus, deep vein thrombosis nor a patent foramen ovale could be detected. Taking all clinical and radiological results into consideration, the current incident was diagnosed as a thromboembolic kidney infarction. As a consequence, lifelong oral anticoagulation was initiated.

BACKGROUND

Friedreich's ataxia is a rare and incompletely understood disease. Cardiac dysfunction is the most frequent cause of death in almost two-thirds of patients affected, most commonly from congestive heart failure or arrhythmia. Renal artery embolism, although not rare in these patients, represents a mayor clinical problem since diagnosis is often delayed or missed.

CASE PRESENTATION

A 37-year-old Caucasian man was referred to our department from an external hospital because of severe abdominal pain of unclear aetiology. The patient suffered from Friedreich's ataxia.^{1 2} His most recent transthoracic echocardiography, performed on the day of admission, showed a dilated cardiomyopathy, impaired left ventricular function with an ejection fraction of 30% and a pulmonary artery pressure of 55 mm Hg; all findings were unchanged from previous examinations. At that time, he lived alone in a residential home and was mobile using a wheelchair.

On arrival at our emergency department, the patient was somnolent and disoriented. His axillary body temperature was 35.7 °C and his arterial blood pressure 125/89 mm Hg, with a pulse rate of 84 rhythmic beats/min. He complained of an increasing, diffuse abdominal pain during the whole day, combined with absence of appetite and vomiting on a single occasion.

His daily medication was perindopril 2.5 mg, torasemide 7.5 mg and esomeprazole 40 mg.

INVESTIGATIONS

On physical examination, we found a tender abdomen with painful epigastric palpation. Normal bowel sounds were auscultated in the upper right and lower left quadrants. The renal bed was free of pain on palpation. On cardiac auscultation, a 2/6 systolic murmur was audible. Lung auscultation showed normal respiration.

Figure 1 Ultrasound of the right kidney shows a normal parenchyma with preserved cortico-medullary differentiation and no focal lesions.

Laboratory findings were slightly elevated C reactive protein (CRP) of 16 mg/l (normal: <5 mg/l), elevated troponin-T-hs of 0.070 µg/l (normal: <0.014 µg/l) and highly elevated pro-B-type natriuretic peptide of 7382 pg/ml (normal: <63 pg/ml). Kidney function parameters were in the upper normal range, with creatinine of 100 µmol/l and a calculated glomerular filtration rate of 73 ml/min. Blood count revealed leucocytosis of 15.6×10^9 /litre. Regarding thyroid function, thyroid-stimulating hormone, f-T3 and f-T4 were in the reference range. Urine analysis showed no pathology.

In abdominal sonography, all organs were normal, especially the kidneys. No free fluid was detected (figure 1).

As the severe pain persisted all night, despite analgesia with paracetamol 1 g, metamizole 1 g and pethidine 75 µg, a contrast-enhanced abdominal CT was performed revealing infarction of the right kidney, which appeared as a hypodense area although kidney arteries and veins were assessed as open (figures 2 and 3).

Figure 2 CT of the abdomen, axial plane: hypodensity of the right kidney with decreased enhancement of the contrast medium.

DIFFERENTIAL DIAGNOSIS

Cardiac dysfunction is the most frequent cause of death in patients affected by Friedreich's ataxia, most commonly from congestive heart failure or arrhythmia. Renal artery embolism is not a rare event in these patients. However, other non-abdominal viscous causes of severe diffuse abdominal pain such as porphyria, familial Mediterranean fever, diabetic ketoacidosis, tocopherol deficiency also had to be excluded. The negative cardiolipin test excluded tabetic crises. No evidence for pre-existing porphyria was encountered for example, quantitative determination of δ -aminolevulinic acid, porphobilinogen and porphyrin in 24-h urine was negative. Further genetic diagnosis regarding, for example, clotting disorder, was also not

Figure 3 Contrast-enhanced CT of the abdomen, frontal plane: non-homogeneous perfusion deficit in the slightly enlarged right kidney.

conclusive. Hypovitaminosis was not present. The patient had no endocrinopathies, for example, no diabetes and no evidence for hypothyreosis or hyperthyreosis.

TREATMENT

The patient was hospitalised without delay at the department of nephrology. Immediate anticoagulation with heparin was initiated and later replaced by lifelong oral anticoagulation with phenprocoumon.

OUTCOME AND FOLLOW-UP

During 11 days of hospitalisation, no complications occurred. Kidney function parameters remained normal. Transoesophageal echocardiography on the fifth day showed no cardiac thrombus and no patent foramen ovale. Deep vein thrombosis was excluded with duplex sonography. Anticardiolipin antibody testing and genetic testing regarding clotting disorders were both negative. Urine tests revealed no evidence of porphyria.

During hospitalisation the patient remained stable regarding cardiological function, while diuretics were adjusted according to the fluid balance.

The patient had already been treated with levofloxacin for pneumonic infection prior to hospitalisation. The antibiotic treatment was continued because of increasing inflammatory parameters in the further course of disease. A CRP increase of up to 200 mg/l was interpreted as an inflammatory response to renal infarction. By discharge, laboratory parameters were significantly decreased.

DISCUSSION

The exact cause of the kidney infarction remains unclear to us. With the underlying disease, we favour the theory of a thromboembolic event with underlying intracardiac thrombus. Although such a cardiac thrombus was never detected in our patient, it has been described in similar patients.^{3 4} Thromboembolism in Friedreich's ataxia has been described in the lung and brain,^{5–7} but never in the kidney.

Abdominal ultrasound did not show any alterations in the affected kidney, as this procedure is very insensitive to parenchymal alterations. Contrast-enhanced CT and radioisotope renal scan are considerably more sensitive.^{8 9}

Learning points

- ▶ Kidney infarction is a differential diagnosis in acute abdominal pain.
- ▶ Consider thromboembolic events in patients with cardiomyopathy, such as that in Friedreich's ataxia.
- ▶ Abdominal sonography may not be decisive in the diagnosis of kidney infarction.

Competing interests None.

Patient consent Obtained.

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