McArdle's Disease Presenting with Acute Renal Failure

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Dear Sir,

Myoglobinuria after exercise is common in McArdle's disease but acute renal failure (ARF) is rare. In the following case, exercise-induced rhabdomyolysis and ARF were actually the presenting manifestations of the disease.

A 23-year-old male patient was admitted for weakness and dark urine. Three days earlier he had suffered a generalized seizure following a tennis match and subsequently noted dark urine. Past medical history was unremarkable except for an episode of exercise-induced pigmenturia at the age of 17. The patient practised several sports including tennis three times a week and never complained of exercise-related muscle pain or cramping. However, since childhood he felt quickly fatigued after sprint running and tended to avoid it. He did not consume drugs, alcohol or any medication. There was no family history of muscle disease.

Physical examination was entirely normal. Serum chemistry was as follows: creatinine phosphokinase 20,875 U/l (normal up to 195 U/l), aspartate aminotransferase 296 U/l (normal up to 40 U/l), alanine aminotransferase 534 U/l (normal up to 40 U/l), lactate dehydrogenase 474 U/l (normal up to 130 U/l), creatinine 858 $\mu\text{mol/l}$ (9.7 mg/dl), urea 42 mmol/l (252 mg/dl), sodium 127 mmol/l, potassium 5.0 mmol/l, calcium 2.05 mmol/l (8.2 mg/dl), phosphate 1.3 mmol/l (4.0 mg/dl), uric acid 765 $\mu\text{mol/l}$ (13.3 mg/dl). Arterial blood gases were normal. Urinalysis revealed 30–40 red blood cells per high power field; no myoglobinuria was detectable at that time.

Despite vigorous intravenous hydration followed by attempted forced alkaline diuresis, the patient remained oliguric and had to be dialyzed on 3 occasions. Serum creatinine was 407 $\mu\text{mol/l}$ (4.6 mg/dl) at discharge 2 weeks after admission and 88 $\mu\text{mol/l}$ (1.0 mg/dl) 3 weeks later. An ischemic forearm test was performed. After 30–45 s, the patient could not move his hand because of severe sustained cramps in his forearm and wrist. Blood lactate levels did not increase with exercise and actually decreased from 1.63 to 1.22 mmol/l.

A biopsy of the left biceps muscle was obtained. No reaction was observed to myophosphorylase compared to a control. Other enzymatic studies including phospho-fructokinase were normal. Light microscopy revealed the presence of characteristic subsarcolemmal PAS-positive vacuoles containing glycogen.

Myoglobinuria occurs in about half the cases of McArdle's disease [1]; it usually follows a particularly intense and/or prolonged physical activity. Myoglobin-induced ARF is a distinctly rare occurrence [2–16]. With only one exception [14], it has only been reported in male subjects, mostly young adults. Muscle symptoms are typically milder and more often have a late onset

(age > 15) in patients who develop ARF than in those who do not [1–16]. Perhaps the existence of some residual myophosphorylase activity as measured by sensitive techniques could be a predisposing factor [17] although more studies are needed to clarify this issue.

Some clinical clues may help to distinguish McArdle's disease from other myoglobinuric states leading to ARF. The typical patient is a young male patient with postexercise pigmenturia, usually with a previous history of exercise-related symptoms. Seizures are particularly common [1]. They tend to precede the pigmenturia [2,4,7], as in our patient. Their exact mechanism is unknown. Serum CPK levels usually remain somewhat elevated after the acute episode of rhabdomyolysis. Finally the family history may be positive.

Identification of subjects with McArdle's disease is important since they may benefit from a high-protein diet [18], avoidance of certain physical activities as well as genetic counseling. This diagnosis should be ruled out in any case of postexercise pigmenturia or ARF.

McArdle's Disease and ARF

697

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