McArdle's Disease Presenting as Acute Renal Failure

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Dear Sir,

McArdle's disease is a primary myopathy related to impaired glycogen breakdown [1]. Myoglobinuria occurs frequently in this disease, yet acute renal failure (ARF) is distinctly uncommon [2]. Moreover, ARF in McArdle's disease in the absence of any precipitating factor has been reported only once in the literature so far [2]. We here report the second such case of acute renal failure in McArdle's disease without any definite precipitating factor.

A 30-year-old male presented to the emergency services of the All-India Institute of Medical Sciences, New Delhi, in February 1994 with a history of high-coloured urine and gradually progressive oliguria for 2 days prior to admission. Simultaneously, he also developed weakness of all four limbs, which rapidly progressed to complete quadriplegia in the next 48 h. On direct questioning, the patient gave a history of easy fatigability and generalised muscle aches for the past 1 week prior to admission. There was, however, no history of any vigorous or prolonged exercise, seizures, alcohol or any other drug intake preceding these complaints. He had had two prior episodes of quadriparesis without any renal dysfunction in the past at the age of 10 years and 14 years, which had completely disappeared spontaneously. One of his younger brothers and his first cousin had expired at the age of 21 years and 30 years, respectively, because of a similar illness. Both had quadriparesis along with oliguria and high-coloured urine, preceded by a history of vigorous exercise, and had expired before they could receive any medical attention. His mother also complained of exercise intolerance and muscular aches since childhood though she had learnt to limit her physical activity lest muscular symptoms be induced.

At admission, the patient had a pulse rate of 54/min, supine blood pressure of 150/80 mm Hg and was afebrile. Physical examination revealed grade 0 power in the lower limbs and 3/5 power in both upper limbs without any muscle atrophy or fasciculations. There was mild tenderness in the quadriceps muscles bilaterally. Investigations at admission revealed a blood urea of 18 mmol/l (110 mg/dl), serum creatinine 513 µmol/l (5.4 mg/dl), sodium 128 mmol/l, potassium 7.4 mmol/l, uric acid 476 µmol/l (8.0 mg/dl), calcium 1.45 mmol/l (5.8 mg/dl), phosphate 3.4 mmol/l (10.5 mg/dl), alanine aminotransferase 620 IU/l (normal up to 40 IU/l),

aspartate aminotransferase 117 IU/l (normal up to 40 IU/l), lactate dehydrogenase 3,690 IU/l (normal up to 130 IU/l) and creatine phosphokinase 48,000 IU/l (normal up to 200 IU/l). Arterial blood gases revealed severe metabolic acidosis with a pH of 7.12. Urinalysis showed 20-30 RBCs per high power field – no myoglobinuria was, however, detectable at this stage. The patient was initially given one peritoneal dialysis and then subsequently put on alternate-day hemodialysis via a right sub-clavian catheter in view of his hypercatabolic state. He received a total of 13 hemodialysis sessions over the next 3 weeks, following which his urine output gradually picked up and his muscle weakness also started improving. Muscle biopsy done on the 22nd day after admission from the right quadriceps revealed excess of free glycogen on both light and electron microscopy. The histochemical staining showed absence of myophosphorylase activity as compared to a control, thus confirming the diagnosis of McArdle's disease. The patient was discharged 6 week after admission with normal renal function and complete recovery of his quadriparesis.

Although myoglobinuria occurs in about half of the cases of McArdle's disease, ARF is rare and has been described, to the best of our knowledge, in only 21 cases to date [2-8]. This may be explained by the limited

exercise capacity of these patients, which is generally not exceeded by them, thus avoiding severe muscle breakdown, profound myoglobinuria and ARF. All cases of ARF in McArdle's disease reported till now, except 1 [2], have been precipitated by vigorous physical activity, including strenuous exercise and heavy lifting, or a grand mal seizure. Our patient is thus the only second such case of McArdle's disease having ARF in the absence of any definite precipitating factor. In conclusion, ARF can rarely occur in patients of McArdle's disease in the absence of any definite precipitating factor for rhabdomyolysis.

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