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SHORT REPORT

Acute renal failure due to rhabdomyolysis in a child with McArdle disease

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Abstract Rhabdomyolysis induced acute renal failure (ARF) is relatively rare in children. We report an 8-year-old boy with McArdle disease and rhabdomyolysis induced ARF after heavy muscle work. Physical examination revealed generalized tenderness on his extremities. Laboratory examinations showed acute renal failure due to myoglobinuria and revealed alanine transaminase 428 U/l, aspartate transaminase 1,400 U/l, blood urea nitrogen 119 mg/dl, creatinin 3.6 mg/dl, uric acid 13 mg/dl, and serum creatinine kinase (CK) 33,766 U/l. Hemodialysis was carried out for ARF. His clinical and laboratory findings improved and became normal in 2 weeks. Enzymatic analysis of the muscle biopsy showed a phosphorylase A level of 129 nmol/s/mg protein (normal: 200–600) and a phosphorylase A+B level of 385 nmol/s/mg protein (normal: 500–1500), which was compatible with glycogenosis type V. As McArdle disease rarely becomes symptomatic and ARF secondary to this condition is very rare, our case represents a rare clinical presentation.

Keywords Glycogen storage disease type V · Rhabdomyolysis · Acute renal failure · Childhood · Treatment

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Introduction

McArdle disease (glycogen storage disease type V; OMIM 232600) is a rare autosomal recessive disease characterized by myophosphorylase (α-1,4-glucan orthophosphate glycosyl transferase) deficiency [3]. The typical features of McArdle disease include exercise intolerance with myalgia, early fatigue, stiffness of exercising muscles, rhabdomyolysis, and myoglobulinuria. Onset typically occurs by teenage years [1].

We report an 8-year old-boy presenting with acute renal failure (ARF) secondary to rhabdomyolysis due to McArdle disease.

Case report

An 8-year-old boy was referred to our department because of acute renal failure. He complained of myalgia, dark urine, and oliguria which had begun after chopping wood. The parents were second cousins. Physical examination

revealed generalized tenderness on the extremities and a systolic cardiac murmur of 2/6 intensity. Laboratory tests revealed no evidence of hemolysis. Serum electrolytes, glucose, bilirubin, and gamaglutamyl transferase (γGT) levels were within normal range suggesting that the increased transaminases were the muscle isoforms. The alanine transaminase (ALT) was 428 U/l, aspartate transaminase (AST) 1400 U/l, blood urea nitrogen (BUN) 119 mg/dl, creatinin 3.6 mg/dl, uric acid 13 mg/dl, and serum creatinine kinase (CK) was 33,766 U/l (normal 0–195 U/l), which were all over the normal limits. Urinalysis showed 1(+) protein and 3(+) hemoglobin reaction with normal microscopic examination. Complement levels, immunoglobulins, blood acylcarnitine analysis, echocardiography, and electromyography were normal. Antinuclear antibody (ANA) and anti-DNA tests and viral markers were negative. Renal ultrasonography showed mildly hyperechogenic kidneys. After admission, intravenous fluid and electrolyte treatment in addition to hemodialysis were started. Ischemic forearm test showed deficient glycogenolysis. Periodic acid schiff (PAS) staining of the muscle biopsy sample revealed an increased amount of subsarcolemmal glycogen (Fig. 1). Muscle phosphorylase A activity was measured as 129 nmol/s/mg protein (normal: 200–600) and phosphorylase A+B as 385 nmol/s/mg protein (normal: 500–1,500). Phosphofructokinase and myoadenylate deaminase levels were normal.

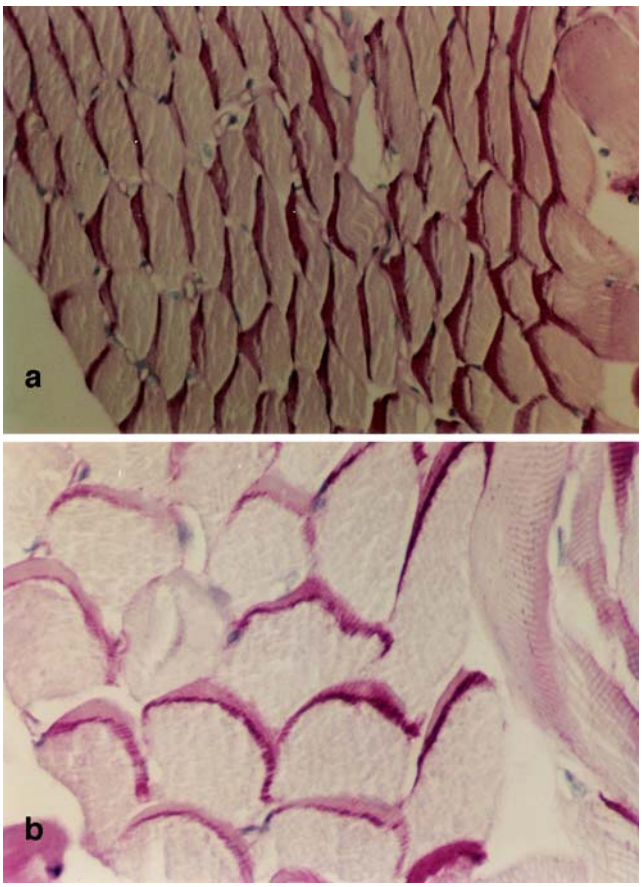

Fig. 1 Subsarcolemmal deposits with crescent formation. **a** H.E.×200. **b** PAS×400

With daily hemodialysis and supportive therapy, clinical and laboratory findings of the patient recovered. Strenuous exercise was avoided. Pyridoxine (30 mg/day) and a protein rich diet was recommended to supply branched chain amino acids as alternative energy sources for muscles.

Discussion

Despite the impaired glycogenolysis in muscles in McArdle disease, only intensive exercise leads to muscular symptoms due to lack of sufficient energy. McArdle disease does not usually become symptomatic during childhood due to higher muscle aerobic capacity [4]. Myoglobulinuria is almost always associated with physical exertion as seen in our patient [1]. The mechanism of renal damage is the tubular precipitation of myoglobin in the glomerular filtrate leading to obstruction. But progression to end stage renal disease has never been reported. In the symptomatic treatment of McArdle disease, limitation of muscular activity in addition to providing glucose and fructose to bypass the metabolic block, a protein rich diet, branched-chain amino acids as alternative fuels for muscles, and pyridoxine (myophosphorylase cofactor) have been suggested [2]. However, during acute renal failure, prompt diagnosis and appropriate treatment with intravenous hydration is the mainstay of the treatment.

Although the onset of McArdle disease is usually seen by the teenage years, inborn errors of muscle metabolism should always be considered among the possible causes of renal dysfunction even at a young age.

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