Intensive Care Med (2001) 27: 1235
DOI 10.1007/s001340100985

CORRESPONDENCE

K. H. Smolle
P. Kaufmann
R. Gasser

Recurrent rhabdomyolysis and acute respiratory failure due to carnitine palmityltransferase deficiency

Received: 30 November 2000
Accepted: 23 April 2001
Published online: 24 May 2001

Sir, Rhabdomyolysis is not uncommon in intensive care units. Recurrent rhabdomyolysis can be caused by extreme physical exercise as a result of cerebral cramps, toxic substances such as alcohol and cocaine, heat-stroke, hyperthermia, and infection (mycoplasma) and, in rare cases, hereditary disease. Symptoms of carnitine palmityltransferase deficiency are noted in most patients during the first or second decade of life in form of myalgias, cramps, muscle stiffness, or weakness [1, 2, 3, 4].

Alternatively, another disorder presenting with exercise intolerance, followed by recurrent cramps, fatigue, and myoglobinuria should be considered in this context: McArdle's disease, a hereditary autosomal recessive muscle phosphorylase deficiency which manifests in juveniles and young adults. In contrast to carnitine palmityltransferase deficiency, however, creatine kinase levels do not completely return to normal between the episodes of rhabdomyolysis in McArdle's disease [5]. Rhabdomyolysis with myoglobinuria may lead to acute renal failure and electrolyte abnormalities. Clinical manifestations can be induced by exertion, exposure to cold, viral infections, and emotional stress.

A 38-year-old patient presented at our intensive care unit with generalized muscle weakness, increasing respiratory insufficiency, and extremely dark coloring of urine. Two days before admission the patient suffered flulike symptoms. Muscle pain and increasing respiratory difficulties caused him to present at the hospital. He suffered similar symptoms several years previously and received artificial ventilation and renal replacement therapy. As a child he had experienced strong muscular pain after extreme physical exercise, which was not investigated. At the time of admission the patient was lethargic and dysarthric and had generalized pain in skeletal muscles. His body temperature was 39.1 °C, heart rate 105 beats/min and respiratory rate at 30 breaths/min. Blood pressure was 109/75 mmHg. Blood analysis yielded the following results: creatine kinase 11.869 U/l on admission and 35.588 U/l 1 day later, myoglobin in serum 66.300 μg/l, creatine kinase isoenzyme muscle-brain 649 U/l, aspartate aminotransferase 1188 U/l, alanine aminotransferase 280 U/l, lactate dehydrogenase 1474 U/l, creatinine 1.1 mg/dl, urea 30 mg/dl, white blood count 10.8×10^3, hemoglobin 16.4 g/dl, C-reactive protein 35 mg/l, hepatitis B surface antigen, hepatitis C virus AB, and human immunodeficiency virus AB negative. Urine analysis showed red supernatant due to myoglobinuria, leukocytes 75 μl, glucose +++ positive, protein 100 mg/dl, and myoglobin +++ positive. The following tests yielded no evidence of active infection: Tick-borne infectious, Ebstein-Barre virus, cytomegalovirus, enteroviruses, influenza, *Borrelia*, *Chlamydia*, mycoplasma, and Hantan viruses. Neurological examination disclosed marked weakness of all major muscle groups, without focal signs. Results of electromyography were normal.

In spite of massive supply of fluids combined with mannitol and bicarbonate infusions, the patient developed oligoanuria and had to undergo continuous venovenous hemofiltration for 5 days and intermittent hemofiltration for the next 20 days before urine elimination became normal. After pressure-controlled mechanical ventilation for 12 days the patient could be weaned in pressure support mode successfully. Second-generation cephalosporins were administered since *Staphylococcus aureus* was found in pure culture in his sputum.

Metabolic myopathy was suspected to be the cause of the recurrent rhabdomyolysis. After complete recovery including normalization of renal function the patient was transferred to the Department of Neurology, Bonn University, for further investigation. A muscle biopsy specimen showed a muscular carnitine palmityltransferase II deficiency; an ischemic exercise tolerance test showed an increase in serum lactate from 0.92 μmol/l to 2.87 μmol/l, and a caffeine-halothane test was performed but showed no disposition to malignant hyperthermia. However, this test cannot exclude such a disposition with certainty.

The patient was advised to follow a high-carbohydrate, low-fat diet and to avoid cold exposure, prolonged fasting, and aerobic exercise. He was also advised to eat small, carbohydrate-rich meals when under physical strain. An metabolic myopathy should be considered in patients with rhabdomyolysis if drugs such as alcohol and cocaine, statins, infectious myopathies, crush, trauma, extreme exertion, and seizures can be excluded.

References

1. Katzir Z, Hochman B, Biro A, Rubinger DI, Feigel D, Silver J, Friedlaender MM, Popovtzer MM, Smetana S (1996) Carnitine palmitoyltransferase deficiency: an underdiagnosed condition? Am J Nephrol 16: 162–166
2. Bertorini T, Yeh Y, Trevisan C, Stadlan E, Sabesin S, Dimauro S (1980) Carnitine palmityl transferase deficiency: myoglobinuria and respiratory failure. Neurology 30: 263–271
3. DiMauro S, DiMauro P (1973) Muscle carnitine palmityltransferase deficiency and myoglobinuria. Science 182: 929–930
4. Sadeh M, Gutman A (1990) Carnitine palmitoyltransferase deficiency: a common cause of recurrent myoglobinuria. Isr J Med Sci 26: 510–515
5. Di Mauro S, Tsujino S (1994) Nonlysosomal glycogenoses. In: Engel AG, Franzini-Amstrong C (eds) Myology. McGraw-Hill, New York p 1554

K. H. Smolle (✉) · P. Kaufmann · R. Gasser
Department of Internal Medicine, Intensive Care Unit, Karl-Franzens University Graz, Auenbruggerplatz 15, 8036-Graz, Austria
E-mail: karl-heinz.smolle@kfunigraz.ac.at
Phone: +43-316-3852447
Fax: +43-316-3857527