CASE REPORT

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An Extremely Rare Cause of Rhabdomyolysis: Emery Dreifuss Syndrome

Rabdomyolizin Çok Nadir Nedeni: Emery Dreyfuss Sendromu

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Abstract

Intense physical activity, medications and trauma are common causes of rhabdomyolysis. However, etiologic factor of rhabdomyolysis can not be determined in a remarkable proportion of the cases. Here, we present a rare case of muscular dystrophy related rhabdomyolysis.

Keywords: Emery Dreifuss syndrome, muscular dystrophy, rhadbomyolysis

Öz

Ağır fiziksel aktivite, ilaçlar ve travma rabdomyolizin sık görülen nedenleridir. Yine de olguların dikkate değer bir kısmında rabdomyolizin nedeni ortaya konamamaktadır. Bu olgu raporunda, muküler distrofi ilişkili rabdomyolizin nadir bir nedenini sunmaktayız.

Anahtar kelimeler: Emery Dreifuss sendromu, musküler distrofi, rabdomyoliz

Introduction

Rhabdomyolysis is characterized by the lysis of skeletal muscle cells and release of intracelular content, including creatine kinase (CK), glutamic oxalacetic transaminase, lactate dehydrogenase, aldolase, the haeme pigment myoglobin, electrolytes such as potassium and phosphates to extracelular fluid (1). The symptoms at admittance are fatigue, fever, tachycardia, nausea, vomiting, dark urine and myalgia. The manifestations of rhabdomyolysis may range from mild electrolyte imbalances to life-threatening acute kidney injury.

Although the etiology of rabdomyolysis is so diverse, muscle necrosis is common pathogenetic mechanism of traumatic and non-traumatic rhabdomyolysis (2). Injuries, heavy exercise, severe dehydration, medications (antipsychotics, colchicine, antidepresants, anticonvulsants, statins), substance abuse (alcohol, eroin, cocaine), ischemia, and viral infections are common causes of rhabdomyolysis.

On the other hand, congenital muscle disorders, lipid and purine metabolism disorders and glycolytic enyzme deficiencies are rare reasons of the situation (3). Acquired causes of muscular dystrophies are immobility, malnutrition and malignancy-related cachexia that is caused by imbalance of synthesis and degradation of myocytes. Additionally there are genetical causes of dystrophies. Both forms of the muscular dystrophies clasically presented with progressive muscle weakness and degeneration.

Emery dreifuss (EMD) syndrome is a X-linked genetical muscle dystrophy (4). The mutations on *FHL1* ve *LMNA* genes are frequently observed in patients with EMD. Skeletal muscle and myocardium are sites of involvement. Manifestations of the syndrome are arythmias, heart

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conduction disorders, palpitation, bradycardia, syncope, heart failure and sudden death. The age at the onset of the symptoms may change from childhood to early adulthood.

Our aim is to present a case of EMD muscular dystrophy patient that admitted with rhabdomyolysis.

Case Report

A 32-year old male had a history of myalgia for 3 days before admitting to emergency department of our medical center. His past medical history, including excessive exercise or medication, was eventless. He had the same clinical situation in the last few months, after an upper airway tract infection. His physical appearance was pale, and has no additional abnormal finding on cardiovascular and neuromotor examination.

Laboratory examination revealed out elevated creatinine kinase (CK: 7000 U/L) and acute kidney failure. On admission, creatinine levels were 3.9 mg/dL and hyperphosphatemia (P: 5.4 mg/dL). Furthermore, liver function tests including alanine transaminase (240 IU/L) and aspartate transaminase (300 IU/L) were higher.

The patient underwent to radiographic studies to exclude renal parenchymal and postrenal abnormalities. Ultrasonographic studies showed no signs of abnormality, except renal parenchymal disease. Fluid resuscitation was initiated and patient was recommended to stay at rest. He required no session of hemodialysis, and acute renal injury was resolved with fluid resuscitation. On the 3rd day of hospitalization, his symptoms were recovered, and laboratory abnormalities turned to normal range.

Because there was no apparent cause of rhabdomyolysis, rare cause of the disorder, such genetic mutations were studied. After consultation with the department of medical genetics, a number of mutations which are associated with rhabdomyolysis were analyzed. Genetical analysis indicated muscular dystrophy of EMD syndrome. He was discharged with the recommendation of regular nephrology outpatient service visits. On his follow-up at the 3rd month of discharge from the hospital, laboratory parameters of the patient were normal, and he had no complaint.

Discussion

The early complications of rhabdomyolysis are compartmant syndrome, hypovolemia, electrolyte disorders, acidosis, hepatic dysfunctions (5). In the advanced stages, patients may experience acute kidney failure or disseminated intravascular coagulation. Although patients may have no apparent symptom, a number of patients may experience life-threatening situation associated with myoglobinuria, extremely elevated CK levels and acute kidney failure,

Classical triad of EMD is muscle contractures, especially elbows and Achilles tendons, muscle weakness and cardiomyopathy (6). In contrast to other muscular distrophies, contractures are early findings. Muscle weakness is commonly observed in the proximal of upper extremities and in the distal of lower extremities after the development of contractures. Patients with EMD usually have moderately increased CK levels which is considered as an evidence of chronic rhabdomyolysis (7). Patients with EMD are under increased risk of cardiomyopathy and first degree heart block.

MM subtype of CK which starts to rise within the 12 hours of muscle injury and generally peaks at 72 hours, is the most sensitive indicator of rhabdomyolysis (5). The CK concentration is proportional to muscle damage, and CK higher than 5 times the upper limit of normal indicates mild rhabdomyolysis.

Another marker of rhabdomyolysis, myoglobin, binds to globulin, and rapidly cleared by kidneys. However, subsequent to muscle damage, myoglobulin levels may exceed protein binding and renal clearance capacity. Serum levels of myoglobin usually resolves in the first 24 hour of injury. Early resolution and false negativity in the presence of high urine nitrite concentration or decreased GFR are the drawbacks of myoglobinuria in the diagnosis of rhabdomyolysis (5).

Muscle biopsy is not a routine diagnostic procedure. However, in patients with suspicion of muscular dystrophy, such as repetitive rhabdomyolysis or co-existing muscle contracture or weakness, within the 3rd month ofresolution of rhabdomyolysis may show fragmentation and necrosis of muscle fibers without the presence of inflmmatory cells strongly suggests muscular dystrophy (8).

Therapeutic options are mainly palliative. First and foremost, preventive strategies such as avoidance of nephrotoxic agents are the mainstay of the therapy. Similarly, volume replacement is also essential to reverse the situation. Effective fluid resucitation that resolves volume loss and stimulates urine output should be initiated. Osmotic diuretics like mannitol acts increasing renal blood flow and preventing the obstruction of myoglobin casts (2). Furthermore, urine alkalinization is

also beneficial to excrete intraceluler contents. Dialysis is required for patients with progressive decline in kidney functions, resistant hyperkalemia, hypervolemia, acidosis or ureamic encephalopathy. On contrary, plasmapheresis has no benefit to resolve the metabolic complications (9).

In conclusion, EMD should be considered in patients presented with rhabdomyolysis that has no apperant risk factor and lasting muscle and cardiac symptoms that can be confirmed by muscle biopsy. After the diagnosis of EMD, routine cardiac screening has crucial importance to establish the development of cardiomyopathy and arrythmias in the long-term.

Ethics

Informed Consent: Written consent was received from the patient.

Authorship Contributions

Concept: H.L., Y.Ç., N.G., E.I.Ş., Design: H.L., Y.Ç., N.G., E.I.Ş., Data Collection or Processing: H.L., Y.Çe., A.M., N.G., Analysis or Interpretation: Y.Ç, E.I.Ş., A.M., A.Ç., Drafting Manuscript: H.L., N.G., A.Ç., A.M., Critical Revision of Manuscript: Y.Ç., E.I.Ş., Y.Çe., Technical or Material Support: Y.Ç., Y.Çe., Supervision: E.I.Ş., H.L., N.G., A.Ç., Final Approval and Accountability: N.G., H.L., A.M., Writing: N.G., H.L., A.M.

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