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FAT EMBOLI SYNDROME IN A CHILD WITH DUCHENNE MUSCULAR DYSTROPHY AFTER MINOR TRAUMA

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Abstract—Background: Fat embolism syndrome is the result of systemic manifestations of fat emboli in the microcirculation. Duchenne muscular dystrophy is a condition that increases the risk of fracture resulting in fat emboli. **Case Report:** We describe a patient with Duchenne muscular dystrophy who exhibited cardiopulmonary, neurologic, and ophthalmologic sequelae consistent with fat emboli syndrome after minor trauma. **Why Should an Emergency Physician Be Aware of This?:** Fat embolism syndrome is a rare but important consideration with significant morbidity and risk of mortality in patients with Duchenne muscular dystrophy after even minor trauma. Early recognition and aggressive resuscitation are crucial to positive clinical outcomes. Published by Elsevier Inc.

Keywords—Duchenne muscular dystrophy; fat embolism syndrome; pediatrics; fat microglobulinemia; tibial plateau fracture

INTRODUCTION

Duchenne muscular dystrophy (DMD) is an X-linked recessive disease that affects one in 3600–6000 live male births that results in replacement of skeletal muscle

tissue with fat due to deletions in the dystrophin gene. This causes a progressive muscle degeneration, weakness, and obesity, which predisposes affected individuals to falls and fractures (1). Loss of independent ambulation begins at age 9–12 years, and patients are usually nonambulatory by age 13–16 years, putting them at higher risk of falls and subsequent fractures (2). Cardiac, pulmonary, and orthopedic complications are most frequent, and without interventions, death from cardiac or pulmonary causes occur by age 20 years (3). Fat embolism syndrome (FES) is a clinical entity that arises from systemic manifestations of fat emboli in the microcirculation most commonly seen after orthopedic trauma (4). Although an alternative biochemical theory exists involving the release of free fatty acids from bone marrow in response to trauma, resulting in hypoxemia and inflammation, the end result is the same—increased capillary permeability and damage to surrounding tissue from inflammatory mediators (4). We report a case of FES in a 15-year-old boy with DMD who sustained a tibial plateau fracture after minor trauma.

CASE REPORT

A 15-year-old boy with DMD presented to the Emergency Department for evaluation of leg pain after a witnessed fall out of his wheelchair onto his left leg. There was no head trauma or loss of consciousness. He had a similar event 1 year previous that resulted

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in a femur fracture requiring internal fixation. In addition to DMD, his past medical history was significant for restrictive lung disease for which he was treated with chronic inhaled steroids. Surgical history was significant only for the previously noted internal fixation of the left femur. He had no known allergies, and his medications included carvedilol, lisinopril, albuterol, and vitamin D.

Initial examination revealed an obese 15-year-old boy in no acute distress. Oral temperature was 36.8°C (98.2°F), heart rate 136 beats/min, respiratory rate 20 breaths/min, oxygen saturation 98% on room air, and blood pressure 137/92 mm Hg. His lungs were bilaterally clear to auscultation without wheezes or rales. He was tachycardic but had a regular rhythm. Abdominal examination was unremarkable. Extremity examination revealed decreased active range of motion and muscle tone consistent with muscular dystrophy. There was mild swelling about the left knee, with minimal ecchymosis. He complained of pain with left knee movement but had full range of motion, as well as transient blurry vision. On initial neurological examination, he was alert and oriented to person and place, and his verbal interactions were at baseline per his parents. Cranial nerves II–XII were intact.

Left knee radiographs showed stable postsurgical changes of the left femur without evidence of acute fracture and a new irregularity of the medial physis of the left tibia (Figure 1). Computed tomography (CT) of the left knee showed a medial tibial plateau fracture with extension into the posterior tibial plateau. Orthopedics was consulted and recommended splinting with outpatient follow-up. During his stay, the child became increasingly tachycardic in the 140–150 beats/min range, above his baseline heart rate of 90–120 beats/min. He was also noted to have a temperature of 38°C (100.4°C) and a new oxygen requirement for saturations in the mid 80s. He was now speaking in only one-word sentences and appeared increasingly lethargic. For these reasons, a chest radiograph (CXR), urinalysis, and blood cultures were ordered in search of an infectious etiology, presuming these changes could be the result of sepsis. A head CT scan was ordered due to altered mental status, and a CT scan of the chest for pulmonary embolism was obtained for his increasing respiratory distress. Urinalysis, CXR, and CT of the head were normal. The CT of the chest did not show a pulmonary embolism, but did reveal diffuse ground glass appearance in both lungs (Figure 2).

The pediatric intensive care team was consulted and the child was admitted for further work-up of his tachycardia, altered mental status, and oxygen requirement. Bilevel positive airway pressure (BiPAP) was initiated due to continued respiratory distress. Azithromycin and



Figure 1. Left anterior-posterior knee x-ray study demonstrating stable postsurgical changes of the left femur without evidence of acute fracture and a new irregularity of the medial physis of the left tibia.

ceftriaxone were started as empiric antibiotic coverage for presumed pneumonia. A magnetic resonance imaging study of the brain was obtained, which revealed

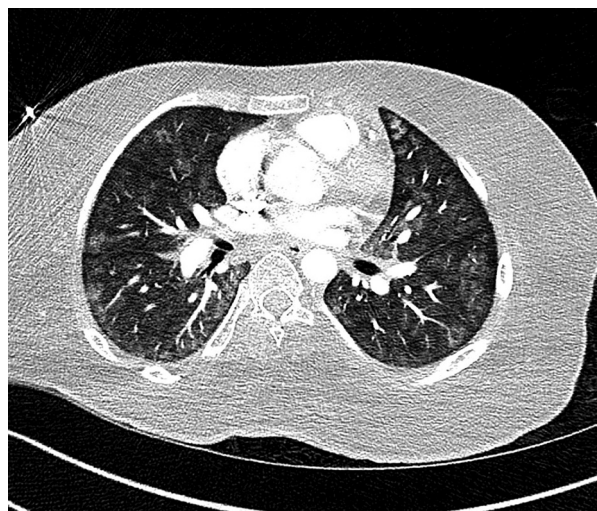


Figure 2. Computed tomography scan of the chest demonstrating ground glass opacities.

multiple punctate areas of hyperintensity in the deep white matter, thalami, corpus callosum, cerebellum, and pons, consistent with microinfarcts (Figure 3). During the patient's stay, the Infectious Disease service was consulted and noted a petechial rash on the child's torso, not previously recognized. Ophthalmology was consulted to evaluate the patient's report of blurry vision. On ophthalmologic examination, his near vision was 20/100 in the right eye and 20/60 in the left eye, with normal intraocular pressure, no afferent pupillary defect, and an unremarkable anterior examination. Dilatated fundus examination revealed scattered nerve fiber layer infarcts with a few areas of intraretinal hemorrhage bilaterally (Figure 4). With the addition of this finding, the diagnosis of FES was confirmed. His neurologic status continued to improve throughout his hospital course; he was slowly weaned from the BiPAP; pressure support and antibiotics were discontinued in the setting of this noninfectious process. Orthopedics placed a cast for his tibial plateau fracture. At time of discharge he still required nighttime use of the BiPAP, but was otherwise back to his baseline status.

DISCUSSION

The first case of FES was described in 1861 after an autopsy of a railway worker that suffered a thoracoabdominal crush injury (5). FES is a rare but well-known entity, thought to be a result of fat droplets released from bone marrow, most commonly after long bone fractures and or-

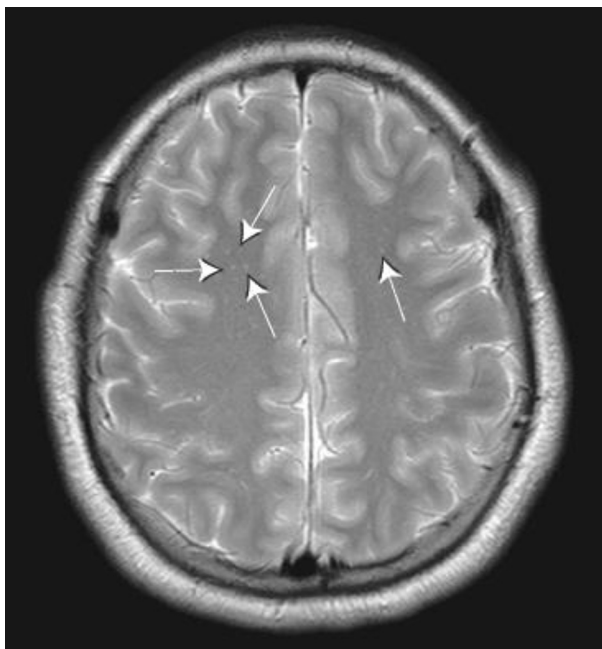


Figure 3. Brain magnetic resonance imaging study demonstrating hyperdensities consistent with microinfarcts.

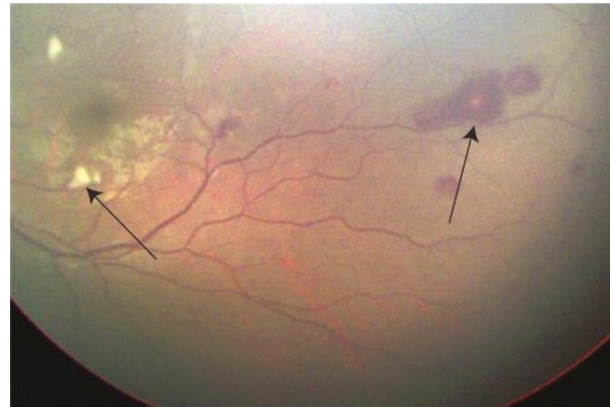


Figure 4. Photo of the left eye dilatated fundoscopic examination demonstrating scattered nerve fiber layer infarcts with few areas of intraretinal hemorrhage bilaterally.

thopedic procedures (6). The majority (up to 90%) of long bone fractures result in fat embolization (7). However, retrospective reviews report a < 1% incidence of FES, and 11–29% in prospective studies (8–13). Many cases go unnoticed and therefore, the true incidence of FES is unknown (14). The small fat droplets are released into venous circulation, pass through the normal cardiopulmonary system including pulmonary capillaries, enter the systemic circulation, and eventually lodge in arterioles, causing microinfarcts.

Clinical presentation may include respiratory symptoms, cerebral signs, a petechial rash, retinal changes, jaundice, renal dysfunction, thrombocytopenia, anemia, elevated erythrocyte sedimentation rate, and fat macroglobulinemia (6). Respiratory symptoms occur in up to 75% of patients with FES and can present as mild hypoxia requiring supplemental oxygen, or acute respiratory distress syndrome requiring prolonged mechanical ventilation (4). Central nervous system signs are usually nonspecific and range from a simple headache to rigidity, disorientation, convulsion, confusion, stupor, and coma (15). Other presenting signs can be nonspecific and include tachycardia, tachypnea, fever, petechial rash, and vision changes (4). Diagnosis can often be difficult, however, there are several criteria that are used to make the diagnosis (12,16). Gurd and Wilson criteria require one major feature, four minor features, and fat microglobulinemia (16). Major features include respiratory insufficiency, cerebral involvement, and petechial rash. Minor features are pyrexia, tachycardia, retinal changes, jaundice, renal changes (anuria or oliguria), thrombocytopenia (drop of > 50% of admission), and high erythrocyte sedimentation rate.

FES is under-recognized in adults and even more so in children, in whom it likely occurs subclinically (6). Because children with DMD tend to have a higher

incidence of falls related to their disease-associated obesity and immobility, they are at risk for falling and fracturing, and therefore, developing FES after even seemingly minor trauma. There are three published case series of patients with DMD who developed FES, with a total of 14 patients (6,17,18). Seven of these cases were fatal, thus demonstrating need for a high level of suspicion, early diagnosis, and aggressive supportive care. Two cases reported visual disturbances but did not document an ophthalmologic examination or visual outcome. There is one published case of a previously healthy 16-year-old girl who sustained a tibial fracture and developed FES with decreased visual acuity and documented nerve fiber layer infarcts in both eyes whose vision recovered completely (19). This case not only highlights features of FES, but also points out the importance of considering this diagnosis in females, as DMD and FES are not limited to the male gender. Our high-risk patient was successfully recognized, diagnosed, and managed through the cooperation of multiple specialty teams, which ultimately led to a positive outcome.

WHY SHOULD AN EMERGENCY PHYSICIAN BE AWARE OF THIS?

Fat embolism syndrome is a rare but important consideration in patients with DMD after even minor trauma. This multisystem condition carries a significant risk of morbidity and mortality without early recognition and rapid management.

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