H97 A Sudden Pediatric Death: A Case of Asymptomatic Muscular Dystrophy

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Learning Overview: After attending this presentation, attendees will better understand: (1) the signs and symptoms of Duchenne Muscular Dystrophy (DMD), (2) its fatal complications, and (3) possible histopathological changes associated with DMD.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by raising awareness of sudden deaths in young children who have not yet lost ambulation due to respiratory complications of DMD.

DMD is an X-linked disorder that affects 1 in 3,300 males born in the United States.[1] A mutation of the dystrophin gene on Xp21 causes membrane instability and loss of calcium homeostasis in skeletal and cardiac muscle fibers. This results in muscle necrosis and subsequent regeneration, with fibrosis and fatty infiltrate replacing muscle when regenerative capacity is reached.[1] Bilateral progressive muscle weakness begins around the age of 4 years; the proximal muscles of the lower limbs are most severely affected. Common signs are "Gowers maneuver," where the child uses their arms to push to a standing position, tip-toe walking, and calf "pseudohypertrophy" from fatty infiltration. Most patients will be wheelchair-bound by age 12, and life expectancy is about 26 years.[2] Fatal complications of DMD include cardiomyopathy and respiratory failure.

Described here is the case of a 3-year-old Arab male with no significant past medical history who was in his usual state of health. He was at home with his mother when he was reported to have complained of stomach pain while eating, turned blue, and became unresponsive. Emergency medical services responded and initiated cardiopulmonary resuscitation. He was intubated at the hospital but was unable to be resuscitated. Of note, his mother had reported that the child had red urine on the day of his death.

At autopsy, external examination showed a normally developed, normally nourished boy with no evidence of traumatic injury. There was no significant calf hypertrophy. Internally, there was bilateral pulmonary edema (combined weight: 293.1 grams, normal: 166 grams). Postmortem toxicology was negative. A postmortem Creatine Kinase-MB isoenzyme (CK-MB) was unable to be obtained due to hemolysis ,and no urine was present at the time of autopsy. Microscopic examination including Movat staining of the heart showed no evidence of fibrosis or cardiomyopathy. The lungs showed scattered foci of atelectasis and there was mild cerebral edema with gliotic changes.

Histopathological examination of the diaphragm, rectus femoris muscle, and psoas muscle revealed myopathic changes consistent with DMD. This included groups of regenerating muscle fibers, small degenerating fibers, and areas of fibrosis and fatty infiltration. A recent diagnostic workup showed that the decedent had tested positive for dystrophin gene mutation. A maternal uncle had died of unknown cause at age 14, and the mother was determined to be a carrier. A creatinine kinase prior to death was 26,040U/L. The cause of death was certified as complications of muscular dystrophy and the manner of death was natural. Myopathy of the diaphragm can cause diaphragmatic paralysis, leading to respiratory failure and sudden death.

A review of the literature reveals that inactivity following loss of ambulation leads to atrophy of intercostal muscles, scoliosis, and chest wall deformity, which contribute to a decline in respiratory function and earlier demise.[3] Unexpected deaths in children with DMD have occurred due to aspiration pneumonia, acute respiratory distress following exercise, cardio-respiratory arrest following minor trauma with no fractures, and multi-organ failure.[4] Less information is available about the mechanism behind these sudden deaths in younger children/teens with DMD and more research is needed to understand the disease progression in different cases.

Reference(s):

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