H183 Sudden Death Caused by Bilateral Diaphragmatic Eventration in Myotonic Dystrophy Type 1

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Learning Overview: After attending this presentation, attendees will understand the pathophysiology and genetic basis of myotonic dystrophy, a multisystem inherited disorder that represents the most common muscular dystrophy observed in adults.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by highlighting a case of sudden death related to respiratory compromise caused by bilateral diaphragmatic eventration and associated restrictive lung disease in a woman with a known clinical diagnosis of Myotonic Dystrophy Type 1 (MD1).

A 48-year-old woman with a known clinical diagnosis of MD1 was found unresponsive in her home. She was pronounced dead at the scene. A clinical diagnosis of MD1 had been rendered at the age of 13 years, although no genetic/molecular testing had ever been performed. At 39 years of age, she was diagnosed with chronic obstructive pulmonary disease and restrictive lung disease.

A medicolegal autopsy was performed, which disclosed a mildly obese (BMI=29.4) adult female. On internal examination, the lungs appeared somewhat hypoplastic, with a combined weight of 470 grams. The diaphragm was intact, but was markedly elevated bilaterally, with associated decrease in pleural cavity volume. Much of the diaphragm was parchment-thin and nearly translucent. Additional gross findings at autopsy included changes consistent with hypertensive and atherosclerotic cardiovascular disease. Microscopic examination of the diaphragm showed deformation and degeneration of skeletal muscle, with fat and fibrous tissue replacement and residual muscle fibers containing numerous central nuclei and rare cytoplasmic vacuolization. The cause of death was ruled respiratory compromise due to restrictive lung disease due to bilateral diaphragmatic eventration due to MD1, with underlying hypertensive and atherosclerotic cardiovascular disease. The manner of death was natural.

Myotonic dystrophy is the most common muscular dystrophy observed in adults.[1,2] The condition is a multisystem disorder, characterized by muscle weakness, myotonia, cardiac conduction abnormalities, and various other manifestations, with cardiac and respiratory issues being responsible for most deaths.[3-5] MD1 is the most common form of the disorder, resulting from an autosomal dominant inheritance of a triplet-repeat disorder.[3,4] The presented case is a bit unusual in that she was diagnosed with the juvenile form of MD1 but experienced only limited clinical manifestations for most of her adult life. Respiratory involvement in the disorder is a complex phenomenon, related to variable involvement of chest wall muscles and the diaphragm, with associated apnea and chronic hypoxemia; restrictive lung disease in MD1 patients is a recognized indicator of cardiac events and risk of death.[6,7] A recently published case of MD1-related death emphasizes the fact that fat-replacement of muscle can contribute to death.[8] In the present case, the marked diaphragmatic eventration, with associated pulmonary compromise, represents another unusual manifestation of MD1. Clinicians and pathologists should remain aware of this rare presentation of MD1.

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Myotonic Dystrophy, Diaphragmatic Eventration, Death