H80 Drowning or Something Else? The Uncommon Birt-Hogg-Dubé Syndrome

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Learning Overview: After attending this presentation, attendees will have learned about an unusual syndrome that must be taken into consideration in cases of diving-related deaths, in particular in non-professional divers, and the diagnostic tools necessary for diagnosis.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by relating the importance of focusing on the need to perform radiological examinations in the event of suspected drowning deaths and, in particular, to focus on a rare, but probably underdiagnosed, pathology.

The case report of a 31-year-old man found dead in the sea of Trieste, Italy, is presented. The suspected cause of death was a diving-related death due to the finding on the body of a diving mask and underwater fins. Before autopsy, due to advanced putrefactive changes, a postmortem Computed Tomography (CT) scan was performed. The results included: absence of air-fluid levels in the sinuses, the sphenoid area, and the mastoid cells; presence of right lung collapse with discrete compression and deviation from the midline of the mediastinum; presence of numerous cystic formations in both lungs. During the autopsy, leaking of multiple air bubbles from the right hemithorax was detected using the conventional pneumothorax test. The right lung appeared significantly reduced in volume with displacement of the mediastinum to the left. The left lung appeared hypo-expanded; there was absence of fluid in the oropharyngeal cavities, in the trachea, and in the large bronchi.

The conclusion of the coroner investigation was the exclusion of a death due to mechanical asphyxiation from drowning. Given the radiological and autopsy finding, the cause of death was a spontaneous pneumothorax in a bullous pulmonary disease during a free dive, likely due to the higher pressure underwater. Considering the radiological characteristics of both lungs, a Birt-Hogg-Dubé Syndrome (BHDS) was hypothesized. This syndrome is a rare autosomal dominant monogenic inherited disorder caused by mutation of the folliculin gene (FLCN) site on the chromosome 17p11.2. It is associated with skin manifestations such as fibrofolliculomas that appear in the third or fourth decade mainly in the upper part of the body, an increased risk of renal cell carcinomas, and the presence of numerous lung cysts, in particular in the basal zones of both lungs. These are associated with a high risk of numerous events of spontaneous pneumothorax starting from the third decade. To prove this hypothesis and more importantly offer a clinical evaluation and genetic counselling to the relatives, it would be necessary to carry out genetic investigations; however, given the high number of mutations of the FLCN gene and that the deceased was a German tourist, no further investigation was carried out.

Autopsy, Postmortem CT, Birt-Hogg-Dubé Syndrome