H150 Sudden Death Related to Aortic Pathology — A Comprehensive Diagnostic Approach

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After attending this presentation, attendees will better understandSudden Aortic Death (SAD) secondary to aortic rupture, dissection, or perforation. The presentation specifically addresses the diagnosis of SAD, including the importance of postmortem imaging and genetic testing.

This presentation will impact the forensic science community by raising awareness of the underlying pathologies that are related to SAD. Also, in a forensic setting, a comprehensive diagnostic approach is needed to identify underlying (hereditary) diseases, which is of paramount importance for relatives of the deceased.

Aortic ruptures or dissections are common causes of sudden death and are, therefore, regularly observed in forensic pathology practice. These cases could be referred to as SAD, in line with the concept of Sudden Cardiac Death (SCD), which is also a major cause of sudden death at all ages, but with a wide range of different pathophysiological backgrounds.

Recent discoveries demonstrate how SAD is directly related to underlying acquired or genetic diseases, such as Marfan-syndrome and many other newly described syndromes. Still, since the immediate cause of death in SAD (e.g., internal hemorrhage) is often easily recognized during autopsy or postmortem imaging, and the underlying pathophysiology is of less importance for purely medicolegal reasons, SAD is not always investigated in-depth in a forensic setting. This may lead to missed diagnoses, especially for hereditary diseases, which in turn leaves family members of the deceased unnecessarily at risk. The recent developments in postmortem radiological imaging for aortic lesions are promising, and it was suggested that, in some instances, a full autopsy is not necessary.

This study compared two series of SAD, one from a clinical setting and one from a forensic setting. First, this study identified differences in the patient characteristics, the diagnoses, and the diagnostic methods. Second, this study used this information to propose a diagnostic approach for forensic pathologists confronted with SAD.

A total of 59 cases of SAD were included, 46 males and 13 females. Thirty-seven were autopsied in a forensic setting and 22 in a clinical setting. The demography of the two groups and the used diagnostic approach differed considerably. For example, postmortem CT-imaging was systematically performed in the forensic group, often complemented with CT-angiography (56.7%), but the clinical setting never used postmortem imaging. Histological sampling was conducted more extensively in a clinical setting.

Comparison of the diagnoses revealed that in the majority of natural deaths, atherosclerosis and/or Cystic Medial Degeneration (CMD) was the principal pathological substrate for the acute event. Both settings had several cases in which an underlying hereditary disease was suspected. This was confirmed in one case in each setting by genetic testing, which found mutations related to Loeys-Dietz syndrome.

These results underscore the importance of an accurate reconstruction of the mechanism of death in a forensic setting. Therefore, for each case of SAD, a full autopsy and thorough histological examination, generally complemented with postmortem imaging and molecular pathology should be recommended.

Aorta, Sudden Death, Postmortem Genetic Testing