H84 An Intrapericardial Rupture of an Aortic Aneurysm in the Anatomic Aortic Arch Variant: A Multidisciplinary Approach

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Learning Overview: The goal of this presentation is to present a multidisciplinary approach to a case of an unexpected sudden death due to rupture of an aortic arch aneurysm associated with an atypical aortic arch branching variant incidentally detected during autopsy. To clarify the cause of death, to perform the postmortem genetic analysis and a systematic evaluation of decedent's family members, different professional experts, such as forensic pathologists, clinical geneticists, and general practitioners, were involved.

Impact on the Forensic Science Community: Little is known in the literature about non-syndromic familial Thoracic Aortic Aneurysms (TAAs); therefore, this presentation will impact the forensic science community by providing a methodical approach in those cases of sudden/unexpected death occurring in asymptomatic individuals with a postmortem diagnosis of Thoracic Aortic Disease (TAD).

According to the literature, aortic arch variants (i.e., variations in the aortic arch vessel branching pattern) are currently viewed as a marker or risk factor for TAD. Furthermore, about 20% of patients with TAA show a family history of similar disease, indicating an underlying significant genetic component. TAAs are usually asymptomatic until acute dissection or aortic rupture occurs; therefore, they often remain undiagnosed until fatal consequences.

A 53-year-old man, who worked as a carpenter, with a negative past medical history, suddenly collapsed at his workplace due to a cardiac arrest. Cardiopulmonary resuscitation maneuvers were immediately performed but were unsuccessful. He was declared dead by medical personnel 25 minutes after the beginning of the cardiopulmonary resuscitation. Before autopsy, the victim's past medical history, circumstances surrounding the sudden death, and the family's medical history were evaluated in collaboration with the general practitioner. The autopsy, performed two days after death, showed no sign of injury, except for traces of emergency medical care procedures. The most striking macroscopic feature was represented by cardiac tamponade, due to the rupture of a saccular aortic arch aneurysm (7.5cm x 8cm), at the site of a complex atheromatous plaque. It was 3cm in thickness, made of several layers of degenerative material. The autopsy also revealed the presence of an aortic arch variant with a two-vessel branching pattern: the first branch was a common origin of the brachiocephalic trunk and left common carotid artery, and the second one was the left subclavian artery. A histological examination was performed, including microscopic evaluation of the site aneurysm rupture and atheromatous plaque samples.

Since the current aortic arch variants associated with the presence of aortic arch aneurysm could be related to the presence of gene mutations that show an autosomal dominant-codominant inheritance with incomplete penetrance, a genetic evaluation of a cluster of most frequently involved mutations was performed, including FBN1, TGFBR1, TGFBR2, COL3A1, ACTA2, MYH11.

These findings suggest that a methodological approach, which involves collective efforts from general practitioners, forensic pathologists, and clinical geneticists, is essential in cases like this, not only to define the cause of death but also to perform postmortem genetic analysis in order to understand the pathophysiology involved in the genetics of aortic aneurysms and a systematic genetic counselling that could reveal disease-causing mutation and potentially prevent further deaths.

Sudden Unexpected Death, Aortic Arch Variants, Thoracic Aortic Aneurysm