G10 A Clinicopathological Correlation of Non- Compaction Cardiomyopathy

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After attending this presentation, attendees will learn about the entity of Non-compaction Cardiomyopathy (NCCM) and how to determine or confirm the diagnosis through postmortem examination.

This presentation will impact the forensic science community by discussing that even though non-compaction cardiomyopathy is uncommon, forensic pathologists should be familiar with this abnormality, not only because it can lead to heart failure, arrhythmias, and embolic events, but also because it demonstrates an increased frequency of familial occurrence.[1]

The deceased was a 46-year-old woman with a history significant for cardiomyopathy, cardiac arrhythmias, congestive heart failure, and alcoholism. An echocardiogram performed two years prior revealed heavy trabeculation consistent with a myocardial NCCM. She suffered from recurrent atrial tachyarrhythmias, including atrial fibrillation with rapid ventricular response, that was refractory to multiple ablation procedures.

While attending a regularly scheduled appointment with her cardiologist, the patient developed tachypnea at rest and atrial fibrillation. She was immediately transferred to the cardiac care unit where she developed increasing dyspnea at rest, productive cough, and lower extremity edema. Chest auscultation revealed atrial fibrillation and bibasilar crackles. Electrocardiogram findings were negative for myocardial infarction. Two days later, the patient spontaneously converted to a normal cardiac rhythm, and she was put on sotalol for rhythm control. A few hours later, the patient experienced a torsades de pointes arrhythmia, a potential complication of sotalol therapy. A successful code was performed; however, the patient aspirated at the time of intubation. Following the code, she remained persistently hypotensive with hypoxemia refractory to medical treatment. Her chest roentgenogram showed extensive consolidation of both lungs. Clinically, the patient remained unresponsive with fixed and dilated pupils. Given the poor prognosis, care was withdrawn and the patient expired.

Autopsy revealed a globally enlarged and dilated heart weighing 424g, which was significantly greater than the sex and body mass adjusted mean of 251g (expected range 171g – 368g), a finding consistent with her history of congestive heart failure. Cross sections of the heart revealed an extensive amount of non-compacted myocardium in the left ventricle, which was most prominent in the distal half and on the lateral wall of the left ventricle. The ratio of non-compacted-to-compacted myocardium was 4.3:1. These findings supported the antemortem diagnosis of NCCM. Microscopically, increased amounts of non-compacted myocardium were seen with variable fibrosis within the non-compacted myocardium. No acute changes were seen. Additionally, the foramen ovale was patent (1.2cm), and multiple old intraparenchymal brain infarcts were identified. The etiology of these infarcts could not be determined with certainty; it is possible that they may have been embolic in origin and associated with the NCCM and arrhythmias and/or associated with the patient's patent foramen ovale.

NCCM is a rare myocardial abnormality with an unclear prevalence. Several studies have determined a prevalence of 0.05% – 0.25%.[1] The age at presentation is highly variable. The cardiac manifestations include heart failure, arrhythmias (atrial and ventricular), and embolic events, all of which were seen in the deceased.[1] In most cases, it is a congenital abnormality due to the arrest of normal embryogenesis of the endocardium and myocardium and is often associated with other congenital cardiac malformations.[2] Familial and sporadic forms of NCCM have been described with both X-linked and autosomal patterns of inheritance.[3]

The mechanism of death in the case presented was aspiration pneumonia secondary to complications of intubation at the time of cardiac arrest. The cardiac arrest was initiated by a torsades de pointes arrhythmia which was most likely induced by the sotalol therapy. NCCM should, therefore, be deemed the underlying cause of death since the intended purpose of the sotalol therapy was to manage the atrial arrhythmia, which is a known complication of NCCM.

In the practice of forensic pathology, NCCM should be considered and recognized as a potential cause of heart failure, fatal arrhythmia, and embolic events including cerebrovascular accidents. Medical examiners must be especially aware of this anomaly since it has been recommended that surviving first-degree relatives be screened by echocardiography given the increased familial association.[2,4]

References:

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2. Weiford *et al*. "Noncompaction of the Ventricular Myocardium." *Circulation*. 2004; 109:2965-2971.
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Noncompaction, Cardiomyopathy, Left Ventricle

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