H3 Congenital Valsalva Sinus Aneurysm Causing Sudden Unexpected Death in a 20-Year-Old Woman

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After attending this presentation, attendees will understand the importance of considering an acquired or congenital cardiovascular disorder as an etiology in sudden and unexpected death.

This presentation will impact the forensic science community by increasing awareness of the possibility of congenital cardiac disorders presenting with a range of clinical symptoms as the cause of sudden and unexpected death.

This is a rare case in which a young woman with recently diagnosed Acute Meloid Leukemia (AML) presents with a pericardial effusion and dies suddenly as a result of a cardiac tamponade secondary to a rupture of a congenital right Valsalva sinus aneurysm.

A 20-year-old woman with high-grade Myelodysplastic Syndrome (MDS) was admitted to the hospital due to increasing respiratory distress. Her MDS converted to AML. While being treated with chemotherapy, imaging studies revealed a significant pericardial effusion. Magnetic residue imaging revealed a massive chronic lobulated pericardial effusion. Cardiac magnetic resonance imaging was performed and revealed a large sinus of Valsalva aneurysm involving the ostium of the right coronary artery. The pericardial effusion occupied one-third of the chest volume and was composed of septations extending from the epicardium to the pericardial wall.

At autopsy, opening of the thoracic cavity revealed a distended pericardial sac containing a 3.0cm thick, yellow-red, encircling layer of partially clotted blood. Both the pericardial sac and epicardial surface of the heart were firmly adhered to the partial clot. The weight of the heart, including the hemopericardium was 1,340gm. Examination of the heart and great vessels revealed a 3.8x2.7x2.5cm partially clotted blood-filled cavity extending 1.0cm below the right coronary artery ostium (the right Valsalva sinus), through the interventricular septum, and into the wall of the left ventricle. This cavity perforated into the right ventricle; however, the thick adherent hemopericardium made it difficult to find the exact perforation defect. This cavity was associated with a tortuous aneurysm which bulged into the right ventricle below the tricuspid valve and into the left ventricle. The right aortic cusp was torn midline from the base. Microscopy of the lesion revealed dense collagen without the aortic elastic layers, as well as erythrocytes, acute inflammatory cells, hemosiderin laden macrophages, and layers of fibrin deposits, consistent with an organizing thrombus and recent hemorrhage.

The majority of natural causes of sudden and unexpected death are due to a cardiovascular disorder. These disorders include arrhythmias, ischemic heart disease, aortic stenosis, pulmonary embolism, and a rupture of a myocardial infarct.[1,2] Congenital cardiovascular malformations are rare and can cause a rupture that leads to a hemopericardium. One rare lesion is an aneurysm of the sinus of Valsalva.

Aneurysms of the Valsalva sinus can be acquired through trauma, degenerative disease, or endocarditis of the aortic valve.[1-3] Congenital aneurysms of the Valsalva sinus are rare, although more common than their acquired counterpart and may be associated with a ventricular septal defect.[1,3,4] There is a 3:1 male predominance and the individual is usually asymptomatic until adulthood.[1,4] This aneurysm results from separation of the aortic media and the aortic valve cusp. This separation is due to the variable thickness of the collagen creating discontinuity, which can dilate, expanding into the cardiac septum or chamber.[4] More than two-thirds of these aneurysms occur in the right aortic sinus, while 25% and 8% involve the posterior and left aortic sinuses, respectively.[1,4]

Approximately 75% of individuals with a congenital aortic sinus aneurysm experience complications, with rupture of the aneurysm being the major complication.[1] The mean age the aneurysm ruptures is 31 years.[1,2,4] Aortic sinus ruptures can involve more than one coronary artery. Ruptures may be large and acute or small and progressive, adding to the diversity of the clinical presentation.[3-5] Surgical repair is often curative, as patients usually die within a year if they don't receive treatment.[4]

The range of clinical symptoms makes aneurysms of the Valsalva sinus difficult to detect. Because of this aneurysm's rarity, it often doesn't make the list of differential diagnoses. An individual may not be aware of having this condition, increasing the chances of being engaged in everyday activities when the aneurysm ruptures. The elusive nature of the condition will most likely result in a sudden and unexpected death requiring the examination of a forensic pathologist.

References:

1. Fealey ME, Edwards W, McMaster K, Carter J. Congenital Aortic Sinus Aneurysm Causing Sudden Unexpected Death in a 56-Year-Old Woman. Am J Forensic Med Path 2009; 30: 195-197.
2. Deshmukh A, Subbiah S, Malhitra S, Deshmukh P, Pasupuleti S, and Modiuddin S. Spontaneous Hemopericardium Leading to cardiac Tamponade in a Patient with Essential Thrombocytopenia. Cardiology Research and Practice 2011: 1-3.
3. Leptidis J, Alozios A, and Gourgiotis S. Fatal cardiac tamponade as the first manifestation of acute myeloid leukemia. Am J Emerg Med 2004: 45-46
4. Goldberg N, and Krasnow N. Sinus of Valsalva Aneurysms. Clin Cardiol 1990; 13: 831-836.
5. Miranda D, Peter A, Osorio J, and Ferreira A. Ruptured Aneurysm of the Noncornary Sinus of Valsalva. *Texas Heart Institute Journal* 2005; 32: 586-588.

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