



G74 Isolated Noncompaction of the Left Ventricle: A Rare Cause of Sudden Death

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This presentation will review the literature and the autopsy findings of Isolated Noncompaction of the Left Ventricle, a rare congenital cardiomyopathy.

A 44-year-old black female with no known medical history was witnessed to collapse suddenly at work. Upon arrival, paramedics found an initial rhythm of ventricular fibrillation; their initial efforts resulted in conversion to sinus tachycardia, which quickly deteriorated into ventricular fibrillation. The patient was taken to a nearby hospital where she was pronounced dead in the emergency room.

Autopsy examination showed a well developed, well nourished black female weighing 158 pounds and measuring 5' 2" in height. Internal examination revealed a 409 gram heart with no evidence of coronary artery disease. The left ventricle measured 1.3 centimeters in thickness, the right ventricle measured 0.5 centimeters, and the septum ranged in thickness from 0.5 to 1.3 centimeters. On cut surface, the left ventricle had a sponge-like appearance. Filling the left ventricle, from the apex to the level of the mitral valve were extensive trabeculations with deep intertrabecular recesses. An area of scarring and hemorrhage was present along the anterior third of the septum. Autopsy also showed pulmonary edema and congestion, emphysematous changes of the apex of the upper lobes of the lungs bilaterally, and nephrosclerosis. Histologically sections from the heart showed trabeculations lined by ventricular endocardial endothelium, which was continuous with the ventricular endocardium. The trabeculations showed areas of endocardial thickening and ischemic changes with myocyte necrosis and hypertrophy of the surrounding myocytes. Sections from the lung showed chronic congestion. Toxicology tests were negative.

Isolated noncompaction of the ventricle (INLV) is a rare congenital cardiomyopathy thought to be caused by an arrest of compaction of the loose meshwork of myocardial fibers during embryogenesis. Noncompaction results in the formation of muscular trabeculations that fill one or both ventricles imparting a spongy appearance. The overall incidence in the adult population is 0.05 percent. Genetic studies have shown an X-linked recessive inheritance pattern with mutations in the gene G 4.5 on the Xq28 chromosomal region associated with INLV. However, the occurrence of INLV in women suggests a possible non-X -linked inheritance pattern.

Patients have been identified ranging in age from 1 week to 71 years. The onset of symptoms, commonly related to depressed left ventricular function, frequently develops during adulthood. The diagnosis in adults is often delayed because the symptoms, which are nonspecific, are similar to more frequently diagnosed conditions such as congestive heart failure. Patients can also present with various arrhythmias. The arrhythmias can be associated with Wolf Parkinson White Syndrome, bundle branch blocks, or ventricular arrhythmias. Some patients present with embolic events that include transient ischemic attacks, stroke, and pulmonary embolism. INLV has specific echocardiographic findings, and it is not until this test is performed that the diagnosis is confirmed.

Isolated noncompaction of the left ventricle is a rare congenital cardiomyopathy affecting both sexes through a wide range of ages, has nonspecific clinical manifestations, and can result in sudden death.

Isolated Noncompaction of the Left Ventricle, Spongy Myocardium, Congenital Cardiomyopathy