G41 The Hidden Side of Sudden Cardiac Death: Forensic Experimental Protocol and Its Applications

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After attending this presentation, attendees will understand the role of gene mutational analysis in sudden cardiac death.

This presentation will impact the forensic science community by discussing the importance of a rigorous genetic testing protocol in sudden cardiac death diagnosis when autopsy examination is not performed.

Introduction: Sudden Cardiac Death (SCD) is death from cardiac causes within one hour of the onset of symptoms. The sudden death of young people is a devastating event for both the family and community. Over the last decade, significant advances have been made in understanding both the clinical and genetic basis of sudden cardiac death. Most commonly, sudden cardiac death can be the first presentation of an underlying heart problem, leaving the family at a loss as to why an otherwise healthy young person has died.

At external examination on a victim of SCD, the features of hypostasis are the most important gross findings. On examination, hypostasis in these cases show, especially, a typical dark red/blue color on the face, neck, and anterior thoracic wall. There is a deep cyanosis on lips and nail beds. The diagnosis is suggested by the absence of external gross injuries. Genetic factors are important factors to the risk of SCD. Coronary artery disease is the major determinant of SCD, and its predisposing genetic role is complex. The other main causes of sudden cardiac death are some hereditary non-structural diseases such as Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT), long QT, and the Brugada Syndrome, as well as structural diseases such as Hypertrophic Cardiomyopathy(HCM) and Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) which cause a significant number of cases of sudden cardiac deaths in the young. Many times for economic reasons, in the cases of SCD judicial authorities do not require an autopsy examination performed by a forensic pathologist. As an unfortunate result, forensic investigation of SCD often involves only the external examination of the victim's body. Especially in these cases, genetic testing may be useful because many mutations in several genes are implicated in arrhythmic syndromes including SCN5A, KCNQ1, KCNH2, RyR2, and genes causing HCM, specifically MYH7 and MYBPC3. In these cases of SCD when autopsy examination is not performed, an important progress in diagnosis depends on the use of a rigorous protocol in order to analyze biological samples of saliva of the victim according to the informed consent of the victim's relatives.

Case Reports: Twenty cases of sudden cardiac death were analyzed. In these cases, autopsy examination was not required. For 18 cases, forensic pathologists made external examination and for two cases autopsy was carried out. In these last cases, the cause of death was thromboembolic death ARVC. In the others, the cause of death remained unknown.

The goal of this study is understanding the importance of investigation protocol in order to investigate gene mutational analysis in both structural and non-structural genetic heart disease including SCN5A, KCNQ1, KCNH2, RyR2, MYH7, and MYBPC3. If the victim's test is positive, this information is important for relatives who might themselves be at risk of carrying the disease-causing mutation.

Conclusions: This protocol has to be applied in all cases where forensic pathologists must identify the cause of death without autopsy examination. The main goals of this protocol is to evaluate exact incidences of cardiac causes of sudden death, counseling for victims' relatives, and potential therapy for genetic mutations carried.

Sudden Death, Autopsy, Genetic Mutation

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