G82 Genetic Testing of Sudden Cardiac Death Victims: From a Forensic to a Multidisciplinary Approach

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After attending this presentation, attendees will learn a practical, ethically, and legally acceptable approach to cases of sudden cardiac death thought to be of genetic etiology.

This presentation will impact the forensic community by demonstrating an interdisciplinary approach in cases of sudden cardiac death believed to be related to channelopathies.

Sudden cardiac death is considered as the most important cause of death in western countries. In cases of sudden, unexpected deaths and especially in young people, a forensic autopsy is required, frequently followed by complementary investigations, in order to determine the cause of death. However, it happens that even after an autopsy is performed in accordance with international recommendations, the cause of death remains unexplained. Such cases, called also autopsy negative sudden deaths, are not rare (6% to 40%) and are often considered to be due to a sudden cardiac arrhythmia.

Thanks to the progress made in molecular biology, it is admitted that most cases of sudden cardiac death of children and young adults are related to genetically determined cardiac diseases. Some of them have a morphological substrate at autopsy as hypertrophic cardiomyopathy. But those related to channelopathies are impossible to detect without genetic analysis. Postmortem genetic testing referred to as molecular autopsy was recently carried out by many authors in cases without morphological explanation of the sudden death and allowed to identify pathogenic mutations described already in clinically known arrhythmic syndromes. However, it is also possible to perform genetic testing to refine the diagnosis of a hypertrophic cardiomyopathy in cases without evident morphological substrate. The genetic cardiac disease may explain the death, but it may also be at the origin of a traffic accident with a loss of car control or drowning. The channelopathies may also be involved in cases supposed to be related to intoxications. Therefore, it is important to consider the genetic screening in forensic investigations.

The legal and ethical aspects of genetic testing in forensic investigation are complex. In Switzerland, the investigating magistrate may mandate genetic testing in the forensic context in order to determine the cause of death. In fact, the particularity of medicolegal autopsy is that during the investigation procedure and in contrast to a clinical context the genetic tests can be carried out without the consent of the dead person or proxy consent. The consent is however necessary for any research activity. Genetic screening is important to establish the cause but also to detect the asymptomatic carriers in order to prevent sudden death in other family members. This prevention involves a multidisciplinary collaboration. In Lausanne, such collaboration was established between services of cardiology, medical genetic, toxicology, and forensic medicine.

This presentation will be illustrated by autopsy cases for which the interpretation of the results of the genetic screening is explained in the light of other autopsy findings. The interdisciplinary collaboration as well as the juridical and ethical aspects of genetic analyses in cases of sudden cardiac death will also be briefly discussed.

Sudden Cardiac Death, Channelopathies, Molecular Autopsy

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