

Swiss Suggestions For A Multidisciplinary Approach To Treating Sudden Cardiac Death In Forensic Medicine

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ABSTRACT

Sudden cardiac death (SCD) is a cardiac condition that is by definition unexpected. A forensic pathologist usually always conducts the inquiry. In these situations, the forensic pathologist has two roles: (1) to quickly and accurately ascertain the cause and manner of death, and (2) to launch a multidisciplinary approach to stop additional deaths among current family members. The district attorney in charge frequently declines additional investigations if it is determined that the death was brought on by 'natural' reasons.

To determine the exact cause of death, additional tests, such as in-depth histopathological investigations and/or molecular genetic analysis, are frequently required. In an effort to unify the methodology used to study SCD, the Swiss Society of Legal Medicine established a multidisciplinary working group involving clinical and molecular geneticists and cardiologists. In to enhance the diagnostic processes and preventive measures for family members who are still alive, the goal of this paper is to bridge the gap between the clinical recommendations for genetic testing of inherited cardiac diseases and the Swiss recommendations for routine forensic post-mortem cardiac examination. The main recommendations are as follows: 1. do forensic autopsies on all SCD victims under the age of 40; 2. gather and store sufficient samples for genetic testing; 3. communicate with the families; and 4. adopt a multidisciplinary strategy incorporating cardiogenetic counselling.

Keywords: sudden cardiac death; forensic autopsy; postmortem genetic testing; genetic counselling

INTRODUCTION

Globally, sudden cardiac death (SCD) is a serious problem for public health. Though the published occurrences are widely believed to be understated [1-4], the precise incidence of sudden death in the younger population is still unknown. Regarding the incidence of SCD in young people or the rate of autopsies, there is no precise information available for Switzerland. Recently, Hofer et al. [5] reported the presumptive values for the canton of Vaud. This study estimates that the incidence of SCD in young people (aged 5 to 39) to be 1.71 per 100,000 person-years (2.73 for men and 0.69 for women). It may be greater, though, as not all instances

of SCD (unexplained deaths, drownings, auto accidents, etc.) are taken into account. In comparison to other nations, the autopsy rate of sudden deaths categorised as ‘diseases of the circulatory system’ was estimated at 47.5% [5].

The rehabilitation of SCD victims and their families remains challenging despite clinical advice on genetic testing [10–12]. In the UK, it was determined that the flow-limiting phase was the availability, or lack thereof, of proband DNA [11,13].

Researchers from the Heart Rhythm Society, the European Heart Rhythm Association, and the European Society of Cardiology developed the current international recommendations on genetic testing for cardiomyopathies and channelopathies [14,15]. These recommendations do not address the practical aspects of our Swiss practise during forensic autopsies or the concerns surrounding cardiogenetic counselling of family members, which is crucial for the prevention and potential treatment of disease.

The working group of forensic pathologists and related cardiologists, clinical and molecular geneticists investigated how the SCD autopsy criteria were decided upon in order to enhance the diagnosis and management of genetically determined cardiac fatalities, as well as the diagnostic processes and preventative measures for living family members. Based on existing recommendations for autopsy procedure and genetic testing for channelopathies and cardiomyopathies [14–18], and taking into account the regulatory environment for forensic activity, the multidisciplinary working group made recommendations for interdisciplinary collaboration and workflow for forensic SCD cases in Switzerland. The multidisciplinary working group made recommendations on interdisciplinary collaboration and workflow for forensic SCD cases in Switzerland based on current autopsy practise guidelines and genetic testing for channelopathies and cardiomyopathies [14–18], taking into account the legal framework of forensic activity.

Aetiology of sudden cardiac death

Coronary artery disease (CAD) is the condition that is most frequently found in SCD patients over the age of 35 [6, 19]. Cardiomyopathies or a morphologically normal heart are the most frequent results in the younger group of those under 35. Approximately 80% of SCD instances are assigned to CAD [15] based on epidemiological statistics from the general population, while 10–15% are caused by cardiomyopathies with a morphological basis such as myocardial infiltrative disorders, arrhythmogenic right ventricular cardiomyopathies, non-compaction cardiomyopathies, hypertrophic cardiomyopathies, and dilated cardiomyopathies. The causes of SCD in the remaining 5–10% of cases include either structurally aberrant congenital cardiac conditions (such as anomalies of the coronary arteries) or arrhythmias without apparent structural alterations, including several genetic arrhythmias like channelopathies [6].

Some genetically based cardiac illnesses, such as HCM, DCM, ARVC, or NCCM [18], which cause sudden cardiac death, are defined by morphological changes that can be seen during autopsy and/or histological testing. Only after molecular genetic investigations may those associated with cardiac channelopathies, such as congenital long QT syndrome (LQTS), Brugada

syndrome (BrS), or catecholaminergic polymorphic ventricular tachycardia (CPVT), be detected [7,12,16]. The majority of these genetic diseases exhibit significant genetic variability and are inherited via the autosomal dominant pathway [14,19].

Management of sudden cardiac death at this time in Switzerland

SCDs and all other sudden unexpected deaths (SUDs), like in many other nations, must be notified to the district attorney's (DA's) office, which then seizes the body and launches an investigation. The major goal of such an inquest is to identify the cause of death, whether it was homicidal, suicidal, or natural. The Swiss Criminal Procedure Code has been uniform throughout Switzerland since January 1, 2011. Forensic pathologists are hired by the district attorney to examine the body. In Switzerland, the initial forensic examination consists of a scene investigation, a comprehensive exterior examination of the corpse, ideally at the site, carried out by professionally qualified medical professionals who assess the scene findings and the medical history. The District Attorney frequently won't order additional investigations, including an autopsy, if third-party involvement is determined to be impossible.

Although it is required by law that the DA be notified of all SUDs, this is not often the case. The death-certifying physician will occasionally declare a sudden death to be 'natural' rather than "undetermined" due to ignorance of the situation or fear of legal repercussions. The next-of-kin may, however, request that such situations go through a clinical autopsy. The major goals of a clinical autopsy conducted by a clinical pathologist are to identify any pathological abnormalities and ascertain the cause of death. The forensic post-mortem cardiac assessment techniques in Switzerland have been a bit erratic. The Swiss Society of Legal Medicine (SSLM) established a working group to develop recommendations for basic standards of autopsy practise for cardiac assessment in order to harmonise the various autopsy methods and raise the overall standard of SCD case treatment. Harmonization of standard forensic autopsy procedures for SCD cases in Switzerland [32]: this advice was posted on the SSLM website.

Currently, sudden cardiac death is defined in a number of different ways. SCD is defined as a sudden unexpected death (SUD) of cardiac causes happening in a short amount of time (often within an hour of symptom onset) in a person with known or unknow heart disease, which is the nomenclature used in forensic pathology in Switzerland.[21] Among newborns younger than one year of age, Sudden Infant Death Syndrome (SIDS) is a subtype of unexplained Sudden Death. SIDS is outside the purview of these suggestions.

Autopsy

For all cases of SCD in patients under the age of 40, the post-mortem cardiac examination should be done in accordance with the recommendations made by the SSLM [19]. A so-called "undetermined" or "functional death" can be postulated (also referred to, in our opinion incorrectly, as a "negative autopsy") if no morphological (macroscopic and microscopic) or toxicological cause of death can be determined and biochemical analyses do not reveal any

significant disorder. A 'functional death' could result from something like an epileptic seizure or an arrhythmic condition. The possibility of genetic testing should be mentioned and the appropriate samples should be collected in anticipation of such testing in the autopsy report of SUD cases as well as in cases where a genetically determined cardiomyopathy is diagnosed by morphological examination (HCM, ARVC, DCM, NCCM).[20] The authority to keep the gathered samples for 5 years should be sought from the DA. If the DA approved the preservation of the samples for a period of five years, the examiner should record the following: "Samples were obtained for potential genetic testing in addition to sample storage for toxicological analyses, etc.

These extra samples will be kept for five years. These materials, specifically EDTA blood or tissue, should first be kept in the forensic medicine lab at a temperature of at least -20 °C (preferably at -80 °C). The DA, who has the authority to do so, can then approve sending the samples to a recognised facility for genetic examination. The results of the autopsy may be crucial for the victim's family clinically if there is a possibility that the person had an inherited heart condition.

Multidisciplinary cardiogenetic counselling and information

If an inherited heart condition is suspected, multidisciplinary counselling should be organised if the relatives contact the forensic pathologist for more information or to obtain the autopsy results, and if the forensic pathologist so desires. In Lausanne, for instance, a brief letter addressed to the family is delivered through the DA; this approach allows for the right not to be informed to be respected. The best way to contact the family is negotiated with the local ethics commission. The multidisciplinary cardiogenetic counselling, which will at the very least include a cardiologist and a clinical geneticist, is encouraged for the forensic pathologist to attend. The proposed course of action, in our opinion, should enable the gap between current Swiss recommendations for routine forensic cardiac examination [23] and clinical recommendations like 'HRS/EHRA Expert Consensus Statement on the State of Genetic Testing for the Channelopathies and Cardiomyopathies' [14] to be closed. The management of these uncommon but complex cases as well as the diagnostic and possibly therapeutic procedures for the living family members will be improved by synchronising autopsy standards with multidisciplinary cardiogenetic counselling.

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