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MÉDICOS FORENSES

# Spanish Journal of Legal Medicine

## Revista Española de Medicina Legal

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### SPECIAL ARTICLE

## Multidisciplinary units, a key element in the study and prevention of sudden cardiac death caused by inherited cardiac conditions<sup>☆</sup>



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Received 24 May 2017; accepted 22 June 2017

Available online 13 February 2018

#### KEYWORDS

Inherited cardiac conditions;  
Multidisciplinary units;  
Sudden cardiac death;  
Forensic pathology

**Abstract** Cardiac diseases often underlie sudden deaths. Sudden death has a great social and economic impact and generates loads of media attention. When it occurs in young people, it is usually due to genetic causes (inherited heart diseases). This group includes cardiomyopathies, channelopathies, and familial thoracic aortic aneurysm/dissection. Inherited heart conditions are rare diseases in terms of prevalence and little knowledge is still available from most of them, so their management requires specialised and multidisciplinary training. Sudden death usually occurs in the out-of-hospital setting, and there has to be a forensic autopsy. These autopsies should be performed according to the minimum quality standards of the European guidelines. The clear genetic background justifies the need for a family screening to enable early diagnosis and also to rule it out in other family members at risk using a multidisciplinary approach offering personalised medicine to the affected families. In addition to medical issues, this scheme enables the provision of early psychological support and genetic counselling to plan new gestations. In Spain there is a great legal void in the approach to sudden death and its prevention, with a lack of homogenisation among the different counties, both at the judicial and healthcare levels due to the variable recognition of acquired skills and to the differences in the judicial and territorial forensic organisation throughout the country. It would be desirable that the consultative panels of both Ministries (Health and Justice) developed a comprehensive plan to ensure the quality in post-mortem studies and a proper subsequent clinical and genetic

DOI of original article: <http://dx.doi.org/10.1016/j.reml.2017.06.007>

<sup>☆</sup> Please cite this article as: Molina Aguilar P, Giner Blasco J, Izquierdo Macian I, Martínez-Dolz L, Barriaes Villa R, Zorio Grima E. Unidades multidisciplinarias en el estudio y prevención de la muerte súbita por cardiopatías familiares. Rev Esp Med Legal. 2018;44:46–52.

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family study in public health services. Thus, the clinical-preventive potential of the autopsies would take shape and become a clear benefit for the affected families and for the society in general. This measure would also favour the teaching and the research of inherited cardiac diseases, which is of paramount importance to improve the current knowledge in this field, so unknown and so devastating.

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## PALABRAS CLAVE

Cardiopatías familiares;  
Unidades multidisciplinarias;  
Muerte súbita cardíaca;  
Patología forense

## Unidades multidisciplinarias en el estudio y prevención de la muerte súbita por cardiopatías familiares

**Resumen** La muerte súbita es fundamentalmente cardíaca y tiene un gran impacto social, económico y mediático. Cuando acontece en personas jóvenes, suele obedecer a causas genéticas (cardiopatías familiares). En este grupo se incluyen las miocardiopatías, canalopatías y el aneurisma/disección aórtica familiar. Las cardiopatías familiares son enfermedades raras en términos de prevalencia y poco conocidas, por lo que su manejo requiere formación especializada y multidisciplinar. La muerte súbita suele acontecer en el ámbito extrahospitalario y es objeto de una autopsia forense. Estas autopsias deberían realizarse de acuerdo a los estándares mínimos de calidad de las guías europeas. El claro sustrato genético justifica la necesidad de un screening familiar para realizar diagnósticos precoces y también para descartarla en otros familiares en riesgo a través de un abordaje multidisciplinar que ofrezca una medicina personalizada a las familias afectadas. Además de las cuestiones médicas, este esquema permite ofrecer apoyo psicológico precoz y consejo genético de cara a planificar nuevas gestaciones. En España existe un gran vacío legal en el abordaje de la muerte súbita y su prevención, con una falta de homogenización entre las distintas comunidades autónomas tanto a nivel judicial como asistencial debido a las características del proceso de transferencia de competencias y a la organización territorial judicial y forense. Sería deseable que, desde los órganos consultivos de ambos ministerios (Sanidad y Justicia) se elaborara un plan de actuación global que asegurara la calidad en el estudio *post mortem* y su continuidad en el posterior estudio familiar clínico y genético. Así, el potencial clínico-preventivo de las autopsias tomaría cuerpo y revertiría en un claro beneficio para las familias afectadas y para la sociedad en general favoreciendo asimismo la docencia y la investigación, tan importantes para el avance del conocimiento en este ámbito tan desconocido y devastador.

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Sudden death (SD) is usually defined as an unexpected death which occurs within an hour from the onset of symptoms, although other time intervals have sometimes been used.<sup>1</sup> It is an event which has a major social and financial impact and which generates a good deal of media attention. The primary cause of sudden death is heart-related, and in particular ischaemic heart disease,<sup>2</sup> with a polygenic and multifactorial origin. Inherited cardiac conditions (ICCs) (channelopathies, cardiomyopathies and familial thoracic aortic aneurysm/dissection) tend to disproportionately affect children and young adults, where they represent the leading cause of sudden cardiac death (SCD).<sup>3</sup> Unlike ischaemic heart disease, ICCs have a clear genetic substrate which justifies the indication for adequate testing of the first-degree relatives of the deceased, with a cascading expansion of the testing circle depending on the results.<sup>1</sup> The most common type of inheritance is autosomal dominant, where 50% of offspring will be affected.<sup>1</sup> This family testing, with clinical and genetic tests, should ideally be carried out by a multidisciplinary team (forensic experts, pathologists, geneticists, paediatricians, basic researchers and cardiologists) which has experience in the field of ICCs,<sup>1,2,4</sup> given their status as rare diseases, their

short course from when they were discovered and the difficulties in establishing definitive diagnoses.

## The forensic autopsy, starting point for family testing

As it is an unexpected event, sudden death should give rise to the opening of a judicial procedure, as the legal system justifies forensic autopsies in these cases. Specifically, Article 343 of the Spanish Law on Criminal Procedure states that, in cases of violent death or suspected crime, an autopsy will be carried out. Furthermore, Recommendation No. (99) 3 of the Committee of Ministers of the Council of Europe, for the methodological harmonisation of medico-legal autopsy rules,<sup>5</sup> specifies that an autopsy should be carried out in the event of a sudden unexpected death.

The medico-legal autopsy fulfils its legal objective of ruling out a violent death but often, despite the efforts of the professional in charge, it is not able to develop its research potential (examining the biological substrate causing the SD in more detail) or its clinical-preventive potential (by allowing the findings obtained in the autopsy to be turned into a

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