

Histopathological study in sudden cardiac death

Estudio histopatológico en la muerte súbita cardíaca

Pilar Molina Aguilar[✉], MD

Institute of Legal Medicine and Forensic Sciences. University of Valencia. Valencia, Spain.

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To the Editor:

In the multidisciplinary approach of sudden cardiac death, it is necessary for every professional to do his/her specific work for accomplishing the complex gear of prevention. In the case of the histopathological study, it is important to approach the diagnosis to the maximum for facilitating the subsequent family cardiac study. Clinical studies that are offered to first-degree relatives are determined by the classification of the index case, and results are complemented by those of the conducted genetic tests (ideally) in blood obtained from the index case and, failing that, in the blood of a relative affected by the phenotype.

Usually, they are classified into the following groups: structurally normal heart (where channelopathies should be suspected), cardiomyopathies (hypertrophic, dilated, non-compacted, arrhythmogenic or non-precised), hereditary valvulopathies (supravalvular aortic stenosis and bicuspid aorta)

and non-atheromatous dissection of the thoracic aorta. In the case of sudden infant death, the histopathological study, along with other supplementary analysis, will allow to establish whether there is a cause of unexplained death, or on the contrary, it is framed as sudden infant death syndrome, and is included in a certain category of the San Diego's classification¹.

The Family Sudden Death Risk-Assessment Unit of the Valencian community has studied a total of 618 cases of sudden deaths in people under 55 years, in the period between 2008 and 2015. As expected, men predominated (499H/119m, 81%) and the most frequent was the ischemic heart disease (53%), followed by the structurally normal heart (24%) and the cardiomyopathies (14%) (**Figure**).

WHAT HAVE WE LEARNED IN THESE EIGHT YEARS?

Ischemic heart disease

Generally, the only finding is a severe coronary arteriosclerosis without thrombosis (73%). The most affected is the coronary anterior descending artery and more involvement of the chronic myocardial affection (43%) than the acute one (ischemia 12%

[✉] P Molina Aguilar

Instituto de Medicina Legal y Ciencias Forenses
Avda. Profesor López Piñero 14
Ciudad de la Justicia 46013. Valencia, España.
E-mail address: pilar.molina@uv.es

and infarction 19%). In some cases, in people under 35 years, where there is only observed a stable plaque with severe stenosis of a vessel, it should be considered the possibility that there is a channelopathy that causes the sudden death. If these cases are defined as ischemic heart disease, the risk of leaving relatives without the possibility of a cardiological study would be a fact. In half of the selected cases, two pathogenic variants were found.

Hypertrophic cardiomyopathy

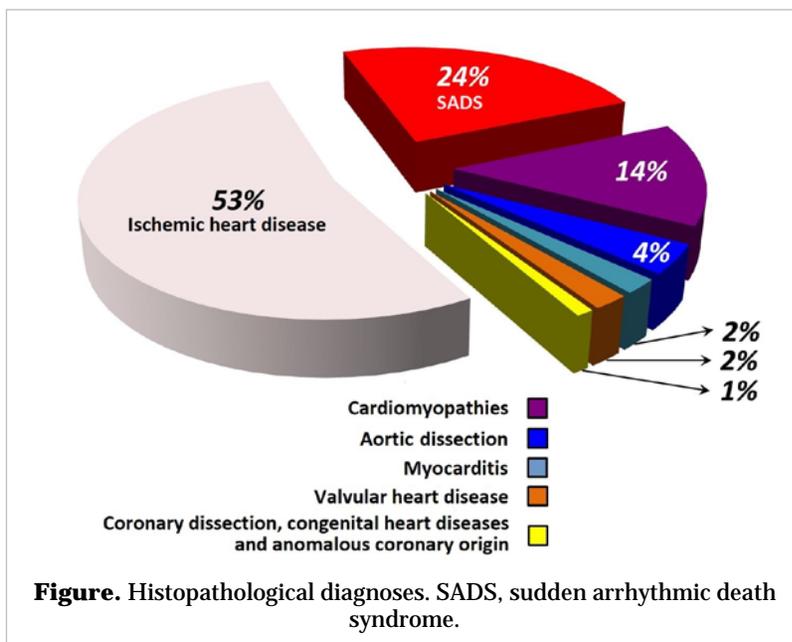
Although the *post mortem* diagnosis of hypertrophic cardiomyopathy is clear, the strategy for genetic studies in our series has enabled budget savings, which would increase previsibly, if considering that families are constantly expanding and they would generate a greater number of cardiological monitoring visits. This expense is minimal once identified the causal mutation that allows later to discard its presence in the relatives, resulting in benefit for patients, if the psychological implications are considered.

Arrhythmogenic cardiomyopathy

Similar results are obtained in the arrhythmogenic cardiomyopathy, with 63% of positive genetic studies: desmosomal genes (DSP [N=8], 57% genes; PKP2 [N=2], 14% and DSG2 [N=1], 7%) and non-desmosomal (DES [N=1], 7%; FLNC [N=1], 7%; TMEM43 [N=1], 7%; SCN5A [N=1], 7%). In our series, it is the cardiomyopathy most associated with sport (39% versus 26% in the hypertrophic cardiomyopathy). There is a predominance of biventricular forms (41%) compared to the left (36%) and right (23%) forms. We have found no myocardial compactation in 14% of cases, with dominant left ventricular (or biventricular) affection and location, most frequently, in the posterior and lateral walls of the left ventricle.

Adipositas cordis

Retrospectively, there have been detected pheno-



types of *adipositas cordis* or cardiac lipomatoses in two index cases of families with Brugada syndrome.

Sudden death in epilepsy and asthma

The molecular autopsy with massive ultrasequencing has identified potentially pathogenic mutations (channelopathies) in 63% of cases, in which the *post mortem* and cardiological family studies had not achieved to precise them, and they were initially classified as sudden death with structurally normal heart, associated with epilepsy or asthma.

CONFLICTS OF INTERESTS

None declared

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