Since there is no “gold standard” to reach the diagnosis of AC, multiple categories of diagnostic information have been combined and the criteria recently updated, to improve diagnostic sensitivity while maintaining specificity. Among diagnostic tools, contrast enhanced cardiac magnetic resonance is playing a major role in detecting left dominant forms of AC, even preceding morpho-functional abnormalities.

The main differential diagnoses are idiopathic right ventricular outflow tract tachycardia, myocarditis, sarcoidosis, dilated cardiomyopathy, right ventricular infarction, congenital heart diseases with right ventricular overload and athlete heart. A positive genetic test in the affected AC proband allows early identification of asymptomatic carriers by cascade genetic screening of family members. Risk stratification remains a major clinical challenge and antiarrhythmic drugs, catheter ablation and implantable cardioverter defibrillator are the currently available therapeutic tools. Sport disqualification is life-saving, since effort is a major trigger not only of electrical instability but also of disease onset and progression.

The presentation will review the current knowledge of this rare cardiomyopathy, suggesting a flowchart for primary care clinicians, general and forensic pathologists and geneticists.

CONFLICTS OF INTERESTS

None declared

REFERENCES

To the Editor:

Sudden cardiac death is defined as the natural death due to cardiac causes occurring suddenly and unexpectedly in an individual with underlying heart disease, known or not, within one hour of the onset of symptoms. Within this, sudden infant death stands out, which is classified into three groups: deaths of explained cause, unexplained death and sudden infant death syndrome (categories I and II of the Legal Medicine Institute of Valencia). The latter is defined as the sudden and unexplained death of a child younger than one-year-old, which apparently occurs during sleep and remains unexplained after a thorough post-mortem investigation, including performance of autopsy, examination of the death scene and review of the medical record.

A percentage of these sudden deaths have a genetic link; therefore, these cases and their relatives require a thorough study to get accurate diagnosis and prevent future events. This fact entailed the creation of multidisciplinary groups to integrally carry out post-mortem and family study. Given the importance of these cases, the Valencian Community created a research group called Sudden Family Death Risk-Assessment Unit (UVRMSF by its acronym in Spanish) in 2008, made up of different specialists in several medicine fields such as: forensics, pathologists, geneticists, microbiologists, cardiologists, pediatricians, biochemists, among others, which include pathology and cytology technician specialists (TEAP by its acronym in Spanish) from the Histopathology Section of the Forensic Pathology Department, Institute of Legal Medicine and Forensic Sciences of Valencia, Spain. This was twofold: properly identify the cause of death and detect sub-clinical family involvement.

The forensic autopsy is performed following the Guidelines for sudden cardiac death from the Association for European Cardiovascular Pathology, with sampling for histological, toxicological, microbiological, genetic and other studies (peripheral blood, vitreous gel, urine), hair sample, nasopharyngeal exudate) and active participation in the cavities and organs dissection during internal examination. The weight of the viscera is recorded, and samples of different tissues are taken for freezing and preservation/fixation in formaldehyde, until further study.

It is also important to review all aspects of the protocol to check that every sample has been accurately taken, labeling them for processing, with a view to further examination, with subsequent control, registration and distribution to the different internal and reference laboratories.

In the laboratory, the TEAP is responsible for recording cases in the sudden death database. They specify the distribution and location of the samples, the data obtained from the corpse removal and autopsy, and record all results. In addition, they refer the samples to other laboratories and are in charge of carving (including a macroscopic description) and photographing the samples, as well as carrying out the cardiac and neurological study, under forensic pathologist supervision.

The TEAP also manages judicial and clinical information collection, which facilitates contact and interview with the family, and their subsequent access to cardiology studies.

In short, the TEAP plays a fundamental role in the different phases of the study and prevention of sudden death. The experience gained over these eight years shows that their participation and active training in autopsied cases of sudden death is crucial for obtaining sufficient and quality material for accurate processing and diagnosis in each of the above-mentioned disciplines, which avoids bias during sample collection and facilitates strict adherence to the protocol, thanks to TEAP direction and inspection.

The support in the subsequent phases of data collection and family contacts, as well as their col-
laboration in the analysis of the results obtained, along with other specialists, further extends the TEAP functions in this field.

CONFLICTS OF INTERESTS

Ninguno

REFERENCES


