Abstract

Cardiogenetics is a dynamically developing field of genetics that allows immediate implementation of laboratory results into practice, leading to improved prevention and treatment for patients and their relatives. A properly set up cardiogenetic care system is an essential mechanism to prevent sudden cardiac death (SCD). Our pilot study contributed to a better understanding of the genetic causes of SCD, in which we examined a total of 100 unrelated SCD victims using molecular autopsy and detected a clear genetic cause in 22 % of them. A major outcome of the study was also the identify of 87 relatives at risk of sudden death who were taken up for clinical follow-up. Finally, the findings led to the development of a set of recommendations for the correct procedure of genetic testing in cases of inherited cardiac disease and sudden cardiac death. In a parallel cohort of 100 cardiac arrest survivors (SCA), we identified the causative variant in 20 % of cases. In 10 of them, this result allowed the diagnosis of arrhythmogenic cardiomyopathy, whose morphological manifestations were below the resolution of imaging methods. The results were published on European Heart Rhythm Association (EHRA) congress and are a promising basis for further forthcoming publications.

Examination of another large cohort of patients with hypertrophic cardiomyopathy (HCM) allowed 2 studies to be conducted that demonstrated that patients found to have a causative variant in the sarcomeric protein gene did not have a different disease course, response to treatment with alcohol septal ablation, or increased incidence of major complications, including sudden cardiac death, compared with genetically negative patients. Thus, finding a pathogenic variant in HCM patients alone does not warrant a change in therapy or overall patient care. The experience of genetic testing has also led to the development of recommendations for the overall management of HCM patients.

In our cohort of patients with aortic syndromes and systemic connective tissue disease, we identified rare variants that have been used in publications expanding the current knowledge of the causes and manifestations of hereditary aortic dissection (TAAD). In these, the diversity of phenotypic manifestations associated with different types of pathogenic variants in the *LOX* gene and the association of pathogenic variants in the *JAG1* gene with the development of isolated aortic aneurysms were examined.

Key words: cardiogenetics, sudden cardiac death, SCD, cardiomyopathy, arrhythmia, aneurysm, aortopathy