

Classification of rare cardiovascular diseases and disorders: challenges in categorisation of different diseases

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Diagnosis and treatment of the majority of common cardiac disorders is based on the results of large, multicentre, randomised trials which are analysed by experts and published in the form of guidelines or recommendations of international cardiac societies. In cases of rare diseases, such methodology is impossible to implement due to the lack of sufficient and large-scale data.

To progress the understanding of rare cardiovascular diseases and disorders (RCDD), a more sophisticated version of the RCDD classification system was needed. Collecting data and exchanging experiences is crucial for the purpose of creating worldwide databases of RCDD. Undoubtedly, such registries facilitate further improvement in the diagnosis of RCDD and subsequent therapy. Having the opportunity to *comment* on the latest version of RCDD classification [1] gave me the chance to think about changes in the categorisation of RCDD. Bearing in mind that the classification of RCDD has never been simple and straightforward, the authors have based the classification of these diseases on common clinical and/or anatomical features, as well as taking into consideration major clinical symptoms and pathologies. This approach is logical and seems to fulfil the criteria for classification of RCDD. Nevertheless, several points require further discussion, and perhaps revision, as the classification itself has certain weak areas, some of which were outlined by the authors themselves in the concluding remarks. In particular, looking from the perspective of a cardiac electrophysiologist, attention should be drawn to the proper inclusion of cardiac arrhythmias.

In comparison with the previous version, the main difference concerns categorisation of cardiac rhythm disturbances. The current version introduces a separate class (class VI – cardiac arrhythmogenic disorders and arrhythmias) for cardiac arrhythmias, which is subdivided into two groups. Previous second subgroup included arrhythmias secondary to or co-existing with another RCDD. In my opinion, the introduction of this subgroup was questionable (and importantly was omitted in the current RCDD classification), since arrhythmia is often a symptom of a disease, not a disease in itself. For example,

a patient presenting with ventricular tachycardia and arrhythmogenic right ventricular cardiomyopathy could previously be classified both in class III group 4 and class VI group 6. This could lead to misclassification of the diseases and affect classification of the RCDD, resulting in inappropriate conclusions on epidemiology, morbidity, and treatment outcomes. The situation could be even more visible in patients with class IV diseases and co-existent arrhythmias.

Moreover, it is worth emphasizing that authors highlighted the use of the term *arrhythmogenic* in class VI, as in the case of some arrhythmogenic diseases, life-threatening arrhythmias are the main symptom of the disease, such as in Brugada syndrome or LQTS syndrome, but sometimes may not occur during the whole lifetime of the patient.

Secondly, similar concerns as the abovementioned applied to previous class of cardiac tumors and cardiovascular diseases in malignancy. Thrombus within a heart chamber is a sign of the disease not a disease in itself and was excluded from the current, significantly improved, version of RCDD classification.

Taking everything into consideration, the attempt by the authors to create an up-to-date and comprehensive study on rare diseases is commendable, considering the frequency of their occurrence, the availability of data, and most importantly, the challenges regarding their proper categorisation. These novel developmental initiatives should prove beneficial in the long-run. With the implementation of registries based on this classification, some diseases may turn out not to be rare but rather underdiagnosed. In such a case, the author's classification of the disease will greatly contribute to progress in the field of modern cardiology.

References

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