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Clinical Classification of Rare Cardiovascular Diseases and Disorders: 2018 Update

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Introduction

Rare diseases and disorders constitute important clinical problems. There are many concerns among physicians while planning the diagnostic and treatment process of such a heterogenous group of patients. These concerns arise not only from the rarity of cases, but also from multiple gaps in knowledge on the management of patients with rare diseases and disorders. The commonly accepted prevalence of rare diseases and disorders is 1 per 2 000 in the general population or less. Incidental prevalence and multiplicity of comorbidities result in an inability to gather enough experience at any single centre. Thus, cooperation and the exchange of ideas is important for the management of patients with rare diseases.

Classification of rare cardiovascular diseases and disorders (RCDD) is crucial for expanding knowledge in the field of RCDD. It consists of an overview of RCDD, facilitates clinical approaches to patients and makes the creation of registries and databases easier. We hope that the updated RCDD classification will aid medical practice through the contribution to progress in diagnostics and therapy. It also serves as a summary of scientific achievements in the field of RCDD. Without the grouping of specific disorders, it is very difficult to create diagnostic and therapeutic algorithms.

The Classification of RCDD was published for the first time in the *Journal of Rare Cardiovascular Diseases* (*JRCD*) in 2013 [1]. RCDD classification was discussed during the 2013 European Society of Cardiology Congress held in Amsterdam (www.crcd.eu/?p=2800) and in international journals, including a recent publication of the European Heart Journal [2, 3]. Clinical classification of RCDD takes into account major clinical symptoms and pathologies and is based on common clinical and/or anatomical features.

Classifications of rare diseases and disorders

Orphanet provides and ensures access to high-quality information on rare diseases and orphan drugs. It was created in 1997 by the French National Institute for Health and Medical Research [4]. It introduced, among other services, the Orphanet rare diseases classification, encyclopaedia and inventory, which includes diseases having a prevalence of 5 per 10 000 or less in the general population [5]. Rare disorders in Orphanet are included in clinical classifications. For example, Brugada syndrome may be found in the molecular classification of channelopathies, Orphanet classification of rare cardiac diseases, and Orphanet classification of rare genetic diseases. However, these classifications, especially in the field of cardiovascular diseases, do not seem to be exhaustive. Each disorder listed in the Orphanet inventory is defined by a unique, permanent ORPHA number. However, these identifiers are attributed randomly by the database [6].

The International Classification of Diseases and Related Health Problems 10th Revision (ICD-10) was developed between 1982 and

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Table. Clinical classification of rare cardiovascular diseases and disorders

Class I – rare diseases of systemic circulation	
Group 1	Anatomical malformations of the arteries
Group 2	Connective tissue disorders causing aneurysmal disease
Group 3	Autoimmune vascular diseases
Group 4	Intimal hyperplasia
Group 5	Spontaneous dissection of the artery
Group 6	Premature atherosclerosis
Group 7	Others
Class II – rare diseases of pulmonary circulation	
Group 1	Pulmonary hypertension
Group 2	Inborn anomalies of the pulmonary vessels
Group 3	Acquired anomalies of the pulmonary vessels
Class III – rare diseases of the heart (cardiomyopathies)	
Group 1	Dilated cardiomyopathy
Group 2	Hypertrophic cardiomyopathy
Group 3	Restrictive cardiomyopathy
Group 4	Arrhythmogenic right ventricular cardiomyopathy
Group 5	Unclassified cardiomyopathies
Class IV – rare congenital cardiovascular diseases	
Group 1	Abnormalities of the position and connection of the heart and vessels
Group 2	Shunts
Group 3	Complex congenital cardiovascular diseases
Group 4	Congenital cardiovascular diseases with concomitant organ dysfunction
Group 5	Grown-up congenital cardiovascular diseases
Group 6	Others
Class V – cardiac tumours and cardiovascular diseases in malignancy	
Group 1	Primary cardiac tumours
Group 2	Metastatic cardiac tumours
Group 3	Inflammatory malformations
Group 4	Cardiovascular complications of oncological therapy
Class VI – cardiac arrhythmogenic disorders and arrhythmias	
Group 1	Primary electrical disorders of the heart
Group 2	Arrhythmias in specific clinical settings
Class VII – rare cardiovascular diseases and disorders in pregnancy	
Class VIII – unclassified rare cardiovascular diseases and disorders	

1989 and was adopted by the World Health Assembly in 1990. It includes only a small fraction of rare diseases [7]. The upcoming and revised ICD classification will have a larger number of specific codes and will include a greater number of rare diseases. The final version of ICD-11 is planned to be introduced in 2018 [8]. The ICD-11 Beta draft is available at https://icd.who.int/dev11/l-m/en.

Methodology of clinical RCDD classification

Since the first clinical classification of RCDD was published, progress has been made in the diagnostics and treatment methods of RCDD [9]. To update the current classification, we performed a search using the Orphanet rare diseases inventory. We have also performed a search of PubMed and Scopus databases for rare cardiovascular diseases according to the wider definition of rare disease proposed by the European Parliament and the Council of the European Union [10]. Moreover, we have included rare cardiovascular disorders in this search. In addition to ICD-10 codes [11], classes, groups, subgroups, examples, and RCDD codes, we have included ORPHA numbers of classified diseases in the Orphanet inventory (available at: http://www.orpha.net/consor/cgi-bin/Disease.php?lng=EN) in our updated classification [5].

General changes

In addition to an extensive literature review, we have introduced appropriate changes as suggested by national and international reviewers and/or readers. The 'rare arrhythmias' class was changed to the 'rare arrhythmogenic disorders and arrhythmias' (RADA) class due to more appropriate terminology, because it includes a spectrum of diseases, disorders and arrhythmias in specific clinical settings. To preserve the clinical and anatomical context of RCDD consecutive classes, the current version of the classification differs with regard to the sequence of classes. The RADA class was transferred from class V to class VI, while the 'cardiac tumours and cardiovascular diseases in malignancy' class was transferred from class VI to class V. In the updated classification of RCDD, significant modification and extension of the RADA classification is provided [12].

RCDD classification and clinical considerations

Clinical classification of RCDD, aside from the rarity of cardiovascular diseases and disorders, takes into account the clinical features and importance of RCDD. It is based on common clinical and/or anatomical features of RCDD. It includes rare diseases of systemic circulation (class I), rare diseases of pulmonary circulation (class II), rare diseases of the heart (class III), rare congenital cardiovascular diseases (class IV), cardiac tumours and cardiovascular diseases in malignancy (class V), rare arrhythmogenic disorders and arrhythmias (class VI), rare cardiovascular diseases

and disorders in pregnancy (class VII), and unclassified rare cardiovascular diseases and disorders (class VIII). Groups of diseases included in the clinical RCDD classification are provided in the Table.

Class I consists of 6 main thematic groups, which are heterogenous in their nature: 1 – anatomical malformations of the arteries, 2 – connective tissue disorders causing aneurysmal disease, 3 – autoimmune vascular diseases, 4 – intimal hyperplasia, 5 – spontaneous dissection of the artery, 6 – premature atherosclerosis and others.

It should be noted that anatomical malformations of the arteries have been previously classified in several smaller classifications addressing arterial territories, e.g. cerebral arteries, coronary arteries or aortic arch [13–16]. Among anatomical malformations of the systemic circulation, we should mention Bland-White-Garland syndrome and other forms of coronary artery anomalies [17][18]. Aneurysmal disease of the aorta, especially with regard to rare genetically inherited aneurysmal formations of the systemic arteries, is poorly defined [19–21]. Along with the DeBakey classification, the Stanford classification is used to separate aortic dissections into those which need surgical repair, and those which usually require only medical management [22]. The Stanford classification divides dissections into two types: type A which affects the ascending aorta and arch, and type B which originate beyond the brachiocephalic vessels [22]. However, these classifications do not address the problem of aneurysmal disease pathophysiology, which in only less than 1% of cases is associated with connective tissue disorders, and as such, is regarded as a minor problem. According to the artery diameter, clinical characteristics, and laboratory or imaging findings, primary vasculitides are classified into seven subgroups and are included in the American College of Rheumatology (ACR) classification [23, 24], as well as the recently updated classification by the European League Against Rheumatism (EULAR) [25, 26]. There are diseases with similar characteristics and overlapping types of vasculitides and new diagnostic modalities are now widely available [27]. Moreover, a number of systemic artery diseases, e.g. intimal hyperplasia, spontaneous dissection of the artery, or premature atherosclerosis are underdiagnosed due to their low prevalence [28, 29]. However, we decided to exclude familial hypercholesterolaemia as a cause of premature atherosclerosis, as it seems to be underdiagnosed rather than rare [30].

Diseases of the pulmonary circulation constitute class II in the clinical classification of RCDD. This class is further subdivided into 3 groups: inborn anomalies of the pulmonary vessels, acquired anomalies of the pulmonary vessels, and pulmonary hypertension (PH). Although in most cases, PH is an acquired disease of the pulmonary circulation, it has been distinguished as a separate group because of its complex pathogenesis and pathophysiology.

Previously, PH was classified [31] according to clinical characteristics and associated risk factors. An updated classification was published in 2013 [32]. Many disorders classified as PH have low prevalence [33], however, not all of them are life-threatening or chronically debilitating. The others can be efficiently cured. An example is PH associated with atrial septal defect which has an estimated prevalence of 0.14:2000 [34, 35]. Some patients with atrial septal defect will develop severe PH, which is a contraindication for curative operation, while some will have persistent PH despite closure of the defect, which is chronically debilitating and will require combined therapeutic strategies. On the other hand, some diseases included in the PH classification are quite prevalent. For example, chronic obstructive pulmonary disease (COPD) affects about 5% of European adults [36], and a significant proportion of these individuals may suffer from PH (between 30% and 70%, depending on the severity of COPD) [37]. Most of them will require smoking cessation and specific COPD treatment, and only a small proportion will develop severe PH (1% of patients with COPD) [37], which is chronically debilitating for the affected patients. These cases are classified as a severe form of low-prevalence PH. Other patients with rare forms of PH, requiring combined therapeutic strategies, are those with two or more overlapping diseases leading to PH. An example of this is PH with atrial septal defect and venous thromboembolic disease. These cases are classified as overlapping PH.

Diseases of the heart constitute class III according to the clinical classification of RCDD. Cardiomyopathies are divided into 5 groups: dilated cardiomyopathy (DCM) [38], hypertrophic cardiomyopathy (HCM), restrictive cardiomyopathy (RCM), arrhythmogenic right ventricular cardiomyopathy (ARVC), and unclassified cardiomyopathies. Our classification proposes an aetiology-based approach to cardiomyopathies which requires an in-depth diagnostic process until the true aetiology of the cardiomyopathy is revealed.

The complexity of classification, pathology, diagnosis, and optimal management of cardiomyopathies has been extensively reviewed and summarized by leading experts in the field in the form of guidelines, review articles, and reference textbooks. The backbone of contemporary cardiology is evidence-based, rigorously prepared guidelines. The area of cardiomyopathies has been covered by several guidelines issued and endorsed by scientific societies including the European Society of Cardiology (ESC), the American College of Cardiology (ACC), and the American Heart Association (AHA). A widely accepted classification of cardiomyopathies in Europe, although not entirely without doubts, comes from the Working Group on Myocardial and Pericardial Disease of the ESC [39]. Discrepancies between the European and American perspectives on the classification of cardiomyopathies are well reflected in the AHA guidelines [40]. European experts underline the importance of the clinical and imaging (preferably echocardiographic) phenotype of cardiomyopathies, which are a starting point in the classification process. In contrast, American experts, while admitting the role of clinical and morphological features of cardiomyopathies, put more emphasis on genetic factors. HCM was initially covered in a pioneering joint publication by the ESC and ACC [41]. At a later date, the American perspective on HCM was presented in joint guidelines endorsed by the ACC/AHA [42]. Experts from the Working Group on Myocardial and Pericardial Diseases of the ESC published the guidelines on the diagnosis and management of HCM. In these guidelines, an accurate model of sudden cardiac death prediction, which is based on robust data, has been presented [43]. As many patients with cardiomyopathies will eventually develop signs and symptoms of heart failure, a discussion of cardiomyopathies is included in the ESC guidelines on heart failure [44].

In the class of 'rare congenital cardiovascular diseases' (class IV), apart from groups of abnormalities of the position and connection of the heart and vessels, shunts, complex congenital cardiovascular diseases, and congenital cardiovascular diseases with concomitant organ dysfunction, a group of grown-up congenital cardiovascular diseases and others are distinguished. The population of adults with congenital heart diseases is heterogenous and requires a multidisciplinary approach to patient care: 15–20% of these cases are complex, rare, and require life-long expert supervision and/or intervention, 35–40% require expert consultation, while the remaining 40% have simple or cured diseases and need little to no specialised care [45]. Patients who have had surgical therapy for congenital heart defects have a different type of haemodynamics and its consequences. For example, tetralogy of Fallot is classified as a congenital heart disease with decreased pulmonary blood flow. After surgery, pulmonary blood flow is usually normalized, but pulmonary valve regurgitation and arrhythmias are the main problems. In some cases, residual ventricular septal defect is detected and such patients may be classified as having a congenital heart disease with increased pulmonary blood flow.

This should be considered in several perspectives regarding the classification of congenital heart diseases [46–48]. A pathophysiological classification, namely, one which is based on the clinical consequences of structural defects impairing the pathophysiology of blood circulation, is frequently used [49, 50]. Some congenital heart defects may occur on their own, while others may occur as a component of various genetic syndromes such as Down syndrome and Turner syndrome, or with other concomitant diseases [51, 52]. The classification of congenital heart diseases has always been challenging. There are two global systems of classification: ICD-10 created by the World Health Organization and International Paediatric and Congenital Cardiac Code (IPCCC) created by the International Congenital Heart Surgery and Database Project of the European Association for Cardio-Thoracic Surgery and the Society of Thoracic Surgeons with the European Paediatric Cardiac Code of the Association for European Paediatric Cardiology [53]. Houyel et al. [45] proposed their own classification based on the IPCCC, regrouping congenital heart diseases into 10 categories. The International Society for Nomenclature of Paediatric and Congenital Heart Disease established a system of nomenclature for cardiovascular catheterisation in congenital and paediatric cardiac diseases, focusing both on procedural nomenclature and on the nomenclature of complications associated with interventional cardiology [54, 55].

Cardiac tumours are very rare and heterogeneous from a histologic point of view [56]. Here, in the class of 'cardiac tumours and cardiovascular diseases in malignancy' (class V) we propose to distinguish primary cardiac tumours, metastatic cardiac tumours, inflammatory malformations, and cardiovascular complications of oncological therapy. The prevalence of primary cardiac tumours is around 0.02%, which corresponds to 1 in 5000 people [57]. These may be associated with genetic syndromes including Carney complex and tuberous sclerosis [58]. Metastatic cardiac tumours are extremely rare and comprise 15.8% of all cardiac tumours [59]. Potential signs, symptoms, and complications depend mostly on the location of the cardiac tumour [60]. However, differential diagnosis is crucial in proper patient management. We have excluded group 3 (Thrombus within heart chambers) from the current class of cardiac tumours and cardiovascular diseases in malignancy. Intracardiac thrombi are signs of the disease and not a disease in itself. To the best of our knowledge, there are no published reports of lone thrombi within the heart chambers.

Cardiac rhythm disorders affect almost all patients. Most of them are benign in their nature, while others, including some RADA, constitute important medical problems and may influence not only the quality of life of affected patients, but may also lead to cardiovascular compromise or even sudden cardiac death. In class VI of the current classification of RCDD, we provide 2 groups of RADA: Primary electrical disorders of the heart and arrhythmias in specific clinical settings. The first group includes not only cardiac channelopathies, such as Brugada syndrome, long QT syndrome, short QT syndrome, and catecholaminergic polymorphic ventricular tachycardia, [61][62][9, 63–67][68] but also among others, congenital complete heart block. [69] The presence of cardiac arrhythmias in different clinical settings is common. However, we decided not to introduce a group of RADA secondary to or co-existent with RCDD, which could include patients with a predominantly arrhythmogenic clinical picture. The presence of such a group could lead to misclassification of the diseases and disorders and influence registries and later conclusions on epidemiology and morbidity. As an example, a patient with ARVC of an autosomal dominant inheritance pattern, due to plakophilin-2 mutations and frequent arrhythmias should be listed in class III (group number 4) [70]. Arrhythmias in specific clinical settings, among other clinical conditions, include patients with arrhythmias as complications of or related to medical treatment which causes difficulties in diagnostics or management [71–83].

Cardiovascular diseases in pregnancy are rare and affect 0.2–4% of all pregnant women [84]. Pregnant women with heart disease are at increased risk for cardiovascular complications [85]. Importantly, virtually all RCDD may be associated with pregnancy. This class (class VII) of RCDD includes two major categories of patients. The first is a pregnancy in patients with a known rare abnormality, while the second category consists of patients with RCDD first diagnosed during pregnancy [86]. This is of great value, since women with known RCDD may benefit from proper preparation before conception to prevent adverse clinical events [87]. The classification of RCDD in pregnancy is difficult. The ORPHANET classification includes only some diseases related to pregnancy, however, there is no data concerning cardiovascular diseases in pregnancy. The clinical classification of RCDD includes the above-mentioned group of patients, so it supplements the ORPHANET classification and may be very helpful for clinicians.

Due to the heterogeneity of RCDD, similarly to the previous classification, we have also distinguished a class of unclassified rare cardiovascular diseases and disorders (class VIII) [88].

Concluding remarks

It should be highlighted that the presented RCDD classification is not exhaustive. It does not include age, which at the time of diagnosis may largely differ between patients. Moreover, it does not

include problems associated with the foetus and specific aspects of care about elderly patients, including polypharmacy, presence of frailty syndrome and cognitive impairment [89, 90]. Furthermore, we do not discuss social, ethical, and moral issues as well as problems associated with end-of-life care and persistent therapy [91]. These aspects significantly influence the well-being and quality of life of patients, which is important, especially in the treatment of chronic diseases [92].

A detailed classification of diseases and disorders included in consecutive classes will be published in appropriate specialty Journals. At the same time, we encourage readers to provide comments and suggestions to further improve the classification system of RCDD.

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