

Clinical classification of rare cardiovascular diseases and disorders: upcoming 2018 update

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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 and makes the creation of databases easier. It 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 (<http://www.crcd.eu/?p=2800>) and in international journals, including in a recent publication in the *European Heart Journal* [2, 3]. Clinical classification of RCDD takes into account the major clinical symptoms and pathologies and is based on common clinical and/or anatomical features.

Since the first clinical classification of RCDD was published, progress has been made in the diagnostics and treatment methods of RCDD [4]. To update the current classification we performed a search using the Orphanet rare diseases inventory, which includes diseases having a prevalence of 5 per 10 000 in the general population or less [5]. 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 and have included rare cardiovascular disorders in our search [6]. In addition to the International Classification of Diseases and Related Health Problems 10th

Revision (ICD-10) codes [7], classes, groups, subgroups, examples, and RCDD codes in the new version, we plan to include ORPHA numbers of classified diseases in the Orphanet inventory (available at: <http://www.orpha.net/consor/cgi-bin/Disease.php?lng=EN>), Table [5]. Furthermore, we plan to introduce appropriate changes suggested by national and international reviewers and/or readers. The 'rare arrhythmias' class will be changed to the 'rare arrhythmogenic disorders' class due to more appropriate terminology, because it includes a spectrum of diseases and disorders. To preserve the clinical and anatomical context of consecutive classes of RCDD, the current version of the classification will differ with regard to the sequence of classes. The 'rare arrhythmogenic disorders' class will be moved from class V to class VI, while 'cardiac tumors and cardiovascular diseases in malignancy' will be moved from class VI to class V. Moreover, in the updated clinical classification of RCDD, significant modification and extension of the 'rare arrhythmogenic disorders' classification will be provided. Due to the presence of arrhythmias in different clinical settings and to minimize repetitions between and within RCDD classes, a group of secondary or coexisting arrhythmias to other RCDD (classes I-V and VIII) will be introduced.

Table. Upcoming 2018 update of clinical classification of rare cardiovascular diseases and disorders

Class	Group	Subgroup	Examples	RCDD code	ICD-10 code	ORPHA number
I – rare diseases of systemic circulation	1 – 10	A – Z	1–10/a–z	I-(1A).(1a)	see*	see**
II – rare diseases of pulmonary circulation	1 – 10	A – Z	1–10/a–z	II-(1A).(1a)	see*	see**
III – rare diseases of the heart (cardiomyopathies)	1 – 10	A – Z	1–10/a–z	III-(1A).(1a)	see*	see**
IV – rare congenital cardiovascular diseases	1 – 10	A – Z	1–10/a–z	IV-(1A).(1a)	see*	see**
V – cardiac tumors and cardiovascular diseases in malignancy	1 – 10	A – Z	1–10/a–z	V-(1A).(1a)	see*	see**
VI – rare arrhythmogenic disorders	1 – 10	A – Z	1–10/a–z	VI-(1A).(1a) or VI-(class)-(1A).(1a)	see*	see**
VII – cardiovascular diseases in pregnancy	1 – 10	A – Z	1–10/a–z	VII-(class)-(1A).(1a)	see*	see**
VIII – unclassified rare cardiovascular diseases	1 – 10	A – Z	1–10/a–z	VIII-(1A).(1a)	see*	see**

RCDD – rare cardiovascular diseases and disorders; ICD-10 – International Classification of Diseases and Related Health Problems 10th Revision. * A sample code from ICD-10, some RCDD are not referenced in ICD-10. ** ORPHA numbers from Orphanet rare diseases inventory, some RCDD are not referenced in this inventory

Modified classes in the clinical classification of RCDD will include (Table):

- Class I – rare diseases of systemic circulation,
- Class II – rare diseases of pulmonary circulation,
- Class III – rare diseases of the heart (cardiomyopathies),
- Class IV – rare congenital cardiovascular diseases,
- Class V – cardiac tumors and cardiovascular diseases in malignancy,
- Class VI – rare arrhythmogenic disorders,
- Class VII – cardiovascular diseases in pregnancy,
- Class VIII – unclassified rare cardiovascular diseases (for diseases and disorders which cannot be unequivocally classified into any other class).

We encourage readers to provide comments and suggestions to further improve the classification system of RCDD. The full version of the first classification is available at *JRCD* website (<https://jrce.eu/index.php/crcd/article/view/78>) [1]. Publication of the updated version is planned in 2018.

References

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