

The Era of Personalized Medicine in Cardiomyopathies

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Learning about cardiomyopathies has become more in-depth in recent years. Thus, several views of specialists from Brazil are present in this edition of *ABC Heart Failure & Cardiomyopathy*, of the ABC Family of *Arquivos Brasileiros de Cardiologia (ABC Cardiol)*, from the most common etiologies to the rarest and ultra-rare ones, with their systemic repercussions, including toxic, genetic, peripartum, stress, diabetes, collagenosis, cardiogenetic interaction, as well as a vision of the future with artificial intelligence and 3D printing in cardiomyopathies.

Advances in imaging such as coronary computed tomography angiography in the non-invasive diagnosis of coronary artery disease, magnetic resonance imaging and its various specific evaluations of cardiomyopathies, nuclear medicine in microvascular disease and non-invasive diagnosis of transthyretin amyloidosis, and genetic tests have facilitated the recognition of different phenotypes. Moreover, based on the genotype of a given case, an entire family can also be identified related to preexisting clinical syndromes. Interaction between specialties and heart teams have improved care, disease management, and assistance of affected families. Accordingly, follow-up with a geneticist and genetic counseling are imperative in the era of personalized medicine.

Genetic sequencing of patients in the context of advances in cardiac imaging has indicated a new perspective for these concepts. The “MOGES” classification of cardiomyopathies, since 2014, has emphasized extracardiac manifestations, familial inheritance pattern, and genetic etiology.¹ Recent clinical studies have described cohorts with specific variants that show subtypes of different phenotypes. Indeed, in the coming years, new classifications of cardiomyopathies and clinical conduct are plausible. Follow-up by multidisciplinary teams specializing in cardiogenetics demonstrates better communication and management of variants of uncertain significance that may be relevant in the clinical expression of cardiomyopathies and may reclassify them. Furthermore, today we can rely on in vitro techniques with targeted gene identification to assist families with variants.²⁻⁵

Genetics is a useful tool in diagnosis, risk stratification, and family management. Currently, there are guidelines with well-established recommendations regarding the application of

genetics and subtypes of cardiomyopathies, which may lead to new treatment opportunities, in addition to more assertiveness in risk stratification.⁵ Accordingly, there are initiatives such as the Renômica Project, led by Dr. Adriana Carvalho, a professor at the State University of Rio de Janeiro, through the Brazilian National Institute of Cardiology. This is a research program that studies causes of hereditary cardiovascular genetic diseases in the Brazilian population within the Unified Health System (SUS).

Additionally, it is worth mentioning the work undertaken for more than 4 decades of studies through the São Paulo Research Foundation (FAPESP), by Dr. Eduardo Krieger and the entire team of the Instituto do Coração (Incor). Their work concentrates on the development of research activities focused on the identification of molecular markers associated with the genesis of cardiovascular diseases and new therapeutic approaches for cardiac regeneration. These are examples of a collaborative network within the SUS for the advancement of teaching and research in genetics.

At the Antônio Pedro University Hospital of the Faculty of Medicine of the Fluminense Federal University (UFF), there is the Cardiogenetics Outpatient Clinic, led by the geneticist Dr. Raquel Germer and the pediatric cardiologists Dr. Aurea Grippa and Dr. Ana Flávia Malheiros Torbey. Also at UFF, there is a 3D printing project led by Professor Claudio Tinoco Mesquita at the Healthscience laboratory. These are examples of initiatives that collaborate with teaching and research in cardiomyopathies.

From an objective point of view, amyloidosis would be the first cardiomyopathy in this paradigm of personalized medicine. There is clear evidence that barriers have been broken in diagnosis and treatment, from measuring free light chains, positive Congo red in polarized light, and mass spectrometry. From liver transplantation to amyloid tetramer stabilizer, RNA interference, antisense oligonucleotides, and gene editing, there are many ongoing studies and, consequently, prospects for better prognosis and survival.⁶⁻⁸

In this context, specialized centers are needed to monitor these patients with cardiomyopathies. However, it is necessary to consider that research and treatment still require high costs. Particularly, rare diseases warrant special attention and multidisciplinary follow-up. This scenario led to the idea to prepare a book about two rare etiologies of cardiomyopathy, *Amyloidosis and Fabry Disease: A Clinical Guide* published by Springer, with Dr. Diane Ávila and Dr. Humberto Villacorta as editors. This book counted on the participation of leaders of major centers from Brazil and worldwide, with scientific research incentive from the UFF Postgraduate Program in Cardiovascular Sciences.

With all due honor and respect, Professor Claudio Rapezzi, an important scientific collaborator in cardiomyopathies,

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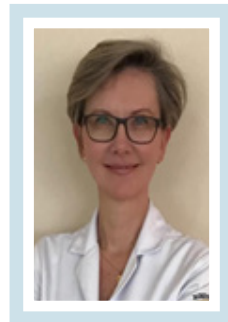
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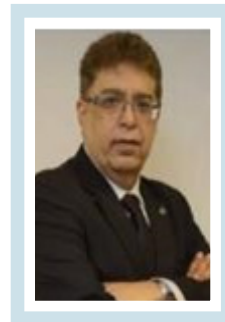
passed away in 2022, having contributed numerous works on cardiogenetics.

On behalf of Dr. Lídia Zytynski Moura, the editor-in-chief of *ABC Heart Failure & Cardiomyopathy*, of Dr. Evandro Tinoco Mesquita and Dr. Marcus Simões, guest editors

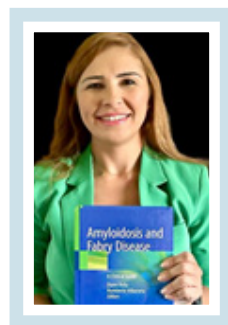
on behalf of the board of the Cardiomyopathies Study Group (GEMIC), we would like to thank all the authors of this edition and Dr. Diane Xavier de Ávila and Dr. Vagner Madrini Junior for their collaboration in putting together this edition.



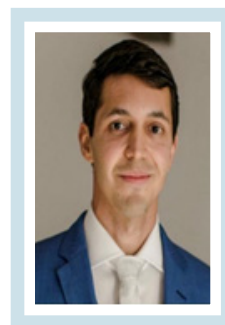
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