

Dear Friends and Colleagues, Dear Readers,

We are pleased to deliver at your hands the second issue of your quarterly *Journal of Rare Cardiovascular Diseases*. The feedback we have received, after launching the *Journal* in October 2012, has convinced us that the *Journal* fills an important gap in the needs of the community of physicians who look after the “unwanted” and “troublesome” cardiovascular patients – the patients with rare cardiovascular diseases (RCD).

Consistent with our editorial policy, the present issue brings a mixture of research, clinical reports, and updates on the events important for RCD Community.

To-date growth differentiation factor 15 (GDF-15) has been known as a marker of developing heart failure and as a risk-stratification tool in patients with heart failure, including those on resynchronization therapy. In this issue of the *Journal*, Professor Mizia-Stec and collaborators present a multi-centre study that indicated, for the first time, that patients after successful repair of aortic coarctation demonstrate lower levels of GDF-15 than age- and gender-matched subjects. This work importantly supplements recent data on potential roles of GDF-15 in risk stratification in patients after surgical correction of congenital heart defects and as a marker of abnormal function of the Fontan circuit in patients with univentricular hearts.

Clinicians always value patient reports, and I would like to encourage you to check this issue of the *Journal* for practical challenges (and some solutions) in a 39 year-old woman with an atypical variant of Klippel-Trenaunay syndrome and progressive thromboembolic pulmonary hypertension (Dr Poreba and colleagues), a 48 year-old lady with partial anomalous pulmonary venous connection (Dr Wrotniak and colleagues), a 29 year-old woman with pseudomyxoma elasticum

(Dr Kanazirev and colleagues), or a middle-aged lady with rapidly progressive idiopathic pulmonary arterial hypertension irresponsive to what has become standard treatment (Dr Drabik and colleagues).

In this issue of the *Journal*, we have also provided space to a maturing draft of the first classification of Rare Cardiovascular Diseases. This classification, published simultaneously in the textbook of rare cardiovascular diseases: “Rare Cardiovascular Diseases: From Classification to Clinical Examples” is currently under a second round of international Experts’ review and the time is now right to share it with the Readers of the *Journal* and ask for your feedback. Rare cardiovascular diseases require multidisciplinary knowledge and multi-specialty diagnostic and therapeutic decisions. As Professor Ségolène Aimé stated in the first issue of the *Journal* “worldwide sharing of information, data, and samples to boost research is currently hampered by the absence of an exhaustive rare diseases classification” (*J Rare Cardiovasc Dis* 2012;1:2). As an attempt to answer this need, the current proposal is aimed at facilitating recognition of RCDs and grouping the expertise in the main fields of RCDs. This proposal, encompassing rare diseases whose major pathological mechanisms affect the cardiovascular system, is not only based on the Centre for rare Cardiovascular Diseases (CRCD) experience of over 300 patients consulted in the years 2006–2013, but it also takes into consideration the majority of publications on RCDs available through PubMed. The Classification, in its present form, cannot be considered a finished piece – and we do look forward to your feedback. All contributions will be acknowledged.

We do hope that you will continue in your interest in the *Journal* and with your support for Authors and the team of Editors. Please feel free to tell us what you

like and what you may not like (if so) in the Journal. Voice your opinion on how we can further improve on rarediseases@szpitaljp2.krakow.pl. Do consider communicating in the *Journal* your own challenges in diagnostic and therapeutic decision-making in an RCD patient from your practice – and share your experience in a Case Report. Or submit an Original Contribution including an analysis of a series of RCD patients, focused on a specific diagnostic or management issue. Or consider combining your experience with the already published data – and submit a review. All these types of scientific communication play an important role in the exchange of thus-far limited information on how to best manage RCD patients. Also, by contributing to the *Journal* you will mark your contribution to the largely abandoned field that is being shaped and organized with new knowledge and new standards.

And last – but not least: we look like to welcome you at the 3rd ESC Symposium on Rare Cardiovascular Diseases at the European Society of Cardiology (September 2nd 2013, Room Vienna). Apart from the opportunity to hear on the hot topics in the field of rare cardiovascular diseases, you will have a unique chance to directly interact with your colleagues who care for the “orphan” RCD patients. Moreover, we are happy to announce that the first textbook of rare cardiovascular diseases: “Rare Cardiovascular Diseases: From Classification to Clinical Examples” will be made available to the ESC Symposium Participants.

Thus see you at the 3rd ESC Symposium on Rare Cardiovascular Diseases in Amsterdam, and in the meantime I look forward to hearing from you via email or the *Journal* webpage!

Piotr Podolec

Editor-in-Chief

Journal of Rare Cardiovascular Diseases