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EDITORIAL

"Join us in making voice of rare diseases heard" motto of the Rare Diseases Day, 29th February 2016

Dear Readers,

Just a few days ago, on 29th February we celebrated for the nine time the Rare Diseases Day. This year the main theme was "Patient Voice" as it is high time to recognize the crucial role of patients, their families and care-givers in the whole process of delivering better care. We all believe that the Patient's Voice is stronger when receiving appropriate training, is vital because rare diseases patients are experts in their disease, and is increasingly present and respected in the medicines regulatory process, during which patients bring real-life perspective to the discussion. Since 2008 in hundreds of cities all over the world, thousands of events dedicated to rare diseases are taking place. Krakow with its two centers for diseases – one in the University Hospital and the other our Centre for Rare Cardiovascular Diseases in John Paull II Hospital are bright spots on the polish map. This hear, we have joined our efforts and together organized the 3rd Conference on Rare Diseases and addressed numerous important topics. However, there ae still many, more to be resolved in the hopefully bright future.

As the long-awaited vernal equinox has finally came this week, we shall eagerly storm out of crowded cities and search for more sun and space. This is always true and repeats for eons. This also means that the first quarter of the year will be finished soon and also that the first issue of the Journal in 2016 has been completed and ready for inspection. The 14th issue of the Journal starts with the Review article on arrhythmias during pregnancy, clinical situations that we are almost always puzzled. The Original article on nitric oxide signaling is so far unique in several years of journal history. Its uniqueness lies in sophisticated methodology used typically in basic studies on cardiac muscle physiology. The center of the Journal occupies four clinical cases presented and commented by the managing teams. The final chapter is a report on the 3rd Conference of Centre of Rare Cardiovascular Diseases that taken place to commemorate the Rare Diseases Day.

The state-of-the-art Review on arrhythmias during pregnancy is written by doctor Sylwia Wisniowska-Smiałek and guided by doctor Agata Lesniak-Sobelga who is a local expert in all pregnancy-related problems. This is really neglected topic by many but yet from time to time everybody is confronted with a scenario of pregnant woman seeking medical attention due to arrhythmias. The authors provide a complete overview on this field and help us with decision-making process. Certainly, not all arrhythmias are truly dangerous and proper management does not mean automatically sending everybody to the tertiary centers. Quite opposite, well-educated and conscious specialist should stratify the patients into those who should be seen by an expert and those who can be treated and observed locally. Otherwise creation of an unnecessary stress on already agitated women and her family alongside with improper utilization of limited resources make more damage than good. This article will definitely help and is a must to read.

The Original article written by doctor Marcin Kunecki and colleagues is an experimental study on the role of nitric oxide in the ischemic post-conditioning of human cardiac muscle. Although basic studies have not been so far explored by the Journal, perhaps a bit of refreshment and look from the different angle will be interesting. The authors using quite complicated laboratory methodology tried to address the hypothesis that ischemic post-conditioning provide cardio-protection in NO-dependent mechanism in the human myocardium. The main conclusion of the paper is that blockade of NO synthesis has negative effects on myocardial function as well as attenuates the beneficial effect of ischemic post-conditioning. Possibly, the readers of the Journal are not used to sophisticated methods taken directly from the laboratory, nevertheless, it is educational to have a short glimpse on basic science.

As has already been mentioned the central place is typically reserved for clinical cases of rare cardiovascular diseases. The first case is authored by doctor Pawel Pro-

chownik and colleagues from our center and is about long-term consequence of urgent percutaneous balloon valvuloplasty due to severe aortic stenosis diagnosed in infancy. The long-term follow-up is presented, which shows gradual deterioration of functional status due to increasing aortic insufficiency and development of secondary pulmonary hypertension. The difficult decisions regarding the optimal patient's management along with informative discussion are clear advantage of this case. Next two cases come from our long-term collaborators in Katowice and both are authored by professor Zbigniew Gąsior and colleagues. From time to time we read with great interest a description of tumors primarily located or metastased to the heart. Authors from Katowice present a patient who had malignant right popliteal fossa tumor that eventually spreads into lungs and left atrium. After difficult and risky operation the tumor of 4 cm in dimeter was successfully removed from the left atrium. The case is accompanied with short discussion and nice CT and echo pictures. The second case from Silesian team is focused on an unexpected misplacement of the atrial pacemaker lead into left atrium through anomalous left subclavian vein draining into left atrium. Such a clinical scenario may be dreadful and potentially dangerous if left undetected. Moreover, it is one of numerous examples how complex and unpredictable can be cardiac anatomy. It is a great lesson that there is not such a thing as a routine procedure and we should always be vigilant. The last case is written by doctor Kamil Bugala and colleagues from our hospital and is about a rare variant of apical obstructive hypertophic cardiomyopathy. Although hypertophic cardiomyopathy is the most frequent form of cardiomyopathy, nevertheless, its apical variant is uncommon in Europe, being much more prevalent in Asian countries. The authors demonstrated long-term follow-up with gradual increase of left ventricular-aortic gradient, which was negligible 15 years ago but rose up to 90 mmHg in recent examination. This is truly interesting and worrisome as the reason for the gradient is mid-ventricular obstruction that is not amenable to invasive gradient-reduction therapy.

This issue of the Journal is closed by the report, written by doctor Natalia Dłużniewska and co-authors, from the 3rd Conference of Centre of Rare Cardiovascular Diseases. The crucial aspect of the Conference were combined discussions and brain-storms with government and National Health Fund representatives. This is the only way forward as without formal help from the governing institution, the progress in the rare diseases area is not possible.

We hope that the current issue of the Journal will be truly interesting and worth-reading.

Piotr Podolec
Editor-in-Chief
Journal of Rare Cardiovascular Diseases