

# “A rare day to put rare diseases in the spotlight!” “With research possibilities are limitless”

## Mottos of the Rare Disease Day

### Dear Readers,

Just few days ago, on 28<sup>th</sup> February, the tenth edition of Rare Disease Day was launched by EURORDIS and its Council for National Alliances to attract thousands people all over the world to come together to advocate for more research on rare diseases. Rare Disease Day goes globally, where all stakeholders, including patients and families, patient organizations, politicians, carers, medical professionals, researchers and industry come together to raise awareness of rare diseases through thousands of events all over the world. Among many topics, patients' involvement in research is getting momentum. Thanks to patients' actions, such as advocacy for research, funding research, governance of research, and last but not least participate in research, actually resulted in more research. It has been numerously underlined that research in rare diseases is a key factor as it brings hope to the millions of people living with rare disease across the world and their families. There are numerous types of research and frameworks that are important to rare disease community, including: infrastructures such as registries (databases of patients with their clinical and genetic information) and biobanks (catalogues of human biological samples), basic research to identify the cause and mechanisms of rare diseases, translational research, which focuses on using the conclusions of basic research to develop therapeutics for patients living with a rare disease, clinical research, when medicinal products are tested in humans through clinical trials, and finally research into quality of life, working conditions, social needs, integration at school, as well as multidisciplinary education of social service providers. Hopefully, all those noble initiatives will move the field forward for the good of all patients and their families.

In this first issue of the Journal in 2017, we start with an interesting Review on cardiac sarcoidosis. The Original paper comes from the study group involving several centers, specializing in the treatment of pulmonary

hypertension (PH) and is about non-invasive assessment of hemodynamics in PH. Then, as usual, we have several clinical cases of rare cardiovascular diseases with informative discussions and overview of the problems. Finally, there is a report from the annual meeting of the European Association of Cardiovascular Imaging (EACVI) – EuroEcho.

The Review article is on an important but frequently forgotten and neglected problem of cardiac manifestations during the course systemic sarcoidosis. Although symptomatic cardiac involvement is relatively rare as it occurs in approximately 5% of patients with systemic sarcoidosis, nevertheless, asymptomatic cardiac damage can be as high as in 25% of patients. The authors provide detailed diagnostic criteria of cardiac sarcoidosis alongside with diagnostic flow-chart that can may prove particularly helpful for the managing physicians. Further, a nice overview of sarcoidosis-specific treatment is presented, that may be interesting for cardiologists who are rather unfamiliar with those modes of treatment. Finally, the authors also comment on cardiac-specific manifestations of sarcoidosis and provide in-details description of optimal drug management strategies and as well as invasive procedures.

Doctor Remigiusz Kazimierczyk and colleagues from Białystok, Lublin and Łódź present results of the pilot study on role of electrical cardiometry in non-invasive assessment of hemodynamics in patients with PH. Relying on measurements of the thoracic electrical bio-impedance, the authors estimated cardiac output, cardiac index as well as fluid status in patients with PH. They enrolled 23 patients and found significant correlations between invasive and bio-impedance based measures of cardiac hemodynamics. The authors concluded that although thoracic bio-impedance is not going to replace right heart catheterization, which is still the gold standard of hemodynamics assessment, nevertheless

with this new technique it is feasible to obtain accurate results of cardiac output. Perhaps, not ready yet for routine clinical utilization, however, this technique being non-invasive and relatively simple is a promising tool for the assessment of pulmonary circulation at the bedside or even in the outpatient care.

As has been numerously reiterated, the backbone of the journal is constituted of rare cardiovascular diseases cases. The first case is authored by doctor Agnieszka Sarnecka and colleagues from our center and is about unique trans-catheter closure of peri-membranous ventricular septal defect (VSD) in patient after previous primum atrial septal defect closure, prosthetic aortic and mitral valves replacement. The managing team percutaneously closed VSD through the aortic prosthetic valve with AMPLATZER™ VSD Occluder. The authors present a nice overview of the problem in the Discussion section. The next case, presented by doctor Monika Różewicz-Juraszek et al. from Warsaw, describes the history of adult women with systemic mastocytosis that eventually ended with surgical aortic valve replacement. Initially, the managing team after the diagnosis of aortic stenosis during the course of mastocytosis, aimed for conservative approach. Obviously, the main worry was a spontaneous degranulation of mast cells and an anaphylactic reaction, that

could be triggered by severe stress related to surgical procedure. However, after two years the patient became symptomatic and she was referred to surgery. Importantly, the authors present in-detail a perioperative protocol based on administration of steroids and other medications, that allowed for an uneventful and successful operation. Doctor Mariusz Gierba from the hospital in Nowy Targ with the cooperation of colleagues from our center, describe the patient with clinically silent, right and left ventricular outflow tract obstructions. The authors present very informative facts about the coexistence of those two anomalies and possible methods of treatment. Moreover, nice figures depicting the pathologies are very helpful to understand the pathologies and challenges of treatment.

The last part of this issue is the report, written by doctor Sylwia Wiśniowska-Śmiałek, who actively participated in the EuroEcho meeting, organized by EACVI, in December 2016 in Leipzig, Germany. As always, EuroEcho attracts world-wide specialist in cardiovascular imaging and is a place where cutting-edge technologies meet with clinics.

We sincerely hope that as in previous years, the next issue of the Journal will be a help in navigating uncharted waters of rare diseases.

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