

Journal of Rare Cardiovascular Diseases: “Conquering new territories” – a new educational program on rare diseases for medical acolytes

Dear Readers,

Nowadays, we witness dramatic changes in the perception of rare diseases. Thanks to the European Union (EU) regulations but more importantly due to increasing pressure from the spontaneous patients’ organizations the Rare Cardiovascular Disease (RCD) area of medicine is quickly evolving. It is no longer good will or gestures of politicians but rather compliance with the EU regulations that shape the landscape of rare diseases in Poland. Moreover, the awareness and education on rare diseases will be implemented in the medical students curriculum. Here, in Krakow we are in the forefront in the education process of young medical acolytes as we have successfully prepared multi-disciplinary course on rare diseases together with the Professor Tomasz Grodzicki, Dean of the Faculty of Medicine Jagiellonian University and numerous Partners and Friends from other departments of Jagiellonian University Medical College, such as from Neurology, Hematology, Gastrology, and many others. Furthermore, we will soon start formal classes with students and will have a chance to verify “theory in practice”. This are exciting times for all involved in rare diseases and we believe that this Journal has a chance to become the “tribune” for those positive changes...

Continuing with the tradition, we start with the Review article entitled. This work comes from Dr Anna Tyrka supervised by Dr Grzegorz Kopeć; both are very active in our RCD Center pulmonary hypertension (PH) team. In their article the authors give us an interesting overview on quality of life (QoL) studies. They underline that typical parameters (end-points) poorly correlate with patient’s personal benefit. Therefore, the novel approach to many diseases and particularly to chronic ones, as is PH, is to use patient-reported outcomes (PROs) that provide patient-centered rather than investigator-centered measurements. Lastly, they elaborate a little on the only PH-specific QoL questionnaire – CAMPHOR which has been proven to correlate

with 6-minute walking test and as well as other commonly used QoL questionnaire – SF-36.

The original paper comes from Dr Agata Lesniak-Sobelga who has been for years an Expert in the management of pregnant women with cardiac problems. This time Dr Lesniak-Sobelga and co-workers published a longitudinal, observational study on pregnant women with bicuspid aortic valve (BAV). Over 26 years they have treated an impressive number of 89 BAV pregnant. The main conclusion is that cardiac complications can be expected in women with moderate-to-severe aortic stenosis or severe aortic insufficiency on the basis of BAV. Reassuringly, all women delivered healthy baby and not a single patient had pregnancy-associated aortic dissection.

As in the previous issues of the Journal, the third part is composed of four exceptional clinical cases, this time exclusively on myocardial and pericardial diseases. We start with the paper from our long-term Partners from Lithuania on the management of middle-aged man with Fabry disease. Doctor Plisine, supervised by Dr Egle Ereminiene, provide an interesting description of diagnostic methods and treatment, enriched with pictures of angiokeratomas, they used to manage their patient with Fabry disease. Next, we have two papers from our group specialized in myocardial diseases. In their first case dr Pawel Rubis and colleagues describe extremely difficult case of an adolescent with severe dilated cardiomyopathy (DCM) on the basis of Duchenne muscular dystrophy. The authors should be particularly credited for very detailed and educational overview of this niche problem. This part is worth-reading both for young adepts and experienced physicians. The second case from this group touches equally difficult and rare problem – the transition of compensated hypertrophic cardiomyopathy towards end-stage or burn-out phase. They describe the patient in critical state with left ven-

tricular ejection fraction of 15% whom previous measurements were supra-normal. Those two cases are also important from the other reason which is the necessity for exhaustive work-up of all cases with DCM as in one case the true reason for DCM was Duchenne muscular dystrophy and in the second – hypertrophic cardiomyopathy. The last forth case, authored by dr Barbara Widlińska and co-workers, is about elderly patient with accidentally diagnosed large mass in the pericardium. Surprisingly, despite large size of the pericardial tumor the patient was asymptomatic. High-quality pictures illustrating a huge pericardial mass provide an additional strong point of this paper.

The final part of this issue is a report, written by Dr Pawel Rubis, from the 2nd International Conference on Rare Cardiovascular Diseases that was organized by the Krakow CRCDD between 16th and 17th October 2014 at the John Paul II Hospital premises. The event attracted numerous guests from European and national RCD centers and showed once more that direct meetings of experts in the field provide an important supplement to the Journal and RDC teleconferences.

As usual, this is only a glimpse of the real content of the current issue. Of value for the Readers and their Patients. This is what "it's all about". Isn't it?

Piotr Podolec

Editor-in-Chief

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