

Journal of Rare Cardiovascular Diseases: *All that glitters is not gold.* Randomized Controlled Trails versus real-life patients: Challenges of a maturing Journal

Dear Readers,

Is time friend or foe? No one has the right answer. With this issue, we are celebrating an important landmark in the Journal's history. On this occasion, we have looked back at our first two years with a pinch of nostalgia. Before I move to encouraging you to look into the content of the 8th issue, let me first of all thank you sincerely for your support expressed not only in words spoken when we are fortunate to get to see some of You, Our Readers, but also in writing and – most importantly – expressed by the fact our your continued (and expanding) submissions to the Journal.

The 8th issue marks a steady pace of the progressive growth of the Journal. With rare cardiovascular disease (RCD) patients, by the nature of the disease, we will not get much help from the randomized control trails (RCTs) that we so used to in contemporary cardiology. RCD patients are either absent or greatly under-represented or in RCTs and yet someone has to take care of them (and this 'someone' should be able to do it well)...

As evidenced in the Journal of rare Cardiovascular Diseases over the last two years, the RCD patients tend to be far more complex than those who get evaluated and serve as a basis for level-one evidence analyzes. The RCT evidence, therefore, is poorly applicable to our real-world patients with rare cardiovascular disease. Therefore an exchange of expert views seems to be the right path to making educated and "best" diagnostic and therapeutic decisions in RCD Patients. The Journal provides a unique platform for such information exchange and mutual education.

The issue begins with a review on Cardiac Tumors prepared jointly by the Krakow group under the lead of our Krakow expert Dr. Monika Komar and Italian Expert Dr Giovanni La Cana with whom the Krakow

RCD Center closely collaborates. The authors provide robust and up-to-date data on the epidemiology, classification, pathology, clinics as well as diagnostic and therapeutic procedures. What comes, to me, as the baseline message from this work is that every cardiac tumor is unique and the management strategy may be completely different in the apparently similar tumors. I believe it is a 'must-read'.

Another article – important on several levels – comes from the war-torn Libya, where brave people like doctor Olivier De Neini maintain their drive to carry on with high-quality research. Doctor De Neini studied hereditary hemorrhagic telangiectasia (HHT) also known as Osler-Weber-Rendu disease. Using written enquirers he precisely defined the prevalence and annual incidence of HHT in the population of Benghazi, Libya. The main result of the study is the HHT is far more prevalent in the Arabic population in comparison to historically reference population from Northern Europe. Perhaps, the main explanation is consanguinity still common in traditional Arabic communities.

In this issue, there are also four exceptional (what is more 'typical' that 'exception' in RCD?) clinical cases that I would like to draw your attention to. Dr. Jakub Stępniewski and colleagues present complicated history of a young adult with the common truncus arteriosus. Following exhaustive diagnostic work-up, the managing physicians came to the conclusion that the best approach to this patient will be close follow-up. Would you agree with their argumentation? The second case report comes also from the area of congenital heart diseases and is authored by Dr Monika Smas-Suska supervised by Dr Lidia Tomkiewicz-Pająk who is a local leader in this subject. They present in great detail the history of adolescent who was diagnosed with

the Tetralogy of Fallot and subsequently underwent numerous surgical corrections. During comprehensive diagnostic work-up it turned out that apart from several cardiac abnormalities, this patient also presents a serious coronary anomaly: a single artery originated from the left sinus of Valsalva and giving right coronary artery and later during its course bifurcates into left anterior descending and circumflex artery. Undoubtedly, several high-quality pictures of various cardiac pathologies shed extra light on this complex case. The third case, authored by Dr Pawel Rubis and colleagues, moves away from congenital heart diseases towards myocardial diseases. The authors present a two generation family with the Fabry disease. The affected member of the family are father and surprisingly his daughter, which is highly unusual as typically female are only carriers of the mutated GLA gene as the disease is chromosome X-transmitted. At the end the authors provide an interesting and up-to-date overview on various aspects of Fabry disease, which is worth reading, especially for younger readers. The last case comes from the group specialized in pulmonary hypertension (PH), who this time tackled the so far unresolved problem of PH-related to end-stage left ventricular systolic failure. As their seriously ill patient had been turned down from heart transplantation (HTX)

due to severe and fixed secondary PH, as a last resort they started him on oral Sildenafil. The authors provide robust evidence on the clinical and hemodynamic improvement of this patient during follow-up, which is reassuring and a good news for all those patients who are not candidates for HTX.

The issue is closed with a report on the 4th Symposium on Rare Cardiovascular Diseases that was organized once more by our RCD Center during European Society of Cardiology Congress 2014 in Barcelona, Spain. The report is by a Chairman of the Symposium, Dr Pawel Rubis. The Keynote Lecture, entitled "Coronary anomalies and the risk of sudden death" was delivered by internationally renown Professor Gary Webb (Cincinnati, Ohio, US), uniquely an Expert in both pediatric and adult rare cardiovascular diseases. The high turn-up of participants confirmed the unmet need for such focused meetings on an international level.

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The Editorial Team and I remain yours,

Piotr Podolec

Editor-in-Chief

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