

2nd Symposium on Rare Cardiovascular Diseases – ESC Munich 2012: Chairperson’s Perspective

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In response to interest generated by the 1st Satellite Symposium on Rare Cardiovascular Diseases (Paris, ESC 2011), a session devoted to managing patients with rare cardiovascular diseases was organized this year again, at the annual congress of the ESC, by the team of the Krakow Centre for Rare Cardiovascular Diseases.

The 2nd Satellite Symposium on Rare Cardiovascular Diseases, part of the official Programme of the 2012 ESC Congress, was held in the ESC Munich Messe (Berlin Hall) on 28 August 2012. The session, moderated by Professor John GF Cleland (Kingston-upon-Hull, UK) and Dr Piotr Musiałek (Krakow, PL), was opened by the Director of the Centre for Rare Cardiovascular Diseases in Krakow, Professor Piotr Podolec. Professor Podolec acknowledged the crucial role of the EU Project MRPO 08.02.00-12-424/10 in establishing the Krakow Centre and in making it possible to organize the Symposium. He briefly characterized the key activities of the Krakow Center for Rare Cardiovascular Diseases in 2011–2012. The key focus has been education and patient tele-consultations within a newly-established network that brings together the Krakow Centre’s physicians, the Partner Centers and the leading European experts in various aspects of rare cardiovascular disease. Introduction of regular, structured tele-consultations enabled to determine an optimal management for a significant series of “orphan” patients – in the context of best knowledge and expertise, taking into consideration local feasibilities in Partner Centers. Professor Podolec also characterized the registries coordinated

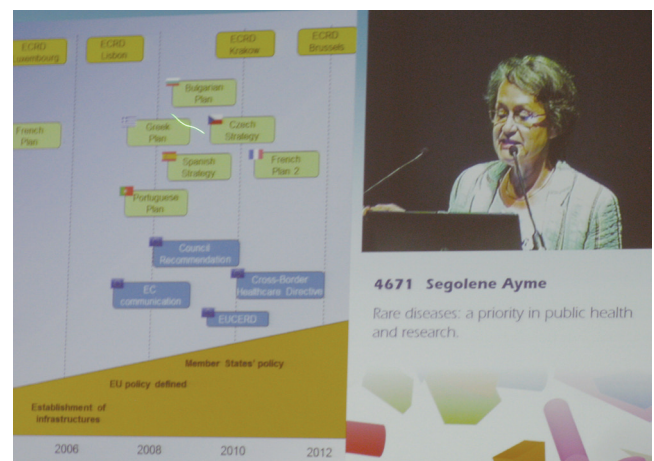
by the Krakow Centre including in particular pulmonary hypertension, grown-up congenital heart disease, rare cardiomyopathies and rare cardiac rhythm abnormalities patients. Professor Podolec announced the first issue of a newly-launched peer-reviewed *Journal of Rare Cardiovascular Diseases*, and concluded with inviting the participants of the symposium to the First International Conference on Rare Cardiovascular Diseases – “Raising Awareness” (18–19 October 2012, Krakow, Poland).

The Keynote Lecture entitled ‘Rare Diseases: a Priority in Public Health and Research’ was given by Professor Ségolène Aymé, ORPHANET Director and Head of the European Union Committee of Experts on Rare Diseases (EUCERD). Professor Aymé explained why managing patients with rare diseases has become a burning medical and public health problem that is currently listed within top EU priorities. She also described the (greatly varying) current situation in the EU member states with respect to rare diseases support structures and patient access to diagnostic tests and expert care. Professor Aymé acknowledged the role of the Krakow Center for Rare Cardiovascular Diseases as a leading center officially recognized by ORPHANET.

The Symposium included two case-based debates centered on determining optimal treatment steps (and their sequence) in a pregnant patient with Turner syndrome and an aortic aneurysm, and in a ‘rare-within-rare’ (to use Professor Nazzareno Galié’s words) patient with pulmonary hypertension causing important diagnostic and



Professor P. Podolec (far right) opened the Symposium and briefly described the key activities of the Krakow CRCD, including coordination of several registries. The session was moderated by Professor JGF Cleland (Hull) and Dr P. Musiałek (Krakow)



Professor S. Aymé (Paris), ORPHANET and EUCERD Director, in the symposium’s Keynote Lecture explained why rare diseases have become a top priority in EU public health and research, and acknowledged a leading role of the Krakow CRCD



Dr L. Tomkiewicz-Pajak (Krakow) proposing stage-by-stage management for a pregnant patient with Turner syndrome and a large aortic aneurysm. Professors P. Hoffman (Warszawa), Z. Gasior (Katowice), and P. Podolec (Krakow) during a break; in the back Dr G. Kopec (left) and Dr P. Rubis (right)

therapeutic challenges (presenters: Dr Lidia Tomkiewicz-Pajak and Dr Grzegorz Kopec, Krakow, discussants: Professor Ségolène Aymé, Paris, Professor Eloisa Arbustini, Pavia, Professor Nazzareno Galié, Bologna, Professor Zbigniew Gasior, Katowice, Professor Piotr Hoffman, Warszawa, and Dr Pawel Rubis, Krakow). The vivid discussion that followed illustrated the upcoming role for strengthened expert networks in determining the most appropriate diagnostic tests and optimal management in rare disease patients. The currently existing inter-country differences (causing a geographic 'luck' ...or 'lack of luck' for a particular patient in access to diagnostic and therapeutic modalities and expert care) became yet again very apparent. Importantly, the discussion demonstrated the necessity to fill many important gaps in existing knowledge; a step that requires setting up common, well-controlled multi-center registries and therapeutic programs. This led to a debate on the role of industry and EU research structures in funding programs devoted to patients with rare cardiovascular disease. In relation to the data presented earlier by Professor Aymé, Professor Cleland raised the issue of a 'desired' balance between the number of Rare Disease Centers (per country? per million population? disease-specific?) and the quality of care related to experience in managing particular group of patients. The debate, going beyond the scheduled programme of the symposium (feasible because of some extra time secured in advance), brought Professor S. Aymé back to the podium. Professor Aymé concluded the debate by stressing the role of preparing high-quality multi-centre grant applications that can successfully bid for EU research funds.

The final presentation of the symposium (Dr Jakub Podolec, Krakow) encouraged everyone to become a frequenter at the Centre for Rare Cardiovascular Disease webpage (www.crcd.eu) and highlighted the role of information technology in networking the rare disease centers and experts. This was bridged to the key aims of the *Journal of Rare Cardiovascular Diseases* – a platform for communicating research and exchanging experience in managing the 'unwanted' patients.

The session, with the lecture hall found nearly too small to accommodate the high turn-up of participants, was attended by 237 doctors from 19 countries including not only Europe but also Australia, India,



Professor N. Galié (Bologna) in a discussion with Professor S. Aymé from Paris (left) and Professor E. Arbustini from Pavia (right) on management steps in a patient with a rare form of pulmonary hypertension; this led to realization that more data are desperately awaited – a need that can be met only through well-coordinated registries and common research programs

South Africa, Brazil and USA. The Symposium had true participants rather than just 'audience'; the Participants were extremely focused and interactive, clearly united through a common goal. After official closing of the session, as many participants stayed in the room to exchange contact addresses and join *ad hoc* interactions and discussions.

In conclusion, the content of the session and the level of participation highlighted the importance of the subject of rare cardiovascular disease in today's cardiology and the burning need to find the right diagnostic and therapeutic answers for the "unwanted" patients. Congratulations on the concept of the Symposium – and many thanks to all those who worked hard to organize the session and to make it a success. A motto of the Krakow Rare Cardiovascular Disease Conference that follows the Munich ESC symposium is 'Because fear is driven by lack of knowledge'.

Good luck then – for 2013 and beyond – in breaking the existing barriers and erasing the fear by filling knowledge gaps through a common endeavour!



The Session was attended by 237 doctors from 19 countries including not only Europe but also Australia, India, South Africa, Brazil, and USA. This high turn-up of participants made the Berlin Hall almost too small to house the Symposium

Photographs by Dr B. Sobien (CRCD, Krakow)