Like in previous years, also this year the Krakow Center for Rare Cardiovascular Diseases organized the 4th Satellite Symposium during the ESC Congress in sunny Barcelona, Spain. This consecutive endeavor was only possible because of the EU Project MRPO 08.02.00-12-424/10, which is under realization by the Krakow Center for Rare Cardiovascular Diseases. Our session on rare cardiovascular diseases has gradually matured over the years and as in the case of last two ESC Congresses in Munich and Amsterdam, was officially included in the Program of the Congress. The Symposium took place on Sunday, 31st of August in the premiere lunch time between 1 p.m. till 1:45 p.m. in the Cairo room. It was planned that our long-term Partner Professor Sabine Pankuweit from Marburg, Germany, the international Expert and author of the guidelines on myocarditis, co-chair the session. However, just before the Congress an acute flu-like infection stopped her from going to the ESC. Fortunately, the inventor of the famous “Alfieri stitch” – Professor Ottavio Alfieri from Milan, Italy kindly agreed to substitute the missing Chairmen. The second chairmen was Doctor Pawel Rubis from the Krakow Center for Rare Cardiovascular Diseases, Poland.

The session program was organized according to the same principles that worked out during recent Congresses. In short, after welcoming and introductory lecture, was a main topic lecture, and finally we had two presentations of rare clinical cases that were presented by the managing physician and commented by two experts. Naturally, the session was opened by the Director of the Krakow Center for Rare Cardiovascular Diseases, Professor Piotr Podolec. This year Professor Podolec presented data on the registries that are currently underway in the Krakow Centre. Moreover, he stressed the usefulness of the newly developed Classification of Rare Cardiovascular Diseases, which was presented last year, and proved to be the “tool” we needed to conduct those registries. On his final slides Professor Podolec presented the impressive number of patients with rare diseases that were included in the registries, e. g. in the registry of rare diseases of the pulmonary circulation (class II according to CRCD Classification) there are 223 patients, in the registry of congenital diseases (class IV) are 421 patients, whereas in the pregnancy registry (class VII) are 25 women with newly discovered significant heart problems. Although the Centre has already registered the substantial number of rare cases, Professor Podolec stressed that this is just a beginning and that we will carry on with recruiting more patients. As in previous years, he warmly encouraged all interested in the rare cardiovascular diseases to join our network for the sake of all those “neglected” patients.

The Keynote Lecture, entitled “Coronary anomalies and the risk of sudden death” was delivered by the Professor Gary Webb, uniquely an Expert in both pediatric and adult rare cardiovascular diseases, from Cincinnati, US. In the beginning Professor Webb defined cor-
onary anomalies into following categories: abnormal origin of coronary artery, which he subdivided into left or right coronary artery arising from pulmonary trunk, coronary fistulas, anomalous origin of coronary artery, being left artery from right aortic sinus (ALCA) and vice versa right artery from left sinus (ARCA), and lastly anomalous course of coronary artery. Anomalous origin of coronary artery is present in 0.2% of the general population and ARCA is more common but ALCA is more dangerous. Professor Webb went on that coronary anomalies are responsible for approximately 20% of sudden cardiac deaths (SCD) in young athletes. Later, Professor Webb hypothesized on the mechanisms of death in coronary anomalies and he provided a few explanations, being acute angle take-off of the artery, abrupt closure of the slit-like coronary orifice, compression of the anomalous artery between aorta and pulmonary trunk, and spasm of the artery. Professor Webb commented on the landmark paper on this subject by Basso and colleges, who identified 27 SCDs due to coronary anomalies in athletes from US and Italy. Great majority (23 cases) were because of ALCA and only 4 due to ARCA. Moreover, 25 of deaths were during intense exertion and remaining 2 just after strenuous exertion. Going into practical aspects of the management strategies, Professor Webb reported than less than one-third of patients have premonitory symptoms, and standard as well as exercise ECGs are seldom abnormal. Finally, he concluded that patients with wrong sinus coronary malformation, less than 35 years old who are symptomatic and have evidence of myocardial ischemia should be operated, whereas in case of those who are asymptomatic and have no evidence of ischemia the decision regarding operation is far less difficult and uncertain. Surely, the surgery should be abandoned in those above 35 years with no symptoms and no evidence of ischemia.

After this fascinating lecture, we proceeded with two case-based presentations on rare diseases and on the optimal management strategy. The first case on the young adult with massive tumor located in interventricular septum was presented by Doctor Jakub Stepniewski from Krakow Centre. The tumor was diagnosed right after the birth due to abrupt onset of ventricular arrhythmia, which was successfully terminated with Flecaïnide, and was regularly observed for the next 22 years. At present, the patient started complaining about palpitations but overall her clinical status is good and she does not have any limitation of exercise tolerance. However, the regularly performed echocardiograms revealed gradual increase in tumor size, which is now 30 × 35 mm. At the end of his presentation Doctor Stepniewski asked the most important question whether the patient should be operated or not. Certainly, there is no better Expert than Professor Ottavio Alfieri, the brilliant cardio-surgeon, who tried to tackle this problem. In the beginning, Professor Alfieri said that it is patient's symptoms that should be primarily taken into account whether to proceed with the surgery. If the patient is symptomatic and the tumor is resectable, the operation is recommended. This scenario should probably be applied also to those patients who are oligo- or asymptomatic but the weight of evidence is smaller. However, if the tumor is not resectable, as is probably in our patient as it occupies the whole septum, the decision is far more complex. The other issue, mentioned by Professor Alfieri, is good clinical status of the patient which favors rather conservative treatment. Given the fact predisposition to ventricular arrhythmias, the patient should be probably implanted with cardioverter-defibrillator (ICD). The second Speaker was Doctor Jakub Podolec from Krakow Centre who presented an overview of cardiac tumors and mode of treatments. He underlined that cardiac tumors are indeed rare diseases with the incidence of 0.02%. Great majority of tumors are myxomas, which represent 70% of all tumors. Interestingly, it seems that annual incidence of tumors has risen over last two-three decades. The historical approach to the cardiac tumors that divides them into benign and malignant have some faults, nevertheless, it may be useful on the daily basis. At autopsy studies, more than three-quarter of tumors are benign with high prevalence of myxomas, whereas the remaining are malignant tumors with angiosarcomas or rhabdomyosarcomas being the most common.

The second case on the complicated management of adult patient with single ventricle was presented by Doctor Lidia Tomkiewicz-Pająk. Her patient was diagnosed in childhood with single ventricle with residual inter-ventricular septum and sub-valvular pulmonary stenosis and at that time her parents declined the surgical operation. Over the years her clinical status has gradually deteriorated and at present she is NYHA class II, hypoxic.
(SpO2 89% on air), and with central cyanosis. Both imaging and invasive studies confirmed severe sub-valvular pulmonary stenosis with the right ventricular pressure almost equal to systemic pressure. Everyone clearly acknowledges that this is extremely difficult patient to manage, therefore, we asked for the opinions of our Experts. Doctor Eva Delmo-Walter started to consider if there is any possibility for surgical treatment for this patient. Firstly, the Speaker identified several critical targets of surgery, which are preservation of systolic and diastolic function, preservation of valvular function as well as optimization of pulmonary circulation. Later, on the diagram, Doctor Delmo-Walter depicted possible surgical interventions, which were sub-classified into definitive and palliative. Doctor Delmo-Walter went on to discuss the Fontan operation which is never an ideal method of treatment and in most cases result in developing heart failure symptoms or protein-losing enteropathy over the years. Leaving aside Fontan tract, Doctor Delmo-Walter focused on other surgical methods, such as ventricular septation and ventricular assist devices in which Berlin Centre has enormous experience. In summary, she concluded that this patient is a candidate for ventricular septation with some residual fenestration and relief of sub-valvular pulmonary stenosis, which hopefully should be a definitive treatment. The final presentation was delivered by Doctor Grzegorz Kopeć who discussed the non-surgical options of treatment. Doctor Kopeć started that whatever methods of treatment are to be chosen, the patient should be enlisted for elective heart transplantation as she may rapidly deteriorates. In the meantime, she should be conventionally managed with classic, heart failure approved, triple neuro-hormonal blockade of beta-blocker, ACE-I and mineralocorticoid antagonists, however, he stressed that solid evidence exists only in patients with systolic dysfunction. Lastly, Doctor Kopeć mentioned about device therapy of ICD and cardiac resynchronization therapy which might be considered in this patient.

Like in previous sessions, also this time the lecture hall was too small to accommodate the high turn-up of participants from all sides of the world interested in rare cardiovascular diseases. Unfortunately, for the sake of time the discussions were very short, otherwise we could have spent next few hours tried to find solutions for those difficult cases. However, many participants stayed long outside the room to carry on with the ad hoc debates and discussions.

In summary, this event was both interesting and educational and a high turn-up of participants confirmed the necessity of such meetings. Far from Congress’s highlights and pivotal trails, recruiting thousands of patients, we proved once again that rare cardiovascular diseases can be equally fascinating and important. Sometimes not the number of studied patients or millions spend on large trails but in-depth knowledge and dedication to patients are crucial factors for success. Ongoing discussions after the session with all those interested in this topic, many of whom took part in this meeting for a fourth time, is a perfect example that it was not a waste of time. Today, once called “orphan” diseases community gained more friends …
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Journal of Rare Cardiovascular Diseases (JRCD) is an international, quarterly, peer reviewed journal that keeps cardiologists up to date with rare disorders of heart and vessels. Topics covered include congenital heart defects, cardiomyopathies, rhythm abnormalities, rare forms of arterial hypertension, pulmonary hypertension, cardiac tumors and other rare diseases affecting heart and vessels such as connective tissue diseases, metabolic disorders, neuro-muscular diseases another unclassified rare diseases.

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