Orphan
RARE DISEASES POLAND
Polish National Forum for rare diseases therapy

EUROPLAN
European Project for Rare Diseases National Plans Development

POLAND

EUROPLAN NATIONAL CONFERENCE

in the framework of the EU Joint Action RD-ACTION

Warsaw, 14 December 2017

FINAL REPORT

RD ACTION
DATA AND POLICIES FOR RARE DISEASES

Co-funded by
the Health Programme
of the European Union

EURORDIS
RARE DISEASES EUROPE

FOREWORD

The EUROPLAN national conferences or workshops are organised in many European countries as part of a coordinated and joint European effort to foster the development of comprehensive National Plans or Strategies for Rare Diseases addressing the unmet needs of patients living with a rare disease in Europe.

These National Plans and Strategies are intended to implement concrete national measures in key areas from research to codification of rare diseases, diagnosis, care and treatments as well as adapted social services for rare disease patients while integrating EU policies.

The EUROPLAN national conferences/ workshops are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS–Rare Diseases Europe. **Rare Disease National Alliances and Patient Organisations have a crucial role to shape the national policies for rare diseases.**

The strength of EUROPLAN national conference/ workshop lies in its shared philosophy and format:

- **Patient-led**: National Alliances are in the best position to address patients' needs;
- **Multi-stakeholders**: National Alliances ensure to invite all stakeholders involved for a broad debate;
- **Integrating both the national and European approach to rare disease policy;**
- **Being part of an overarching European action** (project or Joint Action) that provides the legitimacy and the framework for the organisation of EUROPLAN national conferences/workshops;
- **Helping national authorities adhere to the obligations stemming from the Council Recommendation of 8 June 2009 on an action in the field of rare diseases.**

Since 2008, National Alliances and EURORDIS have been involved in promoting the adoption and implementation of National Plans and Strategies for rare diseases. Altogether, 41 EUROPLAN national conferences took place in the framework of the first EUROPLAN project (2008-2011) and the EU Joint Action of the European Committee of Experts on Rare Diseases – EUCERD - (2012-2015).

Within RD-ACTION (2015-2018), the second EU Joint Action for rare diseases, National Alliances and EURORDIS continue to get involved in a coordinated European effort to advocate for and promote integrated national policy measures that have an impact on the lives of people living with rare diseases.

The EUROPLAN national conferences or workshops taking place within RD-ACTION focus on specific themes identified bythe National Alliances as the most pressing priorities to tackle with national authorities. These thematic priorities are addressed in sessions where all the stakeholders discuss relevant measures to be taken or ways to sustain the full implementation of already approved measures.

Each National Alliance prepares a final report on the national workshop, based on a common format such as the one that follows.

GENERAL INFORMATION

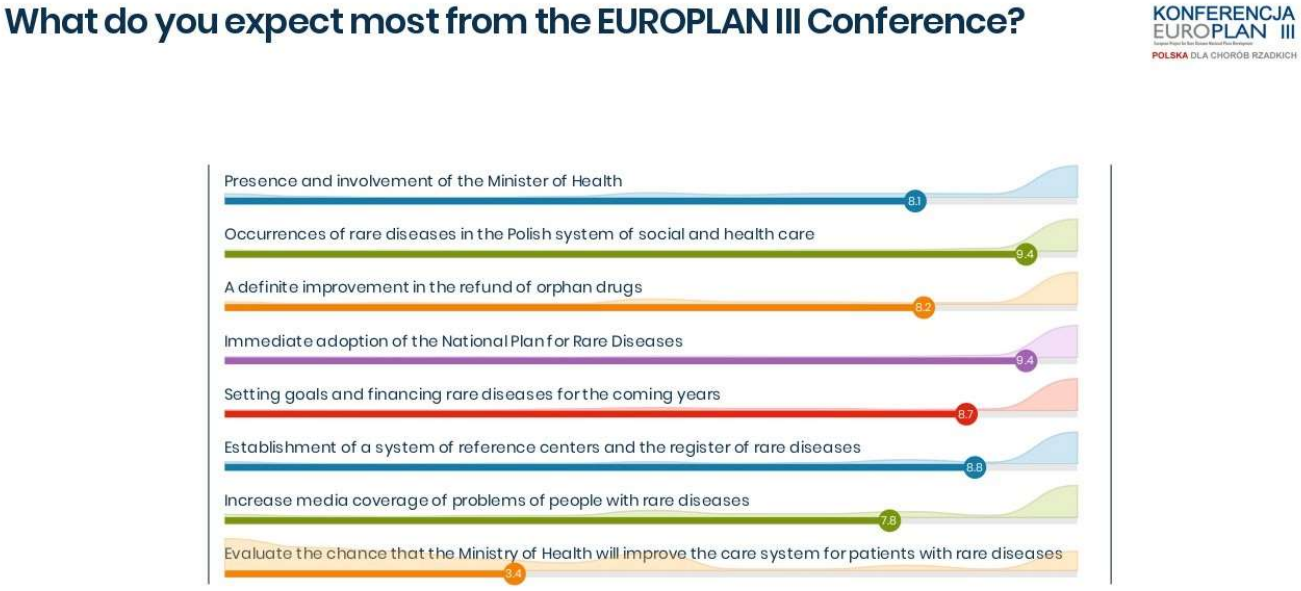
Country	POLAND
National Alliance (Organiser)	The Polish National Forum for rare diseases therapy - ORPHAN (KFO)
Date & place of the national workshop/conference	December 14, 2017 Warsaw
Website	www.europlan2017.pl
Members of the Steering Committee	• Mirosław Zieliński - KFO President • Stanislaw Maćkowiak - President of the Federation of Polish Patients • Prof. dr hab. n. med. Anna Kostera-Pruszczyk • Prof. dr hab. n. med. Jolanta Sykut-Cegielska • Prof. dr hab. n. med. Anna Latos-Bieleńska • Prof. dr hab. n. med. Jolanta Wierzba • Prof. dr hab. n. med. Joanna Chorostowska-Wynimko • Dr Michał Jachimowicz - MAHTA
List of Themesaddressed	• Plenary session on the Plan for rare diseases • The system of reference centers for rare diseases • Diagnostics, a register of rare diseases and scientific research in rare diseases • Education and social policy in rare diseases • Treatment of rare diseases and access to orphan drugs
Appendix	1. Programme of the conference 2. List of participants

FINAL REPORT

On December 14, 2017 at the Novotel Hotel in Warsaw, the 3rd Europlan Conference was held under the motto "Poland for rare diseases". The conference aimed to draw attention to the lack of proper treatment and diagnosis of patients suffering from rare diseases as well as the lack of adequate financing of orphan drugs from public administration bodies. The main appeal of the patient and medical environment is the introduction of an appropriate National Plan for people with rare diseases. The plan, as recommended by the Council of the European Union, should have been introduced in the Member States by the end of 2013 at the latest. Poland currently, still does not have a National Plan for rare diseases in place, however in December 2017 the Ministry of Health presented a draft document leading the future plan. As announced, the inter-ministerial and public consultations of this material will begin soon.

The EUROPLAN III conference was held with the participation of representatives of organizations and foundations associated in the National Orphan Forum ORPHAN, health care specialists, physicians in the field of rare diseases, representatives of public administration and the media.The conference gathered 123 registered participants.

The opening of the Conference was preceded by a public electronic survey available on the www.europlan2017.pl portal from December 1, 2017. The following survey results were presented and commented during the plenary session.



The conference began with greeting the guests by the President of the National Orphan Forum, Mr. Mirosław Zieliński, together with the President of the Federation of Polish Patients Mr. Stanisław Maćkowiak. The Deputy Minister of Health Marek Tombarkiewicz was also present at the conference opening.

Mr. President Miroslaw Zieliński recalled the history of the development of Europlan Project and actions taken for the benefit of people with rare diseases. He expressed hope for reliable cooperation with the Ministry of Health in the area of building the National Plan for Rare Diseases, emphasizing the role and involvement of specialists in the field of rare diseases in the creation of the plan.

Deputy Minister of Health, Mr. Marek Tombarkiewcz, thanked for the invitation on behalf of Minister of Health Konstanty Radziwiłł, who could not participate in person and briefly summarized the draft national plan prepared by his department, calling it a "working version". He assured that work on it will be continued, and expressed willingness to cooperate with the expert community, as he admitted the draft was not extensively consulted with specialists, but prepared exclusively by his department. The Minister also pointed out that the draft requires inter-ministerial consultations, which take time and delay the implementation of the final versions of the project.

PANEL I - "The system of reference centers for rare diseases - where are we?"

Prof. dr hab. n. med. Anna Kostera-Pruszczyk opened the first part of the scientific and educational module, with a lecture titled "European Reference Network on the example of the Neuromuscular Networks: EURO NMD". The professor brought up the issues of existing cooperation between European institutions and the necessity of its expansion.

Professor Kostera-Pruszczyk turned the attention to European cooperation in the field of rare diseases, recognizing the exchange of experience and research results as crucial for improving patient care and effective treatment. She referred to her own professional experience, as she works at the only center in Poland which collaborates with other European institutions in the field of neuromuscular diseases. The professor recognized the attitude of medical staff towards the patients as a key element for proper functioning of medical centers, and emphasized the need to strengthen the trust between them and specialists.

She stressed the need for openness of doctors towards patients which consequently increases trust in the medical staff, and is the basis for development of an effective Reference Network in Europe. Joint research exchange and free flow of information between specialists from medical facilities across the continent were also mentioned as essential factors for effective European cooperation, which have a huge impact on the accuracy of diagnoses and treatment efficiency.

The main aim of the European Reference Network is to strengthen international cooperation and create a platform for information exchange and knowledge sharing. At the end, the professor recognized the necessity to introduce a national plan for rare diseases, and called for the government to show more involvement in helping patients who suffer from them.

Prof. Dr hab. n. med. Jolanta Sykut-Ciegielska gave a lecture on “Centers and reference networks in rare diseases on the example of metabolic medicine”. The professor, in her lecture, emphasized the necessity to create reference centers dealing with rare diseases and the creation of the whole system of functioning of such centers which, as she pointed out, is one of the most important pillars of correct treatment for patients. Based on the already existing but informal reference centers in Poland, it is considered necessary to create a formal system of reference centers as well as a major expert center that would gather all information on rare diseases and treatment of individual cases, and would also be an opinion-forming and auxiliary body associating patients and specialists.

According to the Professor, coordinating such activities would allow greater effectiveness in diagnosing diseases and providing appropriate treatment to patients. The establishment of formal reference centers and determining their scope of activities and competences will minimize current challenges regarding the lack of free information flow between facilities and sharing research results, and will increase joint development of treatment strategies for patients. As an example of a properly functioning reference center system, the Professor indicates France, which currently operates 131 reference centers. The Professor presented the problem of the lack of a formalized system of reference centers in the field that deals with metabolic diseases. Indicating rare diseases of metabolic origin, the best way to diagnose is to screen newborns which, if a congenital malformation is found, will allow to adjust the appropriate treatment and slow down the course of the disease in the future.

The professor ending her lecture, postulating for the creation of the National Center for Rare Diseases which will deal with the coordination of the national plan and the supervision of its creation. This body would include medical environments in the field of rare diseases as well as scientific clusters and patients.

At the end of the session devoted to the subject of the system of reference centers for rare diseases, the lecture was led by prof. Dr hab. n med. Jolanta Wierzba on the subject of "Multi-specialty care of a patient's reference center with a genetically determined rare disease".

Referring to her topic, the Professor drew attention to the need for individual treatment of each patient in every aspect of his/her life. The main goal is to allow the patient the possibility to lead a normal life mode, that is why the Professor indicated the multi-specialized care of reference centers as a necessity for full treatment effectiveness. In addition to providing treatment for the patient we must remember about the education of relatives and provide them with psychological assistance.

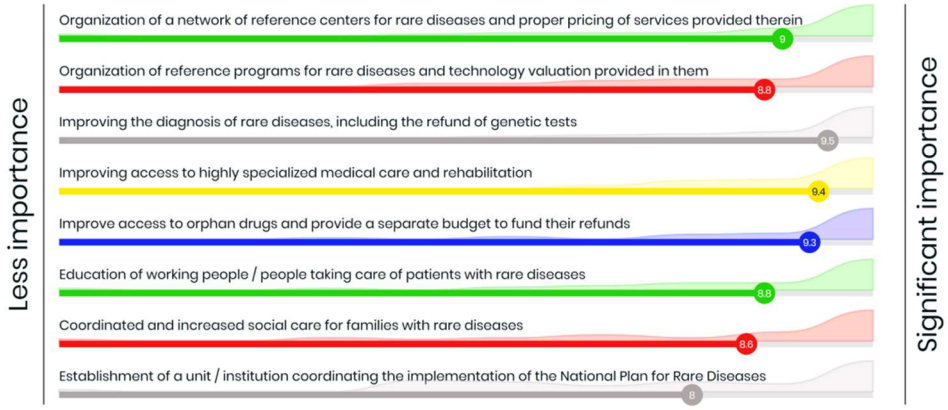
The Professor presenting the diagram of the expected operation of the reference centers recognizes that the center should include specialists such as:

- Geneticist
- Gastroenterologist
- Cardiologist, orthopedist
- Ophthalmologist, ENT specialist
- Neurologist and psychiatrist
- Children's surgeon
- Anesthesiologist
- Physiotherapist
- Speech therapist
- Psychologist

Unfortunately, according to the Professor, the Ministry of Health does not provide funding for such extensive personnel and research. It makes the patient's treatment very difficult, as the patient may be exposed to constant changes of doctors and duplicated tests resulting from the lack of constant monitoring. Professor Wierzba was very focused on a professional team of doctors who, by monitoring the patient from the beginning of the disease, can make right decisions about the course of treatment based on their knowledge. Also, the lack of knowledge about rare diseases due to their individual course is problematic, which is why one should focus on the growth of information campaigns and scientific conferences on rare diseases in order to increase effectiveness in the treatment of rare diseases.

At the end of the module devoted to reference centers, an interactive electronic survey was conducted using MentiMeter® mobile life voting technology. Below are the questions and results of the survey devoted to the evaluation of importance of key chapters/issues of the future National Plan for Rare Diseases:

Specify the importance of priorities in the implementation of the National Plan for Rare Diseases



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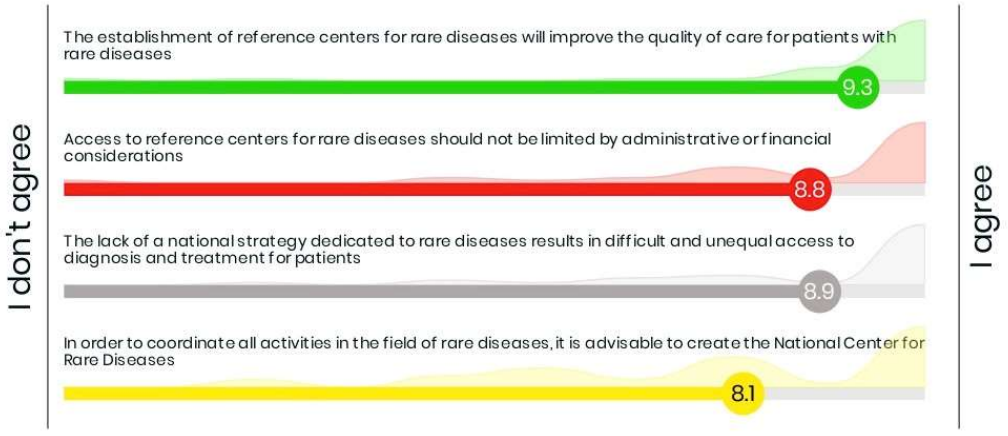
Survey: A system of reference centers for rare diseases - Where are we?

Rate from 1 to 10



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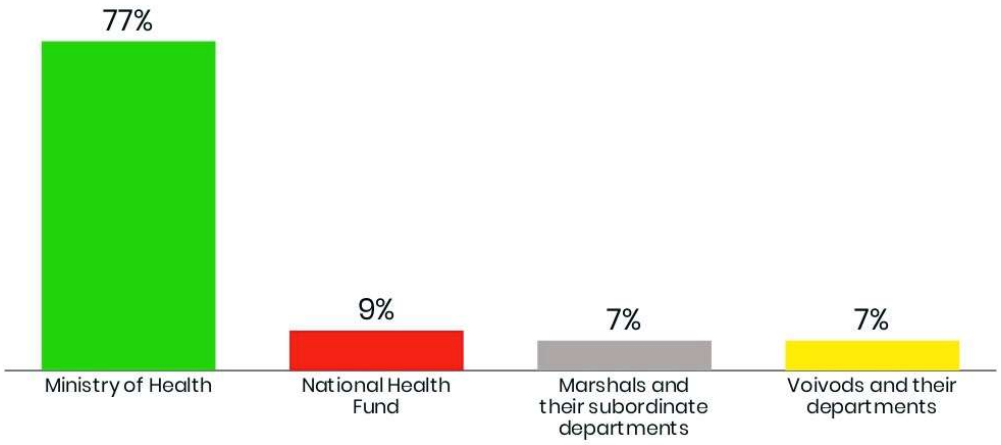
Who should be the administrator of funds allocated to Reference Centers?



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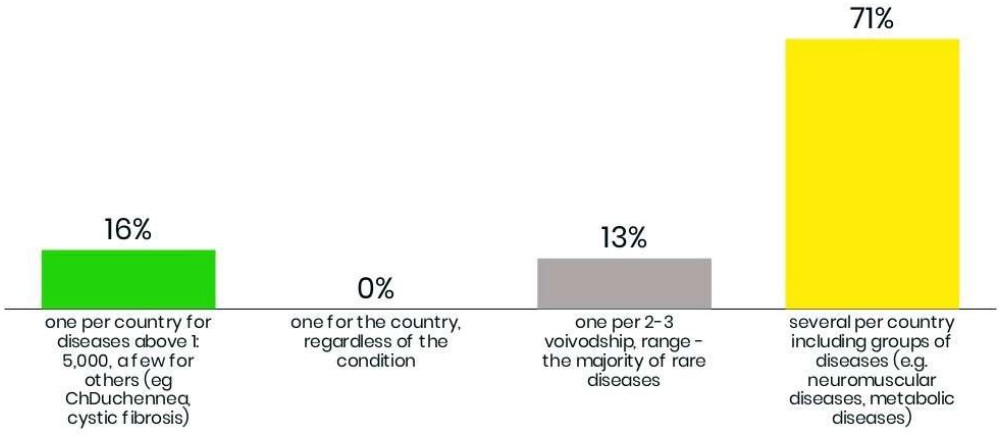
What range should Reference Centers have?



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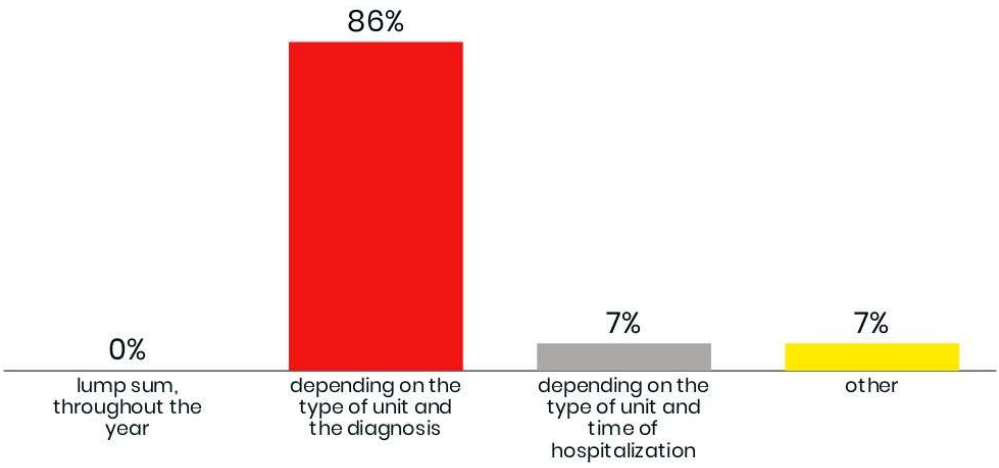
How should Reference Centers be funded?



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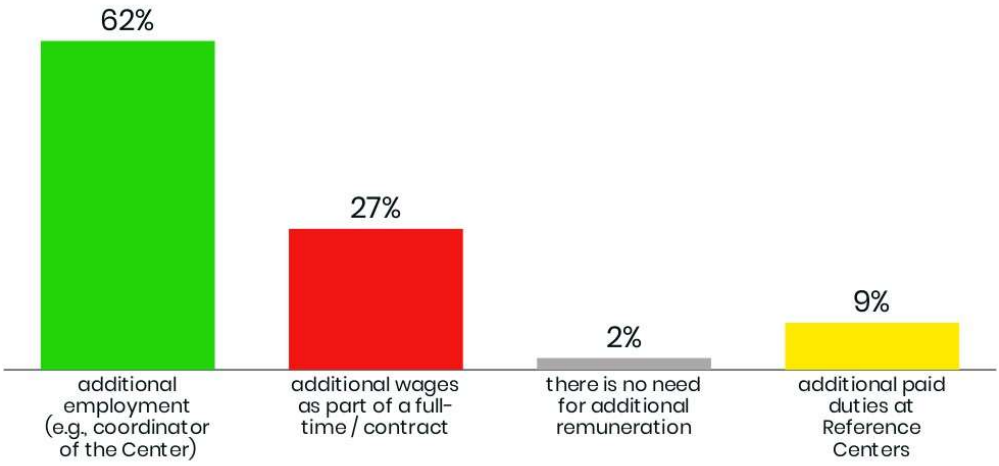
How to pay outside the hospitalization care of Reference Centers (emails, telephone consultations)?



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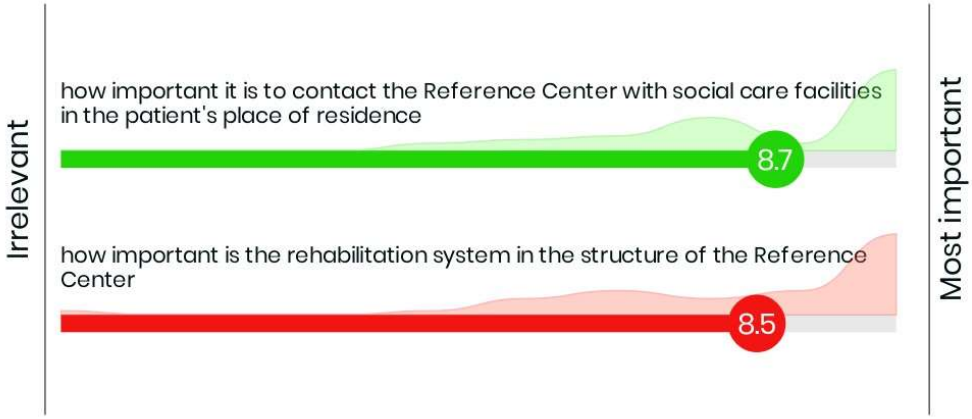

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PANEL II - "Diagnostics, a register of rare diseases and scientific research in rare diseases"

The second module of the conference was opened by Prof. Dr hab. n med. Anna Latos-Bieleńska titled "Registry of rare diseases - how to do it in Poland? "

The main topic of the Professor's lecture is the recognition of the rare diseases register as the foundation for the correct treatment and diagnosis of patients. Such a system must meet certain conditions such as common coding of rare diseases, full identification of rare diseases, stable funding and a team of experts who can use information and use it to protect the health of patients.

Currently, we have three ways to encode rare diseases and one in preparation which in the opinion of the Professor will be the best solution, these are:

- ICD-10 - is a register with 200 diseases from 5-8 thousand of diseases
- ORPHA code - has 20,000 codes, but it is not used in practice by the Polish National Health Fund
- OMIM - this list is used by geneticists in patient records.
- ICD-11 - currently in the preparation phase (not available)

Disease registers are necessary because in addition to the monitoring function they also have an educational and preventive function. In Poland there is the Polish Registry of Congenital Malformations (PRWWR) containing over 15000 children's data with congenital malformations. The Polish Registry of Congenital Malformations is part of the European EUROCAT register, which is one of the three largest consortia in the world.

Keeping a reliable registry brings a lot of benefits in effective diagnosis and treatment. Recording of such cases has a great impact on the education of doctors and allows to include all medical information about the disease, also to contribute to better monitoring of patients and the sharing of observations between specialists. It also contains the results of the tests needed to clarify the treatment. According to the Professor, we have a very good foundation for building a rare diseases registry, one of them is a large PRWWR which contains a lot of information about congenital defects, as well as Polish law that organizes the process of creating medical records. Unfortunately, the Professor also indicates problems when creating this type of registry in the form of the current bad coding of diseases and the lack of appropriate genetic tests that contribute to incomplete knowledge of a given disease, and too tedious procedure for registering the disease by a doctor.

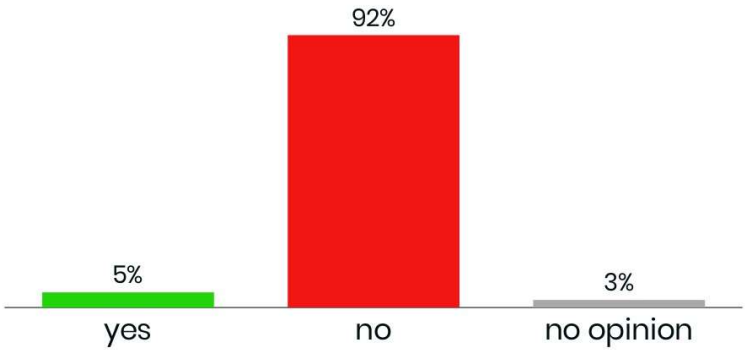
As a solution to the above-mentioned problems, the Professor suggests that the following actions be taken:

- Appointment of a coordinator for Rare Diseases Registry;
- indication of the platform for the database and supplementing it with Polish National Health Fund (NFZ) data;
- streamlining disease registrations by physicians;
- ensuring stabilization in the registry financing.

At the end of Panel II there was an electronic survey related to the panel's theme.

Survey: Diagnostics, a register of rare diseases and research in rare diseases

Do you agree that you should only be diagnosed in the direction of those rare diseases for which pharmacological or substitution treatment is availabl



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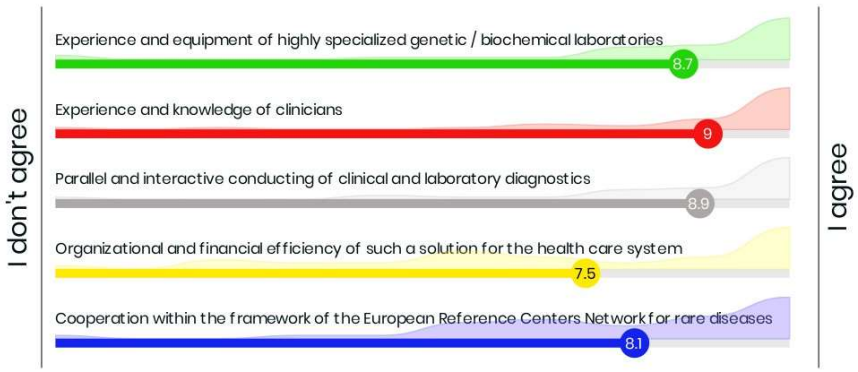
Diagnosis of rare diseases should be carried out by highly specialized reference centers due to



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What problems in the diagnosis of rare diseases in Poland require the most urgent activities?

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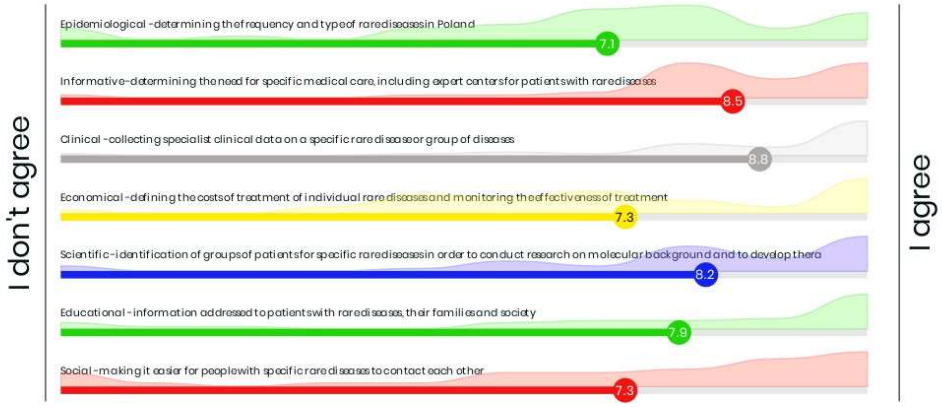
What functions of the Register of Rare Diseases is the most important?



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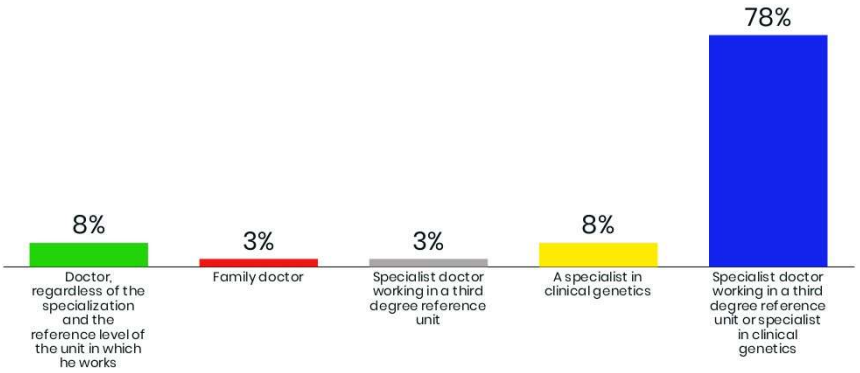
Who should give ORPHA and OMIM codes after recognizing a specific rare disease?



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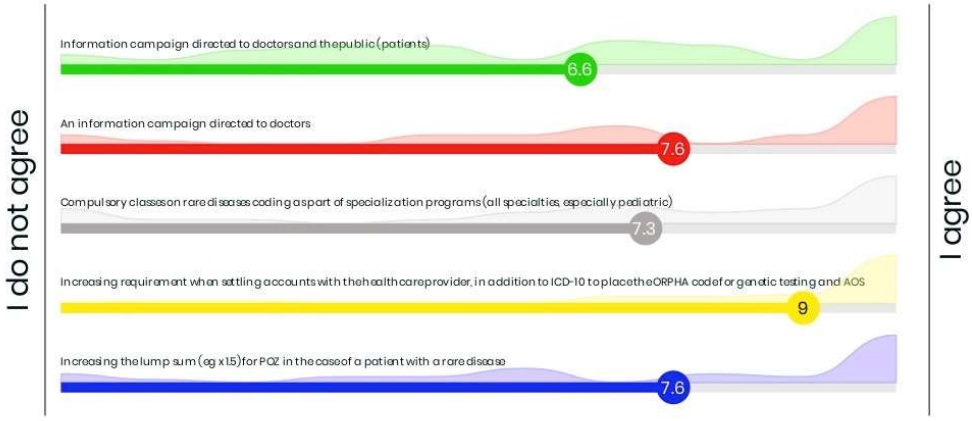
What actions do you consider to be the most effective for popularizing the use of ORPHA codes?



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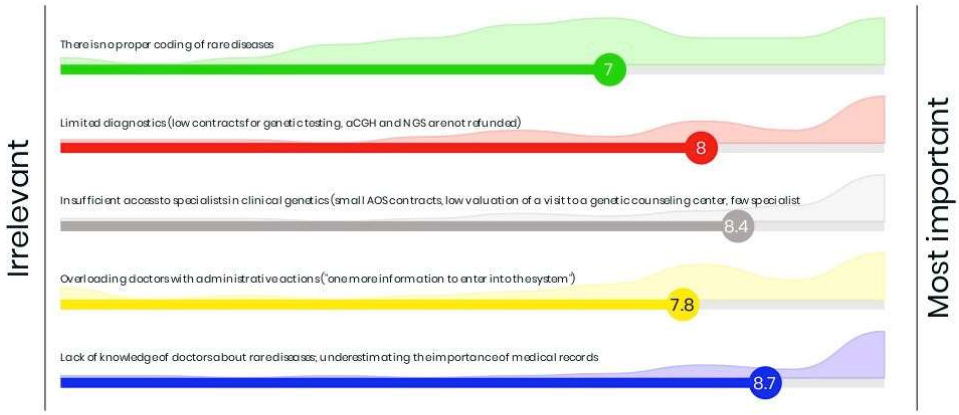
Which barriers introduce the Register of Rare Diseases as the most important?



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PANEL III - "Education and social policy in rare diseases"

Mr. President Stanisław Maćkowiak - Federation of Polish Patients led a lecture on the subject "Integrated social support system for patients with rare diseases and their families"

In his lecture, Mr. President Stanisław Maćkowiak focused on the social aspect of creating the National Plan for Rare Diseases. The initiator of the idea was the European Union, recommending the Member States to introduce a plan and its implementation in the field of treatment of rare diseases. The recommendation envisages the introduction of a rare diseases register, guaranteeing appropriate treatment and assistance to families of sick people as well as pursuing educational and scientific goals in this area. The implementation of these assumptions should lie in the matter of public administration bodies. One of the pillars of the national plan is the creation of an information portal about rare diseases that would be widely

available to any interested citizen. In the social aspect, the national plan should assume improvement in the process of applying for the recognition of each degree of disability and benefits linked to various degrees of disability.

Currently, this process is very bureaucratic and the decision to grant disability living allowance is decided by a commission which often does not have full knowledge about the patient's illness. The introduction of such a plan will help rare disease patients as well as their relatives, apply for due benefits and find a proper specialist issuing opinions on the degree of disability.

It would be also much easier to establish an efficient inter-ministerial cooperation to facilitate patients' access to information about their situation and which actions they need to take. Ministries should also pay more attention to social education in this area, raise the qualifications of health care workers and subsidize research on rare diseases. At the end of Panel III, an electronic survey was held related to the panel's theme.

Survey: Education and social policy in rare diseases

Rate from 1 to 10

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After the survey, a press conference was held on topics discussed at the conference with the participation of journalists and following persons answering the journalists' questions:

- President Mirosław Zieliński
- Attorney Paulina Kieszkowska-Knapik
- Prof. Dr hab. n. Med. Anna Kostera-Pruszczyk
- Prof. Dr hab. n. med. Jolanta Sykut- Ciegielska
- President Stanisław Maćkowiak

PANEL IV - "Treatment of rare diseases and access to orphan drugs"

President Mirosław Zieliński - "Orphan drugs - a challenge or an obligation?"

In his lecture, Mr. President Mirosław Zieliński discussed the issues of treating patients with orphan drugs and their method of financing and accessibility, defining them as "medicinal products intended for the diagnosis, prevention and treatment of rare diseases". He pointed out the main problems of orphan drugs which are accessibility and high price. Unfortunately, the problem is also the Polish law, which does not distinguish orphan drugs from the common ones, which results in serious restrictions on reimbursement of orphan drugs, considering them as not economical, with insufficient Evidence Based Medicines documentation proving clinical effectiveness.

The main problem in Poland is the lack of a general national plan for rare diseases, as orphan drugs are subject to the same reimbursement process as any other more widely used drug, for example: insulin for people with type 1 diabetes. Therefore, a large majority of orphan drugs due to low demand by patients than other diseases do not pass the reimbursement procedure carried out by the health department. The President emphasized the need to amend the Reimbursement Act and create an individual reimbursement process for orphan drugs and to fund them from the state budget.

Ph.D Michał Jachimowicz - "The specificity of the assessment of medical technologies used in rare diseases"

In his lecture, Dr. Michał Jachimowicz approached the economic outlook on the orphan drug distribution process. He pointed out the first problem, namely the development of orphan drugs is a very long process and for many companies also not profitable. In case of orphan drugs, there is a need to pay attention to the requirement of individual research, which hinders and extends the process of creating orphan drugs. Orphan drugs cannot be generalized. However, they are treated in that way because of the profitability and uncertainty of their efficacy.

Doc. Anna Potulska - Chromik - "Non-pharmacological treatment of rare diseases based on Duchenne muscular dystrophy"

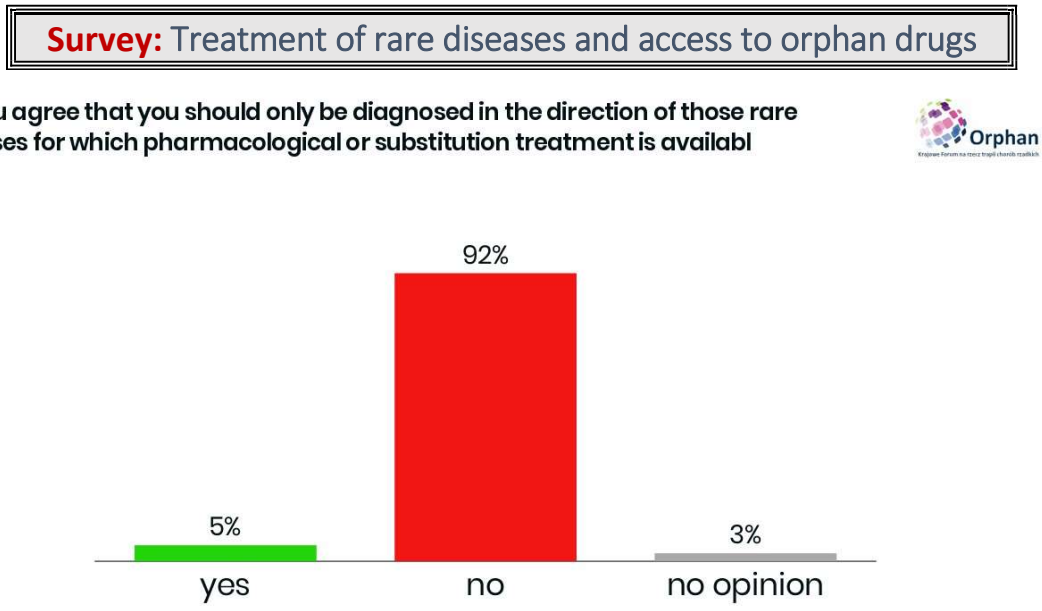
Ms. doc. Anna Potulska - Chromik presented methods of non-pharmacological treatment and care for people with rare diseases. As important elements, she recognized the general care of patients in physical and mental matters. Care must be individualized and directly assigned to a specific patient. On the example of Duchenne's disease, the effects of which lead to the loss of self-mobility, rehabilitation can begin even before the loss of mobility and effectively maintain the function of locomotion despite the development of the disease. Physiotherapeutic treatment can also improve the quality of life of the patients affected of Duchenne's disease through the selection of appropriate orthopedic equipment. It is also necessary to pay attention to other methods of therapy such as stretching exercises, breathing exercises, cardiac care, proper diet and constant monitoring of the patient's course of disease.

Attorney Paulina Kieszkowska - Knapik - "Legal barriers in access to orphan drugs"

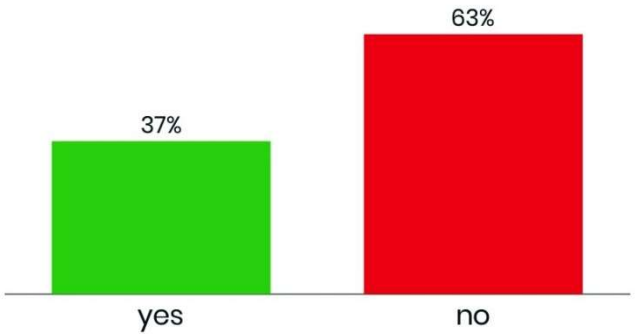
The lawyer's lecture began by presenting the constitutional rights of every citizen and the right to protect their health. She also stressed the state's welfare function and the obligations to provide everyone with access to health care. At the outset, she stressed that the definition of a rare disease as well as ultra-rare disease is not in Polish law, this is a significant problem because without legal definitions, it is also difficult to define orphan drugs and assign them a special charter for easier access to them. We must look for legal definitions of orphan drugs and diseases in the sources of European Union law. The lack of the definition of orphan drugs puts them in the same legal position as common medicines, therefore, the reimbursement process cannot be considered to meet the reimbursement requirements because the difference in their use is diametric. As a way out of the situation, she considers the introduction of a separate reimbursement procedure for orphan drugs. Currently in Poland, statutory changes are being implemented to improve the situation of patients with rare diseases and facilitate their access to medicines. Amendments to acts regarding orphan drugs and rare diseases are:

- The act on openness of public life - transparency of reimbursement proceedings
- Amendment of the Reimbursement Act
- Amendment to the Act on benefits

At the end of Panel IV, an electronic survey was held related to the panel's theme



Is the assessment of surrogate endpoints in clinical trials for orphan technologies the basis for inferring their effectiveness?



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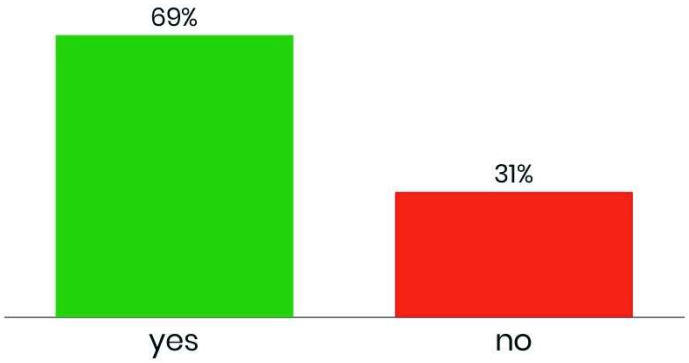
Can surrogates evaluated in orphan technology clinical trials be predictors of hard endpoints?



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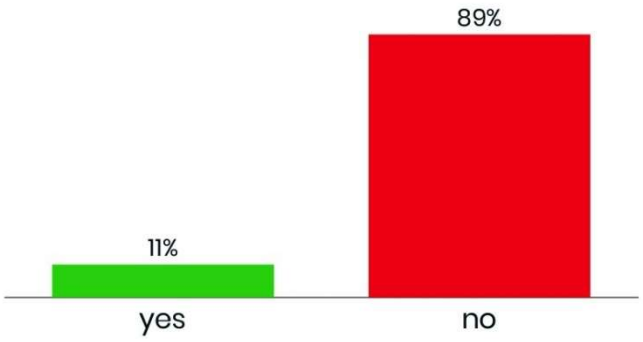
In the case of technologies used in rare diseases, should the availability of high-quality clinical trials be expected?



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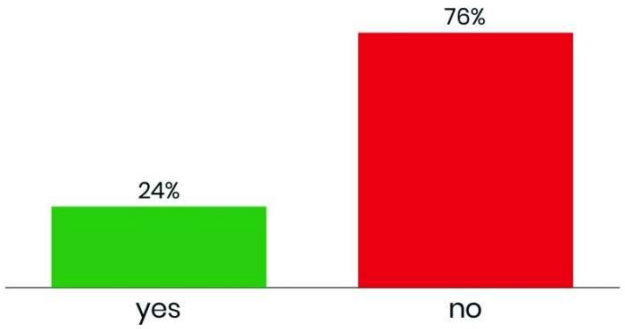
Is the economic assessment of drugs used in rare and ultra-rare diseases before the decision on their reimbursement is justified?



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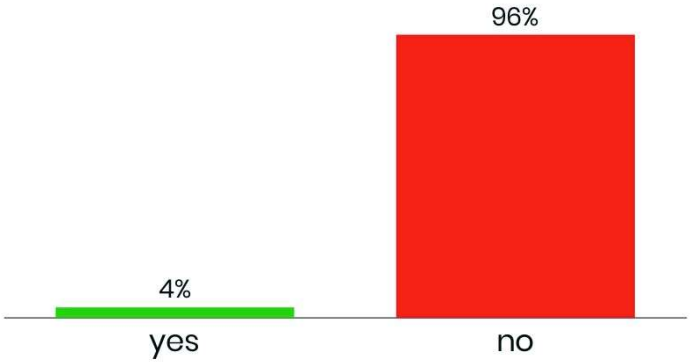
Is it appropriate to use the same cost-effectiveness threshold for QALY in common disease as in the rare economic evaluation of orphan drugs?



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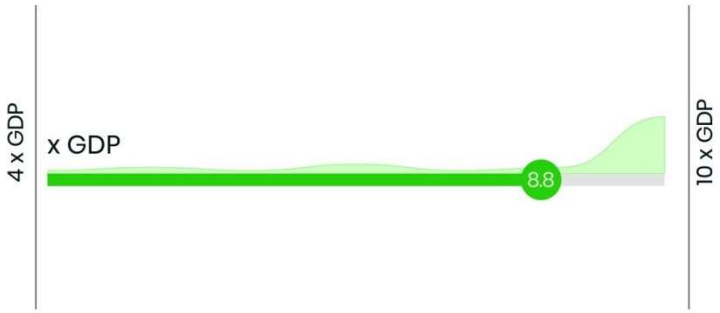
If for the orphan drugs the profitability threshold for QALY should be higher than 3 x GDP used in common diseases, it should be:



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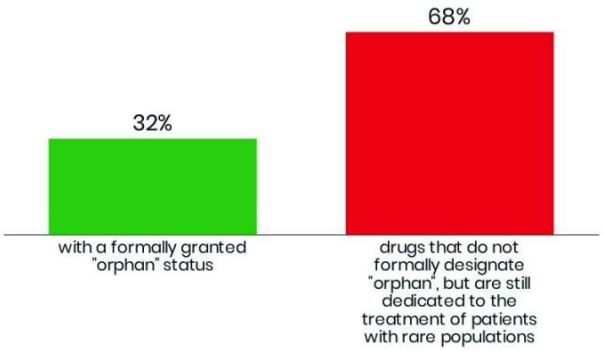
Should separate provisions for rare diseases be applied to:



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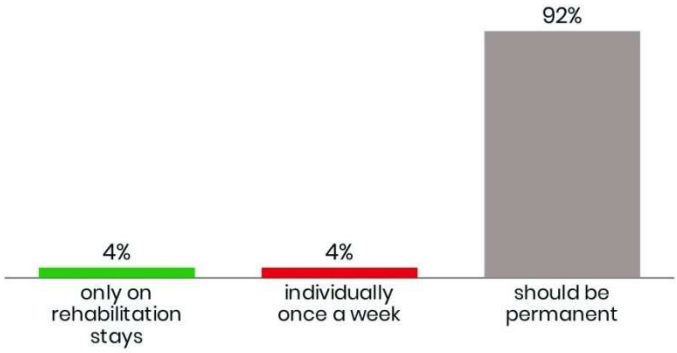
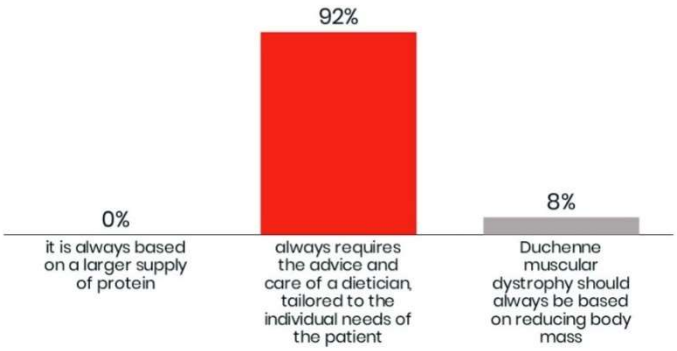
Rehabilitation in rare diseases should take place



26

Dietary treatment of patients with rare diseases:



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Conclusions

As the conference was fully dedicated to the shape of the future Plan for Rare Diseases, the conclusions from each session concentrated on underlining the gaps or different approaches in the draft version of such plan presented by the Ministry of Health.

The expectations of the National Orphan Forum ORPHAN, health care specialists and rare diseases physicians as to the shape of the plan were much broader than suggested by the MoH. That led the National Forum Orphan to the decision to hold a workshop in mid-February in order to prepare the position paper that will be submitted to the MoH shortly before the Rare Diseases Day 2018 (28 February). After the Ministry will acknowledge the recommendations of the experts group “Poland for Rare Diseases”, a second workshop will be organized in early April to agree upon the final shape and content of the Polish National Plan for Rare Diseases.

Appendix 1

Conference Agenda

9:00-9:30	**Registration**
9:30-10:00	• Opening of the Europlan III Conference – plenary session
	• Mirosław Zieliński - President of the National Orphan Forum • Marek Tombarkiewicz - Deputy Minister of Health Republic of Poland
10:00-11:30	A system of reference centers for rare diseases - where are we?
	• Prof. dr hab. n. med. Anna Kostera-Pruszczyk European Reference Networks (ERN) on the example of the Neuromuscular diseases Network: EURO NMD
	• Prof. dr hab. n. med. Jolanta Sykut-Cegielska: Reference centers and networks in rare diseases on the example of metabolic medicine
	• Prof. dr hab. n. med. Jolanta Wierzba *Multi-specialized care in a patient reference center with a genetically determined rare disease*
	• Area assessment (electronic voting)
	Coffee break
11:50-13:00	**Diagnostics, rare diseases register and scientific research in rare diseases**
	• Prof. dr hab. n. med. Joanna Chorostowska-Wynimko *Diagnostics of rare diseases - the key to proper treatment and medical care*
	• Prof. dr hab. n. med. Anna Latos-Bieleńska ***Registry of Rare Diseases - how to do it in Poland?***
	• Area assessment (electronic voting)
13:00-13:30	**Education and social policy in rare diseases**
	• Prezes Stanisław Maćkowiak – Federacja Pacjentów Polskich *An integrated social support system for patients with rare diseases and their families*
	• Area assessment (electronic voting)
	Lunch
13:30	**Press conference with participation of :** • Mirosław Zieliński • Mecenas Paulina Kieszkowska-Knapik • Prof. dr hab. n. med. Jolanta Sykut-Cegielska • Prof. dr hab. n. med. Anna Kostera-Pruszczyk • Stanisław Maćkowiak
14:40-15:40	**Medication of rare diseases and access to orphan drugs**
	• Marcin Czech - Deputy Minister of Health of the Republic of Poland • Mirosław Zieliński *Orphan drugs - a challenge or an obligation?*
	• dr Michał Jachimowicz The specificity of the assessment of medical technologies used in rare diseases
	• doc. Anna Potulska-Chromik *Non-pharmacological management of rare diseases on the example of Duchenne's muscular dystrophy*
	• Maecenas Paulina Kieszkowska-Knapik Legal barriers in access to orphan medicines
	• Area assessment (electronic voting)
15:40-17:40	PGE Polska Company for Rare Diseases - Plan for rare diseases Public consultation - Discussion • Deputy Minister of Health of the Republic of Polanddr Marek Tombarkiewicz • Mirosław Zieliński • Stanisław Maćkowiak
	Official cocktail

Appendix 2

List of Participants

Polish National Forum ORPHAN Patient Associations	5 27
Ministry of Health	2
Public institutions	7
Health care providers	17
Laboratories and pharma industry representatives	12
Research Centres	6
Media and PR	15
Others	32
Total	123