EUROPLAN
European Project for Rare Diseases National Plans Development
Coordinated by the Italian National Centre for Rare Diseases
Italian National Institute of Health
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Orphan
RARE DISEASES POLAND
Polish National Forum for rare diseases therapy

EURORDIS
Rare Diseases Europe

POLAND

EUROPLAN NATIONAL CONFERENCE

FINAL REPORT

27-28 September 2013, Warsaw

EUCERD
Joint Action

FOREWORD

The EUROPLAN National conferences are aimed at fostering the development of a comprehensive National Plan or Strategy 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 are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS – the European Organisation for Rare Diseases. For this purpose, EURORDIS nominated 10 EURORDIS-EUROPLAN Advisors - all being from a National Alliance - specifically in charge of advising two to three National Alliances.

EUROPLAN National conferences share the same philosophy, objectives, format and content guidelines. They involve all stakeholders relevant for developing a plan/strategy for rare diseases. According to the national situation of each country and its most pressing needs, the content can be adjusted.

During the period 2008-2011, a first set of 15 EUROPLAN National Conferences were organised within the European project EUROPLAN. Following the success of these conferences, a second round of up to 24 EUROPLAN National Conferences is taking place in the broader context of the Joint Action of the European Committee of Experts on Rare Diseases (EUCERD) over the period March 2012 until August 2015.

The EUROPLAN National Conferences present the European rare disease policies as well as the EUCERD Recommendations adopted between 2010 and 2013. They are organised around common themes based on the Recommendation of the Council of the European Union on an action in the field of rare diseases:

1. Methodology and Governance of a National Plan;

2. Definition, codification and inventorying of RD; Information and Training;

3. Research on RD;

4. Care - Centres of Expertise / European Reference Networks/Cross Border Health Care;

5. Orphan Drugs;

6. Social Services for RD.

The themes "Patient Empowerment", "Gathering expertise at the European level" and "Sustainability" are transversal along the conference.

EUROPLAN II

POLAND

27-28 September 2013, Warsaw

Rare Diseases Awareness Raising Campaign

Giant Poster on the

Warsaw Palace of Culture and Science

Part I – General Information

Country	**Poland**
Date & place of the National Conference	**27-28 September 2013, Warsaw**
Website	**www.rzadkiechoroby.pl/EUROPLAN**
Organiser	**Polish National Forum for Rare Diseases Therapy "Orphan"**
Members of the Steering Committee	1. Polish National Forum Orphan - Mirosław Zieliński - www.rzadkiechoroby.pl 2. Association Arsvivendi - Stanisław Maćkowiak - www.fenyloketonuria.org 3. Foundation "Knowing to Help" - Krzysztof Swacha - www.umiecpomagac.org 4. Foundation "Matio" - Paweł Wójtowicz - www.mukowiscydoza.pl
Names and list of Workshops	**Panel I: Methodology and Governance of the National Plan** **Panel II: Definition, codification and inventorying of RD** **Panel III: Research and education / information and training** **Panel IV: Centers of Expertise / European Reference Networks/Cross Border Health Care** **Panel V: Social Care** **Panel VI: Orphan Medicinal Products**
Workshop Chairs and Rapporteurs	**Panel I: Methodology and Governance of a National Plan** Introduction: Expectations towards Health System and EUROPLAN Evaluation: Miroslaw Zieliński - President of the National Forum for Rare Diseases Therapy **Panel II: Definition, codification and inventorying of RD** Introduction: Prof. Jolanta Sykut-Cegielska - The Children's Memorial Health Institute

	Expectations towards Health System and EUROPLAN Evaluation: Stanisław Maćkowiak - President of the Polish PMU Association Ars Vivendi **Panel III: Research and education / information and training** Introduction: Prof. Krystyna Chrzanowska - The Children's Memorial Health Institute Expectations towards Health System and EUROPLAN Evaluation: Maria Libura, President of Prader-Willi Association **Panel IV: Centers of Expertise / European Reference Networks/Cross Border Health Care** Introduction: Prof. Małgorzata Krajewska-Walasek - The Children's Memorial Health Institute Expectations towards Health System and EUROPLAN Evaluation: Krzysztof Swacha - President of the Foundation "Knowing to Help" **Panel V: Social Services for RD** Introduction: Dorota Korycińska - President of the Alba Julia - the Association of patients with the Recklinghausen disease and related phacomathosis group Expectations towards Health System and EUROPLAN Evaluation: Paweł Wójtowicz - Cystic Fibrosis Association Matio **Panel VI: Orphan Drugs** Introduction: Łukasz Pera - Specialist at the Drug Department Policy at the Ministry of Health Expectations towards Health System and EUROPLAN Evaluation: Mirosław Zieliński (NA) and Dr Krzysztof Łanda - President of the Watch Health Care Foundation
Additional Workshop - Day II - The debate on "Rare Diseases in Poland - where do we go?"	
Annexes :	**Annexe 1** - Program of the first day of the Conference EUROPLAN II - Rare Diseases, Poland **Annexe 2** - Program of the second day - The debate on "Rare Diseases in Poland - where do we go?" **Annexe 3** – Participants List **Annexe 4** - Core Indicators - Definitions and associated answers **Annexe 5** - Pictures of the event

Part II - Main Report

Plenary Report - Opening Session

Chairman: Mirosław Zieliński - President of the National Forum for Rare Diseases Treatment „Orphan"
Host speaker - via teleconference: Yann Le Cam - Managing Director of EURORDIS

Mirosław Zieliński, President of the national „Orphan" Forum welcomed patients, representatives of EURORDIS, Yann Le Cam, Chief Executive Officer of EURORDIS and representatives of the Italian Centre for Rare Diseases of the Italian Institute for Health (ISS), scientific researchers, physicians, professors and philosophers active in the field of rare diseases. He thanked all the persons involved in drafting assumptions for the National Plan for Rare Diseases, including Doctor Jacek Graliński, former Head of the Rare Diseases Team in the MOH Cabinet.

He referred to EUROPLAN I Conference of 2010 held in Cracow. There, the majority of the activities executed during the last three years which are being continued until today, were initiated. EUROPLAN is a very precious initiative aimed at the evaluation of achievements of EU member countries regarding development and implementation of National Plans. In Poland this evaluation has an accessory dimension for actions undertaken by all stakeholders, including the government. As the result of the Conference the report including all the recommendations will be drafted. Implementation of the plan should start from the beginning of 2014, although the chances for that are reduced. We count on the activity of all participants of the conference and categorization of expectations towards decisions makers.

Yann Le Cam recalled that in 2010 he participated in EUROPLAN I Conference in Cracow. EURORDIS was established in 1997 in order to build a community of patients suffering from rare diseases and highly values its cooperation with the National Orphan Forum in Poland. In 2008 we joined our efforts within the EURORDIS framework. The main purpose was to offer a better care to the patients. Starting from 2010, a big progress has been achieved in EU with respect to rare diseases. It is good that in Poland a Team for Rare Diseases was created which developed broadly a consulted strategy - a seed for the National Plan. Poland is considered as a large and important EU country and many times it was a leader in development of innovative solutions in medicine. The Polish example should become a pattern for other countries of Central and Eastern Europe. Conclusion - within the framework of the EUROPLAN Project Poland, along with other EU countries, is in a position to develop a good model for diagnosing and treating patients with rare diseases, which can serve well for the future generations.

During his presentation M. Zieliński reminded that the first actions regarding rare diseases in Poland were executed in a chaos. He cited the legal sources that created the basis of and indicated the need for an approach to the complex issue of rare diseases in EU and increased access to the therapy through reimbursement of possible largest number of medicines used for treatment of rare diseases in EU member countries. The most important document was the Recommendation of the EU Council of 9 June, 2009. In Poland the work on the National Plan started in 2011. After development by experts and patients' organizations, a draft was presented to the current Minister of Health Mr. Bartosz Arłukowicz and then it was sent to the Team for the Rare Diseases at the Ministry of Health. By the end of December of 2012 a final version was developed and it was approved by the Ministry Board in April 2013. From this time the work of the Team is in a dead point because the Team was deprived of its Head, whose task was to coordinate work of the team. In consequence neither inter-ministerial nor social consultations required for such document were held. The National Forum for Rare Diseases Treatment „Orphan" believes that in order to have an appropriate rank and meaning, the document should be enacted as a decision of the Council of Ministers. It is not too late now; however a leader of the process is needed, as well as a pressure from patients' organizations. The pressure is stimulating and inspiring. Recognizing the fact that in 2004 Poland joined an exclusive club named the European Union we are obliged to act.

At the end of the Plenary Session Mirosław Zieliński briefly presented assumptions and the course of the community and media campaign „Hope in Genes", which accompanied the EUROPLAN II Conference in Warsaw and across the country.

Report of Workshops

Theme 1 - Methodology, Management and Implementation of the National Plan for Rare Diseases

Introduction: Dr Krzysztof Łanda - President of the Watch Health Care Foundation
Systemic expectations and EUROPLAN evaluation: Mirosław Zieliński - President of the National Forum for Rare Diseases Treatment „Orphan"

We recognize the need to establish a unit coordinating actions for patients suffering from rare diseases. It is necessary to create a National Rare Diseases Centre as an agenda of the Minister of Health. Problem of rare diseases in Poland pertains to over 2 million of citizens. This is an important segment of a health care as a whole. 75% of those 2 million are children. This important area requires a systemic approach and coordinated management. We recognize the necessity of establishing a unit coordinating actions aimed at rare diseases patients. Predicted main tasks of the future Centre:

1. Maintaining rare diseases register;
2. Implementation of the National Plan;
3. Monitoring and assessment of the National Plan implementation;
4. Accreditation of reference centers;
5. Place for affiliation of Coordinating Team for ultra-rare diseases (qualifications for treatment program);
6. Presenting opinions regarding reimbursement applications i.e. participation in works of the Agency for Assessment of Medical Technologies (AOTM) and Economic Committee;
7. Representation of Poland in the work of the Commission Experts Group on Rare Diseases (former EUCERD) - it would be advisable that the Head of the Center participate in the meetings of the Expert Group as a representative of Poland;
8. Social education and shaping up/influence on the medical education program.

During the discussion it was established that, despite social campaigns, the awareness level as well as the knowledge regarding reference centers are insufficient. If the National Plan will be adopted, then every year a conference should be held with participation of the medical-scientific groups, patients and decision-makers to assess implementation level. It is necessary to develop a better consultation and meetings system regarding the assessment of the Plan implementation including, for example, consultations with patient organizations at least once in a quarter. Most of the EU member countries will keep the deadlines for the set-up of the National Plans however not Poland. The resolution of the Council of Ministers should be adopted as soon as possible.

Debrief Session Conclusions:

I. Development of Regulations/Laws

The task of elaborating a National Plan (NP) for Rare Diseases (RD) was assigned to the advisory committee set-up by the Ministry of Health - the Rare Disease Task Force (RDTF). After many meetings throughout 2012, and with support from the National Forum for the Therapy of RD "ORPHAN", the Parliamentary Group on RD, and the Partnership for the NP for RD, the RDTF presented the final version (draft 12.2.) to the Ministry of Health (MoH) as scheduled, in December 2012. According to the project plan, the final paper was intended to be signed by the Minister of Health before June 2013, even though at the time the conference took place, this signature is still pending. So although there was an official decision to produce a NP, there is no regulation/legislation as yet.

II. Establishment of Coordination Mechanisms

As above, the task of elaborating the NP was executed by the RDTF. This is a multi-stakeholders committee of 16 members, all appointed by the Ministry. The group must be chaired by a representative of the MoH, and unfortunately for a number of months the

committee has remained without a permanent Chair. Other groups have provided valuable inputs to the Plan development - as above, the Parliamentary Group on RD as well as the Partnership for the NP for RD (the multi-stakeholders think-tank assembled to support the plan elaboration). ORPHAN has also contributed substantially to producing proposals.

Establishment of a budget for developing the plan/strategy

There was no specific indication of a budget being provided to support the development of the NP.

Theme 2 - Definition, Codification, Diagnostics and Inventorying of Rare Diseases

Introduction: Professor Jolanta Sykut-Cegielska - The Children's Memorial Health Institute
Systemic expectations and EUROPLAN evaluation: Stanisław Maćkowiak - President of Ars Vivendi Society

The definition of rare diseases is the one based on Recommendations of the European Council of June 2009. This is based on frequency of occurrence of specific disease in the total number of births. It is recommended by EU not to use the term of ultra-rare diseases, since it can divide patients while they all are equal. The definition is necessary to classify diseases and this in turn is necessary to create a register. It is also important for the statistics. Codification of rare diseases is difficult due to many different systems preferred by specific countries. For example ICD-10, ICD-9 systems are partially used in Poland, however these systems include only 200 diseases while currently over 7000 are known. Every day a new rare disease is discovered worldwide. Then it is necessary to update these systems and a new ICD-11 system is planned. However, it will be ready only in 2017. The task of updating should be implemented by such organizations as WHO or ORPHANET. It is also necessary to draft new regulations regarding introduction of new codes for rare diseases. It is recommended to create them based on the existing Orpha Code, because most of the rare diseases are classified there. Currently this is the most comprehensive system for coding of rare diseases, so it should be universally introduced also in Poland along with the national system. A proper training of the medical doctors and academia is also important. The best solution would be unification of the existing systems.

However, it is necessary to first determine the tasks - what do we need such a register/system for? Does it have to be used for implementation of social and economic goals, what would be its impact on the state budget, how to ensure its conformity with the requirements of personal data protection? Good example of register is the system introduced in France. It includes a small amount of sensitive information about the disease and the patient and it is aimed mostly on assessment of the population of patients suffering from a specific disease. Over 130 reference/experts centers are active there and the access to limited information is ensured for many. Access to the detailed information is provided to the limited number of persons.

Diagnosis: Very weak knowledge regarding rare diseases and their diagnosis among family doctors creates a problem in Poland. It is necessary to assess access to diagnosis not only to examinations because this access is also on a very low level. It is necessary to check and make public how many clinical doctors in our country can diagnose rare diseases. It is necessary to improve access to genetic, prenatal and screening tests. The positive fact is that in Poland about 70% of infants are screened for metabolic dysfunctions and soon 100% will be reached; moreover, there is an important increase of pre-symptomatic diagnostic. The quality of assessment of screening tests also should be improved. The system works relatively well; however, it is necessary to assess its efficiency.

Also monitoring of diagnosed patients is important. In Poland access to the DNA tests is poor. They are expensive and not everyone can afford them so they should be co-financed by the state budget. The number of skilled doctors who could correctly interpret DNA tests is low. It is necessary to analyze accessibility to such tests also for family members of diagnosed patients. It is necessary to identify and recognise the doctors who already encountered rare diseases not only in children but also in adults in the society. Standards of informing patients on rare diseases among doctors are bad and they should be improved. Understanding, empathy and psychological support are necessary.

Debrief Session Conclusions:

III. Codification and Inventorying of Rare Diseases

Poland currently uses ICD-9 and ICD-10 in its healthcare system, as well as MESH and OMIM codes among clinicians.

The team elaborating the NP hopes to adopt the Orpha Code alongside with the current coding systems: this would be beneficial as it is more specific than ICD-10 for RD and would yield a greater amount of data.

IV. Registries for Rare Diseases

There is no national registry of RD; however, and at present, some preparatory works started considering the presence of the RD registry within the Polish health informatics system. The Centre of Information Systems has a responsibility to facilitate the Parliament Act of April 2011 on information systems in healthcare (this is a regulation on the national policy on registries and data collection). It states that all registries have to comply with strict requirements, including data safety and protection. Despite such complexities, the need for a RD registry was emphasized several times throughout the conference and at the debrief session following the EUROPLAN conference - there is a recognition that registries are essential to improve data on RD and enable a more accurate estimate of the budgetary requirements of the NP (one cannot assess the full needs of RD patients and organize healthcare adequately without an understanding of the true scale).

The Ministry has pledged to explore the prospect of creating a national RD registry, although details are not yet available. Main needs in this respect appear to be:

- Technical and legal tools must be adopted in order to protect patients' data confidentiality, and to guarantee safe access to users (especially MDs and in particular geneticists);
- A stable source of funding;
- Links to European and international databases/registries.

Theme 3 - Scientific Research and Education / Information and Trainings on Rare Diseases - Research on Rare Diseases

Introduction: Professor Krystyna Chrzanowska - The Children's Memorial Health Institute

Systemic expectations and EUROPLAN evaluation: Maria Libura - President of the Polish Society for Support for the Persons Suffering from Prader-Willi Syndrome and Director of the Interdisciplinary Studies Institute for Rare Diseases of Łazarski's Academy.

It is necessary to create a kind of help desk receiving general information and then after some time providing answers and contacting respective centers. A team should be financed by the public resources. It must be in touch with all clinical centers, because in the light of rapidly growing knowledge gathering all necessary information is not possible. It is important to divide the helpdesk in separate units, one for patients and one for doctors who have some doubts and may contact the helpdesk. The redirection system would make their lives easier.

Some organizations of patients provide e-mail services for all rare diseases, but there is not a specialist who would be capable to answer all questions. In many cases several days are needed to provide an answer. The helpdesk would make sense if the questions could be also redirected to the respective specialist.

The most important and urgent actions to be taken in Poland according to the participants of the discussion include:

1. Training of employees of the health service in recognizing cases of rare diseases;
2. Introducing training modules regarding rare diseases for the medical students;
3. Establishing research and development programs dedicated to rare diseases;
4. Ensuring long term financial stability for research;
5. Ensuring systemic administrative support for the centers willing to obtain considerable international grants.
6. Mapping of existing research.

It is the task of the Minister of Health to first of all provide legal possibilities and designate the reference centers for treatment of specific rare diseases. It is a very difficult task.

The next element of the action is to create a database catalogue for rare diseases. A great number of new diseases is not corresponding with the classification commonly used worldwide (WHO ICD-10). It is not possible to include all the names of the rare diseases to many registers. Therefore creating a separate register and its dynamic development is a priority for the Minister of Health.

Debrief Session Conclusions:

V. Building a research program for RD

Currently there is no research program specifically for RD in Poland, nor specific funds were allocated for RD research. Researchers must compete with non-rare diseases on calls. There is significant activity in European research programmes, and Poland is an observer of the E-Rare 2 project (the Polish partner for E-Rare ERA-NET is the National Centre for Research and Development.) A key difficulty highlighted during the conference is the lack of awareness of current RD research projects taking place in Poland and/or at the EU level with Polish involvement. Similarly, too often, researchers seem only to learn of possible calls for funding after the deadlines have passed. The possibility of establishing a webpage providing both sets of information, on the MoH site or elsewhere, was discussed.

Theme 4 - Reference Centers, European Reference Networks / Cross-border healthcare

Introduction: Professor Małgorzata Krajewska Walasek - The Children's Memorial Health Institute
Systemic expectations and EUROPLAN evaluation: Krzysztof Swacha - President of the Foundation "Knowing to Help"

Chapter IV of the draft National Plan describes planned actions aimed at creation of reference centers for rare diseases. The question - who would be responsible for establishment and coordination of actions of such centers - still remains open. Currently operational diagnostic centers are not accredited with the Ministry of Health. It is necessary to establish a National Center for Rare Diseases which, beside its diagnostic and coordination functions, will also be a center certifying activities of other reference centers. In case of rare diseases, the knowledge of family doctors in many cases is insufficient. Consultations aimed at establishing such centers with patients' organizations are necessary. Patients' organizations suggest bigger engagement of the European Union financial resources in the creation of reference centers in Poland.

Doctors should have a better access to a platform enabling exchange of expertise with partners from other countries. Unfortunately in many cases they are not interested in such activities due to the lack of funds for participation in international congresses and seminars.

The "Highly specialized" approach of the centers diagnosing and treating rare diseases is yet another problem. Having in mind costs, comfort and security of the patients suffering from rare diseases and in particular children, it is necessary to improve the care obtained at home. The Ministry of Health does not react to the numerous initiatives and requests from the patients' organizations. Patients' organizations should establish better contacts with pharmaceutical companies who can also play an important role in increasing awareness of rare diseases.

Most of the patients rely on reference centers, but the reference doctors should also be commonly accessible. All this should be coordinated by the National Center for Rare Diseases. Unfortunately, the establishment of such a center is a political decision. Such decision should be included in the National Plan. Activity of such a center should be financed from the state budget. It should combine tasks related to health protection, social care and education. It should provide rare disease patients with coordinated multiphase and complex care.

Unfortunately, the fund for rare diseases will have to be established based on the political decision. We should approach the Commission Experts Group on Rare Diseases (former EUCERD) for assistance in determining the position of such center since the EUCERD adopted a set of indicators pertaining to the creation and operation of such institution. In this respect it is advisable to consider the EUCERD Recommendations on quality criteria for Centres of Expertise in EU Member States.

Debrief Session Conclusions:

VI. Reference Centers

There is no official designation policy as yet for centers of expertise (CE) for RD, and therefore there are no official CEs for RD. Around 10-15 centers have a reputation for expertise in a given field and provide diagnostic services and treatment to a varying degree. The "National Plan for Rare Diseases - the roadmap" proposes 4 CEs. There was a suggestion that the team may be able to look at the example of Polish genetics centers here.
The MoH is apparently defining the CE designation criteria, although there appeared to be some uncertainty amongst the conference delegates as to the specifics of this designation process. The need for the MoH to select robust, meaningful criteria was emphasized by many conference delegates, as was the importance of multidisciplinarity. Patients' organizations expressed a strong desire to contribute to the high-level discussions on how to designate Polish CEs for RD. One possibility presented at the conference was to have collaborating Centers

which fulfill strict criteria for a CE, and then Associated Centers, which house expertise without meeting all criteria.

VII. European Reference Networks

Several unofficial centers of expertise participate in international networks, but until the EC publish the Delegating and Implementing Acts, it is difficult to assess how CEs will in fact interact with the first ERNs.

Theme 5 - Social Care

Introduction: Dorota Korycińska - President of the Association of Patients suffering from Recklinghausen Disease and Other Phacomathosis Group Systemic expectations and EUROPLAN evaluation: Paweł Wójtowicz - President of the Foundation for Mucoviscidoze Patients and Their Families - Matio

Social care in rare diseases is still in its infancy in our country. The teams of doctors determining the level of inability do not have a clue about rare diseases neither have they received any information in this respect. For instance, the lack of an efficient system in Poland consists in placing patients affected with Huntington in psychiatric wards. Patients suffering from rare diseases have the right to benefit from programs for disabled persons. Many of the rare diseases do not meet the criteria adopted in Poland for determining inability.

In Poland, disability is determined in levels (3 levels), in many European countries in percentages. A person of the first level of disability is in fact not entitled to any social security benefits.

One of the elements to increase capacity to diagnose and early recognition are from the one side prenatal and genetic (screening) tests and from the other side to educate medical doctors to early recognize symptoms that may indicate that some health problems might be present. The question is how to teach doctors about each tiny symptom to allow them to determine whether they indicate a disease. The idea of the screening system is not only for early therapy but also for early inclusion into the general system: early diagnostics, early directed therapy, early rehabilitation and early social placement. In summary, in order to improve the Polish system regarding access to social care, there is the need for educating the doctors on rare diseases and the impact of the disease on the autonomy of the patient.

Theme 6 - Orphan Medicinal Products

Introduction: Łukasz Pera - Specialist in the Department of Drug Policy in the Ministry of Health
Systemic expectations and EUROPLAN evaluation: Mirosław Zieliński - President of the National Forum for Rare Diseases Treatment „Orphan"

Moderator: Dr Krzysztof Łanda - President of the Watch Health Care Foundation

Poland must not lag behind other EU countries however it is currently the case. The number of orphan drugs and technologies will be increasing and it is today that we have to start their inventory. The legal framework regulating access to treatments for patients suffering from rare diseases in Poland is not good. Since many years, the lack of an egalitarian approach to reimbursement of orphan drugs and technologies remains the main problem.

It is a constitutional duty of the state to ensure equal access to health protection funded from public resources. The question that bureaucrats ask in a case of rare diseases is: Is this a drug or technology of proven efficiency, meaning that the benefits from the treatment overcome the costs borne by the state in case of reimbursement?

Profitability is the relation of the treatment cost to the therapeutic effect. Different countries have different thresholds of profitability reflecting their welfare. Poland is not yet an attractive market for major pharmaceutical companies because worldwide the producers set the price based on price per QALY in the 5 wealthiest states. In case of Poland this level is so far inaccessible. The bureaucrats keep on asking the question: "What will be the result of the therapy?" However, without funding expensive therapies there is no progress on innovations.

The Agency for Assessment of Medical Technologies (AOTM) must take into account the "3xGDP per capita =QALY ratio" because this is the law in Poland. If the price of a drug is above that threshold set in the legislation, the drug is not reimbursed. However, the criteria used in economic analysis for drugs treating common diseases cannot be applied to orphan drug/ new technologies.

If during the assessment of the current technology we stick to the economic analyses, these should not be a decisive factor in making decisions on reimbursement. The notion of "intervention impact" is also subjective. The decision to register the drug or medical technology in the EU or in Poland is also important because it is always based on solid grounds like COMP evaluation prior to OMP designation. Therefore AOTM should broadly consider other aspects, including remaining elements of the "basket", because in many cases OMPs are very profitable, effective and enable decrease suffering of patients and their families, being not that costly as therapies themselves.

3xGDP per capita =QALY - why the Ministry of Health is so selective in applying this criterion? The Ministry should not be bound to this criterion by the law on reimbursement. It is necessary to strive for creating an egalitarian and solidarity-based budget. For example, article 12 item 13 should not apply to orphan drugs and technologies, which gained the orphan status based on other regulations. The MoH should not only look at the price of a drug but at its effectiveness.

The Ministry of Health should retreat from 3xGDP per capita=QALY in the case of a first medical technology of proven efficiency for a specific indication. In the event of a new active substance proposing an alternative to the first medical technology, the economic analyses based on 3xGDP per capita =QALY could then be applied.

For example, in case of advanced Parkinson disease (APD) resistant to traditional oral treatment, the treatment by DBS (Deep Brain Stimulation) is not a competition for Duodopa medical technology because Duodopa is the orphan medical technology of proven efficiency indicated for the very rare group of patients with advanced Parkinson disease. This is a proof to support the thesis that in such case, economic analysis must not be applied because the specific indication is not in competition with other therapies and it should be considered as a new indication.

The lack of presence of the decision makers of the Ministry of Health in this extremely important part of the Conference regretfully suggests that rare diseases are not a priority for them. Also there is no egalitarian approach in the draft text that is being prepared amending the law on reimbursement.

However, the presence of the representatives of the Parliament is a good sign. This may assist to better determine priorities. With regard to the Polish government the European Commission is best placed to try to invite the government to enforce the suggested solutions.

Debrief Session Conclusions:

VIII. OMP (Oprhan Medicinal Products) Access and Policy

OMP access was a central issue during the conference. The system of drug reimbursement changed on 1 January 2012 due to the Reimbursement Act of May 2011. The Minister of Health is not able to introduce reimbursement of a new drug without a prior official request from the Marketing Authorization Holder. 14 ODs are registered and reimbursed out of the 65 ODs that have received a Marketing Authorization from the EU. It was noted that the price of the drugs which are available was rather lower than that of most EU countries. There was some debate as to whether it is appropriate to distinguish here between what one may term ‘ordinarily rare’ diseases and ultra-rare diseases. As new medicinal products become available in country, the MoH announces this in a bimonthly list.

Additional Workshop

Day II - The debate on "Rare Diseases in Poland - where do we go?"

On September 28 2013 during the second day of EUROPLAN II Conference, a debate "**Rare diseases in Poland - where do we go**" was held. The debate tackled philosophic, scientific and medical aspects. Respected experts representing medicine, bioethics and pharmacotherapy as well as representatives of the Polish Parliament (Sejm) and national patients' organizations took part in the debate. During the meeting the discussion was held and some aspects regarding efficiency of orphan technology and drugs (ATA) and regulatory impediments in access to the therapy, as well as perspectives for implementation of the National Plan for Rare Diseases were discussed in details.

These are important issues which since long time raise many doubts and lead to the unnecessary social tensions. The purpose of the debate was to elaborate unified approach and recommendations for decision makers in the health sector in order to support them in their efforts aimed at improving scope and medical standard recommendations elaborated by stakeholders of the healthcare system focusing on rare diseases. The following conclusions and recommendations reflect directions striving to egalitarian treatment of rare diseases strongly and loudly formulated in the entire Europe, by patients, scientific and medical circles and by organizations and EU administrations offices. The presented approach, fully supported by the Polish National Forum for Rare Diseases Therapy "Orphan"and its member organizations, reflects the will and need to implement the EU priorities regarding rare diseases. The recommendations resulted from the public debate to provide support to the Minister of Health in proper implementation of state policy regarding rare diseases, as determined by the **EU Council Recommendation of June 8, 2009 regarding actions in the area of rare diseases (2009/C 151/02). Participants of the debate and their audience wait for actions and legislative changes in appropriate regulations resulting from these recommendations.**

Participants of the debate:

1. Barbara Czaplicka - Member of Parliament, Head of the Parliamentary Committee for Rare Diseases
2. Janina Okrągły - Member of Parliament, Deputy Head of the Parliamentary Committee for Rare Diseases
3. Lidia Gądek - Member of Parliament, Member of the Parliamentary Committee for Rare Diseases
4. Prof. Wojciech Cichy - I Pediatric Department - UM Poznań, Head of the Scientific Council of MATIO
5. Prof. Zbigniew Szawarski - National Institute for Public Health - PZH
6. Prof. Mieczysław Walczak - Head of the Coordination Team for Ultra-rare diseases
7. Gabriela Ofierska - Agency for Assessment of Medical Technologies
8. Dr Krzysztof Łanda - President of the Watch Health Care Foundation
9. Dr Marek Migdał - Institute „Children Health - Memorial", Member of the Rare Diseases Team at the MoH
10. Dr Paweł Miśkiewicz - Polish Association of the Orphan Drugs
11. Mirosław Zieliński - President of the National Forum for Rare Diseases Therapy
12. Stanisław Maćkowiak - President of the Federations of Polish Patients

Conclusions and recommendations resulted from the debate

1. The National Plan for Rare Diseases should be firstly budgeted in the following areas:

a) Establishment of the rare diseases register;
b) Establishment of the National Center for Rare Diseases as a main coordinating unit, certifying reference centers and monitoring the implementation and execution of the National Plan;
c) Reimbursement of orphan drugs and technologies.

That does not mean that the remaining activity areas covered by the National Plan pertaining to such topics as diagnosis, reference centers for rare diseases and their accreditation, proper pricing of highly specialized procedures, rehabilitation, education and social care should be abandoned.

On the contrary, adopting the concept of limited budget for the plan proves honesty in approach to the above mentioned topics and recognizing the fact that their artificial separation from the specific actions implemented by the competent authorities would be costly and challenging from the point of view of the recent accounting in the National Health Fund.

Implementation of the recommendations and actions in these areas included in the National Plan should have secured sources of financing from the budgets of specific ministries.

2. **Establishment of the National Center for Rare Diseases as an agency of the Ministry of Health is necessary.** The problem of rare diseases in Poland pertains to over 2 million citizens and creates an important segment of the health care. 75% of those 2 million are children. Such an important area requires a systemic approach and coordinated management. Predicted main tasks of the future Centre:

   - Maintaining a rare diseases register;
   - Implementation of the National Plan;
   - Monitoring and assessment of the National Plan implementation;
   - Accreditation of reference centers;
   - Place for affiliation of an existing Coordinating Team for ultra-rare diseases (qualifications for treatment program);
   - Presenting opinions regarding refund applications i.e. participation in the work of the Agency for Assessment of Medical Technologies (AOTM) and Economic Committee;
   - Representation of Poland in the work of the Commission Experts Group on Rare Diseases (former EUCERD) - it would be advisable that the Head of the Center participate in the meetings as a representative of Poland;
   - Social education and shaping up/influence on the medical education program.

3. **It is justified to separate the Fund for Rare Diseases out of the resources of National Health Fund (the payer).**
   The purpose of this Fund would be to reimburse orphan drugs and medical technologies (currently approximately 150 million Polish zlotys per year) and delegating this task to be implemented by the State budget (permanent provision in the budget law allocated within the funds of the Ministry of Health). The proper estimated “opening” budget of the fund is approximately 200-350 million Polish zlotys per year. However, the amount allocated to the Fund would be a political decision resulting from egalitarian and solidarity based approach to the rare diseases. The number of drugs will increase, currently out of 65 medical orphan products only 14 are reimbursed in Poland. Such low level and stagnation will create gaps in the future and result with the fact that Poland will lag behind in level of the access of citizens to the orphan drugs in EU. Implementation of the priority task regarding equal access to drugs for EU citizens with respect to orphan drugs and technologies will be threatened.

   It is necessary to underline that expensive therapies using orphan drugs have nothing to do with the employment and salaries on which the income of the National Health Fund is based (in Poland, people or employers pay the monthly health fee - it is 58% of a gross

salary and part of that money goes to the National Health Fund to pay for health services and reimbursement of drugs). Financing of expensive drug therapies has also nothing to do with the social insurance and it is a function and duty of the state resulting from an egalitarian treatment of rare diseases. This is expression of the solidarity of the state with those who for all their life time suffer from rare diseases. Separation of the dedicated Fund for Orphan Drugs is the only mean to make the level of the amounts designated for rare orphan drugs therapies independent from the economic condition of the National Health Fund.

4. **Revising criteria for properly assessing effectiveness of orphan therapies/ technologies**

Currently, at the EU level, works regarding amendment of the Directive on Transparency are ongoing. This will result in the fact that, starting from 2014, reassessing HTA will be banned in case of drugs and technologies which previously obtained the status of orphan medical product issued by COMP at EMA. Assessment of health technologies (HTA) applied for orphan drugs and technologies should have a scope different from the one currently applied by AOTM. The assessment of orphan drugs must consider the fact of their small quantity. The research for more efficient drugs is underway, so Poland, like other countries, should participate in this process by supporting research - the cost of the research is taken into consideration in pricing the orphan drugs. The currently applied HTA assessment process makes the assessment of clinical efficiency versus cost effectiveness of the orphan drug or technology unclear. In real terms, such assessment leads to lack of resources for reimbursement. This leads to uneven national HTA assessments across the EU; as a result, in some countries, like in Poland, citizens have more limited access to orphan medicinal products than citizens living in other EU countries. In many cases, the result of the Polish assessment of a drug turns out that this drug is considered inefficient and clinically ineffective in Poland, whereas it is assessed effective in many other European countries.

It is advisable that in Poland, with respect to orphan technologies and drugs, as the result of an egalitarian treatment of rare diseases, the scope of assessment being used by ATOM in the reimbursement process be limited in relation to orphan drugs and technologies. There is no justification for economic analysis for orphan drugs and technology. The evaluation should only indicate the impact on the budget of the payer and relate to the requested reimbursement records. This should apply to orphan drugs and technologies, which are the first medical technology that has proven effectiveness for specific indications. With each subsequent element / active substance, economic analysis based on 3xGDP per capita=QALY could be applied. This approach would be consistent with the logic of the EU Regulation 141/2000, which establishes a 10-year drug market protection in the case of the registration of the substances or technologies as an orphan by COMP. In cases

when a specific drug or technology proves not to be efficient in case of a specific patient, and despite the treatment no significant and continued progression of the disease would occur, then the treatment should be discontinued and the Polish reimbursement system provides for such solution. Therefore, in such individual cases and only in such cases, a competent Coordinating Team, and not the general regulations set by the reimbursement system, could decide to discontinue reimbursement of the therapy.

5. Recommendations for price and reimbursement of orphan drugs and technologies

It is recommended to amend the law (Reimbursement Act), so that ATOM, the Economic Committee and the Minister of Health are not bound by the statutory level indicator QALY when assessing and making the final reimbursement decisions in relation to orphan drugs and technologies. The use of the limitations pertaining to clinical cost-effectiveness in relation to rare diseases must not take place. For the evaluation and decisions concerning reimbursement, an egalitarian approach should be taken instead of an utilitarian one, as it is the case in relation to ordinary diseases where necessary. Thus, the constraint resulted from art. 12 item 13 of the Reimbursement Act requiring considering the threshold limit three times GDP per capita as an acceptable cost of obtaining an additional year of life, should not be applied in relation to orphan drugs and technologies. Currently, the threshold is around 111,000 Polish zlotys. As a consequence, a vast majority of orphan drugs and technologies do not reach the patients in Poland. Of the 65 drugs approved by the European Committee for Orphan Medicinal Products (COMP) at the EMA, only 14 are reimbursed in Poland. Moreover, one should remember that the reimbursement of orphan drugs is also a social investment in the development of science that in the near future will certainly result in increased number of newly invented drugs and therefore lowering price of these expensive costs.

Annexe 1 - Programme of the first day of the Conference EUROPLAN II - Rare Diseases, Poland

EUROPLAN II - RARE DISEASES, POLAND
WARSAW SEPTEMBER 27, 2013 - Hotel NOVOTEL

	8:30-9:30	**Registration of the participants**
1	9:30-10:00 **Conference Opening** Irys Room	1. Mirosław Zieliński - President of the Polish National Forum for the Rare Diseases Therapy 2. Igor Radziewicz - Winnicki - Undersecretary of State in the Ministry of Health 3. Yann Le Cam - EURORDIS Chief Executive Officer (Teleconference)
2	10:00-10:30 Plenary Session I Irys Room	**EUROPLAN Project - introduction, project presentation** 1. Yann Le Cam - CEO EURORDIS - teleconference 2. Mirosław Zieliński (Polish Rare Diseases National Alliance-(NA)
3	10:30-12:00 **Panel I** Irys Room	**Methodology and Governance of a National Plan** Introduction / Expectations towards Health System and EUROPLAN Evaluation: Miroslaw Zieliński (NA)
	12:0-12:30	**Coffee Break**
4	12:30-14:00 **Panel II** Irys Room	**Definition, codification and inventorying of RD** Introduction: Prof. Jolanta Sykut-Cegielska - The Children's Memorial Health Institute Expectations towards Health System and EUROPLAN Evaluation: Stanisław Maćkowiak - President of the Polish PMU Association Ars Vivendi
5	12:30-14:00 **Panel III** Gerbera Room	**Research and education / information and training** Introduction: Prof. Krystyna Chrzanowska - The Children's Memorial Health Institute Expectations towards Health System and EUROPLAN Evaluation: Maria Libura, President of Prader-Willi Association
	14:00-15:00	**Lunch**

6	15:00-16:30 **Panel IV** Irys Room	**Centres of Expertise / European Reference Networks/Cross Border Health Care** Introduction: Prof. Małgorzata Krajewska-Walasek - The Children's Memorial Health Institute Expectations towards Health System and EUROPLAN Evaluation: Krzysztof Swacha - President of the Foundation "Knowing to Help"
7	15:00-16:30 **Panel V** Gerbera Room	**Social care** **Social Services for RD** Introduction: Dorota Korycińska - President of the Alba Julia - the Association of Patients with the Recklinghausen Disease and Related Fakomatozis Group Expectations towards Health System and EUROPLAN Evaluation: Paweł Wójtowicz - Cystic Fibrosis Association Matio
	16:30:17:00	**Coffee Break**
8	17:30-18:45 **Panel VI**	**Orphan Drugs** Introduction: Łukasz Pera - Specialist at the Drug Department Policy at the Ministry of Health Expectations towards Health System and EUROPLAN Evaluation: Mirosław Zieliński (NA) and dr Krzysztof Łanda - President of the Watch Health Care Foundation
	Plenary Session II Irys Room	**Podsumowanie Polskiej Konferencji EUROPLAN II** Moderator: Chairman of the Rare Diseases Group Panelists: Stanisław Maćkowiak, Krzysztof Swacha, Maria Libura, Paweł Wójtowicz, Miroslaw Zieliński • **Closing of the Conference:** Miroslaw Zielinski (NA) • Barbara Czaplicka - Member of Parliament - Chairwomen of the Parliamentary Rare Diseases Task Force
	18:45	**Dinner**

Annexe 2 - Programme of the second day

Debate on "Rare Diseases in Poland - where do we go?"
Rare Diseases - Poland
Warsaw - September 28, 2013 - Hotel Novotel

1	9:30-10:00	**Opening and summary of the EUROPLAN II Conference.** **Methodology and purpose of the debate** • Mirosław Zieliński - President of the National Forum for Rare Diseases Therapy
2	10:00-11:30 Session I	**Round Table: National Plan for Rare Diseases - budgeting and coordination of the implementation, Orphan Medical Products Fund.** **Moderator:** Stanisław Maćkowiak - President of the Federations of Polish Patients **Participants of the debate:** 1. Barbara Czaplicka - Member of Parliament, Head of the Parliamentary Committee for Rare Diseases 2. Janina Okrągły - Member of Parliament, Deputy Head of the Parliamentary Committee for Rare Diseases 3. Lidia Gądek - Member of Parliament, Member of the Parliamentary Committee for Rare Diseases 4. Prof. Wojciech Cichy - I Pediatric Department - UM Poznań, Head of the Scientific Council of MATIO 5. Prof. Zbigniew Szawarski - National Institute for Public Health - PZH 6. Prof. Mieczysław Walczak - Head of the Coordination Team for Ultra-rare 7. Gabriela Ofierska - Agency for Assessment of Medical Technologies 8. Dr Krzysztof Łanda - Watch Health Care Foundation 9. Dr Marek Migdał - Institute „Children Health - Memorial", Member of the Rare Diseases Team 10. Dr Paweł Miśkiewicz - Polish Association of the Orphan Drugs 11. Mirosław Zieliński - President of the National Forum for Rare Diseases Therapy
	11:30-12:00	***Coffee Break***

3	12:00-14:00 Session II	**<u>Round Table:</u> Philosophy towards OMP: Rare Diseases and HTA as well as the issues of access to orphan medical products and technologies** **Moderator:** Mirosław Zieliński - President of the National Forum for Rare Diseases Therapy **<u>Participants of the debate:</u>** 1. Barbara Czaplicka - Member of Parliament, Head of the Parliamentary Committee for Rare Diseases 2. Janina Okrągły - Member of Parliament, Deputy Head of the Parliamentary Committee for Rare Diseases 3. Lidia Gądek - Member of Parliament, Member of the Parliamentary Committee for Rare Diseases 4. Prof. Wojciech Cichy - I Pediatric Department - UM Poznań, Head of the Scientific Council of MATIO 5. Prof. Zbigniew Szawarski - National Institute for Public Health - PZH 6. Prof. Mieczysław Walczak - Head of the Coordination Team for Ultra-rare 7. Gabriela Ofierska - Agency for Assessment of Medical Technologies 8. Dr Krzysztof Łanda - Watch Health Care Foundation 9. Dr Marek Migdał - Institute „Children Health - Memorial", Member of the Rare Diseases Team 10. Dr Paweł Miśkiewicz - Polish Association of the Orphan Drugs 11. Stanisław Maćkowiak - President of the Federations of Polish Patients
	14:00-14:15	**Conference closing** Mirosław Zieliński - President of the National Forum for Rare Diseases Therapy Barbara Czaplicka - Member of Parliament, Head of the Parliamentary Committee for Rare Diseases
	14:15	**Lunch**
	16:00	**Social event: Rare diseases are common".** Installation of the poster on the Palace of Culture and Rare Diseases Agents Street Action.
4	17:00	**Internal Debrief Session (organizers, selected decision makers, EURORDIS)**

Annexe 3: Participants List

KONFERENCJA EUROPLAN
WARSZAWA 27-28.09.2013
RZADKIE CHOROBY POLSKA
European Project for Rare Diseases National Plans Development
RARE DISEASES POLAND

based on the registration provided on the Conference website www.rzadkiechoroby.pl/EUROPLAN

#	Name	Surname	Title	Organization
1	Magdalena	Knefel	Rada KFO	Krajowe Forum Orphan
2	Dorota	Siemczuk		
3	Marek	Parowicz	wiceprezes zarządu	Polskie Stowarzyszenie na Rzecz Osób z AHC ahc-pl
4	Małgorzata	Szufa	członek	Polskie Stowarzyszenie na Rzecz Osób z AHC ahc-pl
5	Krystyna	Parowicz	członek	Polskie Stowarzyszenie na Rzecz Osób z AHC ahc-pl
6	Mirosław	Zieliński	Prezes	Krajowe Forum na rzecz terapii chorób rzadkich
7	Katarzyna	Wertheim-Tysarowska	dr	Instytut Matki i Dziecka
8	Przemysław	Sobieszczuk	Prezes Zarządu	Stowarzyszenie Debra Polska Kruchy Dotyk
9	Danuta	Lis	Prezes	Polskie Stowarzyszenie Choroby Huntingtona
10	Emilia	Iwaniuk		Fundacja Umieć Pomagać
11	Jolanta	Zagórska		
12	Dorota	Korycińska	prezes zarządu	Stowarzyszenie Alba-Julia
13	Marek	Zagórski		
14	Joanna	Bubak	Prezes Zarządu	Sarko Stowarzyszenie
15	Danuta	Wojtowicz	Asystentka Zarządu	Sarko Stowarzyszenie
16	Tomasz	Antos	V-ce Prezes Zarządu	Sarko Stowarzyszenie
17	Waldemar	Janiec	Prof. dr hab n. med	emeryt
18	Robert	Śmigiel	dr hab, Adiunkt	Katedra Genetyki Uniwersytetu Medycznego we Wrocławiu
20	Stanisława	Gdula	zastępca Prezesa	Stowarzyszenie Rodzin z Ataksją Rdzeniowo-Móżdżkową"Ataksja"
21	Franciszek	Gajek	Prezes Stowarzyszenia	Stowarzyszenie Rodzin z Ataksją Rdzeniowo-Móżdżkową"Ataksja"
22	Marek	Karwacki	dr n. med.	Instytut Matki i Dziecka
23	Elżbieta	Blum	PREZES ZARZĄDU	STOWARZYSZENIE NA RZECZ DZIECI I

				MŁODZIEŻY Z DYSPLAZJĄ EKTODERMALNĄ ORAZ ALERGIĄ "JESTEŚMY"
24	Joanna	Łaba	lekarz/prezes	TRISO-OPOLSKIE STOWARZYSZENIE na RZECZ ROZWOJU DZIECI Z TRISOMIĄ 21
25	Aleksander	Łaba	konsultant	TRISO-OPOLSKIE STOWARZYSZENIE na RZECZ ROZWOJU DZIECI Z TRISOMIĄ 21
26	Andrzej	Mazurek		Swedish Orphan Biovitrum Sp. z o.o. Oddział w Polsce
27	ANNA	Doboszyńska	Kierownik Kliniki Pulmonologii UWM	Uniwersytet Warmińsko-Mazurski
28	Piotr	Tołwiński	Medical Manager	Orphan Europe Przedstawicielstwo w Polsce
29	Katarzyna	Janiszewska	Asystentka Zarządu	Sarko Stowarzyszenie
30	Wiesław	Zacher		Polskie Stowarzyszenie na Rzecz Osób z AHC ahc-pl
31	Łukasz	Pera	Naczelnik	Ministerstwo Zdrowia
32	Dorota	Hedwig	Prezes	Polskie Towarzystwo Walki z Mukowiscydozą
33	Anna	Skoczylas-Ligocka	Dyrektor Zarządzająca	Polskie Towarzystwo Walki z Mukowiscydozą
34	Marcin	Mleczko	Dyrektor Biura	Fundacja MATIO
35	Przemysław	Marszałek		Fundacja MATIO
37	Ewa	Wróbel	Rzecznik Prasowy	Fundacja MATIO
38	Igor	Radziewicz-Winnicki	Wiceminister	Ministerstwo Zdrowia
39	Jerzy	Zacher	chory	Polskie Stowarzyszenie na Rzecz Osób z AHC ahc-pl
40	Krzysztof	Swacha	Fundator	Fundacja UMIEĆ POMAGAĆ
41	Katarzyna	Parafianowicz	Wiceprezes Zarządu	Fundacja Serce Dziecka
42	Ewa	Michalska	członek Rady Fundacji	Fundacja Serce Dziecka
43	Barbara	Bieniasz		
44	Alicja	Morze		
45	Katarzyna	Ziaja	magister pielęgniarstwa	
46	Martyna	Latała		
47	Wojciech	Oświeciński	Prezes	Stowarzyszenie Rodzin z Chorobą Gauchera
48	Mieczysław	Walczak		
49	Małgorzata	Krajewska-Walasek	prof. dr hab. n. med./kierownik Zakładu GenetykiMedycznej IP-CZD	IP-CZD

50	Artur	Nowicki	Doradca	Krajowe Forum na Rzecz Terapii Chorób Rzadkich
51	Anna	Kostera-Pruszczyk	dr hab. med.	Klinika Neurologii Warszawski Uniwersytet Medyczny
52	Kamil	Dolecki	Prezes Zarządu	Stowarzyszenie Pomocy Chorym na Mięsaki „SARCOMA”
53	Roman	Michalik	prezes	Stowarzyszenie Rodzin z Chorobą Fabry'ego
54	Joanna	Girulcka-Michalik		Stowarzyszenie Rodzin z Chorobą Fabry'ego
55	Stanisław	Maćkowiak	Wiceprezes	Krajowe Forum ORPHAN
56	Bożena	Dembowska-Bagińska	Dr hab n med/ Prof nadzw	Instytut Pomnik Centrum Zdrowia Dziecka
57	Mirosław	Bik-Multanowski	Dr hab med	Katedra Pediatrii UJ, Uniwersytecki Szpital Dziecięcy w Krakowie
58	Marek	Migdał	Dr n.med	Instytut „Pomnik-Centrum Zdrowia Dziecka”
59	Marcin	Karpiński		
60	Bogumiła	Kania	członek Zarządu Stowarzyszenia	Mazowieckie Stowarzyszenie Osób z Chorobą Parkinsona
61	Wojciech	Kania	członek Stowarzyszenia	Mazowieckie Stowarzyszenie Osób z Chorobą Parkinsona
62	Paweł	Wójtowicz	Prezes Zarządu	MATIO Fundacja Pomocy Rodzinom i Chorym na Mukowiscydozę
63	Maria	Kubaszewska-Rogal		internetowa grupa wsparcia-ziarniniak Wegenera
64	Aleksandra	Baran	Wolontariusz	Umieć pomagać
65	Maria	Libura	Dyrektor ISInCR	Uczelnia Łazarskiego
66	Aleksander	Janiak		Biuro Rzecznika Praw Obywatelskich
67	Beata	Kusy	Wolontariusz	Umieć pomagać
68	Agnieszka	Grzybowska	Country Manager Genzyme	Sanofi-Aventis
69	Łukasz	Kołodziejczyk	Wolontariusz	Umieć Pomagać
70	Gabriela	Ofierska-Sujkowska		
71	Aleksander	Wiechowski	członek Stowarzyszenia/przedstawiciel Zarządu na Konferencję	Mazowieckie Stowarzyszenie Osób z Chorobą Parkinsona
72	Dorota	Sieńko	pacjent	Stowarzyszenie Gauchera
73	Julita	Sarnowska-Sieńko		Stowarzyszenie Gauchera
74	Ewa	Rogozińska		Stowarzyszenie Rodzin z Chorobą Fabry’ego
75	Dariusz	Rogoziński		Stowarzyszenie Rodzin z Chorobą

				Fabry'ego
76	Krystyna	Chrzanowska	Prof. dr hab. n. med.	Instytut „Pomnik-Centrum Zdrowia Dziecka"
77	Tamara	Cierpiałowska	dr n. hum. adiunkt	Instytut Pedagogiki Specjalnej, Uniwersytet Pedagogiczny w Krakowie
78	Jacek	Sztajnke	Prezes	Fundacja Parent Project Muscular Dystrophy
79	Marek	Surowiec	Naczelnik Wydziału Oceny Dokumentacji Badań Klinicznych	Urząd Rejestracji Produktów Leczniczych, Wyrobów Medycznych i Produktów Biobójczych
80	Marta	Domańska		
81	Krzysztof	Łanda	Fundator	Fundacja Watch Health Care
82	Jan	Kawalec	Prezes Zarządu	Stowarzyszenie Marfan Polska
83	Joanna	Sobieszczuk	członek stowarzyszenia/opiekun	Stowarzyszenie Debra Polska Kruchy Dotyk
84	Agnieszka	Laskowska		
85	Paulina	Gil-Kulik	mgr	Zakład Genetyki Klinicznej
86	Alicja	Niedojadło		
87	Jolanta	Karwat	mgr dietetyk, doktorantka	Zakład Genetyki klinicznej UM w Lublinie
88	Agnieszka	Nosalik	Mama dziewczynki z SMA I/II	
90	Iveta	Spolnikova	Prezes	Fundacja Salemander
91	Igor	Nowak		
92	Katarzyna	Butryn	doktorant	Uniwersytet Jagielloński
93	Anna	Jankowska	skarbnik / matka dziecka chorego	Oddział regionalny PTChN-M w Gdańsku
94	Agnieszka	Ługowska		Instytut Psychiatrii i Neurologii
95	Agnieszka	Zakrzewska		
96	Łukasz	Zakrzewski		
97	Ewa	Kobierska		Polskie Stowarzyszenie Pomocy Osobom z Zespołem Prader Willi
98	Mirosława	Górna		
99	Ewa	Górna		
100	Diana	Saniewska		
101	Edyta	Saniewska		
102	Lidia	Kotuła	lekarz	SPSK 4
103	Anna	Wydrych		
104	HALINA	SPIRYDOŃSKA		Polskie Stowarzyszenie na Rzecz Osób z AHC ahc-pl
105	Anna	Boguszewska-	Prezes Zarządu	Genomed

		Chachulska		
106	Paulina	Gmaj	Członek Zarządu	Stowarzyszenie Pomocy Chorym na Mięsaki SARCOMA
107	Adam	Komar	Prezes Zarządu	Fundacja Potrafię Pomóc na Rzecz Dzieci Niepełnosprawnych z Wadami Rozwojowymi
108	Ksawery	Szczepanik		
89	Ela	Malarowska		
109	Hanna	Milczarek	Koordynator	FPP
110	Karolina	Paciorek	Specjalista ds Komunikacji	FPP
111	Jarosław	Geryń	Przedstawiciel Abbvie	
112	Olga	Shulga		
113	Elżbieta	Oleksiak	koordynator Projektu "Wsparcie osób z zespołami uwarunkowanymi genetycznie"	Polski Związek Niewidomych
114	Izabella	Różańska		
115	Karolina	Zwierzchowska		
116	Mariola	Kowalska	Prezes	Fundacja Równi Wśród Równych
117	Jolanta	Marchewicz		
118	Maciej	Ptasiński	prezes	Stowarzyszenie Pacjentów z Chorobą Pompe w Polsce
119	Marcelina	Bednarz	członek	Stowarzyszenie Pacjentów z Choroba Pompe w Polsce
120	Anna	Sułek		
121	Wioletta	Krysa		
122	Ewelina	Elert-Dobkowska		
123	Marta	Rajkiewicz		
125	Marcin	Karpiński		
126	Iwona	Stępniak	specjalista neurolog, w trakcie specjalizacji z genetyki klinicznej	Instytut Psychiatrii i Neurologii
127	Zbigniew	Szawarski	Profesor	Narodowy Instytut Zdrowia Publicznego - PZH
128	Barbara	Czaplicka	Poseł na Sejm RP, Przewodnicząca Parlamentarnego Zespołu ds. Chorób Rzadkich	Platforma Obywatelska
129	Wojciech	Cichy	Profesor	Uniwersytet Medyczny w Poznaniu
130	Adam	Kraszewski	radca prawny	Kancelaria GESSEL
131	Agnieszka	Bemowska		
132	Joanna	Hryć	właściciel	Stricte PR Sp. z o.o.

133	Paweł	Woźniak	Dyrektor Zarządzający	Komtur Polska sp. z o.o.
134	Małgorzata	Rogaszewska	Medical Manager	ORPHAN EUROPE
135	Lidia	Gądek	Poseł na Sejm RP	Platforma Obywatelska
136	Małgorzata	Szczygielska		NUTRICIA Polska
137	Luiza	Matejuk	Pani	Vitaflo
138	Adrian	Baciu	Consultant	PR
139	Emilia	Chętnik	Biolog	
140	Patrycja	Maćkowiak		Komtur Polska sp. z o.o.
141	Artur	Kędziora	Członek Zarządu	TO BE Group
142	Renata	Czuba		
143	Jolanta	Wierzba	dr med	Uniwersytet Gdański
144	Urszula	Klajmon-Lech	dr adiunkt	Wydział Etnologii i Nauk o Edukacji, Uniwersytet Śląski
145	Joanna	Kowalska	dr	Uniwersytet Warszawski
135	Bożena	Sławiak	Poseł RP	Sejm RP
146	Magda	Czapka		
147	Irena	Wojda		
148	Sławomir	Kowalski	pacjent	osoba prywatna chora na cystynozę
149	Małgorzata	Pacholec	dyrektor	Polski Związek Niewidomych
150	Janina	Okrągły	Poseł na Sejm, Wiceprzewodnicząca Parlamentarnego Zespołu ds. Chorób Rzadkich	Platforma Obywatelska
151	Karolina	Ziora-Jakutowicz	lekarz	IPiN
153	Krystyna	Spodar	dr n med	Centrum onkologii - Instytut
154	Vlasta	Zmazek	EURORDIS representative	
155	Victoria	Hedley	EURORDIS representative	
156	Amalia	Egle Gentile	EURORDIS representative	
157	Ariane	Weinman	EURORDIS representative	

Annexe 4 - Core Indicators - Definitions and associated answers

Indicator	Area of Council Reg. (2009 /C151/02)	Short Answer	Detailed Answer	Votes	Type of indicator	Comments
Existence of Regulations/Laws, or equivalent official national decisions that support the establishment and development of a Rare Diseases (RD) Plan	1	YES	**YES**, existing, fully embedded in a regulation/law/official national decision	**0**	Process	In Poland the works on the National Plan have begun from 2011. After development by experts and patients' organizations, a draft was presented to the current Minister of Health Mr. Bartosz Arłukowicz and then it was sent to the Team for the Rare Diseases at the Ministry of Health. By the end of December of 2012 a final version was developed and it was approved by the Ministry Board in April 2013.
			YES, existing, partially embedded	**25**		
		In progress/ development		**32**		
		NO		**13/9 abstain**		
Existence of a RD advisory committee	1	YES	**YES**, exists and meets regularly and includes all relevant stakeholders	**1**	Process	The task of elaborating a National Plan (NP) for Rare Diseases (RD) was assigned to the advisory committee set-up by the MoH - the Team for the Rare Diseases. Also the Parliamentary Group on RD was created in 2012.
			YES, exists but partly functioning and includes all relevant	**14**		From April 2013 the works of the Team are at a dead point because the Team was deprived of the Head, whose task was to coordinate its

			stakeholders			works. As a consequence, neither inter-ministerial nor social consultations required for such documents were held.
			YES, exists and meets regularly but does not include all relevant stakeholders	**5**		
			YES, exists but partly functioning and does not include all relevant stakeholders	**40**		
		NO		**3/17 abstain**		
Permanent and official patients' representation in plan development, monitoring and evaluation	6	YES	Yes, at all stages	**0**	Process	
			Yes, but only as observers	**6**		
			YES, but only consulted before the final document is approved	**22**		
		NO		**2/42 abstain**		
Adoption of EU RD definition	2	YES	**YES**, the NP/NS measures are applied	**32**	Process	

			using the EU definition			
			YES, but the NP/NS measures are applied using a different definition	**0**		
		NO	*Please, specify the definition used in the NP/NS*	***14***		
Existence of national policy for establishing Centers of Expertise for RD	4	YES	Yes, existing, fully implemented	**0**	Process	
			Yes, existing, partly implemented	**0**		
		In Progress/ development		**0**		
		NO		**55**		
Number of national and regional Centers of Expertise adhering to the national policy	4	*Number*	*Number of CEs complying with the national policy*	**-**	Outcomes	All participants of the session (56) agreed that there are no such Centers and thus such statement should not be a subject of voting.
			Number of CEs/million inhabitants	**-**		All participants of the session (56) agreed that there are no such Centers and thus such statement should not be a subject of voting

			Number of CEs fulfilling EUCERD criteria	**-**		All participants of the session (56) agreed that there are no such Centers and thus such statement should not be a subject of voting
Participation of national or regional centers of expertise in European Reference Networks	4	Number of CEs participa-ting in ERNs as full members		**-**	Outcomes	If there are no such centers, they cannot participate in any network. Thus it cannot be subject of voting.
		Number of CEs participa-ting in ERN as associated members		**-**		If there are no such centers, they cannot participate in any network. Thus it cannot be subject of voting.
NP/NS support to the development of/participation in an	2	YES	Yes, national	**0**	Process	
			Yes, regional	**0**		

information system on RD		NO		**0**		
		Participa-tion in Orphanet Joint Action	YES, participates in Orphanet JA and produces information in national language(s)	**26**		
			YES, participates in Orphanet JA and does not produce information in national language(s)	**0**		
			NO	**0**		
Existence of Help lines for RD	2 i 6	YES, supported from by public funding	Yes, only for professionals	**0**	Process	
			Yes, only for patients	**0**		
			Yes, both for professional and patients			
		. YES, supported by private funding	Yes, only for professionals	**0**		
			Yes, only for patients	**0**		

			Yes, both for professional and patients	**0**		
		YES, supported by private and public funding	Yes, only for professionals	**0**		
			Yes, only for patients	**0**		
			Yes, both for professional and patients	**0**		
		NO		**32**		
Knowledge, Classification/Coding, Registries and Research						
. Existence of a national policy for developing, adapting and implementing clinical practice guidelines	2	YES	**YES**, a policy exists for developing CPGs	**0**	Process	
			YES, a policy exists for adapting CPGs	**0**		
			YES, a policy exists for implementing CPGs	**0**		
		NO		**29**		
. Type of classification /coding used by the health care system	2	*Type of coding system used*	ICD-9	**48**	Process	Coding systems ICD-9, ICD-10 and OMIM are used in Polish health-care system
			ICD-10	**48**		

			OMIM	**48**		
			SNOMED			
			MESH			
			ICD-O			
			Others			
		ORPHA code is used in addition to existing system	Yes	**48**		
			NO	**0**		
. Existence of a national policy on registry and data collection on RD	2 i 3	YES	**YES**, for national/centralized registry and data collection	**13**	Process	
			YES, for regional registry and data collection	**5**		
		NO		**33/6 abstain**		

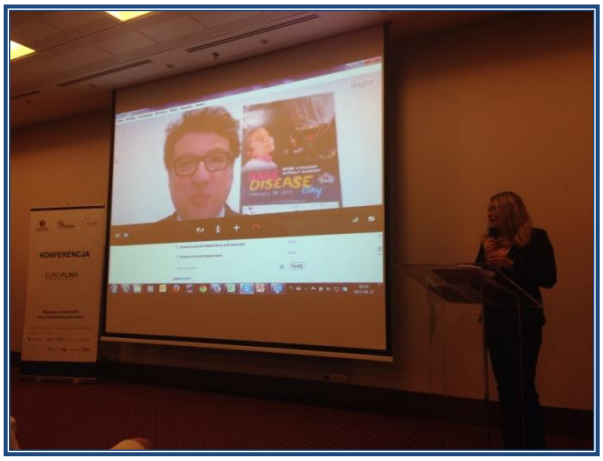
. Existence of a RD research programmes/ projects in the Country	3	YES	Yes, specific research program	**0**	Process	
			YES, specific PROJECTS for RD within general research program	**25**		
		NO		**0**		
. Participation in European and international research initiatives	3	YES	Yes, E-RARE	**28**	Process	
			Yes, IRDiRC	**0**		
			Yes, others	**26**		
		NO		**0**		
. Number of Orphan Medical Products (OMPs) with a European Union marketing authorisation and available in the country (i.e. priced and reimbursed or directly supplied by the NH system)	5	*Number: 14*			Outcomes	This data is based on the National Health Payer records and cover only so-called therapy/medicine programs, mostly for metabolic disorders.
. Existence of a governmental system	5	YES		**1**	Process	

for compassionate use of medicinal products		In progress				
		NO		**70**		
Social Services						
. Existence of programs to support in their daily life RD patients integration	6	YES	**YES**, people living with RD can access general program for persons with disability	**8**	Process	
			YES, there exist specific actions to enable real access for people living with RD to general social/ disability programs (e.g. training, guidelines for social workers, etc.)	**0**		
			YES, there exist specific programs for people living with RD	**0**		
		In progress				
		NO		**20**		
FINANCIAL SUPPORT INDICATORS ***(IMPLEMENTATION OF THE PLAN/STRATEGY)***						

. Existence of a policy/decision to ensure long-term funding and/or sustainability of the measures in the RD plan/strategy	7	YES	**YES**, a policy/decision to ensure long-term sustainability	**0**	Process	
			YES, a budget exists for the plan	**0**		
		In progress		**0**		
		NO		**75**		
. Amount of public funds allocated to the RD plan/strategy	7	*Number*	Value		Outcomes	No such funds allocated
			Value / million inhabitants			
			Value available partially: only for funds allocated exclusively to National Plan (N/A for funds allocated in the general budget)			
			N/A: it is incorporated in the general budget			
. Specific public funds allocated for RD research	3	YES		**0**	Process	
		In development				
		NO		**30**		

. Public funds specifically allocated for RD research actions/projects per year since the plan	3	*Number*	Value		Outcomes	No such funds allocated
			Value available partially: only for funds allocated exclusively to National Plan (N/A for funds allocated in the general budget)			
			N/A: it is incorporated in the general research funds			

Annexe 5 - Pictures of the event




Narodowy Plan dla Chorób
Rzadkich – mapa drogowa
KONFERENCJA


KONFERENCJA
EUROPLAN
Rzadkie Choroby – dokąd zmierzamy?
Warszawa, 28 września 2013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Orphan
EURORDIS
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