Warsaw, Feb 29, 2012

SYSTEMIC ASSUMPTIONS
for the development of the
NATIONAL PLAN FOR RARE DISEASES for 2013-2017

The systemic assumptions for the development of the National Plan for Rare Diseases have been established as part of an initiative of the National Forum for Therapy of Rare Diseases to provide the Rare Diseases Task Force at the Polish Ministry of Health with information, opinions and expectations collected in 2011 during consultation meetings with patients and medical and scientific groups. The initiative was launched following the EU Council recommendation on the involvement of patients and their representatives in the political process and promoting the activities undertaken by patient groups and associations to support rare diseases. The following parties were involved in the preparation of the systemic assumptions:

- Members of the umbrella patient organization ORPHAN National Forum for Treatment of Rare Diseases:

  1. Ars Vivendi Polish Association for Patients with Phenylketonuria and Rare Diseases (*Polskie Stowarzyszenie Pomocy Chorym na Fenyloketonurię i Choroby Rzadkie "Ars Vivendi"*)
  2. "Knowing How To Help" Foundation for Patients with Mucopolissacharidosis (*Fundacja „Umieć Pomagać" na rzecz osób chorych na Mukopolisacharydozę*)
  3. Polish Neuromuscular Diseases Association (*Towarzystwo Chorób Nerwowo-Mięśniowych*)
  4. Association for Families with Gaucher's Disease (*Stowarzyszenie Rodzin z Chorobą Gauchera*)
  5. Association for Families with Fabry's Disease (*Stowarzyszenie Rodzin z Chorobą Fabry'ego*)
  6. MATIO Foundation for Families and Patients with Cystic Fibrosis (*MATIO Fundacja Pomocy Rodzinom i Chorym na Mukowiscydozę*)
  7. Debra Poland Fragile Touch Association (*Stowarzyszenie Debra Polska Kruchy Dotyk*)
  8. Save a Life Association (*Stowarzyszenie Uratujmy Życie*)
  9. Polish Association for Patients with Prader-Willi Syndrome (*Polskie Stowarzyszenie Pomocy Osobom z Zespołem Prader-Williego*)
  10. "Here We Are" Association for Support of Children and Youth with Ectodermal Dysplasia and Allergy (*Stowarzyszenie na rzecz dzieci i młodzieży z dysplazją ektodermalną oraz alergią "Jesteśmy")*
  11. Dina Radziwiłłowa Child's Heart Foundation (*Fundacja Serce Dziecka im. Diny Radziwiłłowej*)
  12. Polish Huntington's Disease Association (*Polskie Stowarzyszenie Choroby Huntingtona*)
  13. Alba-Julia Association for Patients with Recklinghausen's Disease and other Neurocutaneous Syndromes (*Alba-Julia - Stowarzyszenie Pacjentów z Chorobą Recklinghausena i innymi schorzeniami z grupy fakomatoz*)
  14. Polish Association for Patients with Wilson's Disease (*Polskie Stowarzyszenie Pacjentów z Chorobą Wilsona*)
  15. Polish Association for Patients with Pulmonary Hypertension and their Friends (*Polskie Stowarzyszenie Osób z Nadciśnieniem Płucnym i Ich Przyjacół*)
  16. "J-ELITA" Polish Association Supporting People with Inflammatory Bowel Disease (*Polskie Towarzystwo wspierania osób z nieswoistymi zapaleniami jelita "J-ELITA"*)
  17. Association for Patients with Tuberous Sclerosis (*Stowarzyszenie Chorych na Stwardnienie Guzowate*)
  18. Equal among the Equal Foundation (*Fundacja Równi Wśród Równych*)

19. and other organizations cooperating with the National Forum for Treatment of Rare Diseases.

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- Stanisław Maćkowiak: Area A: Classification and Register of Rare Diseases
- Krzysztof Swacha: Area B: Diagnostics of Rare Diseases
- Mirosław Zieliński: Area C: Healthcare
- Paweł Wójtowicz: Area D: Integrated Social Assistance
- Magdalena Knefel: Area E: Information, scientific research and education

On behalf of Polish rare disease patients and their families, we would like to thank all involved in the work on this document for their precious input and the time devoted to the patient initiative in connection with the initiation of work the National Plan for Rare Diseases. We hope that the National Plan which has been long-awaited by patients, their families and physicians will be developed and approved for implementation not later than by the end of 2012 as promised by the Minister of Health. Polish patient organizations are willing and available to cooperate on this initiative.

Kind regards,

Mirosław Zieliński
President
National Forum for Treatment of Rare Diseases - ORPHAN

I. General Outline of the Plan

1. Background

Rare diseases are disorders which are very uncommon to occur; they are usually genetic, follow a chronic and severe course and are in most cases detected during childhood. Due to the rarity of their occurrence and the difficulty in their detection as well as the lack of public awareness, so far little has been known about these diseases. Patients suffering from rare diseases are particularly isolated and vulnerable.

Rare diseases have been granted a special status in the European Union legislation on April 29, 1999 (No. 1295/1999/EC) when the European Commission adopted a programme of Community action on rare diseases within the framework for action in the field of public health (1999-2003). It is pointed out in the European Parliament and the Council Regulation No. 141/2000 dated December 16, 1999 that patients suffering from rare diseases should be entitled to receive the same quality of treatment as other patients.

Pursuant to the decision adopted jointly by the European Parliament and the EU Council (No. 1350/2007/EC) establishing a second programme of Community action in the field of health (2008 – 2013), **COUNCIL RECOMMENDATION of June 8, 2009 on action in the field of rare diseases (2009/C 151/02)** was issued for member states to develop and implement national plans in the field of rare diseases to ensure access to high quality medical care for rare disease patients, including diagnostics, treatment and rehabilitation as well as access to orphan drugs.

The list of rare diseases is very long; currently (as of Jan 20, 2012) there are 5954 diseases registered in the European Orphanet database. Patients suffering from particular diseases have distinctively different needs; however, the ultimate objective is to ensure that the healthcare system provides and facilitates access to professional and comprehensive medical care for patients, particularly in medical centres offering relevant experience and a complete range of diagnostic and treatment services.

2. Rare Disease Definition

It is currently estimated that there are approximately 6000 recognized rare diseases. They affect from 6% to 8% of the population. Following the rare disease definition recommended by the European Union, a disorder is considered in Poland as a rare disease if it affects no more than 5 in 10,000 people.

3. Structure of the future National Plan

It is suggested that a European standard for a national plan be set according to the outline worked out as part of the Europlan Project developed at request of the European Commission based on the report of the Polish EUROPLAN Conference and the experience of other national EUROPLAN Conferences. Based on panel discussions conducted in 2010-2011 as part of consultation meetings, it was agreed that the following target structure of the detailed outline of the Polish National Plan for rare diseases should be recommended:

A. Classification and Register of Rare Diseases

- Codification of Rare Diseases
- Register of Rare Diseases

B. Diagnostics of Rare Diseases

- Widespread use of screening
- Widespread use of genetic testing

- System of Centres of Reference

C. Healthcare for Rare Disease Patients

- Increase accessibility of highly specialized medical care
- Increase accessibility of drug treatment
- Improve the patient rehabilitation system

D. Integrated Social Assistance for Rare Disease Patients and their Families

E. Information, scientific research and education in the field of rare diseases

- Research development and support
- Medical education
- Social education

F. Plan Implementation Monitoring

II. Detailed Outline of the Plan

A. Classification and Register of Rare Diseases

1. Codification of Rare Diseases

The codification of disease entities may help to identify and classify rare diseases. This will help to adopt uniform decisions such as, for instance, a certificate of patient's disability irrespective of the makeup of the adjudicating panel. Decisions that are appealed (due to fact that members of the adjudicating panel lack sufficient knowledge on rare diseases) and referred to a higher level of authority result in additional costs, both for the patients and the state. An efficient process of decision making to determine the degree of disability is necessary for each country to be able to use the allowances and privileges in other areas in accordance with the EU Council recommendation respecting local rights. An adequate classification and codification are required to increase public awareness of the existence of RDs and their recognition in national healthcare systems. The classification and codification will be used for statistical analyses and scientific purposes (e.g. the frequency of occurrence in the Polish population). Using a single source for collecting and updating the reference database, e.g. the ORPHANET database, should be considered.

A special Codification of Rare Diseases (CRD) should be implemented in Poland as soon as possible. The International Classification of Diseases and Related Medical Problems (ICD-10) is the basis and the point of departure for European programmes on rare diseases. Significant effort should be applied to ensure that the classification model that is going to be developed in Poland takes into account what has been established so far at the European level to make sure that the Polish classification will comply with/cooperate with the future European models.

Task 1: Appoint a working team responsible for developing the Polish model of Codification of Rare Diseases. The model should be consistent with rare disease codification systems which are going to be developed in Europe. Close cooperation with the operators of the Polish ORPHANET database is recommended.

Task 2: After the concept for the Polish Codification of Rare Diseases has been developed, the Minister of Health should pass new laws or amend the existing laws to implement uniform rare disease codes in the existing system for codification of disease entities.

Role and Involvement of Patient Organizations:

- actively participate in the Team's work to develop the Polish model of Codification of Rare Diseases;
- participate in the drafting of legal documents implementing the system for Codification of Rare Diseases.

2. Register of Rare Diseases

It is a matter of utmost priority to develop a national register of rare diseases/patients with a diagnosed rare disease. This is a very urgent task because adequate progress of work on many areas covered by the future national plan depends on how soon the register is developed. A common and uniform register will enable full monitoring of patients with a diagnosed rare disease (irrespective of the method of treatment), and will also allow to gather information on natural history of diseases, complications, prognosis and the results of treatment. Recognizing rare diseases which have their codes in the ICD by officially introducing them to the Polish healthcare system should guarantee

quality of health benefits assigned to these diseases (and not to individual symptoms) and access to medicinal products, and serve as a basis for reimbursement of costs (according to the codes). The register will make it easier to monitor the prevalence and incidence as well as the fatality rate for individual disease entities, and to determine geographic location of patients. It may be indispensable to precisely determine where in Poland it is most reasonable to establish specialized care centres (diagnostics, access to specialists, rehabilitation) due to the number of diagnosed cases. It will also help to quickly identify patients from the risk group (especially where it is possible to determine with high probability that a patient is a carrier for a specific disease) to undertake prevention measures (social and educational campaigns, support for general practitioners, paediatricians, gynaecologists) which, in the long run, may lead to a reduction of births occurring to high-risk groups. The register, whether kept as a category or as a group of chronic diseases, may take into account/predict the dynamics of potential changes in the register as new diseases are discovered and described to reflect the modifications in the ICD in 2014.

The works on the Polish register of rare diseases/patients diagnosed with a rare disease should be coordinated with the measures taken in connection with the works on the Codification of Rare Diseases. The register should be available via a central Internet platform with defined multilevel access structure, i.e. a general database and overall information on disease entities/treatment and methods of rehabilitation should be generally accessible, whereas the access to specific levels of the database, for instance patient databases containing personal data, statistical and epidemiologic data, tests and results of clinical trials, should require a central authorization. It is recommended that the register be also used as a source of information for patients where they can find contact details to centres of reference for any specific disease entity, learn about their entitlement to social care and social assistance, find contact data to patient organizations for patients with a particular disease, etc.

Task 1: Appoint a Working Team responsible for developing the Polish model of register of rare diseases/patients with a diagnosed rare disease (objective and subjective scope of the register). The works should be initiated based on the disease entities defined in the ORPHANET database. The model should be consistent with similar registers of rare diseases which are going to be developed in Europe. Close cooperation with the operators of the Polish ORPHANET database is recommended.

Task 2: After the concept for the Polish register of rare diseases and patients with a diagnosed rare disease has been developed, a contractor should be hired to develop and launch the register.

Role and Involvement of Patient Organizations:

- actively participate in the Team's work to develop the Polish model of a register of rare diseases/patients with a diagnosed rare disease;
- after the register has been created, cooperate with regard to completing the information database and promoting the register among patients and physicians.

B. Diagnostics of Rare Diseases

Description of issues, needs and challenges:

The period between the emergence of the first symptoms and the final diagnosis may last from a few months up to a few years or even decades. Such delay in the diagnostics of rare diseases is common and may have tragic consequences. A diagnose is the basis for appropriate treatment and care. There is a special group of diseases which have to be identified before the clinical symptoms occur because an early diagnosis based on screening tests gives doctors a chance to treat and prevent disability, abnormal development or even death. On the way to correct diagnostics of rare diseases there is a range of node points (obstacles), including identification of rare disease symptoms (familiarity with the symptoms) by doctors and limited access to laboratory tests as well as the lack of organized

information for healthcare professionals and patients, which are responsible for the delay and errors in the diagnostics. The implementation of the plan should quickly bring about tangible changes. The following must be done to ensure that the changes take place:

- provide rare disease patients and their families with an equal access to highly specialized clinical and laboratory diagnostics comparable to the access available to patients with diseases commonly occurring in their country and ultimately in the European Community;
- continue and extend population screening programmes for rare diseases based on reasonable epidemiologic and therapeutic evidence;
- provide facilitated and general access to active genetic counselling (determining genetic risk of family members, prenatal and pre-implantation tests for patients with incurable genetic diseases), for instance, by taking greater advantage of the developments in the field of pharmacogenetics (in a broad sense of the term);
- provide a separate scheme for the financing of highly specialized clinical and laboratory diagnostics from national budget taking into account the distinctive nature of rare diseases;
- allow for the possibility to allocate funds from the national budget to support a target-oriented national programme.

The diagnostics of rare diseases (at a clinical and laboratory level) is aimed at early detection of diseases at an initial stage by identifying patients suffering from the specific disorder in an entire population or a high risk group (e.g. proband families).

Providing a final diagnosis of a disease is usually a multidisciplinary process and requires the use of specific diagnostic methods relevant for a particular disease entity. Due to the fact that physicians are generally not familiar with the symptoms of rare diseases, the diagnostics of RDs should be conducted under the supervisions of multidisciplinary expert teams, including physicians, biochemists and molecular geneticists, preferably in specialized centres of reference (or competence centres); cf. section on Centres of Reference.

Overall objective: provide widespread access to quick diagnostics for all rare disease patients to allow patients to begin adequate therapy within a medically acceptable time limit in collaboration with centres of reference. In addition to improving the healthcare of the patients, it is also necessary to conduct research in the field of diagnostic tests and new methods of treatment.

Detailed objectives:

1. Widespread use of screening

Screening tests are used to detect diseases at an initial stage in a population or a high risk group, and they must be confirmed by other, more precise tests relevant for a particular disease entity. This is why screening tests are closely related to genetic (including prenatal) and diagnostic tests. For instance, new-born screening for cystic fibrosis has been common practice in Poland since 2009. The detection rate of this disease has significantly increased (214 new cases have been identified since 2009).

It seems necessary to extend the scope of carrier screening, both among women who are not mothers yet and those in the immediate environment of whom rare diseases have been registered. Especially, individuals from the high risk group (ancestors and lineal descendants) should have the screening test available before taking the decision to have another child. Screening tests should be a common practice, particularly for diseases which are treatable with registered orphan drugs.

Task 1: Evaluate the results of population-based screening and selective screening tests carried out in Poland and develop guidelines for screening based on:

- the register of rare diseases currently being developed, and the development and distribution of questionnaires;
- the analysis of the number of patients with rare diseases detected in Poland through population-based screening (Phenylketonuria (PKU), hypothyroidism (CH), cystic fibrosis (CF), rare metabolic disorders detected by MS/MS, new-born hearing impairment and other) and the future disposition of patients (availability of specialized care at the stage of diagnosis verification, quality of life);
- the analysis of the number of patients with rare diseases detected by selective screening (future disposition, quality of life);
- the analyses referred to above should be used to provide guidelines with uniform indications as to which diseases are screened for and when and where the tests should be performed. It is necessary to ensure that the National Health Fund (NFZ) provide financing for the screening tests for genetic and metabolic disorders diagnosed so far.

How to accomplish the task:
It is necessary to develop analyses and guidelines using funds from the budget for implementation of the National Plan and for the National Health Fund (NFZ) to develop its own guidelines.

Role and Involvement of Patient Organizations:
Collect opinions from relevant parent associations on the positive and negative aspects of the existing solutions and practices; identify rare diseases which do not have parent associations in Poland according to the ORPHANET classification; promote the activities of non-governmental organizations oriented at reaching such patients and their families.

Task 2: Improve the availability and the quality of clinical diagnostics of rare diseases by taking the following measures:

- estimate the number of rare disease patients (actual number and expected/predicted number) in specific domains of medicine/specialties each year based on the above-mentioned analyses (Task 1);
- identify specialized physicians, centres, wards and clinics currently specializing in clinical diagnostics of rare diseases in different areas of medical specialty;
- disseminate the information obtained within a network of centres of reference;
- the analyses referred to above should be used to provide guidelines with uniform indications as to which diseases can be clinically diagnosed and when and where clinical tests should be performed. It is necessary to ensure that the National Health Fund (NFZ) provide full financing of the diagnostic tests for genetic and metabolic disorders diagnosed so far.

How to accomplish the task:
It is necessary to develop expertise using the funds from the budget for implementation of the National Plan and for the National Health Fund (NFZ) to develop its own guidelines.

Role and Involvement of Patient Organizations:
Actively participate in the assessment of accessibility and quality of clinical diagnostics of rare diseases from the point of view of a patient and a patient's family (based on own experience) to provide information on the difficulties and suggestions on how to improve the existing system.

Task 3: Improve the availability and the quality of laboratory diagnostics of rare diseases by taking the following measures:

- estimate the number of diagnostic laboratories, laboratory equipment and staff (hospital and scientific) involved in biochemical diagnostics of rare diseases in Poland;
- estimate the availability of molecular diagnostics of genetically conditioned rare diseases (analysis of mutation in specific genes);

- estimate the availability of molecular diagnostic procedures carried out abroad in laboratories across Europe and around the world (scope of testing, estimate of the number of labs) and the number of the required DNA analyses which are not available or very limited (the method of financing such tests must be assessed);
- the analyses referred to above should be used to provide guidelines with uniform indications as to which diseases may be detected using molecular diagnostics and when and where molecular diagnostic tests should be performed. It is necessary to ensure that the National Health Fund (NFZ) provide full financing of the diagnostic tests for genetic and metabolic disorders diagnosed so far.

How to accomplish the task:
It is necessary to develop expertise using funds from the budget for implementation of the National Plan and for the National Health Fund (NFZ) to develop its own guidelines.
Role and Involvement of Patient Organizations:
Actively participate (interviews and comments) in the development of the assessment of availability and quality of laboratory diagnostics of rare diseases in Poland;

2. Widespread use of genetic testing

Genetic testing should have a priority status in the National Plan, and the access to genetic tests should be considered as standard for rare diseases. Genetic tests should be promoted among all individuals with a diagnosed rare disease case in the family as well as for all those related to a rare disease patient in direct line, i.e. siblings and their children. It is necessary to maintain cooperation with European reference networks developed based on a network of specialized diagnostic laboratories. Centres of reference should be used to develop genetic tests and provide genetic counselling, both before and after tests are performed. This is a matter of major importance for early treatment planning and conscious planning of births in the future.

Task 1: Assess the availability of molecular tests and other diagnostic tests for families with high genetic risk by taking the following measures:

- analyse the availability of genetic tests for proband families where mutation in genes responsible for rare diseases has been confirmed by a molecular or enzymatic diagnosis (access to specialized care, quality of life) with regard to specific disease groups and disease entities;
- assess the level of practical accessibility of prenatal and pre-implantation diagnostics of rare genetic diseases with a fatal course;
- develop a model for active genetic counselling taking into account ethical, religious, legal and financial aspects;

How to accomplish the task:
It is necessary to develop the above-mentioned analyses, assessment and model using funds from the budget for implementation of the National Plan.

Role and Involvement of Patient Organizations:
Assess the availability of molecular tests (DNA analysis) and active genetic counselling for families with genetic risk living in Poland based on own experiences and, possibly, carry out a questionnaire study in proband families.

3. Establishing a System of Centres of Reference

It is a matter of priority to determine the tasks and the role of centres of reference for rare diseases in the Polish healthcare system. Centres of reference should be provided with a precisely determined scope of activity, a separate area of activity (central or regional) and adequate funds to perform their tasks. Also, the needs in terms of quantity and order of establishing regional or national centres of

reference have to be determined. Ultimately, there should be a multidisciplinary team of specialists in each centre of reference (their number and scope of competence should be precisely determined) as well as high-tech equipment and, if necessary, medical facilities for patients and their families (clinics, wards, or possibly hotel accommodation, etc.). It should be possible for clinics and scientific institutions specializing in research on a specific rare disease/ group of diseases with documented scientific accomplishments in that field to become part of the future reference centres.

It is essential that the established/appointed centres offer diagnostics, rehabilitation and drug treatment or direct supervision if treatment (e.g. therapy/drug programmes) is provided by a healthcare provider (directly or by a subcontractor) close to a patient's home. The centres should collaborate closely with the Rare Diseases Task Force and Orphanet Polska with regard to the register of rare diseases being currently developed and with the Coordinating Team for Ultra Rare Diseases with regard to therapy/drug programmes. Cooperation and knowledge exchange between the centres that are going to be established in Poland and other European centres is desirable. They should be connected by a portal-based IT network, preferably the Polish ORPHANET. The number of specialists (or entities) able to establish centres of reference in each region or in areas with the highest number of diagnosed cases must be determined. It is also important that the centres of reference that are going to be established adopt a multidisciplinary approach to the complex and diverse issues connected with rare diseases.

Centres of reference should, first and foremost, be the place where specialists verify initial diagnoses of rare diseases given to patients based on the results of population screening, genetic family testing, selective screening of symptomatic patients and differential clinical diagnostics across the country. A final diagnosis at a centre of reference should be based on standardized and specific clinical and/or laboratory diagnostics (enzymatic or metabolic biochemical tests, histopathologic examinations and other highly specialized tests, and analysis of the molecular basis).

Centres of reference should also be the place where specialists quickly refer patients for specific treatment, provide long-term monitoring of the results of their treatment and coordinate patient care to ensure that patients and those around them maintain an optimal quality of life. The access to centres of reference may not be limited due to any organizational, administrative or financial reasons of the state, and it should compare to the access to general medical care in the country. Centres of reference should cooperate closely with scientific institutions conducting research on etiopathogenesis of particular rare diseases and the methods of their treatment. Collaboration between centres of reference and relevant non-governmental organizations and parent groups is desirable, as well as their participation in scientific research and educational and informational programmes conducted across the country.

In their relations with previously diagnosed and treated patients, centres of reference should essentially support patients in the permanent monitoring of the course of their disease and should fulfil their obligations resulting from rehabilitation recommendations. It is necessary to manage the healthcare pathways (access to specialized physicians, rehabilitation specialists, psychologists). It is also important that the existing centres (as independent units or hospital wards) and newly established entities develop "How to live with ……" guides for rare disease patients (and their families).

As regards costs and procedures executed at centres of reference, a detailed estimate of costs financed/reimbursed by the National Health Fund (NFZ) needs to be prepared. Due to their specialization in rare diseases, centres of reference should receive higher point estimates for the services provided and the tasks fulfilled than in the DRG system.

Task 1: Determine the need for centres of reference for rare diseases (type of centre, estimated number of patients, staff, laboratory equipment, national/regional level, scientific accomplishments, educational/informational programmes) and identify the existing centres of reference for rare diseases, their location, scope of activity, staff,

equipment, availability to patients, level of information provided to physicians by taking the following measures:

- determine in which domains of medicine such centres or specialists are lacking, estimating the costs of establishing/assigning reference centres and the need for centres of reference for rare diseases across the country;
- work out a target and uniform model for a centre of reference;
- draw up a common application form for healthcare centres to apply for the status of a centre of reference taking into account the current status and limitation of activity of individual centres of reference for rare diseases, the scope of their activity (number of patients per year), staff, apparatus and room equipment, method of financing, major difficulties and shortages;
- based on the above, draw up a national list of centres of reference for rare diseases (whose standards of reliability have been verified) and begin work on their recognition at the national level (official form of accreditation based on uniform terms and conditions that healthcare entities must meet to obtain the status of a centre of reference);
- determine the current scale of services provided by European centres of reference to Polish patients with specific rare diseases;
- work out the target model of communication between general practitioners and highly specialized diagnostic and treatment centres;
- ensure visibility of the scope of activity and contact data to centres of reference on the ORPHANET platform defined and selected for purposes of the plan and initiatives taken in connection with the plan;
- facilitate patients' communication with the future centres of reference by developing a plan to support communication projects with IT tools (e.g. telemedicine, info lines) and to widespread the knowledge and access to the Polish ORPHANET information portal.

How to accomplish the task:

- The Rare Diseases Task Force and professionals specializing in rare diseases should organize a workshop to determine the model for a centre of reference and to commission a survey for potential centres of reference to apply for the status of a centre of reference;
- It is necessary to issue general terms and conditions that healthcare entities must meet to be awarded the status of a centre of reference for rare diseases;
- Support the functionality of the Polish ORPHANET database, including IT support and appointing a team to coordinate performance of tasks resulting from the National Plan;

Role and Involvement of Patient Organizations:
Actively participate in the identification of non-formal centres of reference for rare diseases available in Poland and domains and/or physicians specialized in specific rare diseases by means of direct contact with physicians and diagnosticians; participate in the planning of the future network of centres of reference; assist with conducting the survey.

C. Health Care for Patients with Rare Diseases

Description of issues, needs and challenges:

The health care for rare disease patients incorporates three basic pillars:

- highly specialized medical care
- treatment with available orphan drugs
- specialized rehabilitation

Providing patients with rare diseases with such scope of services is a serious challenge for the health care system. Rare disease treatment requires greater funds than the treatment of patients suffering from common disorders. This approach to rare disease therapy is commonly accepted in the EU. Similarly to what is being done in other EU member states, the initiatives to improve the health

situation of patients suffering from rare diseases in Poland should take into account the need for larger funds to provide rare disease patients across the EU with a similar level of health care.

Although the issue of rare diseases treatment receives insufficient attention, it should be noted that in the recent years much progress has been made in Poland to change the status quo. Due to increasing interest in the medical care of rare diseases, more and more serious discussions on how to improve the standards of treatment are conducted by medical groups. Political groups also seem to be noticing the issue of rare diseases as an aspect of health care that requires much more attention and improvement of the standards of treatment and coordination of medical procedures to take into account the distinctive nature of rare disorders. In the last few years, the access to extremely costly orphan drug treatment has significantly improved, particularly in terms of reimbursement of treatment costs of many genetic disorders as part of the existing therapy programmes. For a few years now, there is a Rare Diseases Task Force working at the Ministry of Health whose main responsibility is to provide recommendations on how the state's policy on treatment and care for rare disease patients should be shaped. Although the activities of the Task Force are extended in time and require increased intensity, they will lead to the development of the Polish National Plan for Rare Diseases recommended by the European Commission. In 2010, the Head of the National Health Fund appointed a Coordinating Team for Ultra-Rare Diseases which is responsible for the qualification and monitoring of the use of enzyme replacement therapy in the treatment of ultra-rare diseases.

However, in spite of the fact that the issue of rare diseases is acknowledged by the health care system, rare disease patients still experience many significant difficulties in the healthcare system. In order to improve and enhance the health care system, it is necessary to make fundamental changes in each of the aforementioned health care areas, both in terms of how the distinctive nature of rare diseases is perceived and the methods and schemes of treatment and management of patient treatment.

Medicine has not yet found efficient drug treatment for the great majority of rare diseases. From several thousand rare disorders that exist, the registered specialist drugs (so-called orphan drugs) are available for only a few hundred of them. Due to the multidisciplinary character of this type of disorders, rare disease patients require a coordinated, specialized and comprehensive medical care.

Detailed objectives:

1. <u>Increase the availability of highly specialized medical care</u>

The system of financing outpatient and hospital medical care that is currently in place does not take into account to a sufficient extent the complexity and the heterogeneous nature of rare diseases. The existing cost-estimates for medical procedures do not take into account the actual workload required from a healthcare provider to perform procedures and services for rare disease patients. This often leads to a situation where patients are refused medically recommended treatment because the full cost of such procedure cannot be covered. The cost of treating a rare disease is always higher than the cost of treating a common disorder.

Task 1: Update the cost-estimates for highly specialized medical procedures by taking the following measures:

- develop and implement cost-estimates for individual specialized procedures taking into account the challenges connected with patient care for rare disease patients, such as increased workload and scope of services;
- develop and implement cost-estimates for non-medical specialized services (psychological and psychosocial care, nutrition counselling, home care) based on correct estimates taking into account the challenges connected with patient care for rare disease patients, such as increased workload and scope of services;

- develop cost-estimates and introduce the possibility to use drug treatment at a patient's home due to economic and health-related reasons (so-called "home care" model used in many EU countries) provided that the home-care solution is only available to patients from the "stable group" (patients using the drug for long periods without complications); disseminate information on drug treatment and ensure that the use of drug treatment at patients' homes due to health-related reasons becomes a widespread standard;

Task 2: Increase spending on specialized medical care by taking the following measures:

- hold a debate for members of different ministries and the Polish Sejm to discuss the possibility to increase the funding currently available to the National Health Fund (NFZ) for highly specialized treatment of rare diseases by additional funds allocated directly from the national budget. The result of the debate should be reflected in future Polish budget acts.

Role and Involvement of Patient Organizations:

- participate in works on legislation changes to introduce the home-care model to treatment/drug programmes;
- actively participate in the debate in connection with the promotion of the concept of co-funding the costs of highly specialized treatment from the national budget.

2. Increase the availability of drug treatment

There is still much work to be done to ensure that orphan drugs registered by COMP/EMA are widely available in Poland. An obstacle preventing a widespread access to highly specialized drug therapies, in addition to financial limitations of the payer (National Health Fund), is the fact that the efficacy of orphan drugs registered and available in the EU is frequently questioned. This is because the statutory procedure for health technology assessment (HTA) currently used by the Agency for Health Technology Assessment in Poland (AOTM) with regard to requests for reimbursement of drug therapy costs does not take into account the specific criteria for assessment of rare diseases which are generally recognized in all EU members states.

Additionally, the efficacy of medicinal products is usually questioned based on the fact that there is little scientific evidence that confirms the efficacy of these products, which results in negative recommendations being issued by AOTM on reimbursement for orphan drug treatment. Yet, little scientific evidence of the efficacy of such drugs is typical in the case of rare diseases and should not be used as a criterion for questioning the efficacy of these products. Due to that reason, drugs for rare disease therapy have been grouped into a separate category in the central drug register at the EU level. The orphan drug status is assigned based on detailed tests and (obviously) a limited number of trials. The limitation results from the rare occurrence of these disorders in a global scale which, in extreme cases, may affect a population of no more than a few hundred.

As regards the financing (reimbursement) of extremely costly drug treatment, currently the costs are covered by the National Health Fund only. Since the approach to providing medical and social care to patients with rare diseases is based on solidarity and not general insurance, it is advisable to increase the current funds available to the National Health Fund could by allocating additional funds directly from the national budget. A similar solution has been successfully implemented in some EU member states.

Also, as regards the costs of drug treatment that is currently reimbursed or is going to be reimbursed in the future, the procedure should be rationalized in order to account for situations where treatment must begin immediately to save a patient's life.

Since, as a rule, due to its medical efficacy drug treatment is aimed at restoring patients' ability to function relatively well in society, the descriptions of therapy/drug programmes should be more flexible in order to account for such aspects as patient's life situation, migration and life-related needs and the necessity to administer drugs close to a patient's home. The programmes should be

beneficial for patients and their families, not stressful. Allowing more flexibility in the specification of therapy/drug programmes should prevent previous situations from recurring, such as when the National Health Fund would refuse to cover the costs of services that had been provided by hospitals based on strict and unrealistic programme descriptions.

Task 1: Adapt Polish legislation to the EU concept of how orphan drugs should be treated and used by amending the act on reimbursement of medications by:

- removing the obligation for AOTM to assess the efficacy of products with COMP status of orphan drug for which reimbursement is requested;
- removing (not applying) the criterion determining the cost threshold per extra year of life gained to orphan drugs when taking decisions on reimbursement for orphan drugs (in other words, 3xGDP rule does not apply to orphan drugs);

Task 2: Increase healthcare spending on state-of-the-art drug treatment by taking the following measures:

- hold a debate for members of different ministries and the Polish Sejm to discuss the possibility to increase the funding currently available to the National Health Fund (NFZ) for highly specialized treatment of rare diseases by allocating additional funds directly from the national budget. The result of the debate should be reflected in future Polish budget acts.

Task 3: Accelerate the decision-making process with regard to drug treatment and implement patient-oriented descriptions of treatment/drug programs by taking the following measures:

- implement a so-called rapid response procedure taking into account the necessity to immediately begin a life-saving drug treatment covered under a drug programme;
- modify descriptions of treatment/drug programmes so that they take into account such aspects as patient's life situation, migration and life-related needs and the necessity to administer drugs close to a patient's home ("user friendly" programme) and so that healthcare providers consider the performance of contracted treatment/drug programmes as a sign of trust and distinction and do not run the risk of financial losses as a result of the payer's refusal to reimburse the costs.

Role and Involvement of Patient Organizations:

- participate in the works on amending the act in connection with the performance of task 1;
- participate in the debate in connection with the promotion of the concept of co-funding the costs of rare disease treatment from the national budget;
- participate in the preparation of rapid response procedures and the works on amending legislation with regard to patient-oriented treatment/drug programmes.

3. Improve patient rehabilitation system

Rehabilitation contributes to improving patients' quality of life. Currently, rare disease patients have a limited access to public rehabilitation services; they are only available as part of hospital treatment. Public rehabilitation services are not performed at patients' homes, unlike in many European countries.

Patients suffering from some progressive diseases require rehabilitation at a stage when the symptoms are still scarce. Most patients are provided with periodic rehabilitation services, i.e. there are long periods between rehabilitation cycles when the service is not provided. The essence of rare disease patient rehabilitation is regularity of the rehabilitation therapy so that patients do not lose what has been achieved thanks to the therapy.

Rare diseases lead or may lead to motor and/or intellectual disabilities. Some patients remain disabled during their entire life. This is why it is so important to develop a rehabilitation programme according to a patient's age (early support of child development, developing sensory integration

trails) as a necessary element of disease treatment. Recommendations should also include additional support activities such as exercises at a swimming pool, manual skill development classes, movement activities at kindergartens, schools, integration centres, and continuous speech therapy.

Post-surgery rehabilitation needs are equally important. In the case of rare diseases, rehabilitation, in a broad sense of the term, should be part of patient-guaranteed care (motor, psychological, therapeutic and social rehabilitation) as an integral element of the overall system of support provided to patients and their families. If a rare disease has been diagnosed, it is necessary to facilitate the access to rehabilitation wards/centres for the patient because adequate rehabilitation is another crucial element of patient treatment in addition to pharmacological treatment (many times unavailable).

Task 1: Develop and stimulate widespread use of a model procedure for patient rehabilitation therapy and access to specialized rehabilitation therapy by taking the following measures:

- Develop a standard procedure for patient rehabilitation therapy taking into account a patient's age, health condition and capability for each diseased classified in the future rare disease register;
- Include the standard rehabilitation procedure applicable to the classified rare diseases in the patient-guaranteed care package;
- Provide training to rehabilitation specialists on the therapy of a selected group of rare diseases in collaboration with centres of reference;

Role and Involvement of Patient Organizations:

- Collaborate with determined associations to define the standard rehabilitation procedure for specific disease entities;
- Participate in the preparation of legislation amendments to include rehabilitation therapy in the patient-guaranteed care package;
- Collaborate with determined associations to develop a relevant training programme for rehabilitation specialists.

D. Integrated Social Assistance for Rare Disease Patients and their Families

Background/ description of issues, needs and challenges:

The situation of patients with rare diseases and their families is very difficult and complex. The families are to a large extent excluded from participation in social life and have to cope with many financial problems. The solution to these problems lies in integrated social assistance for patients and their caregivers as well as collaboration between all institutions involved in patient support. The existing forms of social assistance available to rare disease patients are insufficient and depend on whether a patient has obtained a certificate of disability or meets the income criteria. A benefits package should comprise services and measures that fully solve all problems causing exclusion of patients and their families from participation in the normal activities of the society. It is important that the initiatives outlines in the National Plan equal the health, educational and social status of rare disease patients to allow them to be able to use their constitutional "right to equality", "access to public health care" or "support from the state" in practice, not just in theory. In this context, all social assistance measures provided for in the Plan should be part of an integrated health, social and educational support package which will contribute to the enforcement of the rights guaranteed by the Constitution. In order to offer patients comprehensive social assistance, it is necessary to establish a closer cooperation between such national intuitions as, for instance, the Ministry of Labour and Social Policy, the Ministry of National Education, the Ministry of Health, Municipal Social Assistance Centres (MOPS), District Family Assistance Centres (PCPR), the Social Insurance Institution (ZUS), local governmental units, governmental institutions and non-governmental organizations. Due

to the complexity of problems resulting from rare diseases, it is necessary to, first of all, increase reimbursement for drugs, patient care products and rehabilitation equipment from state funds provided to insured patients suffering from diseases defined herein, and to increase attendance benefits.

An example of the actual problems that rare disease patients face are the special educational needs which have been defined in Polish education law without taking into account the complex needs of children and youth with rare diseases. Many times child patients do not receive the necessary support in spite of the serious and complex disorders they suffer from because their disease and needs do not correspond to the recommendations. Medical information on rare diseases and their course is not given the required importance in educational practice (or is disregarded altogether). The result of this approach is that, for instance, children whose disease inevitably leads to deterioration of eyesight are not taught relevant skills early enough, or children with hypothalamus disorders causing disturbed control of appetite are referred to eating disorder treatment, or children with documented congenital disturbances in cognitive and social development are treated as if their behaviour was a result of parenting errors, whereas children with progressive muscular dystrophy are sent away from schools to attend individual classes at their home, which additionally contributes to lowering the standard of their education and deepening their social exclusion. The approach to rare diseases with progressive neurological changes should have been changed in the educational system long time ago. Children with progressive neurological changes are expelled from special schools and kindergartens for failure to achieve satisfactory therapeutic progress. If a student suffers from a rare progressive neurological disorder, it is hard to expect learning progress. The only solution offered to such children is individual classes at home without being able to interact with peers. This situation is completely due to the fact that the education law fails to meet the needs of these children and allows schools to “get rid of” such unpromising students easily.

Teachers often lack adequate preparation to support students with rare diseases and they do not know where to seek help (it often happens that there is no centre to provide relevant information). In order to be able to work with a child suffering from a rare disease, a teacher should obtain relevant knowledge from professionals specializing in other areas, such as medicine, clinical psychology, etc., who usually work in other institutions. Currently, interdisciplinary collaboration is usually limited to a single centre or is based on consultation with external organizations. Decisions on the need for special education are many times issued separately from certificates of disability and medical diagnosis and do not take into account the natural course of a disease, which often requires that actions be taken before a student displays all of the deficits caused by the disease.

Another important issue is the fact that certifying physicians determining disability and, for instance, declaring patients unfit for work do not possess relevant knowledge on rare diseases. Certificates of disability are sometimes issued to children with a rare genetic disease for a period of 1 year only, while it is known that genetic diseases last a lifetime and do not reverse. Even though it is extremely difficult to travel with a disabled child, especially one that cannot walk, certifying authorities take un uncompromising stance and insist that a Disability Commission must review each case once every few years. There is a strong need to recognize the fact that taking care of rare disease patients is a “full-time job” and to increase the amount of attendance benefit and allowance for parents taking care of their children. Also, there are no clear-cut national criteria for declaring disability of patients that account for the fact that patients require the assistance of a carer which is an aspect taken into consideration when determining entitlement to attendance allowance. It should be noted that not every patient with a rare disease is disabled, which is a serious problem for a whole group of diseases. Reliefs and entitlement to benefits, e.g. a tax relief for patients requiring rehabilitation is only available to patients with a certificate of disability. It is very difficult for adult patients with rare diseases to claim a social pension due to their inability work. Many times they manage to obtain the right to a pension only after appeal in the second instance. Decisions are made based on patients’ looks, and not their medical documentation.

In terms of social assistance, there are many different problems and issues that need to be solved by adapting solutions which are already used in the EU, for instance:

- increase the amount of child care benefit for parents/one designated parent who were/was professionally active before the child was diagnosed and now do/does not work (not due to their fault) to take constant care of their disabled child/children (based on relevant certificates) and:
- increase the amount of benefits per each child with a rare disease;
- adapt the amount of carer's allowance per each child in the family diagnosed with a rare disease to reach the European Union standard;

It is also recommendable to adapt the solutions that are currently being developed to solutions already used in some member states, for instance:

- the amount of attendance allowance is related to national minimum pay. In the European Union, it corresponds to 70% of national minimum pay (at the most recent rate or a rate valid on the day the disease was diagnosed) and it is paid per each disabled child and not per family;
- extra allowance is paid if there are neurological changes/intellectual disability and, most of all, severe mental retardation;
- the parent who quits his/her job to take care of the child is entitled to retirement/disability pension;
- additional retirement/disability pension contributions payable for each subsequent child with a disease;
- housing policy for people with disabilities;
- a base of public housing with no architectural barriers for people with disabilities;
- household allowances: the cost of water supply, gas, garbage removal, etc. are partially paid by the state;
- medication/rehabilitation is provided at no charge/at a partial cost;
- child caregiver/assistant;
- allowances for appropriate foodstuffs, depending on the type of disease as in the case of hospitalization of diabetes patients.

Social assistance is just one area covered by the package. The situation of patients who are unable to perform activities of daily living and require assistance from other people also needs to be improved. These are patients with motor and mental disabilities, behavioural disturbances and those whose health condition is very serious. In such cases it is the patient's family that provides care, and as such also requires systemic support.

Currently, the act on social assistance provides for so-called "care services" and the position of "assistant to a person with disabilities". Unfortunately, there are many problems associated with the existing solutions:

- the current definition of a professional "assistant to a person with disabilities" does not include certain care-related activities (such as, for instance, transferring the patient to a wheelchair, assisting the patient to use the bathroom, put on the outerwear, etc.), taking into account the distinctive nature of rare diseases, the advanced stage of such disorders and dependence of rare disease patients on their carers;
- people providing care services usually lack adequate medical training which is an important aspect of rare disease patient care. A carer should be trained for first aid and have psychological credentials (medical care and social assistance);
- there are no systemic solutions with regard to the position of an assistant to a child with disabilities;
- the services are not fully reimbursable and depend on the income of the family; also, they are not provided to families at a required level.

Also, it is a good idea to refer to the concept of independent living presented in the Madrid Declaration proclaimed in 2002 by the European Congress on People with Disabilities: *"For a person with disabilities, living independently means to be able to take equal control over their lives and make the same choices every day as any non-disabled person: own house, financial security, personal assistant".*

The profession of a personal assistant to a person with disabilities (hereinafter PAPD) has not been yet recognized in the Polish system for support of people with disabilities; however, forms of support equivalent to the services provided by a personal assistant to a person with disabilities can be found in various legal regulations, although that specific term is not used to describe such services. It seems reasonable to first regulate the status of a PAPD by adding this profession to the Polish Classification of Occupations and Specializations for Labour Market Needs and to develop standards for this profession similar to the ones currently used in the EU. Services provided to people with disabilities by a PAPD should be a very important element of support to compensate their disabilities.

The PAPD services should be reimbursable irrespective of the professional status of the parents due to the fact that the patient requires constant care. The patient's family should be provided with psychological assistance and financial support according to the EU conditions and recommendations.

In order to offer patients integrated social assistance, a new occupation of "Carer for a family of a chronic rare disease patient" needs to be created. This would be a task combining the aspects of medical care and social assistance in connection with patient organizations and centres of reference. A carer's job would be to supervise comprehensive and constant care for the patient and the patient's family. S/he would act as a liaison between medical and social aspects of care to provide the patient and the patient's family with optimal support, and could also be a source of information for the patient's family.
A carer should collaborate with medical professionals (including attending physician) to coordinate the medical care provided (schedule appointments and consultations, adjust the dates of medical examinations, assist during appointments, hospitalization, help solve any problems that may occur during the patient's stay at a medical centre, particularly if the patient is disabled), as well as with social assistance institutions to supervise the social assistance provided (determine the family's financial needs, plan which benefits are required, act as a liaison between the family and the institution when applying for assistance, inform the family about the possible forms of assistance, ensure quality and continuity of social assistance and provide any other support in that respect).

Today, buying a house is a big challenge for any family in Poland. The families of patients with disabilities have many expenses related to the disability alone, and they still have to buy a house and pay for architectural modifications to accommodate the patient's needs. There are practically no standards for building houses that meet the needs of people with disabilities. A house must be arranged in a way that it is possible for a wheelchair to move around, and there must be enough space for rehabilitation equipment. After buying a house, the family needs to pay extra costs to make architectural modifications, including, for instance, widening doorways and corridors for easier wheelchair access, remodelling bathrooms, removing threshold from exterior door, installing handrails, non-slip flooring and ramps; and has to pay more money for larger parking spaces. The Home for Each Family Programme (*Rodzina na swoim*) helps average families buy their own house; however, it lacks elements that are relevant from the point of view of a disabled person's family such as:

- taking into account the cost of removing architectural barriers, including lifts and ramps, wider door without thresholds, remodelling bathrooms;
- increasing the area limit for a disabled person's family;

Commune authorities should acknowledge the need to offer public housing without architectural barriers to the poorest families. The issue of adapting a public house to accommodate the needs of a disabled person could by handled by the Government Plenipotentiary for People with Disabilities

upon consultation with a local organization with experience in removing architectural barriers according to the needs of persons suffering from a specific disease. At the same time, the process to develop building standards according to specific types of disability should be launched.

Overall objectives:

The overall objective that need to be met in the area of integrated social assistance is to seek out opinion of patients and their representatives regarding social policies on rare diseases, and to facilitate the access of patients to updated information on social assistance, promote the initiatives taken by patient organizations, such as consulting and counselling, exchange of information and best practices, developing networks, or establishing contact with the most isolated patients.

Patient organizations have wide experience in fighting the effects of disability through implementation of their programmes as set forth in the organization's charter, often running the risk of losing their liquidity. It is a good idea to use this experience, and this is why patient organizations should participate in social policy decision-making processes with regard to rare diseases.

Social benefits, on the other hand, should be equally available to patients and their caregivers, and they should facilitate integration between patients and non-disabled groups of people at school and at the workplace. State and local institutions should collaborate closely to support the integration of social assistance that meets the patients' needs.

The state should support initiatives taken by organizations to provide patients with information and social assistance in a more effective manner. The state should also provide facilitated access to programmes for patients/people with disabilities, including increasing spending on social assistance and social programmes for patients, and allow for assistance programmes to be developed by non-governmental organizations and the private sector.

Detailed objectives:

1. <u>Integrate collaboration between state organizations (social assistance institutions and units) to support rare disease patients</u>

Task 1 collaboration between national and local government organizations on social assistance programmes

Integrated and comprehensive collaboration between the Ministry of Health, the Ministry of Labour and Social Policy, the Ministry of National Education, local government units, municipal/commune social assistance centres, District Family Assistance Centres (PCPR) and non-governmental organizations supporting rare disease patients will result in a better and more efficient social assistance for patients and will allow them to exchange information on key issues and programmes available for patients and their families. The collaboration should be developed together with the establishment of social assistance programmes to meet the needs of rare disease patients and their caregivers.

Role and Involvement of Patient Organizations: State administration organizations may appoint non-governmental organizations to provide information on available programmes and delegate certain duties and competence of the state in the field of social assistance to NGOs. Patient organizations should collaborate between each other to improve provision of information and to ensure coordination of measures and understanding of the topic and the issue of rare diseases.

Task 2: collaboration between national and local government organizations with regard to the disability evaluation process and the process of granting disability pensions to patients who are unfit for work.

The collaboration between District and Regional Disability Evaluation Boards, the Ministry of Health,

the Ministry of Labour and Social Policy, the Social Insurance Institution (ZUS) and local government units and organizations will improve the quality of the disability evaluation process and the process of granting disability pensions to patients who are unfit for work. The initiatives taken as a result of this collaboration should be implemented through educational and information programmes for certifying physicians and by increasing the level of standardization of procedure for disability evaluation boards and the Social Insurance Institution (ZUS) as well as by enhancing the collaboration between ministries in order to develop clear-cut criteria and standards for evaluating disability and unfitness for work of rare disease patients and to work out a definition of such disability/unfitness for work.

Role and Involvement of Patient Organizations: Collaboration to develop educational programmes, determine more precise criteria and point out to government institution what the potential irregularities may be; involvement in advisory and informational activities.

Task 3: Collaboration between the Ministry of Science and Higher Education, the Ministry of National Education, the Ministry of Health, government administration and local government units and organizations to ensure better understanding of the needs of rare disease patients in the education system

The provisions of Polish education law should define special educational needs (SEN) in a way that will recognize and embrace rare diseases and ensure adequate financing and support of the special educational needs of children and youth suffering from such diseases. The existing definitions of disability and special educational needs should be reformulated to reflect the current state of medical knowledge, particularly with reference to rare genetic disorders. A decision on the need to provide a child with special education should be made taking into account information resulting from medical diagnosis and the child's certificate of disability, instead of being based exclusively on the result of tests and other psychological and pedagogical diagnostic tools. A pedagogical diagnosis and a pedagogical intervention programme based on the diagnosis should take into account the medical diagnosis and the course of the disease certified by medical documentation. Teacher training courses should provide information on rare diseases. The ultimate objective is to include training on rare diseases in the curriculum of all teaching courses (and not just special education courses) as well as teachers' continuing training. Centres of reference should be established to provide knowledge on rare diseases not only from the field of medicine but also developmental psychology, speech and language therapy and education. The care of children with rare diseases should be provided by multidisciplinary teams made up of medical experts, education professionals working with the child and the child's parents/legal guardians.

Role and Involvement of Patient Organizations: Patient organizations should be involved in the process of providing information on irregularities and developing standards to accommodate patients' needs, and support schools to better understand rare diseases and potential issues and educational needs of students with such disorders. Patient organizations should also be engaged in advisory and informational activities and collaborate between each other to improve provision of information and to ensure coordination of measures and understanding of the topic and the issue of rare diseases.

2. <u>Improve available reliefs and social assistance programmes for patients with rare diseases and their caregivers</u>

Task 1: Increase the rate and change the rules for granting attendance allowances

The existing criteria for granting attendance allowances should be changed; the rates of allowances should be increased, and the group of potential beneficiaries should be expanded, particularly to include carers of patients who are not disabled. A carer for a person with disabilities should be entitled to higher retirement/disability pension contributions. Also, it needs to be decided whether taking care of a child with a rare disease by the parents should be considered as a "full-time job" as provided for in the act on "assistants".

Role and Involvement of Patient Organizations: Patient organizations should assist in the development of the system and in defining the needs of patients according to specific disease groups and engage in advisory and informational activities.

Task 2: Intensify support for disabled patients with rare diseases and provide complementary forms of assistance

It is essential to define the profession of a "Personal Assistant to a Person with Disabilities" and determine the scope of PAPD's responsibilities. New *ad hoc* regulations should facilitate implementation of PAPD programmes taking into account PAPD's tasks in connection with care-related activities (such as, for instance, transferring the patient to a wheelchair, assisting the patient to use the bathroom, put on the outerwear, etc.), taking into the distinctive nature of rare diseases, and the need to provide the personal assistance service to patients with motor and/or intellectual disabilities and children.

Ultimately, the PAPD service should be available to patients depending on the degree of the patient's disability, irrespective of the work situation and income of the disabled patient and/or the patient's legal guardians. The PAPD service should be provided by specialized local institutions attached to social assistance and administration centres.

Additionally, in order to offer patients integrated social assistance it is necessary to create a new occupation of "Carer for a family of a chronic rare disease patient". This would be a task combining the aspects of medical care and social assistance in connection with patient organizations and centres of reference. A carer's job would be to supervise comprehensive and constant care for the patient and the patient's family. S/he would act as a liaison between medical and social aspects of care to provide the patient and the patient's family with optimal support. A carer would collaborate with medical professionals (including attending physician) to coordinate the medical care provided to the patient (schedule appointments and consultations, adjust the dates of medical examinations, assist during appointments, hospitalization, help solve any problems that may occur during the patient's stay at a medical centre, particularly if the patient is disabled), as well as with social assistance institutions to supervise the social assistance provided (determine the family's financial needs, plan which benefits are required, act as a liaison between the family and the institution when applying for assistance, inform the family about the possible forms of assistance, ensure quality and continuity of social assistance and provide any other support in that respect).

Role and Involvement of Patient Organizations: Patient organizations have vast experience in providing the PAPD services. National administration institutions may appoint patient organizations to perform specific public objectives (social programmes). Patient organizations may assist in developing the system and defining the needs of patients with groups of diseases as well as engage into advisory and informational activities.

Task 3: Improve the housing situation of disabled people with rare diseases

The provisions of Polish building law should take into account the needs of the disabled in housing construction regulations by setting standards for barrier-free homes. The standards should provide specifications taking into account the need to install lifts and ramps and build more spacious bathrooms, wider passageways for wheelchairs and wider doorways without threshold, etc. There are different types of obstacles observed with regard to removal of architectural barriers in buildings shared by many families, for instance blocks of flats administered by housing co-operatives or tenants' associations. New legislation should further facilitate removal of architectural barriers.

The national housing policy should take into account the higher costs of barrier-free housing and improve the financing of removal of architectural barrier. At the moment, there is no programme providing supplementary financing for the purchase of barrier-free houses. The only solution offered are programmes for removal of barriers in houses already owned by disabled people and they are difficult to access. Consequently, the market for specially adapted housing is very small and the price of such houses is higher. It seems reasonable to make relevant changes in the Family on Its Own Programme (*Rodzina na swoim*) to take into account:

- the cost of removing architectural barriers, including installation of lifts and ramps, widening doorways without threshold, bathroom modifications;
- higher size limit for homes where families with disabled family members live;

Commune authorities should acknowledge the need to offer public housing without architectural barriers to the poorest families. The issue of adapting a public house to accommodate the needs of a disabled person could by handled by the Government Plenipotentiary for People with Disabilities upon consultation with a local organization with experience in removing architectural barriers according to the needs of persons suffering from a specific disease. At the same time, the process to develop building standards according to specific types of disability should be launched.

Role and Involvement of Patient Organizations:
Suggest changes in the existing housing system to ensure facilitated access to specially adapted housing for people with disabilities and their families; help to define the needs of patients according to specific disease groups and to determine building standards.

E. Information, scientific research and education in the field of rare diseases

Background/ description of issues, needs and challenges:

A better understanding of rare diseases will make it possible to improve the diagnostics and treatment of these disorders, and this is why scientific research in this field should be considered as priority. At the same time, due to the distinctive nature of rare disease, it is difficult to conduct research, which is why and it is necessary to adopt a multidisciplinary approach and use the existing resources.

Although a relatively large amount of scientific research on rare diseases is observed, the existing structure does not facilitate coordination of the research work. It is important to extend and strengthen scientific collaboration within the European Union as there are EU funds available for short- and long-term research projects, scholarships and scientific excursions. The differentiating factor of rare diseases is their interdisciplinary nature as they combine many different fields of study. Further research will, without a doubt, help us better understand these disorders and, consequently, improve the quality of life of RD patients.

Professionals, including physicians of different specialties, psychologists, nutritionists and other specialists engaged in providing comprehensive patient care do not have equal access to information on rare diseases. Another problem is the level of education before and after obtaining the diploma which usually varies as well. Therefore, it is necessary to make improvements in education given to physicians and extend medical curriculum to provide comprehensive knowledge on rare diseases. General practitioners lack knowledge on how to detect and treat rare diseases and there are no postgraduate education programmes available in the field of rare diseases. Also, the exchange of information on rare diseases between medical professionals is insufficient, particularly with regard to emergency situations and has an impact on correctness of diagnosis and further treatment.

Social education and patient-doctor communication are a key element of the National Plan. Patients and their families find it difficult to obtain information on rare diseases. The support that patients and their families receive comes from patient associations which bring together people suffering from specific rare diseases. The fact that patients and their families have to deal with the disease and related problems on a daily basis makes them specialists in that field. There are approximately 50 patient organizations in Poland bringing together people with rare diseases and one umbrella organization composed of 18 associations for rare disease patients (Orphan National Forum for Treatment of Rare Diseases). Non-governmental organizations are ready to actively collaborate with national institutions. It is necessary to develop comprehensive information materials for patients and national institutions in collaboration with patient associations, and undertake sustainable and systematic regular measures to communicate with the society. The fact that public opinion is unaware of the existence of rare diseases is a major problem which contributes to the isolation of patients and their families as a result of lack of understanding from the society and doctors. Information on rare diseases and initiatives taken in that regard does not reach selected recipients and the public opinion to a sufficient extent.

General objectives:

- Streamline research on rare diseases through promotion of research work in that field and relevant coordination with this regard;
- Improve medical education in the field of rare diseases and facilitate exchange of medical information;
- Develop information addressed to patients, public institutions and the society;

Detailed objectives:

1. <u>Stimulate scientific research on rare diseases</u>

Task 1: Ensure recognition of the issue of rare diseases by the National Centre for Research and Development responsible for commissioned research projects

Incorporate rare diseases in the National Framework Programme of the National Centre for Research and Development on clinical research and research on the influence of diagnostic and treatment methods on patients' health condition and quality of life (with the support from the Ministry of Science and Higher Education and the Ministry of Health); determine priorities with respect to research on rare diseases to increase the number of research projects carried out by research institutes;

Task 2: The Commissioned Research Project Team at the National Centre for Research and Development to launch a competition for research projects on rare disease epidemiology;

Task 3: Develop a system to coordinate the existing and newly built databases for purposes of research work being carried out and establish rules for information sharing (in collaboration with the Healthcare Information Systems Centre (CSIOZ) and Inspector General for Personal Data Protection (GIODO);

Task 4: Draw up a list of national science centres conducting research on specific rare diseases; assess the centres using such criteria as publications on rare diseases, number of patients examined, and collaboration with clinical centres.

Task 5: Support the development of multidisciplinary research conducted by multiple institutes and promote Poland's participation in international research projects by drawing up and publishing a list of projects being carried out and research centres participating in the project in all EU states; create a national grant fund to pay the costs of participation of Polish institutes in international research projects;

Task 6: Promote the development of independent clinical research by providing public support and obtaining EU funding;

Role and Involvement of Patient Organizations:
actively participate in the identification of research institutions conducting research on rare diseases in Poland and research areas and/or expert scientists carrying out projects/research at research centres attached to universities; provide research centres with relevant information; invite research centres to participate in future campaigns; jointly organize seminars/lectures to demonstrate the problems, scale and scope of conducted research work;

2. Medical education: the education system and specialization courses

Task 1: Improve the level of education of medical students with regard to rare diseases by extending medical curriculum developed by the Ministry of Science and Higher Education to provide seminars and lectures on subjects related to rare diseases (5^{th} and 6^{th} year of study), particularly as part of the following courses: paediatrics, internal medicine, pulmonology, gastroenterology, neurology, and psychiatry.

Task 2: Improve the level of post-graduate education by developing compulsory specialization programmes and courses on rare diseases at the Medical Centre of Postgraduate Education (CMKP). The courses will be certified, and physicians participating in such courses will receive financial support. Additionally, support will be provided to develop narrow specialties in the field of rare diseases (e.g. metabolism paediatrics);

Task 3: Provide training programmes in the field of rare diseases to physicians and scientists as part of continuing education;

Task 4: develop and implement validated recommendations for diagnostics and treatment of rare diseases to allow specialized centres to exchange bets practices;

Task 5: draw up and provide general access to a list of diseases, drugs and centres to minimize the time spent waiting for available treatment (Orphanet); develop and promote an encyclopaedia of rare diseases;

Task 6: develop scientific exchange and international internships programmes and involve students and young physicians in scientific research on rare diseases;

Role and Involvement of Patient Organizations:
Actively participate in education initiatives (seminars, exhibitions, lectures) for students in final years of study and develop relevant materials (in collaboration with the Ministry of Health and the Ministry of Education); actively participate and monitor postgraduate education; participate in the development of training programmes (Internet, telemedicine); actively participate in developing best treatment practices with strong support from national and local units of government administration.

3. Social education and patient-doctor communication

Task 1: Develop information base available over the phone and via the Internet; make the Polish database/platform Orphanet the point of reference providing access to information on rare diseases for patients and physicians; create Orphanet medical encyclopaedia for patients and medical professionals providing detailed information on each identified disease;

Task 2: Develop information on rare diseases for specific groups of recipients, such as public institutions, teachers and social assistance professionals; create new online resources for medical professionals (how to diagnose a disease or detailed information on the healthcare system; guides on what to do in emergency situations, etc.), social assistance professionals (administrative procedures regarding education, professional integration and financial aids), and for patients;

Task 3: Create a grant fund to support educational initiatives taken by patient organizations;

Task 4: Plan and launch a social campaign on rare diseases across the country in collaboration with patient organizations and based on previous initiatives;

Task 5: Promote the plan for rare diseases among medical professionals, patients and the society; inform medical groups, patients and patient associations on progress in the implementation of the Plan and the measures taken.

Role and Involvement of Patient Organizations:
Actively participate in the development of a national portal for rare disease patients (diagnosis, treatment, rehabilitation, disability evaluation process, treatment centres); monitor social campaigns together with public administration units; organize themed conferences for government officials, scientists, physicians and patient organizations to provide information on the current state of affairs (latest research news, changes and amendments to treatment programmes, launch of new treatment, information on rehabilitation, etc.) according to the National Plan Assumptions.

F. Plan Implementation Monitoring

The National Plan should be administered by the Ministry of Health. The Ministry should coordinate and monitor the implementation of the tasks defined in the plan for each specific area. Due to the large scale of the project, it is advisable to establish a special Department of Rare Diseases at the Ministry of Health.

The implementation of the Plan can only be successful if relevant funding and supervision are ensured. There should be a clearly defined source(s) of financing for each area of the National Plan. The National Health Fund (NFZ) should not be the only organization financing implementation of the Plan. Supplementary funds to pay the costs arising from implementation of the Plan should be provided from the national budget, the budget of relevant ministries and the EU. Extra funds should also be obtained from non-budgetary sources, including the EU designated subsidies. It is important to use the funds allocated for the execution of the Plan to implement the EU recommendations on rare diseases.

It is necessary to establish a monitoring system at all levels from local to central based on a central patient information database (e.g. electronic competence centre).

It also needs to be considered if the monitoring service should be sub-contracted to an independent entity. The monitoring requires constant and systemic collaboration with designated patient organizations which should participate in the process as recommended by the EU.

ADDITIONAL COMMENTS:

1. This document is based on the existing programmes on rare diseases and reflections arising from the experience of associations who deal with patients on a daily basis. The concepts outlined herein present the priority issues which are most apparent and important for patient organizations and medical groups. The development of the National Plan for rare diseases is a major undertaking. It requires detailed planning of all measures taken to support a group of patients that has been neglected for many years.

2. Due to the fact that the Plan requires involvement of different ministries, representatives of all competent and relevant ministries should be engaged in the plan development process as soon as possible.

3. In order to ensure proper implementation of the future plan and efficient monitoring of measures taken in that regard, the use of tables for specific issues regulated by the plan is recommended, for instance based on the following model:

Task performance schedule

Task performance	2013				2014				2015				2016				2017			
	quarter				quarter				quarter				quarter				quarter			
	1	2	3	4	1	2	3	4	1	2	3	4	1	2	3	4	1	2	3	4
Task no. and description																				
Task no. and description																				
Task no. and description																				

Plan implementation costs

Task performance costs	2013	2014	2015	2016	2017
Task no. and description					
Task no. and description					
Task no. and description					
TOTAL					

Implementing/partner entity

Task	Implementing entity	Partner entity	Monitoring entity
Task No. and description			
Task No. and description			
Task No. and description			