



Ministerul Sănătății Publice



Allianța Națională pentru Boli Rare Românie

# ROMANIAN NATIONAL PLAN FOR RARE DISEASES 2010- 2014



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**Romanian National Alliance for rare Diseases**  
**Romanian Prader Willi Association**

**THE PURPOSE for developing the National Plan for Rare Diseases**

Improving the quality of life for people affected by rare diseases in Romania, through equal access to early diagnosis, quality treatment and rehabilitation services for people with rare diseases.

*„HEALTH IS A FUNDAMENTAL HUMAN RIGHT, WHETHER THE DISEASE IS  
COMMON OR RARE”*

**PREAMBLE:**

A rare disease is a disease affecting less than 5 in 10.000 people, in the European Union. The synonym “orphan disease”, particularly used in France, provides rare diseases with political and social dimensions, trying to convey the patients’ request to be taken into account, no matter how rare the disease may be. Rare diseases are also generally orphan from the point of view of treatment, diagnosis and adequate care.

In 1995, the WHO categorized 5000 rare diseases. Today, there is talk of over 8000 such diseases. Depending on the disease, it can register between a few to a few thousands.

Numerous and complex, they are hard to know for the medical professional and the health system official. They concern all medical specialties, and require an interdisciplinary coordinated approach, having various degrees of gravity, depending on the disease and the patient.

They can manifest at birth or during the first years of childhood. In over 50% of rare diseases,

the first clinical signs occur in adult age and are generally accompanied by physical and/or sensory impairments, which are severe and cause an important disability.

Considered to be unprofitable by pharmaceutical laboratories, they are rarely the object of scientific research, they lack treatment and diagnosis may spread over many years. The economic and social support of these diseases is still incomplete.

80% of rare diseases have genetic cause. They are generally hereditary and are passed on from one generation to another. They may also be the result of a spontaneous mutation, new, with no family history.

Among the rest of 20%, there are infectious diseases and others caused by various factors (environment, etc).

**Rare diseases are also different from the point of view of gravity and manifestation.** Life expectancy of rare disease patient is significantly reduced. Many of these diseases are complex, degenerative and cause chronic invalidity, while others are compatible with a normal life if diagnosed on time, tracked and/or treated adequately. They affect physical, mental, behavior and sensory abilities and cause invalidity. Some invalidities are often accompanied by numerous **functional consequences** defined and poli-disabilities or pluri-disabilities). They enhance the feeling of isolation, may represent a **source of isolation** and may reduce education, professional and social possibilities.

Medical and scientific knowledge of these diseases is still in embryonic phase. Of the 8000 diseases, only a small fraction benefit from a deeper understanding of the physiological mechanisms involved. Most of them do not benefit from a specific treatment. Only in a few cases, care measures allow the improvement of patients' quality of life.

As rare diseases affect over 25 million people in Europe, the EU Health Commission seems more and more preoccupied to revive research in this field, to create an educational climate for the medical professional, patients and general population, with the declared purpose to provide the conditions for a correct and early diagnosis of these diseases, avoiding their recurrence in

family and ensuring a better life for these patients.

For this purpose, Patients' support groups – Patients' associations are accepted and encouraged, having the most important successes in advocacy work and managing to influence policies and stimulate medical research, due to the “personal involvement”. These are established and managed by parents of rare disease children or by patients themselves, they effectively collaborate with specialists and they are “the engine of research” in the field of rare diseases.

There are many types of patient group organization in the world, from tiny groups to large organizations, that may be focused on supporting patients with a particular disease or a number of diseases, have members in a certain country, or are organized at European or world level.

In such an organization, patients unite their efforts to collect information about the disease, accessible treatment, medical services or existing specialists in the world, and each of their experiences is cherished at its true value.

Estimating a rare disease prevalence of 6-8% in Romania, just as in the other European countries, these diseases affect approximately 1.300.000 people in our country, of which around 1.250.000 patients do not yet have a correct or complete diagnosis, nor treatment or adequate care.

## **ISSUES**

Rare diseases (RD) are life-threatening, often chronic and very complex diseases. The majority of them are genetic, toxic or infectious. They require a global approach based on special combined efforts in order to prevent significant morbidity and premature mortality that might have been avoided, as well as to improve the quality of life and the socio-economic potential of people affected.

The lack of **specific policies regarding RD** and the expertise deficit in this field lead to late and wrong diagnosis and limited access to medical care. This leads to additional physical and mental deficiencies, sometimes the birth of affected siblings, inadequate or even damaging treatments,

as well as loss of confidence in the medical health system.

In spite of all this, some RD are compatible with a normal life if they are diagnosed on time and adequately followed-up. A wrong diagnosis or the lack of diagnosis are the main obstacles in the way of improving the quality of life for thousands of rare disease patients.

The interest shown towards rare diseases is a **relatively new phenomenon** in the majority of EU member states. Not long ago, public health authorities and political decision makers ignored these challenges, because of the division of political debates around the large number of different rare diseases, rather than acknowledging the common aspects of all RD.

National medical care services for the diagnosis, treatment and rehabilitation of rare disease patients greatly differ with regards to availability and quality. Citizens of member states and/or regions of member states have **unequal access** to specialized services and orphan medicines. Certain member states successfully approached some of the issues raised by the rarity of diseases, while others have not yet envisaged possible solutions.

The role of the European Community regarding health is to encourage the cooperation between member states and, if necessary, to support their actions in this field. The specific characteristics of rare diseases – limited number of patients, insufficient knowledge and expertise in the field – individualizes this as a special field with a very high added value at European level.

**A community action program regarding rare diseases, including genetic diseases**, has been adopted covering January 1, 1999 – December 31, 2003. This program defined prevalence as reduced if the disease affects **less than 5 in 10.000 people** in the European Union.

Although this prevalence of 5 in 10.000 seems small, it represents approximately **246.000 people/rare disease in the European Union with 27 member states**.

Based on current scientific knowledge, **between 5.000 and 8.000 different RD** affect a percentage of up to 6% from the total population of EU at a certain moment in life. In other words, **approximately 29 million** citizens are or will be affected by a RD in the European

Union.

On **09.06.2009**, the Council of health Ministries in the European Union adopted a European strategy through which all EU member states are required to implement national plans for rare diseases, by the end of 2013. The Council Recommendation is important as it appeals to a joint action, concentrated at European and national level, having the following objectives:

- To ensure an adequate codification and classification of rare diseases;
- To intensify research in the field of rare diseases;
- To identify Centers of Reference and to include them in European Networks of Reference;
- To support bringing together European level expertise;
- To share evaluations on added clinical value of orphan medicines;
- To encourage patients' initiatives by involving them and their representatives in all stages of decision making;
- To ensure sustainability of rare disease infrastructure.

Adopting a European strategy is the peak of all documents and regulations facilitating the recognition of rare diseases as a public health priority and the concentration of European efforts in this field.

EURORDIS (European Organization for Rare Diseases) and national rare disease patients organizations are in the centre of this process, making known the patients' requests regarding the necessity to have a legal framework for rare diseases. It all started with the success of the public consultations regarding rare diseases in November 2007, followed by adopting the Commission Communicate on rare diseases in November 2008, then adopting the Council recommendations on European actions in the field of rare diseases. Each step demonstrated the vital importance of European actions and cooperation between member states.

Now, that there are political instruments, EURORDIS together with the other organizations interested, will follow the implementation of the Council recommendation both at European and at national level.

Research in the field of rare diseases has played an essential role in identifying the majority of

human genes until now, as well as the discovery of a quarter of the innovative medicines authorized on the EU market (**orphan medicines**). Research in rare diseases proved to be useful for a better understanding of the mechanisms of common diseases such as obesity and diabetes, as these are a model of biological process dysfunctions. In spite of this, research in the field of rare diseases is not only insufficient, but dispersed in various labs in all EU.

## **CURRENT SITUATION IN ROMANIA**

1. Many rare diseases are undiagnosed/diagnosed late because of the fact they are unknown for doctors in the field, due to the small number of specialists in medical genetics and the absence of specialized diagnosis centers;
2. Neither specialists, nor patients have the necessary information regarding the rare disease, to facilitate a correct diagnosis and the application of existing methods of treatment, rehabilitation and integration;
3. There is no national network of specialized medical centers doing identification, diagnosis and follow-up of rare disease patients;
4. Lab tests for the conformation of genetic diagnosis are frequently made abroad, at very high costs;
5. Many of the patients who are identified, are not properly monitored due to lack of protocols and best practice guides;
6. Prevention services that could identify and provide genetic counseling to patients and their relatives are generally lacking;
7. In case of diseases for which there are orphan medicines, they are not available for Romanian patients or when available, they are inconsistently administered;
8. Social services that may improve the lives of rare disease patients are scarce and there are no services yet specialized for rare diseases;
9. There are no statistical studies regarding the frequency of these diseases in Romania;
10. The collaboration with the European network of specialized services in rare diseases is sporadic and not systemized.

## **HOW IS THIS NATIONAL PLAN FOR RARE DISEASES POSSIBLE?**

### **The commitment level of patient organization, the involvement of the Romanian Society of Medical Genetics (SRGM) and the openness of the Health Ministry towards this issue:**

The Romanian Prader Willi Association brought together in August 2007, 32 organizations and groups of patients affected by rare diseases in Romania establishing the National Alliance for Rare Diseases Romania, involving a large group of specialists from the whole country, especially members of the Romanian Society of Medical Genetics.

The involvement of Health Ministry representatives in the work groups and expert groups, as well as the consultation of other ministries during a round table discussion organized through a CEE Trust funded project, lead to the unanimous agreement that rare diseases are and should be considered a public health priority in Romania too.

Community actions will most certainly support member states in providing an effective integration and organization of the limited resources in the field of rare diseases, and they can help patients and professional from various member states to collaborate in order to exchange information and coordinate expertise.

The European Commission will stimulate a consolidated cooperation between EU programs, with the purpose to maximize available resources for rare diseases at community level. At national level, a joint cooperation is needed between these groups, in order to implement the objectives and activities stipulated in this plan, so that they may be developed as joint projects at national level.

## **WORK METHODOLOGY**

The development of the NATIONAL PLAN FOR RARE DISEASES was achieved in six stages:

1. Identification of needs, intervention fields and problems within the work groups created;
2. The development of the plan;
3. The public debate and adjustments stage, according to suggestions expressed during the meetings organized by the Health Ministry Work Groups, Ministry of Labor, National



Medicines Agency, National Authority for People with Disabilities, National Authority for the Protection of children's Rights, representatives of the Bulgarian Health Ministry and EURORDIS (European Organization for Rare Diseases). The comments have been formulated by the Group of European experts and the conclusions drawn during the National rare Disease Conference, in November 2-3 2007: "***Rare diseases, from identifying needs to setting priorities***";

4. Signing of the partnership agreement with the Ministry of Health on 29.02.2008: "**Rare Diseases, a Public health priority in Romania**";
5. Detailing the objectives and the activities of the National Plan for Rare Diseases within the National Committee for Rare Diseases;
6. Inclusion of the National Plan for Rare Diseases in the National Public Health Strategy.

In the first stage, during the meeting organized to reach an agreement and create the Work Group, having 32 representatives of rare disease patients and specialists, the main fields have been identified to become part of the strategic plan. A preliminary list has also been completed, consisting of the main health issues rare disease patients are facing in Romania, a list of problems specialists face in diagnosing and managing rare diseases.

The selection of priority issues to develop the **NATIONAL PLAN FOR RARE DISEASES** was achieved during an active participative process, where all Work Group members attended.

In order to finalize the document, EU statistics have been used, as well as national statistics, existing legislation and specific strategies. More Work Group meetings have been organized and an ongoing communication was held with the group of Experts.

After finalizing the National Plan for Rare Diseases, this became accessible on the [Romanian Prader Willi Association](#) website. In the same time, it was debated with representatives of the health Ministry, and a summary of this plan was sent to the European Commission – DG Sanco, [EURORDIS](#), [ORPHANET](#), main public health institutions in the country. It was also presented in various national and international conferences, explicitly requesting feedback in their fields of competence. Based in these reactions and comments, the final version of the **NATIONAL PLAN FOR RARE DISEASES** was created.

At this point, a [Council Recommendation on action in the field of rare diseases](#), and the NATIONAL PLAN gains a European perspective because it follows the direction of the European Commission regarding national policies for approaching rare diseases (see chapter on **Plans and strategies in the field of rare diseases** and chapter on **Empowerment of patient organisations**)

## **TARGET GROUPS**

- Rare disease patients – approximately 6-8% of the country population, 1.300.000 people;
- Specialists from all levels of health care system, social workers, teachers and other specialists involved in diagnosing and managing rare diseases;
- NGOs in the field, patient organizations;
- The community.

## **GENERAL OBJECTIVES OF THE NATIONAL PLAN FOR RARE DISEASES:**

- 1. To develop an institutional frame**
- 2. To develop services for the diagnosis, treatment, rehabilitation and prophylaxis in the field of rare diseases**
- 3. To improve access to information in the field of rare diseases**
- 4. To develop human resources**
- 5. To stimulate research in the field of rare diseases**
- 6. To increase the role of patient organizations**

## **ACTIVITIES:**

### **1. To develop an institutional frame**

#### ***1.1. Establishment of the National Committee for Rare Diseases - CNBR;***

CNBR brings together specialists, representatives of the National Alliance for Rare Diseases (ANBRaRo), of the Romanian Society of medical Genetics (SRGM), Health Ministry and other ministries and institutions involved. The Committee will meet on a quarterly basis, identify the needs and suggest measures that need to be taken to improve the quality of life for rare disease patients, will set criteria for nominating and assessing reference centers / inventorying resources.

#### **Assessment of current situation (2009-2010)**

There is an emphasis set on criteria that will be considered when accrediting reference / competency centers.

- Equipment – in laboratories and clinical services
- Human resources according to categories of staff
- Possible diagnosis
- Capacity
- Services
- Number of patients/year/disease in the past 5 years
- Institutional organization
- Access of patients to services
- Communication between departments – cohesion between teams of specialists
- Research according to fields
- History of collaboration (experience) with other sections of the same specialty/other specialties, patient associations
- Program management
- Registries
- Staff structure
- Existing infrastructure
- Publications in the field.

## ***1.2. Definition of institutional frame***

A hierarchical structure will be used where, under the coordination of CNBR and Health Ministry, a network is created, consisting of reference centers (national level), competency centers (regional level) and country medical clinics for rare diseases.

### ***1.2.1. Reference Centre – unique in the country for a disease or a group of diseases***

The selection of reference Centers will be made based on a national competition and regular review (every 5 years) of the activities.

Main responsibilities of Reference Centers:

- Administration of the National Rare Disease Registry for the disease or group of diseases for which it is accredited;
- Diagnosis – clinical and via specialized advanced investigations, initiation into treatment and development of indications regarding the follow-up and treatment of patients in competency centers;
- Collaboration in the development of clinical guides;
- Monitoring patients with a specific disease from the specific group of diseases;
- Linking research;
- Coordinating and promoting best practice guides;
- Management of health programs according to the disease group (coordination, planning, assessment both in the field of human resources and that of funding and related issues);
- Organizing the rare disease management process;
- Multidisciplinary approach, expertise, collaboration with patient organizations;
- Defining procedures and protocols to implement screening programs;
- Organizing the prescription and follow-up of very expensive orphan medicines;
- Information/training (patients, health professionals, networks);
- Regularly informing the Ministry of Health, CNBR, National Health Insurance

Institution, Ministry of Labor, Ministry of Education;

- Collaboration with the European Centers specialized in rare diseases;
- **Active collaboration with equivalent European institutions:**
  - Participation to the network or reference centers at European level;
  - Identification of methods of diagnosis and of European reference centers that offer such methods for the diagnosis of rare diseases;
  - Identification of sources of funding to allow the reimbursement of diagnosis and treatment expenses occurred abroad;
  - Stimulating the participation of the Romanian parties to European research projects in the field of rare diseases and co-funding these participations;
  - Active participation to epidemiologic studies developed in the European community to establish specific parameters in various rare diseases.

### ***1.2.2. Competency Centre – provides services at regional level***

Responsibilities of Competency Centers:

- Application of best practice guides agreed with all Reference Centers;
- Monitoring the provision of services and relay of information to Reference centers and County medical offices for rare diseases;
- Organizing/implementing screening;
- Developing a reference specialists database in collaboration with Reference Centers and County medical offices for rare diseases;
- Prevention, diagnosis, treatment, rehabilitation;
- Collaboration in European programs.

### ***1.2.3. Medical offices for rare diseases in each county***

County medical offices for rare diseases are organized at county level – they have at least one genetics specialist.

Responsibilities of County medical offices for rare diseases:

- Implementing screening;
- Identifying, referring, diagnosing and monitoring simple cases. Referring complex cases to Competency Centers;
- Role in informing and educating patients, families, the population;
- Establishing and maintaining links with patient organizations;
- Applying, supervising treatments and rehabilitation/integration procedures;
- Maintain records of patients and resources.

#### *1.2.4. Development of national registries for rare diseases*

Reference Centers are responsible for the establishment of Rare Disease Registries and the development of coherent policies regarding the epidemiologic supervision organized for rare diseases.

##### **Objective:**

To provide the necessary information for the development of a coherent policy in order to epidemiologically supervise rare diseases in an organized manner.

##### **Activities:**

- Collect secure and representative epidemiologic data for rare disease patients;
- Define a set of basic data to be included in the registry – standard collation protocols;
- Inform and involve interested medical centers;
- Inform all participating patients and obtain their consent;
- Collaborate with Rare Disease Registries kept at European level.

The rare disease registries will be developed by:

- Defining epidemiologic supervision methods for these parameters;
- Providing specialized assistance to ensure a correct epidemiologic supervision, such as:
  - Support the effective and correct epidemiologic data collection;
  - Use adequate statistical methods in collaboration with regional public health services;
  - Organize a network for relaying this data to all parties involved;

- Provide technical assistance for the analysis and synthesis of data collected during the epidemiologic supervision of rare diseases;
- Run a global epidemiologic study regarding rare disease caused mortality;
- Collect data regarding the disability degree, possibilities of social, educational professional adaptation of rare disease patients;
- Regularly register the evolution of patients included in the registry, identifying and organizing the assessment of people at risk in the family.

**The definition of strategies and communication protocols in this field, between local, regional and national responsibility levels**

Rare Disease Registries represent essential instruments in the improvement of knowledge regarding rare diseases and the development of clinical research. This constitutes the only means to collate the necessary data to obtain a sufficient sample size for epidemiological and/or clinical research.

Rare Disease Registries will ensure the collection and registration of data according to legislation in force regarding personal data protection.

***1.3. Steps before achieving the institutional framework***

- Establishment of CNBR, with the participation of government bodies with attributions in this field (Health Ministry, Ministry of Education, Ministry of Labor, National Authority for People with Disabilities), patient and professionals organizations;
- Definition of accreditation criteria and assessment procedures for reference centers, competency centers and country medical offices;
- Established criteria become public;
- Centers compile candidacy files
- Files are sent to Health Ministry, which makes the selection and authorization based on the set of criteria.

***1.4. Implementing the nominated network***

In the first instance, existing services will be supported and developed, services which have proved their efficiency. County medical offices for rare diseases will be established with the support of county hospitals.

### ***1.5. Ensuring the on-going funding of the network***

Firm and constant financing will be provided for the reference/competency/county medical centers, so that activity blockages due to lack of funds can be eliminated. Funding will be ensured from various sources: payment for certain services, national programs, education or expertise activities, epidemiologic studies, etc.

### ***1.6. Evaluation of network***

CNBR will develop an evaluation form.

- Evolution of activities since appointment;
- Regular evaluation and re-evaluation of the centers.

### ***1.7. Updating the legal framework***

The plan is to create an institutional frame to approach rare diseases at European and national level, and to adopt national plans for rare diseases until the end of 2013, in accordance with the European and international legislation and in partnership with EURORDIS, the Council of National Alliances – through the EuroPlan project funded by the European Commission/DG Sanco.

CE recommendations (during the Lisbon Conference, the public debate of the [Council Recommendation on action in the field of rare diseases](#) opened until 14.02.2008) have been taken into account.

Member States have a joint commitment to ensure equal access to high quality medical care, based on the principle of equity and solidarity. But in the case of rare diseases, expertise is limited. Certain centers of reference (also known as expertise or excellence centers in some



states) have developed an expertise and intensely used by professionals from that particular state or even internationally, allowing rare disease patients access to adequate medical care. There are a few such centers in our country as well (although they are not known as expertise/excellence/reference centers). These must be inventoried and supported to become reference centers according to European criteria.

The 2006 report of the Rare Disease Task Force, called [Contribution to policy shaping: For a European collaboration on health services and medical care in the field of Rare Diseases](#) recommends member states to contribute towards the identification of their centers of expertise and to support them financially.

## **2. To develop services for the diagnosis, treatment, rehabilitation and prophylaxis in the field of rare diseases**

### ***2.1. Improving access to a fast and correct diagnosis***

- Information about the existence of reference centers both among specialists and the general population;
- Equipment of reference centers with the necessary equipment and materials for diagnosis;
- Provision of staff qualified and specialized in rare diseases in these centers.

### ***2.2. Development of continuous/multidisciplinary services and rehabilitation services for rare disease patients***

- The establishment and support of rehabilitation centers (rehabilitation services within the centers of competency) specialized on types of disabilities produced by rare genetic diseases;
- Supporting the activity of counseling and information centers;
- Establishment of care recommendations and protocols by Reference Centers;
- Dissemination of best practice models, follow-up provided to the patient and family at

different levels;

- Initiation of a National Plan of Social Services in collaboration with the Ministry of Labor, Social Solidarity and Family;
- Establishment of a **pathway of services (patients' care pathway)** that can be provided (based on the particularities of each disease) and of centers providing these services;
- Performance evaluation of care systems, care methods and psycho-social support;
- Establishment of wards/centers treating a small group of diseases with identical or very similar characteristics;
- Specific training of staff (according with groups of diseases);
- Development and functioning of a **multidisciplinary team** (appointing specialists in the multidisciplinary teams).

### ***2.3. Specialized Social Services***

Existing specialized social services will be supported, and new ones will be initiated. Medical care pathways will be organized for patients, by developing cooperation with experts in country and abroad. A multidisciplinary approach of medical care will be promoted in case of complex and diverse problems such as those caused by rare diseases. The integration of medical and social levels will be encouraged, taking into account patients' and medical staff needs and expectations.

Improving specialized care for rare disease patients:

- Collaboration of reference and competency centers in the diagnosis and care of rare disease patients with the County Social Work Departments, so that the monitoring of rare disease patients can be made by a multidisciplinary team specialized in the patients' real needs;
- Development of a medical care and social work network grouped around reference centers;
- Organization of a transport network to allow the transport of rare disease patients to the reference centers;
- Organization of psychological and social counseling services around reference centers, so

that the best care can be provided to rare disease patients and their families;

- Improving the communication between the medical staff, the paramedics, patients and patient organizations for a better care of rare disease patients;
- Through the collaboration between doctors specialized in rare diseases and County Social Work Departments in order to develop monitoring guides for rare disease patients, to include all possibilities of diagnosis and treatment, as well as all measures necessary for patients' integration into society;
- Development of respite care centers to meet the needs of patients and their families;
- Ensuring the connection between patient organizations and the National Health Insurance System (via the Health Ministry), so that a quick reimbursement system becomes operational for the treatment and care expenses related with rare disease patients.

#### ***2.4. Intensifying efforts in favor of orphan medicines***

##### ***2.4.1. Ensuring availability of orphan medicines and the reimbursement/compensation of orphan medicines costs through the public health insurance system***

- The introduction of orphan medicines on the list of free medicines will be supported;
- The improvement of the medicines provision system for rare disease patients, before the approval and/or reimbursement of new medicines (the so called “compassionate use”);
- The evaluation of the therapeutic added value of all orphan medicines by the Health Ministry and the National Medicines Agency;
- The cost of orphan medicines will be covered from the general health insurance budget, not the budget of hospitals or reference centers;
- Diagnosis and care protocols will be established by the Health Ministry and reference centers; funding for medicines without marketing authorization, nutritional supplements, medicine food, crèmes and bandages, special dental care, clothes (i.e. epidermolysis bullosa), based on current best practice models and patients' needs.

**The recognition of rare diseases and chronic diseases** implies the following approaches:

- The simplification of reimbursement procedures by acknowledging rare diseases as invalidating chronic diseases;
- Adding all specific treatments to the free and compensated list of medicines, including similar products or equivalents from the same therapeutic group, for cases where standards recommended therapy does not work;
- The simplification of reimbursement procedures of transport expenses to and from reference centers;
- Improvement of knowledge about rare diseases and recognition of the particularities of these diseases, by the medical staff working in the national health insurance system, so that unjustified refusal of reimbursement may be limited at maximum.

### **Measures:**

- The Health Ministry sets the criteria based on which a rare disease may be included in the chronic disease list and identifies reference centers able to diagnose and initiate the therapy and care procedures for rare diseases;
- Certifies the use of certain medicines for treating rare diseases and imposes the Health Insurance Departments to reimburse expenses related with these therapies, even if the same medicine used for the treatment of common diseases does not benefit from compensation;
- Develops diagnosis and treatment protocols for rare diseases, so that the reimbursement of expenses is justified;
- Establishes the regional registration of patients with certain diseases and finds solutions for the reimbursement of travel expenses;
- Includes patient organizations representatives in specialized commissions, so that all decision regarding rare diseases are taken with the patients' support.

The social and economic impact of rare diseases lead to considerable efforts in the study of the etiopathology and therapeutic possibilities. Innovative medicines appeared in the past few years, authorized on the EU market as efficient medicine for a series of rare diseases. According to the Recommendation of the EU Pharmaceutical Forum - [Improving access to orphan medicines](#)

for all affected EU citizens – adopted by the member states on October 2<sup>nd</sup> 2008, the main current objective is the promotion of sustainable development of valuable orphan medicines and improving the sustainable access to these medicines for all EU patients.

Great efforts are made for the identification of new medicines, called “orphan medicines”, in the circumstances where they are applicable for less than 2/10.000 patients. At European level, research in this field is controlled by the Committee for Orphan Medicinal Products, within the European Medicines Agency - EMEA. This committee analyzed over 800 medicine proposals during the past 10 years, giving the favorable approval for 50 such products, currently valid in all EU states. These medicines are proved as effective and beneficial for a number of rare and very rare diseases.

**To improve access of Romanian rare disease patients to orphan medicines, we suggest the establishment of a national strategy to provide the necessary treatment and the rehabilitation of rare disease patients, including:**

- Development and dissemination of diagnosis, treatment and care protocols for the main rare diseases (or groups of rare diseases), in collaboration with the Health Ministry, The National Health Insurance System, the Ministry of Labor, Family and Social Protection.
- Biannual updating and dissemination of therapeutic protocols and EMEA list of orphan medicines to the entire medical network, irrespective of the specialization, through National Health Insurance Departments, regional representatives and County Sanitary Departments;
- Facilitate the development and smooth running of the necessary documentation to obtain access to specific medicines for a certain disease;
- Include newly diagnosed patients in the program, right after submitting the necessary documentation by the patient’s doctor and their registration in the rare disease registry, in the valid specific treatment section;
- Reimbursement, in certain situations, of therapy costs for patients that, by meeting all the selection criteria for the specific therapeutic program (according to the documentation filed by the doctor in agreement with the therapeutic protocol), purchased the necessary medicines from their own money. (\*therapeutic emergency, impossibility of on-going treatment due to lack of specific medicine).

- Monitoring of each patient following a specific treatment, based on reports received from the patient's doctor, with the purpose to have an exact evidence of therapeutic compliance and efficiency, and to allow necessary reports be available for the epidemiologic studies and EMEA work groups.

***2.4.2. Supplementation in the number and categories of products included in the health insurance frame contract; diversification of assistive devices***

Collaboration with the Ministry of Labor and National Authority for People with Disabilities in order to prepare joint documents for this purpose.

Completion of the list of devices needed for the rehabilitation/improvement of deficiencies, reimbursed by National Health Insurance System:

- Prostheses;
- Hearing aids;
- Lenses and magnifying glasses;
- Orthosis;
- Compensatory hearing devices and materials
- Tiphlo-technical means;
- Orthopedic devices;
- Walking aids;
- Assistive devices to facilitate daily activities;
- Verticalisators – verticalizing frames
- Different sized wheelchairs for children, with adjustable elements;
- Folding wheelchairs;
- Wheelchairs with optimized and extended motional facilities – i.e. verticalization possibilities;
- Complex orthetic systems and devices;
- Respiratory support devices;
- Financial support of medicine foods for various diseases (i.e. PKU), nutritive supplements;

- Non-adhesive special bandages (EB), nasogastric tubes (EB).

### ***2.5. Development of prevention services for rare diseases***

The implementation of national level neonatal screening for phenylketonuria and congenital hypothyroidism.

Neonatal screening for identifying PKU and congenital hypothyroidism represents a current practice in Europe and proved extremely efficient in preventing disabilities for the children affected. As technology develops, many tests can be made these days, including the test made through automated systems, at a low cost, for a wide range of rare diseases, especially metabolic or genetic diseases in general.

**It is necessary to implement and monitor screening programs for identification of phenylketonuria and congenital hypothyroidism in all counties in Romania, as soon as possible, as stipulated in the National Health Program, under point 11 from the National Health Program for Women and Children.**

#### **Objectives:**

- To introduce a coherent policy for the application of rare disease screening, based on the clear definition of priorities as a result of rigorous analysis of social benefits versus financial efforts required by such a policy;
- To improve diagnostic tests;
- To participate to the development of common European policies in the field of rare disease screening.

#### ***2.5.1. Developing a screening network in the field of rare diseases by organizing and implementing population screening programs:***

- Establishment of a general control procedure to clearly define the implementation stages and evaluation methods for the correctitude of screening methods applied;
- A systematic evaluation before and after the screening program application from all

parties interested (Health Ministry, Public Health Institute, Romanian Society of Medical Genetics, patients organizations);

- Development by the Health Ministry of a methodological guide to evaluate the quality of screening programs;
- Establishment of an independent consultative committee to analyze the efficiency of screening programs and to provide solutions for the improvement of their application;
- Training and developing a team of evaluators to allow the development and implementation of new methods and rare disease screening programs;
- Development of a legal frame and methodology necessary for the implementation of rare disease screening programs.

### ***2.5.2. Improving access to genetic diagnostic techniques and provision of genetic advice:***

#### **Improve genetic testing methods by:**

- Identification of laboratories capable to run cytogenetic tests, molecular cytogenetics and genetics – through collaboration between Medical Genetics Commission from the Health Ministry and the Romanian Society of Medical Genetics;
- Accreditation of these labs according to European standards in force;
- Identification (by the Medical Genetics Commission from the Health Ministry and the Romanian Society of Medical Genetics) of a group of 50-100 rare diseases for which the molecular testing can be made in Romania;
- Constant financial support of these labs for the molecular testing of rare diseases with state budget funds or various sponsorships;
- Identification (by the Medical Genetics Commission from the Health Ministry) and establishment of partnerships with laboratories from the European Community able to run molecular tests for diseases that cannot be tested in Romania;
- Reimbursement of medical services obtained abroad based on approvals from a commission of specialists;
- Provision of correct and competent pre- and post-testing genetic counseling;
- Prenatal diagnosis when there is significant risk of rare disease pregnancy, introducing



certain diseases with high morbidity and/or mortality rates on the list of diseases for which therapeutic abortion is recommended.

**Improving methods of information dissemination to specialists and general public regarding screening programs and methods of diagnosis of rare diseases by:**

- Developing programs to increase the awareness of medical staff and general public regarding screening programs via printed materials and mass-media channels (newspapers, radio, TV);
- Organizing continuous medical education programs for health staff in the field of rare disease screening.

*2.5.3. Participation to the joint efforts of the European Community for establishing common policies regarding rare disease screening*

**3. To improve access to information in the field of rare diseases**

*3.1. Promoting the National Plan for Rare Diseases to the specialists, patients, patient organizations and authorities*

- Development of information brochures about the **National Plan for Rare Diseases** and their distribution to the medical staff, medical centers involved in the care of rare disease patients and patient organizations;
- Dissemination on European level of information regarding the National Plan for Rare Diseases;
- Organization of conferences, seminars and round tables (for example, the EuroPlan Conference in June 2010);
- Involvement of mass-media in the promotion of the National Plan for Rare Diseases.

*3.2. Improving access to general information regarding rare diseases*

An essential element in improving diagnosis and specialized assistance in the field of rare

diseases is the provision and dissemination of exact information, in a form adapted to the needs of professionals and patients.

- Creating and developing secure sources of information and increasing access to these for specialists in the field of rare diseases, by involving specialists organizations and the Health Ministry;
- Creating and developing secure sources of information and increasing access to these for patients and their families (i.e. through patient organization, **Helpline, Information centers for rare diseases, etc**);
- Supporting patient organizations in their efforts of information dissemination;
- Disseminating information about services related with rare diseases for patients and specialists;
- Developing and providing access to a map of services for patients and specialists;
- Improving access to secure information (i.e. Orphanet) in Romanian.

### ***3.3. Sensitizing the general public***

- Support the development of awareness campaigns in order to fight against stereotypes and prejudices faced by rare disease patients and to support the acknowledgement of their rights;
- Cooperation between the National Alliance for Rare Diseases and the Health Ministry in organizing the annual National campaign **International Rare Disease Day**, on the last day of February.

### ***3.4. Information networks in the field of rare diseases***

Priority action points regarding the existing information networks around specific diseases:

- To guarantee the information exchange through existing national and European information networks;
- To develop strategies and mechanisms for the information exchange between interested parties;
- To support best practice exchange and development of measures for patient groups;

- To create and disseminate best practice guides, adapted to patients, for the medical and paramedical staff (psychologists, medical nurses, social workers, physiotherapists, speech therapists, etc) with information on:
  - Diagnostic possibilities
  - Network of laboratories where diagnosis can be certified;
  - Particular methods of care and treatment for rare disease patients;
  - Access to clinical and/or therapeutic trials open in Romanian or international reference centers;
  - Network of centers providing such services.
- Increasing patients', medical and paramedical staff access to information about:
  - Patient care and access to various treatment methods
  - Network of experts in the rare disease field and possibilities of contact;
- Improving access to information for people with special needs (language or social problems, those included in minorities with deficient social integration), by educating and involving staff working in such communities (doctors, medical nurses, social workers, psychologists);
- Counseling for the professional guidance and access to labor market;
- Development of partnerships with phone companies and mass-media trusts to promote the rare disease issues to the general public;
- Information dissemination through Complex Evaluation Services within the Country Departments of Social Work and Child Protection.

#### **4. To develop human resources**

##### ***4.1. Training programs for specialists in various fields regarding ways to approach rare diseases***

#### **Objectives:**

- Introduction of rare disease training courses in the university curricula;
- Experience exchanges;
- Master's Degrees programs;

- Introduction of rare diseases in the curriculum of all medical specialties (presenting the specific pathology in each specialty)
- Development of curricula for the staff employed to look after patients;
- Organization of national and international experience exchanges between human resources that wish to train in this field (those participating to these exchanges then to become trainers in Romania)

### Measures:

- Identification of training needs;
- Identification of potential beneficiaries of training programs (establishing a multidisciplinary team of specialists contributing to the diagnosis and intervention in rare diseases, that could subsequently train other specialists in their field of activity);
- Identification of potential trainers;
- Development of a multidisciplinary team to create the curricula on ways to work with rare disease patients (having a more general characteristic and targeting the basic training of all those involved in diagnosis and intervention);
- Submission of the curricula to the Ministry of Education and Health Ministry for approval and validation;
- Organization of training programs for specialists involved in the rare disease diagnosis and intervention, programs accredited and acknowledged by the Ministry of Education and Health Ministry;
- Organization of workshops having as main objective the acquiring of competences when working with rare disease patients (general workshops for all those involved in diagnosis and intervention and specific workshops, adapted to the participants' field of expertise);
- Development of a credits system for specialization management;
- Selection of students willing to work in the field of rare diseases from universities and their inclusion in professional training programs, to practice in this field;

At the level of each county (or county municipality), **counselors or mediators** will be trained for people diagnosed with rare diseases. The National Alliance for Rare Diseases and the Information center for Rare Diseases will be involved in the training and coordination of these mediators.

The role of counselors / mediators:

- Information and counseling regarding the local, regional, national and European social infrastructure available for the needs identified together with the patient;
- Suggestion of intervention alternatives together with the rare disease patient;
- Provision of assistance for the patient's problems (access to information, documents needed to benefit from rights stipulated by law);
- Organization of information seminars, events (connected to national level events, such as the Rare Disease Day), as well as local;
- Identification of potential partners in the implementation of projects with internal or external funding, counseling for those willing to apply to national/international grants/funding programs.
- Information activities/sessions for groups of various specialties, adapting the information to the respective field (i.e. in case of teachers: educational planning, for family doctors, for children in kindergartens and schools – educational programs, etc).

#### ***4.2. Ensuring specialized staff in employed in the health care system for people with rare diseases***

- According with types of socio-medical services: geneticists, specialist doctors, biologists, medical nurses, laboratory workers, social workers, psychologists, vocational therapists, physiotherapists, speech therapists, special education teachers, sociologists, specialized educators, etc.
- Training courses for all specialists involved in the diagnosis and management of rare diseases, adapting the university curricula, authorizing and accrediting specialization courses, initiating projects on this theme, printing updated educational materials.

#### **Adapting basic medical training:**

- Introducing rare diseases as a compulsory subject in one of the final years of study in

medicine or medical nurses college;

- Introducing seminars on rare diseases in the professional basic training of paramedical specialists (psychologists, physiotherapists, social workers, etc) involved in the care of rare disease patients.

#### **Organization of ongoing medical training in the field of rare diseases:**

- Creation of an adequate number of vacant positions specialized in medical genetics for residents (as 80% of rare diseases are genetic)
- Introduction of modules on rare diseases in the training of specializations involved in the care of these patients (neonatology, pediatrics, psychiatry, neurology, internal medicine, endocrinology, cardiology, dermatology, ophthalmology, ENT, family medicine), in collaboration with the National Center of Training in Health;
- Master's Degree programs in the field of rare diseases;
- Organization of seminars, round tables, post-graduate courses targeted on a specific field in rare diseases, in collaboration with patient organizations, The Romanian Society of Medical Genetics and universities in Romania;
- Development of information materials in the field of rare diseases, available on the websites of all institutions involved in this field (Health Ministry, Romanian Society of Medical Genetics, patient organizations, etc)

#### **Increase the amount of information in the training of paramedical staff involved in the care of rare disease patients:**

- Modification of basic curricula and ongoing post-graduate training of paramedical staff: medical nurses, therapists (vocational, speech, physio), social workers, psychologists.

**Improving the medical information circuit** regarding rare disease patients, respecting the principles of confidentiality.

### **5. To stimulate research in the field of rare diseases**

#### ***5.1. Improving the capacity to access research projects in the field of rare diseases,***

*international partnerships;*

- Organizing training courses on project writing, counseling for accessing national and European funding.

**5.2. Stimulating scientific research in the field of rare diseases in Romania**

Encouraging collaboration in research projects, European and international networks;

- FP7 projects; etc. (rare disease – one of the priorities)

**Considering rare disease as a priority in Romanian medical research:**

- Logistic and financial support for the research projects aiming the identification of new diagnosis methods and new therapy products for rare illnesses by organizing a national contest for research programs in the field;

**Coordinating research efforts in the field of rare disease.**

- Accomplishing a partnership with the competent structures in the research field at the Ministry of Education, Research and Innovation (National Council of Scientific Research in Superior Education, National Authority for Scientific Research, etc.) through the establishment of a multi-annual research plan in the field of rare disease, having financial support and allowing the yearly launch of a competition for research projects.

**Establishing the priority fields in rare disease research:**

- *Epidemiology* – promotion of research projects aiming the analysis of rare disease epidemiology parameters (prevalence, incidence, morbidity, mortality, natural evolution, clinical nosology, etc.).

- *Genetics* – making etiopathogenic studies of rare disease for a better understanding of mechanisms of production, allowing to find new diagnosis methods.

- *Pharmacology* – discovering new therapy drugs, especially acting at cellular or molecular level, should be a long-term objective, unfortunately the lack of interest of pharmaceutical companies could be a major obstacle in rare disease research, imposing new approaches in order to stimulate the drug producers to invest in such a venture.

- *Treatment and care* – forming research teams including qualified staff also from the field of social sciences, in order to study the impact of new therapies and identify new measures for improving care of patients with rare disease.

- *Social research* – assessing the quality of life for patients with rare disease and

evaluate the existing needs of treatment, care and support; starting research projects concerning impact assessment of the patient organization's activities.

**Stimulating pharmaceutical companies to invest in rare disease research:**

- Establishing a partnership between the Health Ministry and pharmaceutical companies in order to produce clinical studies to validate the actions of these drugs, respecting strictly the laws in the field as well as the right of patients with rare disease;

**Identifying research needs in the field of rare disease (at medical, psychological, social, etc. level)**

**Establishing research priorities in the field of RD.**

Collaboration between research centers and patient organizations in order to identify research subjects.

Creating a database with documents relevant in the field of rare disease.

Publishing the research results in scientific magazines with reputation in the field.

Dissemination research results to professionals and patient organizations.

Developing the infrastructure needed for research.

Elaborating animal models for research in rare disease.

Elaborating research, collaboration protocols .

Initiating joint research projects.

Promoting a volunteer and dedicated research policy especially in the field of clinical studies.

Developing some tests for diagnosis.

Collaborating with the Health Ministry on projects such as E-rare / ERA-net.

**6. To increase the role of patient organizations**

***6.1. Supporting the development of patients organizations, ensuring their sustainability re;***

- Financial support of promotion actions, national interest information campaigns e. g. Day of Rare Disease



- Collaboration in organizing national and international events;
- Initiating partnerships in strategic projects.

***6.2. Ensuring the representation of patient organizations at decision making levels within public institutions (national and local)***

- Including the patient organizations' representatives in the Health Ministry special committees
- Including the patient organizations' representatives in all the decisions concerning patients with rare disease and dedicated services.
- Supporting organizations with relevant experience in the field in the process of public utility recognition.