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PROJECTS OF THE BULGARIAN ASSOCIATION FOR PROMOTION OF EDUCATION AND SCIENCE (BAPES)

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RARE DISEASES IN BULGARIA

PERIODIC REVIEW OF THE ACTUAL SITUATION AND TENDENCIES IN THE RARE DISEASES FIELD IN BULGARIA

Methodology

This review aims to describe in details and to analyze the current situation and trends in the field of rare diseases in Bulgaria. It contains 5 main chapters:

- **Methodology and governance of the Bulgarian National plan for rare diseases (2009 – 2013)[1]**
- **Definition, codification and inventorying of rare diseases**
- **Research on rare diseases**
- **Centres of expertise, reference networks and access to orphan drugs**
- **Patient empowerment**

Each chapter is composed of 3 sections:

- **Relevant information from the EU Council recommendation on action in the field of rare diseases[2]**
- **Relevant information from the specific EUROPLAN recommendations[4]**
- **EUROPLAN indicators to evaluate the achievements of rare diseases initiatives[4]**

The project EUROPLAN, which is funded by the European Commission, aims to help Member states in developing national policies to improve access and opportunities for prevention, diagnosis and treatment of people with rare diseases by making recommendations, identifying, collecting and disseminating best practices, elaborating indicators and supporting organizing national rare diseases conferences.

The EUROPLAN indicators, which are used in this review, have been selected to monitor the implementation of national plans and strategies and to evaluate their impact. This set of indicators also contributes to ensure the comparability of data among Member states and the implementation status of the Council recommendation on rare diseases in the EU countries.

Definitions

- ***Rare disease* – a life-threatening or chronically debilitating condition affecting not more than 5 in 10 000 persons in the EU.**
- ***Orphan drug* – a medicinal product which is intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition, affecting not more than five in 10 thousand persons in the Community when the application to EMA is made.**

Used sources and links

1. Bulgarian National plan for rare diseases 2009 - 2013 (genetic, congenital malformations and non-hereditary diseases) (Bulgarian Ministry of Health)
2. EU Council recommendation on action in the field of rare diseases (Directorate-General for Health and Consumers (DG SANCO)
3. EC Communication on rare diseases (Directorate-General for Health and Consumers (DG SANCO)
4. EUROPLAN Project
5. Bulgarian EUROPLAN national conference for rare diseases, 28-30 May 2010, Plovdiv, Bulgaria
6. Information centre for rare diseases and orphan drugs

EUROPLAN
European Project for Rare Diseases National Plans Development

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INTRODUCTION

Rare diseases are a threat to the health of EU citizens as they are life-threatening or chronically debilitating diseases with a low prevalence and a high level of complexity.

It is estimated that between 5 000 and 8 000 distinct rare diseases exist today, affecting between 6 % and 8 % of the population in the course of their lives. In other words, although rare diseases are characterised by low prevalence for each of them, the total number of people affected by rare diseases in the EU is between 27 and 36 million. Most of them suffer from less frequently occurring diseases affecting one in 100 000 people or less. These patients are particularly isolated and vulnerable.

Because of their low prevalence, their specificity and the high total number of people affected, rare diseases call for a global approach based on special and combined efforts to prevent significant morbidity or avoidable premature mortality, and to improve the quality of life and socioeconomic potential of affected persons.

In recent years, rare diseases have become a priority area in public health of the European Union. With several official documents, EU called on Member states to take concrete measures to improve prevention, diagnosis, treatment and rehabilitation of people with rare diseases. The highlight of these initiatives was the adoption of EU Council recommendation on an action in the field of rare diseases (8 June 2009). It invites the Commission to produce, by the end of 2013 and in order to allow proposals in any possible future programme of Community action in the field of health, an implementation report on this recommendation addressed to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions and based on the information provided by the Member states, which should consider the extent to which the proposed measures are working effectively and the need for further action to improve the lives of patients affected by rare diseases and those of their families; and secondly, to inform the Council of the follow-up to the Commission Communication on rare diseases on a regular basis.

In this context, thanks to intensive work, done by the Bulgarian Association for Promotion of Education and Science (BAPES) and National Alliance of People with Rare Diseases (NAPRD), Bulgaria went ahead by adopting a National Program for Rare Diseases 2009-2013 (genetic, congenital malformations and non-heritable diseases) on 27 November 2008.

Chapter 1:
Methodology and governance of the Bulgarian National plan for rare diseases

1.1. EU Council recommendation:

I. PLANS AND STRATEGIES IN THE FIELD OF RARE DISEASES

- *Establish and implement plans or strategies for rare diseases at the appropriate level or explore appropriate measures for rare diseases in other public health strategies, in order to aim to ensure that patients with rare diseases have access to high-quality care, including diagnostics, treatments, habilitation for those living with the disease and, if possible, effective orphan drugs, and in particular:*
  (a) elaborate and adopt a plan or strategy as soon as possible, preferably by the end of 2013 at the latest, aimed at guiding and structuring relevant actions in the field of rare diseases within the framework of their health and social systems;
  (b) take action to integrate current and future initiatives at local, regional and national levels into their plans or strategies for a comprehensive approach;
  (c) define a limited number of priority actions within their plans or strategies, with objectives and follow-up mechanisms;
  (d) take note of the development of guidelines and recommendations for the elaboration of national action for rare diseases by relevant authorities at national level in the framework of the ongoing european project for rare diseases national plans development (EUROPLAN) selected for funding over the period 2008-2011 in the first programme of Community action in the field of public health.

1.2. EUROPLAN recommendations:

- Rare diseases are recognised as a priority in the public health area because of their specific needs.
- Initiatives are stimulated to raise awareness about the dimension of the problem and create joint responsibility.
- A mechanism (e.g. interdisciplinary panel, committee) including relevant stakeholders is established to assist the development and implementation of the National Plan or Strategy.
- An inventory of existing healthcare resource, services and policies is made including those directly addressing rare diseases as well as those from which rare disease patients may benefit (e.g. in the fields of disability, child healthcare, reimbursement, epidemiological surveillance, psychological and social care, etc.).
- Unfulfilled needs of patients are assessed.
- Available options for improving health and social care of people affected by rare diseases at national level are evaluated.
- European collaboration and the European documents in the field of rare diseases are taken into account in the development of the National Plan or Strategy.
- The National Plan or Strategy is elaborated with well described objectives to be achieved within a specific timeframe of limited duration. The general objectives of a National Plan or Strategy are based on the general overarching values of universality, access to good quality care, equity and solidarity.
- The policy decisions of the National Plan or Strategy are integrated, i.e. structured maximizing synergies and avoiding duplications with existing functions and structures of the health care system of the country.
- The policy decisions of the National Plan or Strategy are comprehensive, addressing not only health care needs, but also the other important needs of patients with rare diseases and their families, such as social needs.
- Specific areas are indicated, with priority given to those areas where major needs have been identified in the Member state.

- The allocation of appropriate resources, consistent with own decision-making processes, is important to ensure the feasibility of the actions in the planned time.
- Information on the National Plan or Strategy is made accessible to the public and it is disseminated to patients' groups, health professionals' societies, general public and media, making the plan also be known at European level.
- Measures are taken to ensure the sustainability, transfer and integration of the actions foreseen by the national plan or strategy into the general health system of the country
- The National Plan or Strategy has a duration of three – five years.
- The National Plan or Strategy is monitored at regular intervals using, as far as possible, EUROPLAN indicators. An intermediate deadline is established, after which, an evaluation process is undertaken and possible corrective measures are adopted.
- The implementation of the actions and their achievements are assessed.
- An external audit or evaluation of the National Plan or Strategy is carried out taking into account also patients' and citizens' views.

1.3. EUROPLAN indicators:

Actions	Indicators	Type	Actual state in Bulgaria
Development of regulations / laws	Existence of regulations /laws that support the creation and development of a RD plan	Process	The Bulgarian National Plan for Rare Diseases (NPRD) started on 1 January 2009 and will last for 5 years.
	National / regional	Process	NPRD works on national level.
Establishment of Coordination mechanisms	Existence of a coordination mechanism	Process	National Consulting Council on Rare Diseases (NCCRD) is established to the Ministry of Health (MoH). It consists of representatives of MoH, patients (National Alliance of People with Rare Diseases – NAPRD), medical professionals. It includes also specialist from the Ministry of Finances, Ministry of Labour and Social Policy, State Agency for Child Protection.
	Existence of an expert advisory committee	Process	NCCRD sits once monthly and supervises NPRD progress and implementation.
Establishment of an external evaluation of the plan/strategy procedure	Existence of an external evaluation body / procedure	Process	-
Degree of comprehensiveness	Number of priority areas included	Process	NPRD includes 9 priorities – creating a national registry, expansion of screening programs, introduction of new genetic tests, integrated approach, specialized training of medical professionals, establishment of reference centres for rare diseases, national awareness campaign on rare diseases, support and cooperation with patient associations, cooperation with other Member states of the European Union
Establishing of a budget for developing the plan/strategy	Budget of plan/strategy	Process	NPRD estimated budget is 11.3 millions euro. However, the assigned funds so far are much less.

Chapter 2:
ADEQUATE DEFINITION, CODIFICATION AND INVENTORYING OF RARE DISEASES

2.1. EU Council recommendation:

II. ADEQUATE DEFINITION, CODIFICATION AND INVENTORYING OF RARE DISEASES

- *Use for the purposes of Community-level policy work a common definition of rare disease as a disease affecting no more than 5 per 10 000 persons.*
- *Aim to ensure that rare diseases are adequately coded and traceable in all health information systems, encouraging an adequate recognition of the disease in the national healthcare and reimbursement systems based on the ICD while respecting national procedures.*
- *Contribute actively to the development of the EU easily accessible and dynamic inventory of rare diseases based on the Orphanet network and other existing networks as referred to in the Commission Communication on rare diseases.*
- *Consider supporting at all appropriate levels, including the Community level, on the one hand, specific disease information networks and, on the other hand, for epidemiological purposes, registries and databases, whilst being aware of an independent governance.*

2.2. EUROPLAN recommendations:

- The European definition of rare diseases is adopted in order to facilitate transnational cooperation and community level actions (e.g. collaboration in diagnosis and health care; registration activities).
- The use of a common EU inventory of rare diseases (Orphanet) is promoted in the national health care services and collaboration is carried out to keep it updated.
- Coding of rare diseases is promoted, encouraging their traceability in the national health system.
- Cross-referencing rare diseases is carried out across the different classification systems adopted in the country, ensuring coordination and coherence with European initiatives, such as the reference to the Orpha-code.
- The collaboration in the ICD10 revision process is ensured and ICD-11 is adopted as soon as it becomes available.
- Healthcare professionals are appropriately trained in coding rare diseases.
- Initiatives are promoted at national level for the integrated use of administrative, demographic and health care data sources to improve the management of rare diseases.
- National/regional registries of rare diseases are promoted and supported for public health purposes under the management of the health authorities or other stakeholders. Public/academia partnerships for the establishment and maintenance of registries are stimulated.
- Participation of existing registries and centres of expertise in European registries is fostered.
- Collaboration at European level is ensured for assessing and agreeing on ethical and legal issues (ownership, intellectual property, personal data protection, etc.), scope and use of European/national registries and funding aspects, which can hamper the functioning of many European registries.
- Instruments are identified for combining EU and national funding for registries.

2.3. EUROPLAN indicators:

Actions	Indicators	Type	Actual state in Bulgaria
To officially adopt the EC RD definition (no more than 5 cases /10,000 inhabitants	Adoption of the EC RD definition	Process	NPRD states that the EC RD should be used.
To include the best Rare Diseases	Type of classification used by the health care system	Process	ICD-10

classification currently existing into the public health care related services	Developing policies for recognising RD by the care information systems	Process	Absent and not applicable at the moment. This reflects on the overall poor visibility and awareness for rare diseases in all fields - healthcare planning and allocation of funds, education and training of specialists, public awareness and solidarity.
Defining a surveillance system based on a patient outcomes registry	Registering activity	Process	There are many registries for local use, but they that are not standardized and not related to each other. Rare tumors are included in the National Cancer Registry.
	Number of diseases included	Outcomes	Outside the National Cancer Registry and individual clinical records, there are currently only two specialized epidemiological registry for rare diseases (thalassemia and chronic myeloid leukemia). Both are created and maintained by the Bulgarian Association for Promotion of Education and Science (BAPES).

Chapter 3:
RESEARCH ON RARE DISEASES

3.1. EU Council recommendation:

IV. RESEARCH ON RARE DISEASES

- *Identify ongoing research and research resources in the national and Community frameworks in order to establish the state of the art, assess the research landscape in the area of rare diseases, and improve the coordination of Community, national and regional programmes for rare diseases research.*
- *Identify needs and priorities for basic, clinical, translational and social research in the field of rare diseases and modes of fostering them, and promote interdisciplinary co-operative approaches to be complementarily addressed through national and Community programmes.*
- *Foster the participation of national researchers in research projects on rare diseases funded at all appropriate levels, including the Community level.*
- *Include in their plans or strategies provisions aimed at fostering research in the field of rare diseases.*
- *Facilitate, together with the Commission, the development of research cooperation with third countries active in research on rare diseases and more generally with regard to the exchange of information and the sharing of expertise.*

3.2. EUROPLAN recommendations:

- Specific national research programs for rare diseases (basic, translational, clinical, public health and social research) are established and supported with dedicated funds, preferably for a long period.
- Research projects on rare diseases should be made identifiable and traceable within broader national research programs.
- Specific provisions are included in the national plans or strategies to promote appropriate agreements between Health, Research and Social Ministries/Departments for improving knowledge on different aspects of rare diseases.
- National networks are promoted to foster research on rare diseases. Special attention is given to translational research in order to facilitate the application of new knowledge into rare disease treatment. Compilation and updating of a directory of teams carrying out research on rare diseases should be endorsed when feasible.
- Proper initiatives are developed to foster participation in cooperative international research initiatives on rare diseases, including the EU framework programme and E-RARE.
- Specific technological platforms and infrastructures for rare disease research are established and supported.
- Multi-centre national and trans-national studies are promoted, in order to reach a critical mass of patients for clinical trials and to exploit international expertise.
- Instruments and measures (e.g. centres) are set up to support clinical research on rare diseases.
- Specific programs are launched for funding and/or recruitment of young scientists on rare diseases research projects.

3.3. EUROPLAN indicators

Actions	Indicators	Type	Actual state in Bulgaria
Building a research programme for rare diseases	Existing of RD National / Regional research programmes	Process	NPRD actually do not envisage any official policy to stimulate research on rare diseases. It will only encourage the establishment of partnerships,

			including the future centers of expertise, which also may seek different means of financing.
	RD research programme monitoring	Process	No actions have been taken.
	Number of RD research projects approved by year	Outcomes	-
	Clinical trials funded by public bodies	Outcomes	-
	E-RARE joining	Process	-
	Including public health and social research, in the field of rare diseases	Process	-
	Research platforms and other infrastructures are also funded by the research programme	Process	-
Existence of national policy in support of the recruitment of young scientists / researchers specifically for rare diseases	Number of young scientists recruited every year to work specifically on rare diseases	Process	-
Allocate funds for the RD research programme	There are specific public funds allocated for RD research	Process	Under discussion
	Funds specifically allocated for RD research actions /projects per year	Outcomes	-

Chapter 4:
CENTRES OF EXPERTISE AND REFERENCE NETWORKS. ORPHAN DRUGS AND PROVISION OF TREATMENTS

4.1. EU Council recommendation:

IV. CENTRES OF EXPERTISE AND EUROPEAN REFERENCE NETWORKS FOR RARE DISEASES.

- *Identify appropriate centres of expertise throughout their national territory by the end of 2013, and consider supporting their creation.*
- *Foster the participation of centres of expertise in European reference networks respecting the national competences and rules with regard to their authorisation or recognition.*
- *Organise healthcare pathways for patients suffering from rare diseases through the establishment of cooperation with relevant experts and exchange of professionals and expertise within the country or from abroad when necessary.*
- *Support the use of information and communication technologies such as telemedicine where it is necessary to ensure distant access to the specific healthcare needed.*
- *Include, in their plans or strategies, the necessary conditions for the diffusion and mobility of expertise and knowledge in order to facilitate the treatment of patients in their proximity.*
- *Encourage centres of expertise to be based on a multidisciplinary approach to care when addressing rare diseases.*

4.2. EUROPLAN recommendations:

- Transparent mechanisms of designation of centres of expertise are established ensuring their quality, efficiency and long term sustainability.
- Healthcare pathways are defined and adopted, based on best practices and expertise at national and international level.
- A national directory of Centres of expertise operating in the national territory is compiled, kept updated and published at national level as well as in a common European Inventory (Orphanet).
- Centres able to provide quality diagnosis and care are identified in neighbouring or other countries, where patients or biological samples can be referred to, and networking is promoted.
- Travelling of biological samples, radiologic images, other diagnostic materials, and e-tools for tele-expertise is supported.
- A national framework is ensured on rare diseases screening options and policies.
- Proper performance of newborn screenings prescribed in the country is monitored with appropriate indicators.
- Accessibility to genetic counselling is promoted.
- The quality of genetic testing and other diagnostic tests is ensured, including participation in external quality control schemes at national and international level.
- Collaboration with the EU inventory of the national provision of testing for rare diseases (genetic, biochemical, enzymatic, tissue/cell identification, clinical) published both at national level and on Orpha.net is ensured.
- Centres of expertise provide proper training to paramedical specialists; paramedical good practices are coordinated, in order to serve the specific rehabilitation needs of rare diseases patients.
- An ad hoc coding must be incorporated into the national health system to recognise and appropriately resource the special rehabilitation treatments necessary for people affected by rare diseases.

4.3. EUROPLAN indicators:

Actions	Indicators	Type	Actual state in Bulgaria
Improve the quality of healthcare by defining: appropriate centres with experience on RD; pathways that reduce the diagnosis delay and facilitate the best care and treatments	Existence of a policy for establishing centres of expertise (CoE) at the national/regional level	Process	The requirements and criteria for CoE designation are under discussion. It is expected that CoEs would be located within university hospitals, having in mind better facilities and human resources.
	Number of CoE adhering to the policy defined in the country	Outcomes	0
	Groups of rare diseases followed up in CoE	Outcomes	-
	CoE adhering to the standards defined by the Council Recommendations - paragraph d) of preamble	Outcomes	-
	Participation of national or regional CoE into European reference networks	Outcomes	-
Develop Screening Policies	Number of diseases included in the neonatal screening programme	Outcomes	3 (phenylketonuria, congenital hypothyroidism, congenital adrenal hyperplasia)
	Number of diseases included in the neonatal screening programme properly assessed	Outcomes	3 of 3
Ensure quality of RD diagnosis laboratory	Existence of a public directory/ies of both genetic tests on rare diseases	Process	Under discussion
	Proportion of laboratories having at least one diagnostic test validated by an external quality control	Outcomes	3 of 6
Ensure the mechanism that facilitates ODD access and the reimbursement of their cost to patients after they got the Market Authorisation by EMA	Number of ODD market authorisations by EMA and placed in the market in the country	Outcomes	18 of 60* by June 2010
	Time between the date of a ODD market authorisation by EMA and its actual date of placement in the market for the country	Outcomes	By June 2010 60 medicinal products have OD status by EMA. 18 of them are included in the new positive list of drugs (PLD), Annex 3 and 4, but only 11 of them are reimbursed under MoH Regulation 34. 44 months is the average delay from the EMA market authorization to the inclusion PLD.
	Time from the placement in the market in the country to the positive decision for reimbursement by public funds	Outcomes	
	Number of ODD reimbursed 100%	Outcomes	Only 11. Both patients and medical professionals agree that the actual legal framework is not adequate to the OD

			access.
To develop mechanisms to accelerate ODD availability	Existence of a governmental programme for compassionate use for rare diseases	Outcomes	Under discussion

Chapter 5:
PATIENT EMPOWERMENT

5.1. EU Council recommendation:

VI. EMPOWERMENT OF PATIENT ORGANISATIONS

- *Consult patients and patients' representatives on the policies in the field of rare diseases and facilitate patient access to updated information on rare diseases.*
- *Promote the activities performed by patient organisations, such as awareness-raising, capacity-building and training, exchange of information and best practices, networking and outreach to very isolated patients.*

5.2. EUROPLAN recommendations:

- Advocacy of patients' needs by patients' associations is recognised as an important element in defining policies on rare diseases; the organisation of a national umbrella organisation that represents the interests of all rare diseases patients is encouraged.
- The patients' organisations are involved in decisions making processes in the field of rare diseases.
- Valid information on rare diseases is produced and made available at national level in a format adapted to the needs of patients and their families.
- National information of interest to patients is communicated to EURORDIS for publication in its website.
- Specialised social services are supported for people living with a chronically debilitating rare disease and their family carers.
- Specialised social services are established to facilitate integration of patients at schools and workplaces.
- A directory of centres providing specialised social services, including those offered by patients' associations, is compiled, kept updated and communicated to national, regional and patients' websites and included in the Rapsody network.
- Interactive information and support services for patients are promoted (such as help lines, e-tools etc).
- Information and education material is developed for specific professional groups dealing with rare diseases patients (e.g. teachers, social workers, etc.).
- The activities aiming at patients' empowerment carried out by patients' associations are facilitated.

5.3. EUROPLAN indicators:

Actions	Indicators	Type	Actual state in Bulgaria
Promoting the existence of a RD patients' organisation that represents all RD patients' associations	Number of umbrella organisations specific on rare diseases	Process	The National Alliance of People with Rare Diseases is the only global representative of the rare diseases patients on national level.
	Having a directory of RD patients' organisations	Process	There is an unofficial one on the Information Centre for Rare Diseases and Orphan Drugs (ICRDOD) website.
	Number of patients' associations	Резултати	21
	Number of diseases covered by patients' associations	Резултати	More than 30
Patients' organisations involvement in decisions affecting RD	Permanent and official patients' representatives in plan development, monitoring and assessment	Process	A NAPRD representative is a member of NCCRD and takes part in its sessions.
	Participation of patients' organisations in the development of RD research strategies	Process	No.
	Participation of patients	Process	Patients' opinion should be counted when the

	organisations in the RD centres of expertise designation and evaluation		CoE work is being evaluated.
Support the activities performed by including patient organisations, such as: Awareness raising Capacity building and training Exchange of information and best practices Networking Outreach to very isolated patients	Resource (funding) provided for supporting the activities performed by patient organisations	Outcomes	-
	Support to sustainable activities to empower patients	Outcomes	Rare diseases patient association in Bulgaria hope that the NPRD will help them in their mission to raise the public awareness about the rare diseases topics. Already some of the Bulgarian rare diseases patient associations have strong connections with their European and international equivalents, they take part in joint projects and events. NAPRD is a member and works closely with EURORDIS.
Building and supporting the existence of comprehensive help line for patients	Availability of Help line for RD	Process	ICRDOD help line is the only universal rare diseases help line in Bulgaria. It is also cited on the European Commission website as a quality and reliable source of information about rare disease. However, recently many patient associations have been starting and maintaining different channels of communications (mostly websites and forums), but they are focused on a single specific rare condition.
Compensating disabilities caused by rare diseases	Existence of official programs supporting patients and families with disabilities	Process	Existing, clearly stated, partly implemented and enforced.
	Existence of an official directory of social resources for patients with disabilities	Process	There is such a directory in the framework of the programmes for integration of people with disabilities, but it is not particularly focused on rare diseases.
Supporting rehabilitation programmes	Existence of programmes to support rehabilitation of RD patients	Process	In Bulgaria, there are still no specialized programmes for people with rare diseases. So, these patients are forced to seek alternative in the existing general schemes for rehabilitation and integration of people with disabilities, which are in fact outdated, far from best quality and do not meet European standards and recommendations in this area. They are unevenly distributed across the country and public awareness of them is low. Additionally, patients with rare diseases not always have access to them, because of obstructions from TEMC (territorial expert medical commission), whose legislation is not adapted to the specifics of the rare diseases. At this stage we can talk about the lack of integrative approach and specialized programs for physical and social rehabilitation of people with rare diseases and their families.
Supporting social services aimed at rare disease	Existence of national schemes promoting access of RD patients and their families to	Process	This is one of the main priorities of the NPRD, but at this stage no work has been done on it yet. Most rare diseases patients believe that

patients and their families	respite care services		specialized services for them should not be separated or be in opposition to current programs for people with disabilities.
	Existence of public schemes supporting therapeutic recreational programmes	Process	There are several ones which are part of the national strategies for integration and rehabilitation of people with disabilities, but they are not rare diseases specified.
	Existence of programmes to support integration of RD patients in their daily life	Process	-

SUMMARY

People with rare diseases are anywhere around us. Every day, they and their families face in discrimination, they lack access to adequate treatment, they have been refused opportunities for rehabilitation and integration. National Program for Rare Diseases was a glimmer of hope for these people, but its implementation is constantly held back by bureaucratic obstacles, lack of funding (although allocated such one) and not least, personal ambitions and interests.

All that was found during the Bulgarian EUROPLAN National Conference for Rare Diseases, where the participants agreed on the following suggestions and guidelines for action at national level:

1. Full support of the priorities set out in the Recommendation on rare diseases, adopted on 9 June 2009 by the Council of Ministers of the European Union;
2. Ensuring the National programme for rare diseases with the all the funds, required and assigned for its implementation;
3. Urgent need for legislative initiatives to protect the rights of people with rare diseases and ensure adequate prevention, treatment, rehabilitation and social care;
4. Stimulating the creation of epidemiological registries for rare diseases in Bulgaria;
5. Integrative approach to people with rare diseases and their families;
6. Organizing and conducting a public campaign aimed at creating a fund to stimulate research on rare diseases in Bulgaria.

People with rare diseases and their families continue to hope that the upcoming healthcare reforms will improve their capacity for integration and equal access to adequate and quality medical cares and follow-up.