NATIONAL ALLIANCE OF PEOPLE WITH RARE DISEASES (NAPRD)
Address: 59 Dragan Tsankov blvd., 1000 Sofia, Bulgaria
e-mail: *tomov@raredis.org* ; phone *+359-888-323748*

BULGARIAN ASSOCIATION FOR PROMOTION OF EDUCATION AND SCIENCE (BAPES)
Address: 4 Bratia Sveshtarovi str., 4017 Plovdiv, Bulgaria
e-mail: *info@raredis.org* ; phone *+359-897-858870*

BULGARIAN EUROPLAN NATIONAL RARE DISEASES CONFERENCE
FINAL REPORT

I. General information

Country	**BULGARIA**
Date & place of the National Conference	**28-30 May 2010, Congress Centre of Novotel, Plovdiv**
Website	**www.conf2010.raredis.org**
Organisers	**National Alliance of People with Rare Diseases (NAPRD)** **Bulgarian Association for Promotion of Education and Science (BAPES)**
Members of the Steering Committee	**Mr. Vladimir Tomov, President of NAPRD** **Prof. Rumen Stefanov, President of BAPES** **Prof. Radka Tincheva, Chair of the National Consulting Committee for Rare Diseases (NCCRD) to the MoH** **Dr. Stamen Popov, Association of the Research-Based Pharmaceutical Manifacturers in Bulgaria (ARPharM)** **Dr. Ivelina Yordanova, Medical University of Pleven/ DEBRA Bulgaria** **Mrs. Dorica Dan, EURORDIS**
Names and list of Workshops	**Panel Session 1 – Rare diseases policies and recommendations for actions** **Panel Session 2 – Reporting proposals from the conference workshops and seminars; adopting conference memorandum** **Workshop 1 – Methodology and governance of the Bulgarian national plan for rare diseases** **Workshop 2 – Patient empowerment and specialised services** **Workshop 3 – Definition, codification and inventorying of rare diseases** **Workshop 4 – Research on rare diseases** **Workshop 5 – Rare diseases in the focus of hematology** **Workshop 6 – Rare diseases in the focus of**

	physical medicine and rehabilitation Workshop 7 – Rare diseases in the focus of immunnology Workshop 8 – Rare diseases in the focus of medical genetics and pediatrics Workshop 9 – Standards of care for rare diseases. Centres of Expertise Seminar 1 – First national training seminar for cystic fibrosis Seminar 2 – Second national seminar for thalassemia Seminar 3 – First national meeting of the primary pulmonary hypertension patients in Bulgaria Seminar 4 – Setup meeting of arthrogryposis patient association
Chairs and Rapporteurs of Workshops	PS 1 – Vladimir Tomov, Rumen Stefanov (co-chairs) PS 2 – Vladimir Tomov, Rumen Stefanov, Radostina Simeonova (co-chairs) WG1 – Rumen Stefanov (chair and rapporteur) WG2 – Vladimir Tomov (co-chair and rapporteur), Veska Sabeva (co-chair) WG3 – Ralitsa Yordanova (co-chair and rapporteur), Georgi Iskrov (co-chair) WG4 – Rumen Stefanov, Maria Simeonova (co-chairs), Ivelina Yordanova (rapporteur) WG5 – Valeria Kaleva (co-chair and rapporteur), Yulian Raynov, Stefan Goranov, Petrana Chakarova (co-chairs) WG6 – Troycho Troev, Ivet Koleva (co-chairs), Radostina Simeonova (co-chair and rapporteur) WG7 – Elisaveta Naumova, Mariana Murdjeva, Fani Martinova (co-chairs), Dora Popova (co-chair and rapporteur) WG8 – Maria Simeonova (co-chair), Dimitrina Konstantinova (co-chair and rapporteur) WG9 – Radostina Simeonova (co-chair and rapporteur), Stamen Popov (co-chair)
Attachments (programme, list of participants, etc.)	1. List of participants 2. Conference memorandum

II. General Overview

In recent years, rare diseases have been established as a **priority area** in public health of the European Union. On 27 November 2008, the Bulgarian Council of Ministers adopted the **National Programme for rare diseases (2009 – 2013)** in order to improve prevention, diagnosis, treatment and rehabilitation of people with rare diseases in Bulgaria.

The Bulgarian EUROPLAN National Conference for Rare Diseases and Orphan Drugs was organized in the framework of the EUROPLAN Project, funded by the European Commission and aimed to provide information regarding the various steps in developing a strategic plan, to identify best practices and to exchange models and data on effective strategies for rare diseases. In this context, the event aimed to:

- **present the National Programme** for rare diseases, its specific tasks and opportunities it offers to the medical professionals, patients and society as a whole;
- **make the Bulgarian rare diseases stakeholders familiar with the EU Council Recommendation on rare diseases and EUROPLAN recommendations for actions,** to present and gather proposal on them, to discuss their transferability and adaptation in Bulgaria;
- **determine on expert level the priority rare diseases in Bulgaria** and to draft proposals for actions to improve prevention, diagnosis, treatment and social integration of patients, further training of physicians, all in **collaboration with national consultants and medical scientific societies** in the country.

The overall success of the conference was guaranteed by the good discussions that took place. That was one of the most important things - to listen to the opinion of the stakeholders - and the patients have been very active in that field. People were intersted in national developments, but also in European level actions, recommendations and practices.

The conferece highlights:

- Through the National Alliance, patients now can stand up and ask for their rights. Most of the rare diseases patient associations in Bulgaria are active stakeholders and the big number of proposals, coming from them during the conference means only that they are now determined as never before to make their voice be heard by the rare diseases policy makers. Patients and medical professionals initiated extensive discussions about rehabilitation and recreation of people with rare diseases.

- Very high interest from the medical community and especially hematologists, immunologists, geneticists, pedeatricians, etc. who seemed to be really motivated to cooperate with the other stakeholders in order to make decent progress in the rare diseases field. National plan, Centres of expertise (CoE) and rare diseases research were one of the most discussed topics among them. Most of the medical scientific societies expressed their willingness to participate actively in the implementation of the National Plan's priorities, to help establish the National registry of patients with rare diseases and to create and update guidelines and protocols for rare diseases management.

- Rare diseases patient associations proved to be capable to use their European partnerships in order to achieve significant results on local level. Bulgarian thalassemia major and cystic fubrosis patient associations put in practice their good relations and partnerships with their umbrella organizations (TIF and CF Europe respectively) and managed to hold patient seminars with some of the EU leading experts from Belgium, Italy and Greece attending and presenting the most advanced practices of integrative approach to these two specific conditions. The fact that both these events were attended by more than

100 participants (patients and medical professionals) each means a lot. Additionally, two other seminars took place during the conference – the primary pulmonary hypertension annual patient meeting and a setup meeting of the arthrogryposis patient association. Patients from HAE and primary immunodeficiencies associations have been also very active in the immunology workshop. Here is an interesting fact to mention – one of the main topics of the immunology meeting was the management of primary immunodeficiencies and particularly the missing clinical pathway for them within the National Health Insurance Fund. All agreed that actions should be made as soon as possible in order to give the patients access to treatment. And just 10 days after it, this pathway was actually approved by the National Health Insurence Fund. That's a very good example of collaboration among different rare diaseases stakeholders.

- Very active participation at the conference organization by the medical students. The conference co-organizers have a long history of good partnership with Association of medical students in Bulgaria and this event was not an exception. More than 20 students attended the meeting and it's an excellent sign that the future health professionals will be well introduced in the rare diseases' topics. It's well to mention that many students took part at different workshops' preparation and some very good ideas for common project of patients and students came during the conference.

- The official patron of the conference was the Bulgaria's First lady – Mrs. Zorka Parvanova, who sent a greeting message to the participants, expressing once more her support for the rare diseases cause and gratitude to the NAPRD and ICRDOD, as leading rare diseases stakeholders in Bulgaria. The event was also supported by the National consulting council for rare disease to MoH, University hospital "St. George" and the Bulgarian Scientific society for clinical and transfusional hematology.

- ZDRAVE.net was the official media partner of the event. This webportal is one of Bulgaria's leading healthcare and health system information services. ZDRAVE.net published a series of rare diseases articles before and after the conference which helped a lot to increase the number of conference participants and to promote the event's objectives to wider public.

III. Main Report

Theme 1 - Methodology and Governance of a National Plan / Strategy (NP)

Sub-Themes

1. Mapping exercise before developing a National Plan
2. Development and structure of a National Plan / Strategy
3. Governance of a National Plan
4. Monitoring the National Plan
5. Sustainability of the National Plan

Workshop 1

Chair: Prof. Rumen Stefanov (BAPES; Medical University of Plovdiv; NCCRD)

Rapporteur: Prof. Rumen Stefanov

Date, time and place – 16:30-18:30, 29 May 2010, Paris Hall of Congress Centre of Novotel-Plovdiv

Audience: 23 persons

EUROPLAN Indicators:

ACTIONS	INDICATORS	TYPE	ANSWERS
Development of Regulations/Laws	Existence of Regulations/laws that support the creation and development of a RD plan	Process	Existing, clearly stated and substantially implemented and enforced
	National/regional (percentage of regions)	Process	100%
Establishment of Coordination mechanisms	Existence of a coordination mechanism	Process	Existing, clearly stated and substantially implemented and enforced
	Existence of an expert advisory committee	Process	Existing and meets regularly
Establishment of an external evaluation of the plan/strategy Procedure	Existence of an external evaluation body/procedure	Process	n/a
Degree of comprehensiveness	Number of priority areas included	Process	9
Establishing of a budget for developing the plan/strategy	Budget of plan/strategy	Process	11.3 millions euro

Proposals, arising from the held discussion

1. **Is there an awareness of the situation of RD in your country (epidemiologic figures, dimension of the problem)?**

All the existing data is partial, there is not an integrative framework of the RDs. However, thanks to the activities of the National Alliance of People with Rare Diseases (NAPRD), the Bulgarian Association for Promotion of Education and Science (BAPES) and the newly established National Plan for Rare Diseases (2009-2013) there have been significant progress in this area for the past 5 years.

2. **Is an inventory being made, or a report, of the existing resources and actions on RD (or of which RD patients can benefit) in the national health care and social system?**

Patients have been particularly discontent with the lack of inventory of rehabilitation and integration possibilities. Additionally, the available ones are out-of-date and do not answer the basic needs of the RDs patients. Medical professionals are more interested in getting information about funding of RDs activities and sharing best practices with foreign experts.

3. **Are the unmet needs of RD patients being evaluated?**

The fundamental need is this of the therapeutic treatment, or at lest for these conditions for which there are suitable drug treatment. But with the prospects of the NPRD people became more aware of the rehabilitation possibilities which are inseparable part of the RDs patient daily life. People complained about the lack of it or the inadequate one.

4. **Is there in your country a legal/policy framework in the form of a national plan or strategy created to address the health care and social needs of patients with RD with specific actions? What are the steps to be taken?**

Yes, NPRD.

5. **Is the NP integrated and comprehensive so to respond to all patients' needs?**

Yes, it consists of 9 basic priorities, covering the whole specter of RDs topics – from prevention and diagnostics to international co-operation and patient empowerment.

6. **Is the NP created in the form of a written document with a clear structure?**

Yes. See question 4 above.

7. **What general and specific objectives or priority areas have been / can be identified in your country?**

The NPRD 9 priorities are:

1. Collecting epidemiological data for the rare diseases in Bulgaria by creation of a national register.
2. Improvement of the prevention of the genetic rare diseases by enlarging the current screening programmes.
3. Improvement of the prevention and diagnostics of the genetic rare diseases by introducing new genetic tests, decentralisation of the laboratory activities and easier access to medico-genetic counseling.
4. Integrative approach to the prevention, diagnostics, medical treatment and social integration of patients and their families.
5. Promotion of the professional qualification of medical specialists in the field of early diagnostics and prevention of rare diseases.
6. Feasibility study on the necessity, possibility and criteria for creation of a referent centre for rare diseases of functional type.
7. Organising a national campaign for informing the society about rare diseases and their prevention.
8. Support and collaboration with NGOs and patient associations for rare diseases.
9. Collaboration with the other EU members.

8. **Are the specific actions envisaged in the NP accompanied by clear deliverables and measurable results?**

Yes, but these indicators are not included in the public version of the programme.

9. **Is there a timeline for the achievement of priority actions with specific deliverables?**

Yes, it is published on the MoH's website.

10. **Is there a Steering Committee (SC), or coordination committee, governing panel... governing the implementation of the Plan?**

Yes, the National Consulting Committee for Rare Diseases (NCCRD) to the Ministry of Health (MoH).

11. **Do all stakeholders participate in the governance of the NP – healthcare authorities, patients, healthcare professionals, academics, representatives of the industry, etc.? Do these stakeholders cover all areas of expertise relevant to the NP, such as pharmacology, regulatory, clinical, health and social services, epidemiology, administrative policies, etc.**

NCCRD consists basically of all the stakeholders – healthcare authorities, patients, medical professionals, academics. Additionally, there are representatives from the Ministry of Finances, Ministry of Labour and Social Affairs, State Agency for Child Protection. There are no representatives from the industry.

12. **Is the participation of patients envisaged to all phases of the NP so to ensure that patients are actors in the decision on health care measures directed to them?**

Yes, there is a patient representative, Mr. Vladimir Tomov. He is a member of NCCRD and participates at NPRD all stages of work.

13. **Does the SC meet regularly?**

Approximately once a month.

14. **Does the SC write a status for their activities and the responsibilities of its members?**

Yes, there is an internal MoH order about the aim and responses of NCCRD.

15. **Does the SC write a regular (yearly) report on the achievement of the objectives of the NP and deliverables?**

Yes, it is delivered to the Minister of health.

16. **Is the NP made public in the general content and specific actions?**

Yes and it contains raising-awareness and NPRD presentation public campaigns, scheduled as sub-objectives. That is, of course, to assure that the society is aware of its existence, goals and actions and the RDs topics as well.

17. **Is there a monitoring system for the NP?**

NCCRD is responsible for governing, monitoring and implementing NPRD and it is directly subordinate to the MoH. There is an internal set of monitoring procedures at the MoH, concerning financial issues and implementation reports.

18. **What type of indicators is used to monitor its implementation? Are the EUROPLAN indicators used as a basis for monitoring and evaluating the actions of the NP?**

Both processes and outcomes, they are pretty much the same as EUROPLAN ones. The NPRD was written and approved before the EUROPLAN initiation.

19. **Is the evaluation of the Plan ensured by an external body, i.e. different from the SC?**

No. It's the NCCRD which is evaluating the NPRD progress.

20. **Does the evaluation include also the collection of opinions and satisfaction surveys addressed to patients?**

No. But some proposals evolved to use the NAPRD network of associations in order to get feedback from the patients about the programme's results. That will be considered in near future.

21. **Is there a specific budget attached to the NP? Does it ensure the long term sustainability of its actions?**

Yes, 11.3 millions euro.

22. **What are the main sources of funding of the National Plan?**

The MoH budget is the only source of funding.

23. Are there specific budget provisions accompanying specific actions in certain priority areas e.g. orphan drugs, CoE, diagnosis, research, etc.?

There is budget within the NPRD, planned to support the CoE.

<u>Conclusion</u>

The overall assessment of this workshop varied from very good to excellent. The vast majority of the participants were medical professionals and academic representatives. The coming proposals can be summarized into several groups:

1. full support for the NPRD implementation (including guaranteed funding for its priorities and their realization)
2. more information about the NPRD and RDs services as whole for both patients and healthcare professionals
3. co-operation and partnerships within EU programmes and initiatives in order to implement the best RDs practices

Theme 2 - Definition, codification and inventorying of RD

Sub-Themes

1. Definition of RD
2. Classification and traceability of RDs in the national health system
3. Inventories, registries and lists

Information and training

4. How to improve information on available care for RDs in general, for different audiences
5. How to improve access to quality information on RDs
6. How to ensure adequate training of healthcare professionals on RDs

Workshop 3

Chairs: Dr. Ralitsa Yordanova (BAPES-ICRDOD, Orphanet Bulgaria)
Mr. Georgi Iskrov (BAPES-ICRDOD)

Rapporteur: Dr. Ralitsa Yordanova

Date, time and place – 9:00-10:30, 29 May 2010, Evridika Hall of Congress Centre of Novotel-Plovdiv

Audience: 12 persons

EUROPLAN Indicators

ACTIONS	INDICATORS	TYPE	ANSWERS
To officially adopt the EC RD definition (no more than 5 cases/10,000 inhabitants	Adoption of the EC RD definition	Process	Yes
To include the best Rare Diseases classification currently existing into the public health care related services	Type of classification used by the health care system	Process	ICD-10
	Developing policies for recognising RD by the care information systems	Process	Not existing, not clearly stated
	Developing policies for recognising RD by the care information systems	Process	Not existing, not clearly stated
Defining a surveillance system based on a patient outcomes registry	Registering activity	Process	Multiple RD registries, not standardised
	Number of diseases included	Outcomes	More than 20

Proposals, arising from the held discussion

1. **Is the EU official definition (RD are those affecting up to 5 out of 10 000 person) used in your country?**

Yes, it has been officially adopted through the National Programme for Rare Diseases (2009-2013) (NPRD).

2. **Are there alternative or more specific definitions used instead or in addition?**

No.

3. **What classification system is used in your country?**

ICD10

4. **For which purpose is the classification system used, e.g. surveillance, reimbursement, provision of social support, etc.**

It is used at all the area of the RD management (including reimbursement, social services, etc.).

5. **Is your country prepared to adopt the WHO-led system, the ICD-11, recommended by the EU in the Council Recommendation on RD, when ready (2014)?**

Yes.

6. **What level of awareness and knowledge do healthcare professionals have of the RD classification and codification? What can be done to improve it?**

All the participants expressed their high level of satisfaction about the medical specialists' awareness and professional knowledge about the RDs. But on the other side, most of the patients deeply disagreed on GPs' general education when talking about RDs patients and forwarding them to medical specialists and rehabilitation services. The reasons for that are many, the most important of which are lack of adequate training about the RDs, bad management of the GPs work and lack of motivation for further training.

Parents of children with RDs proposed to organize through the National Alliance of People with Rare Diseases (NAPRD) and NPRD a series of meetings, attended by patients, GPs and medical specialists in order to make the GPs more familiar with the general prospective of RDs and introduce to them some of the more “frequent” RDs, so they could be more prepared to deal with them. It was proposed to make these workshops on a regional level rather than national and to include patients and their families in order to create a more touchable experience.

7. **Are there official lists of RD in your country? Is there an official governmental RD registry? And/or specific RD databases e.g. held by Centres of Expertise? Are there RD surveillance projects or programmes (e.g. sentinel programmes, surveys)?**

There is not an official RDs list in Bulgaria, currently a preliminary version is being prepared.

There is an official RDs registry neither. Its set-up should start shortly after the adoption of the official RDs list.

However, there are existing numerous specific rare diseases registries, ran by the specialized clinics. But they are not standardized and cover only the areas of interest of the particular institution which is managing them. Through the NPRD there will be an unified and standardised National RDs registry which of course will be initiated on the basis of the existing ones.

Currently, one of the conference co-organizers – the Bulgarian Association for Promotion of Education and Science has been creating and managing two RDs registries on a national level – thalassemia major and chronic myeloid leukemia. The outcomes of these projects are available online. Additionally, the gained experience is being used to start other RDs registries. The conference has been particularly productive on this matter with both clinicians and patients showing huge interest in establishing registries for their diseases.

8. **What kind of initiatives should be taken or reinforced in your country?**

See the other questions.

9. **Do these registries and programmes receive government support?**

National RDs registry is planned to be started within the NPRD and will be funded by MoH.

10. **How to ensure, through appropriate funding mechanisms, the long-term sustainability of registries and databases?**

There are already two successful projects in this area. Their model has shown some interesting aspects – to be a successful project (that includes sustainable as well), it must cover all the stakeholders – patients, clinicians, researchers, policy makers and industry representatives. This not only assures the multi-prospectiveness, but also multiplies the possibilities for different types of funding. Additionally, there must be an accent on the registries outcomes – their value can be used anywhere from governmental activities planning to scientific research and international co-operation.

11. **Does your country participate to the development of a EU inventory of RD as recommended in the Council Recommendations on RD?**

Yes, through participation in various EU projects and programmes.

12. **How are these information sources and initiatives publicised?**

Currently, they are publicised only by the RDs stakeholders. All the participants agreed that there is not enough media attention to the RDs topics, even there is negligence towards them and this really doesn't help to raise the awareness among the society as whole.

13. **How to make sure that they reach out to the target audiences? And to the wider public?**

The lack of communication with wider public is a basic issue. Here, two different ways of proposals evolved. Medical specialists said that step-by-step method is the best way of solving that tough situation. The initiatives should start on small targeted audiences and gradually to enrol others. Patients insisted that they have waited too much and some radical changes should be undertaken. Most of them thought that only big protest activities could capture the general public attention and through this the media focus as well. However, all agreed that the "human face" of the RDs problems should be used – all the awareness and publicity activities should focus on real persons with real problems rather than statistics and medical information. There have been proposal to set billboards on high-crowded spaces such as airports, train stations, etc. in order to attract the public attention.

14. **What are the existing information sources in the country? Are they of good quality? Do they receive public funding or Patients Org. funding?**

BAPES starting project was the Information Centre for Rare Diseases and Orphan Drugs (ICRDOD), a free information and education services, dedicated to patients, their families, medical professionals and students. Its web portal has been providing RDs information since 2004 and has been listed in the DG SANCO website as a reliable and quality source of information. It does not receive public or patients organizations funding.

Recently, some of the patients associations have started maintaining their own websites and trying to put the information about their specific condition on it. However, its number is still small and not of high quality.

15. **Is there a national official website for RD in the country?**

No.

16. **Are there help lines for both patients and healthcare professionals? Are they known to the public?**

Yes, ICRDOD maintains a help line for both patients and healthcare professionals. It has been established in 2004 and the number of daily requests has been steadily growing since then. It is the only RDs-orientated help service in Bulgaria. Other patients associations have been developing recently help lines too, but they provide only general information and do not have the necessary RDs background.

Patient representatives proposed to established online expert communities for particular RDs which can be used by both patients and medical professionals. They even cited some tryouts but complained from the lack of interest and motivation from the medical community to establish such kind of directories.

17. **Are there initiatives of centres of expertise and/or patient organisations or programme to stimulate the development of information and educational material for patients or specific publics (teacher, social workers, etc.)?**

Actually, it seemed that patients are more motivated in this than the other stakeholders. Some of them told about their personal experience of co-operation with European patients organizations and Bulgarian medical universities in order to adapt the existing RDs materials and to create new in Bulgarian, so more targeted groups can be reached, such as school teacher, social workers, governmental institutions. That showed once more that only when the different stakeholders act as a group, there can be decent results.

18. **Are existing resources at EU level, Orphanet and Eurordis used: (information on diseases, specialised centres and patient groups, ongoing research projects, clinical trials)?**

Yes, all the EU-level RDs projects and initiatives have been presented in Bulgaria by the NAPRD and BAPES work. Moreover, patient and medical professionals have been motivated to seek a personal contact with common European stakeholders in order to gain experience and to participate at these initiatives.

19. **Are there initiatives to raise awareness on RD such as a RD Day?**

Yes, NAPRD and BAPES have been active participants at RD Day since its very beginning. Since 2008 the scope of the RD Day events in Bulgaria has increased, more different stakeholders have been attracted. Patients deeply agreed on that matter, saying it is a very important activity to catch the public attention on RDs and to establish partnerships with different institutions such as municipality authorities, schools and cultural organizations.

20. **How to ensure that existing and validated international guidelines are used at national level to guide diagnosis and treatment of RD?**

Proposals for more patient control in this area emerged here. Despite the role of the society and NGOs being recognized by the health authorities, their active inclusion has not been a fact yet.

21. **What mechanisms can be put in place to support the exchange of expertise at EU level and the adequate training for all healthcare professionals?**

Patients proposed to use the “personal contact” experience when training young healthcare professionals and students. They said that the only inclusion of RDs topics could not give decent results, more attention-taking ways must be used such as co-seminars for medical students and RDs patients and co-organization of RD Day in Bulgaria.

22. **Is your country supporting the participation of national experts in developing international guidelines to guide diagnosis and treatment of RD at national level?**

No.

Conclusion

The overall assessment of this workshop varied from very good to excellent. The audience consisted of both patients and medical professionals. Before the discussion, participants were introduced to the ICRDOD activities and Orphanet portal. The coming proposals can be summarized into several groups:

1. raising awareness among GPs;
2. wide public campaign in order to make the society more informed about the RDs problems and make people more solider with the RDs patients.

Theme 3 - Research on RD

Sub-Themes

1. Mapping of existing research resources, infrastructures and programmes for RDs
2. Needs and priorities for research in the field of RDs
3. Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects
4. Sustainability of research on RD
5. EU collaboration on research on RD

Workshop 4

Chairs: Prof. Rumen Stefanov (BAPES; Medical University of Plovdiv; NCCRD)
Prof. Maria Simeonova (Medical University of Pleven)
Dr. Ivelina Yordanova (Medical University of Pleven; DEBRA Bulgaria)

Rapporteur: Dr. Ivelina Yordanova

Date, time and place – 9:00-10:30, 29 May 2010, Paris Hall of Congress Centre of Novotel-Plovdiv

Audience: 19 persons

EUROPLAN Indicators

ACTIONS	INDICATORS	TYPE	ANSWERS
Building a research programme for Rare Diseases	Existing of RD National/Regional research programmes	Process	Not specific RD research programme
	RD research programme monitoring	Process	Not existing, not clearly stated
	Number of RD research projects approved by year (if possible yearly starting the year before plan commencement)	Outcomes	N/A
	Clinical trials funded by public bodies	Outcomes	No
	E-RARE joining	Process	No
	Including public health and social research, in the field of rare diseases	Process	No
	Research platforms and other infrastructures are also funded by the research programme	Process	No
Existence of national policy in support of the recruitment of young scientists/Researchers	Number of young scientists recruited every year to work specifically on rare diseases	Process	N/A

specifically for Rare Diseases			
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 since the plan started	Outcomes	N/A

Proposals, arising from the held discussion

1. **Evaluation of RD research resources and infrastructures across different disciplines and sources of funds, both public and private. Considering whether a combination of private and public support is feasible.**

Currently, there is no specific RD research policy defined. The National plan for rare diseases (NPRD) does not object to implement such a one. Instead, it will stimulate the creation of joint partnerships, including the expert centres for rare diseases in order to apply for different sources of funding.

2. **Does a specific national RD research programme with dedicated funds exist? Is there a scope for such programme?**

No. There is a more general national programme, envisaged to stimulate the scientific research in Bulgaria. Despite of not exactly define the term “rare diseases”, it does not excluded the rare diseases from its scope of funding. Actually, medical professionals and academics proposed to set a specialized national fund for RDs research. It may start as a joint public-private initiative with limited resources, but it will focus the attention of the healthcare professionals and researcher to the RDs problems.

3. **What is the scope of patient-driven research?**

Very limited and predominantly orientated in the public health research. Most of the patients, who participated, stated the reason for that to be the lack of precise funding opportunities and motivation for common work from the other stakeholders (particularly the medics).

4. **Assessing needs and priorities for basic, clinical and translational research, as well as priorities for social research in the field of rare diseases**

Patients and healthcare professionals were unanimous here – the RD patient registries are the fundamental need now. Establishing working national registry for rare diseases will be a huge push for all the RDs related research activities.

5. **Biobanks and databases**

That problem has not been selected as a primary objective for the actual moment by Bulgarian RDs stakeholders.

6. **How to make the link between basic and translational research and Centres of Expertise?**

Through NPRD, the National scientific research fund, the specialized Fund for RDs research (if created soon) and the medical scientific societies.

7. **Promoting interdisciplinary approaches to research**

It should be regulated by the official RDs policy from one side and additionally promoted and monitored by the medical scientific societies.

8. **Strengthening the exchanges among patient organisations**

A good way to do not only that, but also to assure a good communication and co-ordination among them is to create a website with possibilities for discussions, exchanges of ideas and contacts and publication of specialized information.

9. **Specific programmes for funding or recruiting young scientists on RD research**

No. Recently, the Ministry of Health (MoH) started discussions among the healthcare professionals and academics how the training of young doctors should be reorganized since the existing methodology is no longer efficient. So, that particular can must be discussed when the new MoH policy is known. Of course, the best way to attract the young scientists' interest is the availability of decent funding and career training resources. Another way is trying organizing it through public institutions with appropriate regulations.

10. How to ensure, through appropriate funding mechanisms, structural and long-term sustainability of research projects and research infrastructures in the field of RDs? In particular in respect of public health and social research, as well as transversal infrastructures.

The RDs patient registries are a very good example because of their mufti-functionality. So, the combination of gathering all the RDs stakeholders and providing multiple deliverable is not the only, but it is the best way to ensure sustainability of RDs research activities.

11. How to foster and support the participation of national researchers and laboratories, patients and patients’ organisations in EU-wide projects?

First of all, they need to be more aware of RDs topics. And after that, a specialized national programme for RDs research must be set-up. As explained already, it is not a huge resource, but a significant help to kick-off more intensive work in this field. Partnerships with all stakeholders on both national and EU level must be sought. And the deliverables from the past familiar projects must be easily available in order to raise awareness particularly among young professionals.

Conclusion

All participants, attending this meeting agreed that there almost no rare diseases research, currently conducted in Bulgaria. In order to foster that process they proposed:

- defining rare diseases as a topic of research within the National Scientific Research Fund
- defining clear criteria of funding rare diseases research by state resources
- establishing industry-based fund for rare diseases research, counting about 5% of drugs marketing funds, used in Bulgaria
- guaranteeing more media coverage of rare diseases research in Bulgaria in order to make it a public cause
- active participation at European rare diseases research project, presentation and implementation of their outcomes at local level.

Theme 4 - Standards of care for RDs - Centres of Expertise (CoE)/ European Reference Networks (ERN)

Sub-Themes

1. Identification of national or regional CoE all through the national territory by 2013
2. Sustainability of CoE
3. Participation in ERN
4. How to shorten the route to diagnosis
5. How to offer suitable care and organise adequate healthcare pathways for RD patients
6. How to ensure in CoE multidisciplinary approaches and integration between medical and social levels
7. How to evaluate CoE

4.1. Orphan Drugs (OD)

8. Future of OD
9. Access of RD patients to orphan drugs Pricing and Reimbursement
10. Compassionate use and temporary approval of orphan drugs. Off label use

Workshop 9

Chairs: Dr. Stamen Popov, Association of the Research-Based Pharmaceutical Manifacturers in Bulgaria (ARPharM)
Dr. Radostina Simeonova (Medical Centre "RareDis")

Rapporteur: Dr. Radostina Simeonova

Date, time and place – 17:00-19:00, 29 May 2010, Business Hall 1 of Congress Centre of Novotel-Plovdiv

Audience: 34 persons

EUROPLAN Indicators

ACTIONS	INDICATORS	TYPE	ANSWERS
Improve the quality of health care 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 at the national /regional level	Process	N/A
	Number of centres of expertise adhering to the policy defined in the country	Outcomes	0
	Groups of rare diseases followed up in centres of expertise	Outcomes	Covering all or most of rare diseases
	Centres of expertise adhering to the standards defined by the Council	Outcomes	0

	Recommendations - paragraph d) of preamble		
	Participation of national or regional centres of expertise into European reference networks	Outcomes	0
Develop Screening Policies	Number of diseases included in the neonatal screening programme	Outcomes	3
	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 61
	Time between the date of a ODD market authorisation by EMA and its actual date of placement in the market for the country	Outcomes	About 636 days
	Time from the placement in the market in the country to the positive decision for reimbursement by public funds	Outcomes	About 1080 days
	Number of ODD reimbursed 100%	Outcomes	11

Proposals, arising from the held discussion

1. **How to ensure that all patient living with a rare disease have access to a CoE in your country or abroad, and support the CoE creation where necessary?**

The criteria framework for the CoE in Bulgaria is being defined now. As it is scheduled and as most of the academicians and medical professionals have proposed the future network will be based (not exclusively) on the university hospitals in Bulgaria. That’s because their relatively good technical base and the high-profiled specialists, working there. An interesting statement, coming from the industry representative, proposed to established super CoE, putting at one place diagnosis, treatement and rehabilitation. However, all the other participants disagreed on it, believing it will be mission impossible.

2. **How to best apply in your country the criteria identified in the report of the EC Rare Disease Task Force, which will possibly become part of the EC Cross-Border Healthcare Directive, art.15, for the designation of CoE?**

Currently the CoE set of criteria is being defined by the NCCRD and MoH. All the EU recommendations on this topic will be considered. However, all the futures directives should have in mind the specifics of the national healthcare systems.

3. **In particular, how to make sure that CoE are –as much as possible- expert of the specific RD in both the clinical and the research field?**

Right now, that's not the most important issue in Bulgaria. The Bulgarian CoE are planned to be much like reference point, a link among the different rare diseases stakeholders. They may provide information about research activities and take part in research projects, but conducting research activity won't be a priority for them.

4. **What are the best structures and solutions suitable to be a CoE in your country? Do they depend on the disease (or group of diseases)? Are they regional or national?**

The envisaged network will include 5-6 centres on national level. The centres will be distributed almost equally in the country, so the distance factor will be well estimated. The centres will be working on all rare diseases field, not only on a separate group of them. And the fact, that they will be established within the university hospitals network will help to easy the connection between them and the medical specialists' and researchers' communities and will definitely increase their visibility for the patients and the society as a whole, the last one being an important issue for the patients, who in their own terms are left "alone" after being diagnosed with a rare disease.

5. **How to ensure, through appropriate funding mechanisms, the long-term sustainability of healthcare infrastructures, in particular Centres of Expertise?**

First of all, they should be actively supported by the MoH, especially during the initial stages. After that, they could possibly be funded by the National Health Insurance Fund (NHIF) and try to establish partnerships with the university hospitals, patient associations and the other rare diseases stakeholders. Interesting proposals on that topic included organizing continuous education events for medical professionals and students and running charity fund-raising activities.

6. **How to foster the participation of Centres of Expertise to European Reference Networks?**

Promoting access to best practices and sharing expertise is a very good option. Of course, it would be good to have it defined in some legal framework in advance with some particular attention on the funding aspects, since the Bulgarian network would be still newly-established and would need some technical and know-how help. That problem could be solved on both national and EU level. But all the rare diseases stakeholders in Bulgaria clearly underlined the idea of the European-level networks, realizing that's the only way to assure sustainable progress in rare diseases' field.

7. **How to support the mobility of patients and/or professionals beyond the national borders?**

That should be regulated exclusively on EU level with all the decisions taken to be valid anywhere in EU. Leaving this problem on the national governments will not only slow the process, but may even stop it.

8. **How to support the mobility of expertise and knowledge to facilitate the treatment of patients in their proximity (including mobility of information through ICT)?**

It should be regulated together with the patients/professionals mobility.

9. **How to map, network and support the laboratories at national level?**

They have been already supported through the NPRD by providing technical support and screening scope being expanded to 3 rare diseases. Genetic laboratories have been designed as an important element of NPRD, especially for the diagnosis and prevention priorities. Additionally, as most of them are within the university hospitals, a reliable connection between them and the CoE is assured.

10. **How to organise DNA and samples exchanges and reimbursement at European and international level?**

It should be organized by following and implementing the best European practices in this particular field.

11. **How to support the development of European guidelines on diagnostic tests and population screening?**

Participating in common regional screening programmes is one good way to do it. Even for small countries, it is the preferable way to organize the screening procedures. Within the EU frame it is not so difficult and through this other rare diseases international activities can be promoted.

12. **What mechanisms to develop in order to support common protocols and recommendations such as European reference opinions on diagnostic tools, medical care, education and social care?**

Good legal base is very important. Also, establishing and running common surveys and feedback tools in order to explore the assessment of different rare diseases stakeholders on these topics may be of great value.

13. **How to develop a system based on the adoption of clinical pathways for the provision of care for RD patients: provide funding for multidisciplinary consultations, cover patient and families transportation costs?**

Only by governmental set of regulations and strategies.

14. **How to link medical expertise of the specialised centres to local medical, paramedical and social care?**

A campaign promoting the NPRD is envisaged to take place soon. It will target all the potentials stakeholders, that’s very important in order to increase the visibility of the rare diseases in general. Additionally, NPRD will certainly link the CoE with the corresponding treatment, rehabilitation and integration services, directories of which will be formed too.

15. **How to establish good cooperation with relevant experts within the country or from abroad when necessary through European reference networks, with the aim to adopt common healthcare pathways based on the best evidence and expertise?**

CoE should rely on best medical specialists in the country. That could be defined through the medical scientific societies in Bulgaria, most of them have already recognized the rare diseases as their priority. Common research projects and organizing joint raising-awareness events are also good ways to set up a good cooperation.

16. **How to ensure multidisciplinary approaches in Centres of Expertise?**

Multidisciplinary approach is a leading aspect of the NPRD. It would be emphasized through all the NPRD’s priorities. When defining CoE criteria, they will include high qualification and rich experience in this field, which of course do mean having already applied multidisciplinary approaches in its activities.

17. **How to ensure integration between medical and social levels?**

There is a representative from the Ministry of Labour and Social Affairs to the NCCRD, so to make sure all social services are promptly used by the NPRD’s priorities. The multidisciplinary approach, mentioned already, will sure include social rehabilitation and integration matters.

18. **How to envisage a system for the evaluation of Centres of Expertise?**

CoE will be followed-up and assessed by MoH.

19. **Would it be based on clinical outcomes or patient satisfaction, or both?**

On both.

20. **How to improve and speed up national procedures for pricing and reimbursement of OD?**

Various proposals came here – a separate national regulation for ODs, decreasing VAT for ODs, etc. However, most of the participants agree that the actual legal base is not appropriate at all and a possible way to solve that problem is to transfer the ODs topics to the NHIF. Right now all this is regulated by the MoH and there is plenty of missing, such as clear criteria for deciding which ODs to reimburse and at what quantity. Patients proposed that rare diseases patients' registries should be used and estimated when defining ODs regulations and planning the public budget. Additionally, CoE should have some active engagement in that process too, serving as link between the governmental institutions and the other rare diseases stakeholders.

21. **In particular, what mechanisms to put in place to use the "clinical added value of orphan drugs" report developed at the EU level (EMA) to base the national decision on pricing and reimbursement in order to minimise delays in access to OD?**

A very interesting mechanism was proposed here. Medical professionals, academicians and researchers should present more actively to the MoH scientific publications and clinical evidences for the ODs' value. Expert positions from the patient associations, medical societies and industry should be included as well.

22. **How to promote a national policy on conditional pricing and reimbursement, based on the EU Pharma Recommendation "Improving access to orphan drugs"?**

See the answers above.

23. **How to foster access to OD through compassionate use?**

A regulation for ODs compassionate use is being currently prepared by an initiative group and will be proposed to MoH soon.

24. **Can drugs be prescribed off label and reimbursed when the evidence of a benefit for the patients exists?**

The prescription and the use of such medicaments is not always an easy job, however it is possible. On the other side, the reimbursement in this particular case is absolutely not possible.

25. **What measures can be studied and put in place to provide treatments other than medicinal products when the evidence of a benefit for the patient exists?**

Maybe some advance physical rehabilitation procedures can be explored. But particularly for the Bulgarian rare diseases patients medicinal products treatment is the most important issue right now. They would be all fully satisfied if they provided with it and combined in some aspect with appropriate rehabilitation and integration programmes.

26. **Number of OD on the market and number of patients treated**

Currently, there are 18 ODs, placed in the Bulgarian market. All of them are listed at the Positive Medicinal List, which should mean that they available and reimbursed to all the rare diseases patients. But that's not reality in Bulgaria. To be reimbursed, they should be included at Regulation 34 of MoH, which describes the so called "expensive medicinal treatment". Currently, only 11 of them are listed there, but for 2 of them there has been any reimbursement yet. Still, it does not mean that the 9 left are fully available to all the rare diseases patients because of the missing prevalence data, unclear criteria and non-transparent procedures for including in this reimbursement list and not at last place – lack of political will to solve these problems.

27. **Obstacles to the availability of OD approved in the EU within the timeframe requested by the regulations (180 days).**

Pharmaceutical companies seem to have lack of motivation to place their OD products in small countries like Bulgaria. The legal framework is far from perfect in Bulgaria, but the fact that almost one third of ODs is already available and part of it reimbursed shows that the problem is solvable. The industry and the national health authorities should be probably pushed by the corresponding EU institutions to accelerate this process.

Conclusion

The workshop was focus on 2 topics – the CoE network in Bulgaria and the ODs. The Centres of Expertise have been interesting as newly emerged topic. All the participants agreed with the preliminary criteria for defining CoE. Additionally, most of the medical professionals suggested that these future institutions should only focus on connecting rare diseases patients to appropriate services and units, they should be very big structures, but have to tend to be compact in order to be more dynamic in prospective. However, they should not be limited when talking about taking part in common national and European projects. Also, industry representatives concluded that CoEs should be involved in some matter in the process of accelerating ODs access in Bulgaria.

The second topic (ODs) was also so attractive for the participants and especially the patients. All the present rare diseases stakeholders expressed their dissatisfaction with the current ODs legal mechanisms which are very slow, with plenty of unclear criteria and rules. Almost unanimously they suggested that all the ODs management should be transferred from MoH to the NHIF with all the interested sides consulted parallelly. Additional proposals included a separate regulation for ODs, different from these for the traditional drugs and lower VAT on ODs. All the participants expressed their support for the EU and EUROPLAN recommendations on that questions and even called on them to be mandatory, not only of recommending character.

Theme 5 - Patient Empowerment and Specialised Services

Sub-Themes

1. Involvement of patients and their representatives in decision-making processes in the field of RDs
2. Support to the activities performed by patient organisations
3. Specialised social services: Respite Care Services; Therapeutic Recreational Programmes; Services aimed at the integration of patients in daily life
4. Help Lines

Workshop 2

Chairs: Mr. Vladimir Tomov (NAPRD; NCCRD)
Mrs. Veska Sabeva (National Council for Integration of People with Disabilities to the Council of Ministers)

Rapporteur: Mr. Vladimir Tomov

Date, time and place – 17:00-19:00, 28 May 2010, Moscow Hall of Congress Centre of Novotel-Plovdiv

Audience: 38 persons

EUROPLAN Indicators

ACTIONS	INDICATORS	TYPE	ANSWERS
Promoting the existence of a RD patients' organisation that represents all RD patients' associations	Number of umbrella organisations specific on rare diseases	Process	NAPRD – official representative of the RD associations on national level
	Having a directory of RD Patients' organisations	Process	Yes
	Number of patients' associations	Outcomes	21
	Number of diseases covered by patients' associations	Outcomes	More than 30
Patients' organisations involvement in decisions affecting RD	Permanent and official patients' representatives in plan development, monitoring and assessment	Process	Yes, clearly established, substantially implemented and participation reimbursement considered
	Participation of patients' organisations in the	Process	No

	development of RD research strategies		
	Participation of patients organisations in the RD centres of expertise designation and evaluation	Process	No
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	N/A
	Support to sustainable activities to empower patients, such as: Awareness raising Capacity building and training Exchange of information and best practices Networking Outreach to very isolated patients	Outcomes	N/A
Building - supporting the existence of comprehensive help line for patients	Availability of Help line for RD	Process	Referred RD help lines
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	Yes
Supporting rehabilitation programmes	Existence of programmes to support rehabilitation of RD patients	Process	In preparation
Supporting social services aimed at rare disease patients and their families	Existence of national schemes promoting access of RD patients and their families to Respite Care services	Process	In preparation
	Existence of public schemes supporting Therapeutic Recreational Programmes	Process	Yes
	Existence of programmes to support integration of RD patients in their daily life	Process	No

Proposals, arising from the held discussion

1. **How to ensure, through appropriate funding mechanisms, patient representativeness in decision-making processes relevant to RDs?**

Patients believed that they are more interested in the overall progress of the NPRD). All the patients associations in Bulgaria are on voluntary basis and they are run by patients and their relatives. They said what motivates them is the possibility for better treatment and rehabilitation. The funding must be in two directions – adequate and quality treatment and rehabilitation (the principle one) and popularization of the RDs topics which will raise the society's awareness about them and make people more comprehensive and solider.

2. **How to support activities performed by patient organisations?**

Partially through NPRD. But patients complained about the lack of support coming from there by now. The more experienced associations said that taking part in common national and European projects is a good way to develop an organization too. Partnerships with European umbrella organizations help them a lot too, patients expressed their particular gratitude to EURORDIS, Thalassemia International Federation and Cystic Fibrosis Europe, the other patients associations should be encouraged too to seek co-operation with their international equivalents.

3. **What mechanisms can be put in place to support patients' empowerment activities and their representativeness in EU-wide instances?**

See question 2 above.

4. **What kinds of programmes exist in your country to support patients and families with RD and/or disabilities in general?**

In Bulgaria, there are no support programmes for people with RDs. There are programmes for patients with disabilities and their families, but they are too general, out-of-date and not available for all persons who need them. Most of the existing programmes are managed and co-ordinated by the municipal authorities, so there is also a difference of the services available on a national level – the majority of the programmes being almost in the biggest cities and a lack of specialized services in the small rural and remote communities. The access to these programmes passes by a certificate of disability, issued by a territorial expert commission (TEK), composed by medical professionals. These commissions define and evaluate the degree of disability. Most of the proposals here were directed to the TEK activities. RDs patients complained about the bureaucracy, incompetence and lack of clear criteria when getting this certificated. Most of TEK commissioners are unaware of the RDs, RDs patients are made to prove evident facts. There are also no clear rules how the RDs patients have to be certificated and even paradoxes of RDs patients in a severe condition not being evaluated by TEK. Patients insisted for renewing of the TEK work guidelines and RDs patients' involvement in the process of control of TEK.

5. **What kind of schemes or programmes do exist supporting access of RD patients to Respite Care Services, Therapeutic Recreational Programmes and services aimed at the integration of patients in daily life ?**

See question 4 above. The existing programmes include personal caregivers support, specialized transport and nutrition services, physical rehabilitation, psychological aid, social integration and professional training and pre-qualification programmes. However, only limited persons have access to them. Most of the services are not fully appropriate for RDs patients, because they are intended for more general health disorders. But the access of RDs patients to them is really hard because of the existing legal framework which does not included RDs specifications.

6. **What can be done to improve their availability and accessibility of such services, including public funding?**

NPRD does include such priority. But by now, there are no steps taken in this direction. Most of the patients agreed that RDs should not be separated by the other disorders in terms of rehabilitation, integration and recreation services. However, they need to be defined and legally included in the existing sets of social services. The methodology of TEK must be absolutely revised and updated, including clear definitions for RD condition and precise criteria how the patients must be classified.

Additionally, since one part of the social programmes is organized on a municipal level, the local authorities have to be introduced in RDs problems and be aware of the existence of such people. That could also be used as a chance to start local partnership between NAPRD branches and different municipalities in order to include the RDs patients in the local life and, as a global objective, to raise the public awareness and solidarity towards RDs.

7. **How are specialised social services financed? By government institutions and budget? By private initiative or patient associations?**

The vast majority are public-financed. Of course, there are some private initiatives, but they are locally concentrated and focused on a very specific target group.

Most of the RDs patient associations in Bulgaria are recently established and they are not financially supported through government programmes. All of them rely on external funding through common projects and activities, on both national and EU level.

8. **What kind of help lines (all diseases) exist in your country to assist RD patients and healthcare professionals?**

There are several groups of help lines in Bulgaria. The institutional ones (MoH, National health insurance fund, Executive drugs agency, etc.) provide general and organizational information. The patients associations (general diseases) provide advocacy support and information about the rights of the patients. Specific information about the RDs for both patients and healthcare professionals is only provided by the Information centre for rare diseases and orphan drugs (ICRDOD) help line services. NAPRD and its member-association do not run a separate help line. Through NAPRD they participated in management of the help line of the Confederation for health protection, but it is a general one and does not provide disease-orientated information. Some of NAPRD members run their own website, collecting useful materials and links about specific rare diseases but they are few.

9. **How to develop or consolidate existing patient-run help line services for RD?**

The participants defined three obstacles here. The basic one is the lack of funding, which solution must be sought by the ways, explained in the previous questions. Additionally, lack of motivation among the patients and the lack of adequate information are also serious problems. The majority of the Bulgarian RDs patients continue to be closed inside their own daily problems and do not want to work more actively for the RDs cause. The proposals here included co-operating with international voluntary corps, present in Bulgaria, establishing more intense contacts with medical students and young medical professionals. Also, more efficient work should be done with the local authorities. Sometimes, it’s better to start solving a problem on a local basis and gradually to do it on a national level later. The lack of the information is caused not by the lack of information resources. European databases, such as Orphanet are now well known among the RDs stakeholders in Bulgaria. But the regular patients do not always know English or other foreign languages, so they are not able to extract the information they need. A proposal to organize a national directory for RDs information has evolved. It should contained information for both patients and professionals.

10. **How to improve the service offered? How to improve their visibility esp. for patients?**

The main proposal here is to have the contacts of NAPRD members, so when a RDs patient has been diagnosed or there are suspicions for RDs, he could easily get in touch with people, who are experienced in these topics.

11. **National measures to establish the 116 European number**

None.

12. **How are help lines financed? By private initiative or patient associations? Is there any government funding?**

Exclusively by private initiatives.

13. How to ensure their long-term sustainability?

The basic chance for long-term sustainability is to participate in common projects with the RDs stakeholder on both national and EU level.

<u>Conclusion</u>

Representatives of 18 RDs patients associations attended this meeting. The proposals can be put into the following groups:

- obligatory ratification of all RDs strategies and action plans by the RDs patients and their representatives
- more intense and efficient public awareness campaign
- inclusion of the RDs into the rehabilitation and recreation services’ legal framework
- appropriate possibilities for governmental funding of the RDs associations’ activities
- creating online directories for RDs topics (medical information, research, social services, etc)

IV. Summary

Over 350 patients, medical professionals, representatives of government and industry participated in the Bulgarian EUROPLAN National Conference for Rare Diseases and Orphan Drugs, part of the project EUROPLAN, funded by the European Commission. Within the conference, a total of 2 panel sessions, 9 workshops and 4 patient seminars was held, in which participants had the opportunity to learn best European practices and recommendations in the field of rare diseases, the priorities and objectives of the Bulgarian National Plan for rare diseases and most importantly - to discuss these issues, to express their proposals in order to implement in an optimal way the policies and strategies for rare diseases in Bulgaria.

Conference participants have agreed on the following general proposals and guidelines for actions at national level:

- full support of the priorities set out in the EU Council Recommendation on rare diseases actions, adopted on June 9, 2009;
- securing the implementation of the Bulgarian National Programme for rare diseases with the approriate funds, previewed in its budget framework;
- need for urgent legislative initiatives to protect the rights of people with rare diseases and ensure adequate prevention, treatment, rehabilitation and social cares;
- encouraging the establishment of epidemiological registries for rare diseases in Bulgaria;
- implementation of an integrated approach to people with rare diseases and their families;
- organizing a public campaign aimed, to fund and to stimulate research on rare diseases in Bulgaria.

V. Document History

Status (Draft/Reviewed/Final)	**Reviewed**
Version n°	**20072010**
Author(s)	**Georgi Iskrov**
Reviewer	**Vladimir Tomov**
File name	**Bulgarian EUROPLAN NC 2010 - Report**

Annex 1

List of participants

	Name	Affiliation
1	Ahmed Alikanov	Association "CF"
2	Albena Dimitrova	Association of patients with HAE
3	Alexander Alexandrov	Association "CF"
4	Alexander Gochev	Association "CF"
5	Alexandra Gritsnelova	OTB
6	Alexandrina Topalova	AMSB
7	Anastas Batalov	University hospital "St. George"
8	Anelia Balabanova	University hospital "St. George"
9	Anelia Licheva	Association "CF"
10	Anelia Stoeva	Association "CF"
11	Anelia Todorova	Bulgarian anti-thalassemia organization
12	Aneta Feodorova	Association "CF"
13	Aneta Ivanova	University hospital "St. Ivan Rilski"
14	Aneta Pankina	AMSB
15	Angel Stoimenov	National hematology centre
16	Angelina Stoyanova	University hospital "St. George"
17	Ani Kodinova	AMSB
18	Anna Filipova	OTB
19	Anna Indjova	Association "CF"
20	Antonia Ivanova	National association "Mucopolysaccharidosis"
21	Antonio Piga	Thalassemia centre, Turin, Italy
22	Anzhela Harizanova	OTB
23	Atanas Banchev	AMSB
24	Atanas Platnarov	OTB
25	Atanas Shivachev	Bulgarian Association "Wilson's Disease"
26	Atanas Stefanov	Association "CF"
27	Aynur Alieva	Association "CF"
28	Basri Dzhebir	OTB
29	Biliana Kitanova	OTB
30	Biljana Petkovska-Popov	SOLPHARM
31	Blaga Baeva	Regional hospital of Gabrovo
32	Boriana Spasova	National Association Primary pulmonary hypertension
33	Boril Mechkov	Association of patients with acromegaly in Bulgaria
34	Borislav Stoyanov	Association "CF"
35	Branimir Kanazirev	University hospital "St. Marina"

36	Brigita Radeva	University pediatric hospital
37	Christina Stefanidou	TIF
38	Dafinka Tancheva	Association "CF"
39	Daniela Virovska	Bulgarian Association "Wilson's Disease"
40	Darko ArizankoskI	AMSB
41	Denitsa Simeonova	AMSB
42	Denka Kostova	University hospital "St. George"
43	Denka Stoyanova	National hematology centre
44	Desislava Abadzhieva	DEBRA Bulgaria
45	Desislava Dimitrova	AMSB
46	Desislava Dimitrova	Association "CF"
47	Desislava Uzunova	NOVARTIS
48	Diana Marinova	OTB
49	Diana Plachkova	Association "CF"
50	Diana Zheleva	Association "CF"
51	Dilian Iliev	OTB
52	Dimitar Stoyanov	Association of patients with acromegaly in Bulgaria
53	Dimitar Stoyanov	OTB
54	Dimitar Vuchev	University hospital "St. George"
55	Dimitrina Dimitrova	OTB
56	Dimitrina Konstantinova	Medical university of Varna
57	Dimo Mitev	AMSB
58	Dimo Ribov	Regional hospital of Burgas
59	Dobriana Panova	University hospital "Queen Giovanna"
60	Dobrinka Mitova	Association "CF"
61	Dobromir Dobrev	Association "CF"
62	Donka Vasileva	National hematology centre
63	Dora Popova	Military medicine academy
64	Dorica Dan	EURORDIS
65	Doroteya Leonkeva	University hospital "St. Ivan Rilski"
66	Dragomir Slavev	OTB
67	Dragomir Tomalevski	Bulgarian anti-thalassemia organization
68	Ekaterina Valcheva	University hospital "St. Marina"
69	Elena Bangieva	AMSB
70	Elena Dimova	OTB
71	Elena Yolovska	Bulgarian Association "Wilson's Disease"
72	Eleonora Hristova	AMSB
73	Eli Hristozova	Medical university of Plovdiv
74	Eli Zheleva	OTB
75	Elina Beleva	AMSB
76	Elina Peteva	University hospital "St. Marina"

77	Elisaveta Naumova	University hospital "Alexandrovska"
78	Emil Aleksov	NOVARTIS
79	Emilia Ilieva	Medicus Alpha Clinic
80	Emilia Peneva	National association of congenital hypothyroidism
81	Emine Adem	OTB
82	Erhan Dzhebir	OTB
83	Ersen Aliev	Association "CF"
84	Evelina Zlateva	OTB
85	Evgeni Genov	OTB
86	Evgeni Hadzhiev	University hospital "Alexandrovska"
87	Evgenia Milanova	OTB
88	Evgenia Minkova	OTB
89	Fani Martinova	University hospital "Pirogov"
90	Gabriela Koteva	OTB
91	Galia Pavlova	National hematology centre
92	Galia Zlateva	OTB
93	Galina Gicheva	Association "CF"
94	Gancho Stamatov	OTB
95	Geno Kalchev	Association "CF"
96	Georgi Goranov	University hospital "St. George"
97	Georgi Iskrov	BAPES-ICRDOD
98	Georgi Koychev	National Association Primary pulmonary hypertension
99	Georgi Mihaylov	National hematology centre
100	Georgi Mitev	Silistra
101	Georgi Petkov	National Association Primary pulmonary hypertension
102	Georgi Tonev	ProVita
103	Gergana Alexandrova	Association "CF"
104	Gergana Atanasova	National Association Primary pulmonary hypertension
105	Gergana Nedyalkova	Association "CF"
106	Gergana Petrova	Medical university of Plovdiv
107	Gergana Razhankova	Association "CF"
108	Gergana Semova	University hospital "St. Marina"
109	Ginka Angelova	Association "CF"
110	Ginka Kolchakova	National Association Syringomyelia
111	Greta Stoyanova	Association "CF"
112	Gyuldzhan Myumyun	OTB
113	Hasan Burnusuzov	University hospital "St. George"
114	Hristina Angelova	Association "CF"
115	Hristina Milanova	Military medicine academy
116	Hristina Shisheva	OTB
117	Hristo Ivanov	OTB

118	Hristo Zaharinov	Medical university of Sofia
119	Hristofor Hristov	CML association
120	Iliana Hristova	Association "CF"
121	Ismet Ismailov	Association "CF"
122	Iva Denkova	Association "CF"
123	Ivan Alexandrov	Association "CF"
124	Ivan Chakarov	University hospital of Stara Zagora
125	Ivan Gogov	Association "CF"
126	Ivan Ivanov	OTB
127	Ivan Kirev	AMSB
128	Ivan Markov	OTB
129	Ivan Mitov	Association "CF"
130	Ivan Mumdzhiev	University hospital "St. George"
131	Ivan Razhankov	Association "CF"
132	Ivanka Galeva	University hospital "Alexandrovska"
133	Ivanka Nenova	University hospital "St. George"
134	Ivanka Pironkova	Association "Gaucher disease"
135	Ivaylo Hristov	Pleven
136	Ivaylo Ivanov	Association "CF"
137	Ivelina Bozhkova	Pazardzhik
138	Ivelina Yordanova	Medical university of Pleven / DEBRA Bulgaria
139	Ivet Koleva	Medical university of Pleven
140	Janet Grudeva	University hospital "St. George"
141	Julieta Kratunkova	Sliven
142	Kalin Mihaylov	Ruse
143	Kalina Kasabova	Association "CF"
144	Kalina Madarzhieva	National hematology centre
145	Kamelia Alexandrova	University hospital "Alexandrovska"
146	Karina Shahinyan	OTB
147	Karlien De Rijcke	CF Europe
148	Katerina Madzharova	AMSB
149	Katerina Nikolova	National hematology centre
150	Katia Gocheva	OTB
151	Katia Sapunarova	University hospital "St. George"
152	Kolyu Sapundzhiev	Association "CF"
153	Konstantin Sotirov	OTB
154	Kostadin Tanchev	Association "CF"
155	Krasimir Kraev	AMSB
156	Krasimira Berbenlieva	AMSB
157	Krasimira Chudomirova	University hospital "St. George"
158	Krasimira Halacheva	Thracian university

159	Krasimira Nedeva	Association "CF"
160	Krastina Manoilova	University pediatric hospital
161	Kremena Velikova	Association "CF"
162	Lachezar Marinov	Medical university of Varna
163	Lidia Stanoeva	OTB
164	Lili Andreeva	OTB
165	Lilia Popova	AMSB
166	Lilia Stoyanova	Crohn association
167	Liliana Grozdanova	Medical centre "RareDis"
168	Liliana Stoyanova	Association of patients with acromegaly in Bulgaria
169	Lyuba Pramatarova	Regional hospital of Burgas
170	Lyubov Chochkova	University hospital "St. George"
171	Lyudmil Simeonov	AMSB
172	Magdalena Abadzhieva	DEBRA Bulgaria
173	Magdalena Ivanova	AMSB
174	Mancho Manchev	Swedish Orphan
175	Manuela Mihaleva	OTB
176	Margarita Orlyova	OTB
177	Maria Boncheva	University pediatric hospital
178	Maria Daneva	OTB
179	Maria Genova	OTB
180	Maria Kamburova	Association "CF"
181	Maria Kedeva	Association "CF"
182	Maria Kuzmanova	Association "CF"
183	Maria Luiza Skerleva	OTB
184	Maria Marinova	Association "CF"
185	Maria Nedeva	OTB
186	Maria Simeonova	Medical university of Pleven
187	Maria Spasova	University hospital "St. George"
188	Maria Stoyanova	OTB
189	Marian Angelov	NOVARTIS
190	Marian Ivanov	Association of patients with HAE
191	Mariana Angelova	Medical centre "RareDis"
192	Mariana Mavrova	Association "CF"
193	Mariana Murdjeva	Medical university of Plovdiv
194	Mariana Pancheva	OTB
195	Mariana Spasova	OTB
196	Mariana Yordanova	National alliance of porphyria patients
197	Marieta Pesheva	Association "CF"
198	Marieta Peycheva	University hospital "St. George"
199	Marin Skerlev	OTB

200	Marin Zhelev	Association "CF"
201	Marleen Moens	UZ Leuven
202	Marta Baleva	University hospital "Alexandrovska"
203	Martin Kozhinkov	Crohn association
204	Martin Pete	AMSB
205	Maya Hristozova	Association "CF"
206	Mesut Habil	OTB
207	Miglena Georgieva	Medical university of Varna
208	Milen Milev	Sofia
209	Milena Belcheva	University hospital "St. Marina"
210	Milena Krasteva	National association of primary pulmonary hypertension
211	Milena Naneva	OTB
212	Milena Slavova	University hospital "Alexandrovska"
213	Militsa Brancheva	AMSB
214	Mima Tsoneva	OTB
215	Mirela Bahova	NAPRD
216	Mirela Rangelova	National hematology centre
217	Monka Todorova	Smolyan
218	Myriam Vreys	UZ Leuven
219	Nadezhda Madzhirova	Medical university of Plovdiv
220	Nadezhda Racheva	Medical university of Sofia
221	Nadia Todorova	Association "CF"
222	Nartsis Kaleva	University hospital "St. George"
223	Natalia Maeva	National association of primary pulmonary hypertension
224	Natasha Stoyneva	Targovishte
225	Nedezhda Gogova	Association "CF"
226	Neli Dirimanova	National alliance of porphyria patients
227	Neli Kostadinova	Bulgarian association for neuromuscular diseases
228	Neli Nedyalkova	Association "CF"
229	Nenad Poljak	SOLPHARM
230	Nevena Miteva	AMSB
231	Nevena Popova	ZDRAVE.net
232	Nikola Myahov	Association of patients with HAE
233	Nikolay Nikolaev	Association "CF"
234	Nikolay Stavrev	University hospital "St. Marina"
235	Nina Petkova	University hospital "Alexandrovska"
236	Ognyan Ognev	NOVARTIS
237	Olia Divizieva	Association "CF"
238	Pavel Gradev	OTB
239	Penka Georgieva	Confederation for health protection
240	Peter Atanasov	National association of primary pulmonary hypertension

241	Peter Indzhov	Association "CF"
242	Peter Nedyalkov	Association "CF"
243	Peter Petrov	Association "CF"
244	Petko Ivanova	Association of patients with HAE
245	Petrana Chakarova	University hospital of Stara Zagora
246	Petrana Musenova	AMSB
247	Plamen Ivanov	OTB
248	Polina Milusheva	Growth hormone defficiency patient association
249	Preslav Kolchakov	National Association Syringomyelia
250	Radoslav Ilchev	ABBOTT
251	Radostina Simeonova	Medical centre "RareDis"
252	Ralitsa Yordanova	BAPES-ICRDOD
253	Rosen Dimitrov	NOVARTIS
254	Rosen Zlatev	Bulgarian Association "Wilson's Disease"
255	Rositsa Georgieva	University hospital of Stara Zagora
256	Rositsa Kichukova	OTB
257	Rositsa Staneva	Association "CF"
258	Rumen Marinov	University hospital of Stara Zagora
259	Rumen Stefanov	BAPES / Medical university of Plovdiv
260	Rumiana Mihova	Association "CF"
261	Rumiana Tarnovska	University hospital "Alexandrovska"
262	Rusina Spasova	National association of primary pulmonary hypertension
263	Sevil Ahmed	National hematology centre
264	Silvia Pashkunova	Sofia
265	Silvia Ruseva	Association "CF"
266	Slav Tsonev	OTB
267	Snezhana Stoyanova	Regional hospital of Haskovo
268	Sofia Skarantavou	TIF
269	Sofka Dayanova	Association "CF"
270	Spas Gachev	Genzyme
271	Spyros Bitsis	TIF
272	Stamen Popova	ARPhaRM
273	Stanimir Georgiev	National association of primary pulmonary hypertension
274	Stanimir Stoev	Association "CF"
275	Stanimir Tsonev	Prader-Willi association
276	Stefan Goranov	University hospital "St. George"
277	Stefka Popova	OTB
278	Stefko Georgiev	National association of primary pulmonary hypertension
279	Stoil Lazarov	Primary immunodeficencues patient association
280	Stoyan Apostolov	OTB
281	Stoyan Spasov	National association of primary pulmonary hypertension

282	Svetla Kostova	Medical university of Pleven
283	Svetlana Atanasova	Association "CF"
284	Svetoslav Stoyanov	OTB
285	Svilen Nikolov	Bulgarian anti-thalassemia organization
286	Tania Panayotova	Association "CF"
287	Teodor Atanasov	Association "CF"
288	Teodora Haralampieva	Genzyme
289	Teodora Pancheva	OTB
290	Teodora Zaharieva	Bulgarian patient forum
291	Tihomir Hristov	OTB
292	Todor Mangarov	National association of primary pulmonary hypertension
293	Todorka Kostadinova	Medical university of Varna
294	Trayana Koleva	Association "CF"
295	Troycho Troev	Military medicine academy
296	Tsetska Bambova	Association "CF"
297	Tsonka Miteva	Medical university of Plovdiv
298	Tsvetanka Raycheva	Growth hormone defficiency patient association
299	Tsvetelina Chorbadzhiyska	Pleven
300	Tsvetelina Panayotova	Association "CF"
301	Tsvetelina Radeva	PNH patient association
302	Tsvetelina Yurukova	NAPRD
303	Vaklin Dayanov	Association "CF"
304	Valentina Filipova	Association "CF"
305	Valentina Petkova	Medical university of Sofia
306	Valentina Peychinova	OTB
307	Valeria Kaleva	University hospital "St. Marina"
308	Valeria Pershina	Association "CF"
309	Valia Kiriakova	NOVARTIS
310	Vania Dechkova	OTB
311	Vania Dobreva	Association of patients with acromegaly in Bulgaria
312	Vania Petrova	Association "CF"
313	Vania Rangelova	AMSB
314	Vania Toteva	National Association Primary pulmonary hypertension
315	Vanushka Markova	Silistra
316	Vasily Mihaylov	Medical university of Pleven
317	Velina Cherkezova	University hospital Stara Zagora
318	Veneta Radeva	OTB
319	Veneta Rusanova	OTB
320	Venko Stanev	Association "CF"
321	Ventseslava Atanasova	NOVARTIS
322	Vesela Stefanova	Medical university of Plovdiv

323	Veselin Boyadzhiev	Medical university of Varna
324	Veselina Goranova	University hospital "St. George"
325	Veselina Nedeva	OTB
326	Veska Ivanova	Stara Zagora
327	Veska Sabeva	National council for integration of people with disabilities
328	Viktor Bashev	OTB
329	Vili Garabedyan	Medicus Alpha Clinic
330	Violeta Antonova	Bulgarian association for neuromuscular diseases
331	Violeta Yotova	Medical university of Varna
332	Violina Velikova	Association "CF"
333	Vladimir Tomov	NAPRD
334	Vladimir Velichkov	OTB
335	Vladislava Atanasova	Bulgarian anti-thalassemia organization
336	Yana Firova	OTB
337	Yanka Mircheva	Association "CF"
338	Yanko Ivanov	Association "Gaucher disease"
339	Yordan Kalchev	AMSB
340	Yordan Uzunov	Medical university of Pleven
341	Yordan Yordanov	Association "CF"
342	Yordanka Petkova	Association of patients with HAE
343	Yordanka Stanilova	Haskovo
344	Yulia Stanoycheva	Association "CF"
345	Yulian Georgiev	National association of primary pulmonary hypertension
346	Yulian Raynov	Military medicine academy
347	Zdravka Gocheva	OTB
348	Zhelyazko Valchinkov	Bulgarian anti-thalassemia organization
349	Zhivka Sirakova	University hospital "Alexandrovska"
350	Zhivka Skerleva	OTB
351	Zlatka Misheva	University hospital "St. George"
352	Zoya Aleksieva	OTB

Annex 2

Bulgarian EUROPLAN National Rare Diseases Conference

28-30 May 2010, Plovdiv, Bulgaria

Memorandum

Over 350 patients, medical professionals, representatives of government and industry participated in the Bulgarian EUROPLAN National Conference for Rare Diseases and Orphan Drugs, part of the project EUROPLAN, funded by the European Commission. The event was co-organized by the National alliance of people with rare diseases and the Information centre for rare diseases and orphan drugs.

Within the conference, a total of 2 panel sessions, 9 workshops and 4 patient seminars was held, in which participants had the opportunity to learn best European practices and recommendations in the field of rare diseases, the priorities and objectives of the Bulgarian National Plan for rare diseases and most importantly - to discuss these issues, to express their proposals in order to implement in an optimal way the policies and strategies for rare diseases in Bulgaria.

Conference participants have agreed on the following general proposals and guidelines for actions at national level:

- full support of the priorities set out in the EU Council Recommendation on rare diseases actions, adopted on June 9, 2009;
- securing the implementation of the Bulgarian National Programme for rare diseases with the appropriate funds, previewed in its budget framework;
- need for urgent legislative initiatives to protect the rights of people with rare diseases and ensure adequate prevention, treatment, rehabilitation and social cares;
- encouraging the establishment of epidemiological registries for rare diseases in Bulgaria;
- implementation of an integrated approach to people with rare diseases and their families;
- organizing a public campaign aimed, to fund and to stimulate research on rare diseases in Bulgaria.