2013 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE

STATE OF THE ART OF RARE DISEASE ACTIVITIES IN BULGARIA

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More information on the European Union Committee of Experts on Rare Diseases can be found at www.eucerd.eu.

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### ACRONYMS

CAT - Committee for Advanced Therapies at EMA  
CHMP - Committee for Medicinal Products for Human Use at EMA  
COMP - Committee on Orphan Medicinal Products at EMA  
DG - Directorate General  
DG Enterprise - European Commission Directorate General Enterprise and Industry  
DG Research - European Commission Directorate General Research  
DG Sanco - European Commission Directorate General Health and Consumers  
EC - European Commission  
ECRD - European Conference on Rare Diseases  
EEA - European Economic Area  
EMA - European Medicines Agency  
ERN - European reference network  
EU - European Union  
EUCERD - European Union Committee of Experts on Rare Diseases  
EUROCAT - European surveillance of congenital anomalies  
EUROPLAN - European Project for Rare Diseases National Plans Development  
EURORDIS - European Organisation for Rare Diseases  
FDA - US Food and Drug Administration  
HLG - High Level Group for Health Services and Medical Care  
HTA - Health Technology Assessment  
IRDiRC – International Rare Diseases Research Consortium  
JA - Joint Action  
MA - Market Authorisation  
MoH - Ministry of Health  
MS - Member State  
NBS - New born screening  
NCA - National Competent Authorities  
NHS - National Health System  
PDCO - Paediatric Committee at EMA  
RDTF - EC Rare Disease Task Force  
WG - Working Group  
WHO - World Health Organization
GENERAL INTRODUCTION

This document was produced by the Scientific Secretariat of the European Union Committee of Experts on Rare Diseases (EUCERD), through the EUCERD Joint Action: Working for Rare Diseases (N° 2011 22 01), which covers a three year period (March 2012 – February 2015).

The present report aims to provide an informative and descriptive overview of rare disease activities at European Union (EU) and Member State (MS) level in the field of rare diseases and orphan medicinal products up to the end of 2012. A range of stakeholders in each Member State/country have been consulted during the elaboration of the report, which has been validated as an accurate representation of activities at national level, to the best of their knowledge, by the Member State/country representatives of the European Union Committee of Experts on Rare Diseases. The reader, however, should bear in mind that the information provided is not exhaustive and is not an official position of the European Commission, its Agencies or national health authorities.

The report is split into six parts:

Part I: Overview of rare disease activities in Europe
Part II: Key developments in the field of rare diseases in 2012
Part III: European Commission activities in the field of rare diseases
Part IV: European Medicines Agency activities and other European activities in the field of rare diseases
Part V: Activities in EU Member States and other European countries in the field of rare diseases
Part VI: Activities at National level in each EU Member State and other European countries in the field of rare diseases

Each part contains the following description of the methodology, sources and validation process of the entire report, and concludes with a selected bibliography and list of persons having contributed to the report.

The present document contains the information from Parts II and V of the report concerning Bulgaria. A list of contributors to the report and selected sources are in annex of this document. For more information about the elaboration and validation procedure for the report, please refer to the general introduction of the main report.
RARE DISEASE ACTIVITIES IN BULGARIA

Definition of a rare disease
Stakeholders in Bulgaria accept the definition of a prevalence of no more than 5 in 10,000 individuals. This definition is officially mentioned in the Bulgarian National Plan for Rare Diseases.

National plan/strategy for rare diseases and related actions
On 27 November 2008, the Bulgarian Council of Ministers approved the National Plan for Rare Diseases – genetic disorders, congenital malformations and non-hereditary diseases (2009 – 2013). The Bulgarian National Plan for Rare Diseases started on 1 January 2009 and will last for 5 years. Bulgaria’s National Plan for Rare Diseases is currently active and consists of nine priorities targeting all rare diseases:

- Collection of epidemiological data for rare diseases in Bulgaria by creation of a national register;
- Improvement of the prevention of genetic rare diseases by enlarging the current screening programmes;
- Improvement of the prevention and diagnostics of genetic rare diseases by introducing new genetic tests, decentralisation of the laboratory activities and easier access to medico-genetic counselling;
- Integrative approach to the prevention, diagnostics, medical treatment and social integration of patients and their families;
- Promotion of the professional qualification of medical specialists in the field of early diagnostics and prevention of rare diseases;
- Feasibility study on the necessity, possibility and criteria for the creation of a reference centre for rare diseases of functional type;
- Organisation of a national campaign to inform society about rare diseases and their prevention;
- Support and collaboration with NGOs and patient associations for rare diseases;
- Collaboration with the other EU members.

A National Consulting Council on Rare Diseases (NCCRD) has been established by the Ministry of Health, to supervise the progress and implementation of the plan. NCCRD includes medical professionals, Ministerial representatives and a representative of the National Alliance of People with Rare Diseases. Although the estimated budget of the Plan is €11.3 million, the assigned funds are much less and are disproportionately distributed (i.e. directed almost exclusively towards genetic testing and screening activities). The estimated budget does not take into account the costs for the provision of clinical services for rare disease patients. Funding for rare disease policies is provided by the Ministry of Health and reimbursements of drugs for rare diseases are covered by the National Health Insurance Fund (defined by Ministerial Ordinance 38). Since 2011 there has been a tendency to gradually transfer all rare diseases treatment coverage from the Ministry of Health to National Health Insurance Fund. However, the Ministry of Health stays a major actor in rare diseases treatment provision through the Ministry of Health-operated Fund for Children’s Treatment and Commission for Treatment Abroad.

The National Plan’s implementation was greatly disrupted by the country’s general economic difficulties and pending healthcare reforms. Nevertheless, individual efforts from medical professionals, university clinics, patient groups and association have greatly contributed for the overall progress of rare diseases issues in Bulgaria. Many new rare diseases activities have been launched by different stakeholders, an excellent rare disease network has been established within the country and rare diseases awareness has been significantly improved. Funded by the National Plan, national newborn screening programmes have been stabilised and improved, which has positively affected the start age of therapy in newborns picked up by mass and selective screening programs, diagnosis and therapy have also moved forward. There is now more mutual confidence and close collaboration among different groups, as well as more active “crosstalk” regarding rare diseases policy (especially rare diseases centres of expertise).

In addition to these measures, the First National Conference for Rare Diseases in Bulgaria (28 to 30 May 2010), organised within the scope of the EUROPLAN project, brought together stakeholders in order to discuss the provisions of the plan and its implementation. The conference participants agreed on the following general proposals and guidelines for actions at national level: to fully support of the priorities set out in the EU Council Recommendation on an action in the field of rare diseases adopted on 8 June 2009; to secure the

1 http://www.conf2010.raredis.org/
implementation of the Bulgarian National Programme for rare diseases with the appropriate funds as defined in the budget framework; to stress the need for urgent legislative initiatives to protect the rights of people with rare diseases and to ensure the adequate prevention, treatment, rehabilitation and social cares; to encourage the establishment of epidemiological registries for rare diseases in Bulgaria; to implement of an integrated approach to people with rare diseases and their families; to organise a public campaign to fund and stimulate research on rare diseases in Bulgaria. The final report of the workshop has been published and is available online for public consultation. Annual rare disease conferences continue to be organised so as to discuss with stakeholders the provisions and advancements of the national plan.

Two major developments concerning the NCCRD and the Bulgarian National Plan took place in 2012. The first one was the Annual National conference for rare diseases, which gathered more than 160 national rare diseases stakeholders to discuss the official designation of rare diseases centres of expertise in Bulgaria. The participation of the Chair of Committee on Healthcare at the Bulgarian Parliament, the Director of the National Health Insurance Fund and leading experts from the Ministry of Health was an encouraging sign for all local rare diseases stakeholders. The second one was that NCCRD board was significantly renewed at the beginning of 2013. Its members are now supposed to take more active role in the EU Cross-Border Healthcare Directive transposition in Bulgaria, especially on the matter of rare diseases and centres of expertise.

Centres of expertise
Currently, there is no official designation procedure for centres of expertise for rare diseases in Bulgaria. The national plan was supposed to carry out a feasibility study on the necessity, possibility and criteria for the creation of a centre of expertise for rare diseases. However, by the end 2012 no such steps have been undertaken.

Nevertheless, there are several academic centres that are specialised as centres of research, treatment and management for rare diseases, i.e. cystic fibrosis, mucopolysaccharidosis, thalassemia major, Gaucher disease and neuromuscular diseases. Treatment with orphan medicinal products is currently reimbursed in these centres, which also manage the provision of very expensive orphan medicinal products. Medical experts from these centres also participate in developing protocols for the National Health Insurance Fund, which serve the treatment of rare disease patients.

Rare diseases centres of expertise for rare diseases were the principle focus of the Bulgarian Third National Conference for Rare Diseases and Orphan Drugs (14-15 September 2012): a panel of experts focused on the EUCERD quality criteria for centres of expertise for rare diseases, with the goal of adopting a set of designation criteria that could be proposed to the Ministry of Health. Given the wide range of rare diseases and problems associated with them, stakeholders agreed that it is difficult to approach uniformly all these issues. However, requirements such as multidisciplinarity, recognised expertise and reputation, networking with other similar national and European structures, cooperation with patient organisations were unanimously supported as criteria for designation of these centres. On the other hand, not all Bulgarian experts believe that the criteria for scientific contribution and participation in clinical trials can be fully met, mainly due to the insufficient human and material resources for these activities in the hospitals here.

In addition to these measures, since May 2009, the Bulgarian Association for Promotion of Education and Science runs highly specialised medical centre for rehabilitation and education of people with rare diseases “RareDis”. The main idea is to upgrade the services of the Information Centre for Rare Diseases and Orphan Drugs, by launching a tertiary-level rehabilitation centre, aimed at improving the quality of life of people with rare diseases.

Registries
The first priority in the National Plan was to provide epidemiological data on rare diseases in Bulgaria through the establishment of a National Registry. The arrangements for the establishment and operation of the registry are within the competence of the Programme’s National Consulting Council for Rare Diseases (NCCRD) within the Ministry of Health. The registry’s tasks include:

- Collecting, summarising, and providing epidemiologic information on the incidence and prevalence of rare diseases in Bulgaria;

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2. Brief report of the 3rd National Conference for Rare Diseases and stakeholders feedback in “Rare Diseases & Orphan Drugs” (October 2012, in both Bulgarian and English) http://raredis.org/pub/Newsletter/Newsletter_12_EN.pdf
- Assisting the preparation of diagnostic and therapeutic protocols and standards for treatment of rare diseases;
- Assisting the Ministry of Health, the Ministry of Labour and Social Policy, and the National Health Insurance Fund in the planning and delivery of expensive treatment and medical care to patients with rare diseases;
- Improving the interaction between health services and patient organisations;
- Providing and publishing data needed for planning and comparison on a regional and national level.

However, the National Registry has still not been established. Still, rare disease stakeholders realise the importance and benefits of registry tools and several registries show the fruit of collaborative efforts. The Bulgarian Second National Conference for Rare Diseases and Orphan Drugs in 2011 was an opportunity to present and discuss these issues. The forum demonstrated a growing interest and motivation for the establishment and implementation of these registries. Consensus was evident among patients and physicians on the need to continue providing support to all the existing epidemiological registries for rare diseases in the country, as an initial step to create the long-expected and so necessary national registry for rare diseases.

By the end 2012 no specific steps for the implementation of this task have been undertaken.

The Bulgarian Information Centre for Rare Diseases and Orphan Drugs (ICRDOD) released a new report in 2012 listing the epidemiological registries for rare diseases in Bulgaria: the report aimed to provide up-to-date and reliable information on the epidemiological registries for rare diseases in the country. Eight nation-wide epidemiological registries concerning rare diseases have been identified by the ICRDOD: the National registry of Patients with Phenylketonuria, the National Registry of Patients with Primary Immunodeficiencies (PID), the National Registry of Patients with Thalassemia Major, the National Registry of Chronic Myeloid Leukaemia Patients, the National Registry of Crohn Disease Patients, the National Registry of Wilson Disease Patients, the National Registry of Gaucher Disease Patients, the National Registry of Mucopolysaccharidosis type 2 Patients. However, this data are not complete and only include registries whose coordinators have provided feedback. The survey will be organised once again in 2013 in order to catch the missing information, as well as the recent developments in this field.

On 28 October 2009, BAPES (Bulgarian Association for the Promotion of Education and Science) was officially given the status of data privacy administrator of rare diseases registries by the Commission for Protection of Data Privacy. Soon after, the collection of epidemiological data for the project “The National registry of thalassaemia major patients in Bulgaria” started. The project is implemented as a result of the common work and cooperation between BAPES, ICRDOD, Medical Centre “RareDis”, Bulgarian Scientific Society of Clinical and Transfusion Haematology, university haematology clinics and thalassaemia patient organisations. A subsequent update and collection of new epidemiological data was organised in March-April 2011, results of Phase III were published in June 2011.

Following this very successful model, BAPES has initiated recently 5 new rare diseases registries. In May 2011 the first results from a joint study of BAPES and Wilson disease patient association were published. The Crohn Disease National Registry is already working and its first statistics were officially adopted in June 2011 at a Crohn Disease national workshop. The Bulgarian Scientific Society of Gastroenterology, Gastrointestinal Endoscopy and Abdominal Echography and the university gastroenterology clinics throughout the country have greatly supported and contributed to both Crohn and Wilson patient registries. Just before the Second National Conference for Rare Diseases in September 2011 the provisional results of two new rare diseases patient registries were announced for Gaucher disease and Mucopolysaccharidosis type 2. The corresponding patient associations have provided data, which were analysed by BAPES. In December 2011 BAPES has reached agreements with the Bulgarian Scientific Society for Clinical and Transfusion Haematology and the Bulgarian Scientific Chirurgic Society for launching two new rare diseases registries for primary myelofibrosis and neuroendocrine tumors respectively. In 2012 epidemiological data for the national thalassemia, chronic myeloid leukemia and Crohn disease registries have been updated, as well as pilot epidemiological studies for myelofibrosis and neuroendocrine tumors have been started. A unique character of all BAPES-managed epidemiological registries for rare diseases is that they involve joint activities by all relevant stakeholders.

The Screening Laboratory of the University Pediatric Hospital in Sofia has also created and is currently maintaining several databases, which are primary sources for analysis and evaluation of neonatal screening programmes in Bulgaria. Several modules are implemented (with no government funding): mass neonatal

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5 http://www.raredis.org/?page_id=2432&mel=4&smel=41&lang=en
6 http://www.raredis.org/?page_id=2447&mel=4&smel=44&lang=en
7 http://www.raredis.org/?page_id=2454&mel=4&smel=43&lang=en
screening registry, primary congenital hypothyroidism, congenital adrenocortical hyperplasia, hyposomatomatropism and Turner syndrome patient registries. There were talks with the Ministry of Health to link the Ministry of Health’s birth registry with the mass neonatal screening registry and to co-work in real time. All the University Pediatric Hospital registries have proved to be very efficient for the respective rare diseases patients’ follow up, as well as medical professional training.

Additionally, some rare tumours are included in the National Cancer Registry, which receives public (governmental) funding.

Bulgaria also contributes to the EUROCARE CF and TREAT-NMD European registries.

**Neonatal screening policies**

One of the national plan’s priorities is to improve the availability and accessibility of the current screening programs. In 1979 mass neonatal screening was introduced in Bulgaria for phenylketonuria, galactosaemia (discontinued in 1993), congenital hypothyroidism and congenital adrenal hyperplasia. Some selective metabolic screening programmes are coordinated by the University Maternity Hospital National Genetic Laboratory in Sofia for the metabolic screening programmes (phenylketonuria), and the University Paediatric Hospital in Sofia for the endocrine screening programmes (congenital hypothyroidism and congenital adrenal hyperplasia). There is logistic coverage of the entire country with more than 130 neonatal structures carrying out blood sampling 3-5 days after birth. Over 90% of neonates are included in existing measures. Ordinance 26 2007 of the Ministry of Health provides equal access to the neonatal screening programmes. However, there exist certain problems, such as postponed mailing of screening cards to centralised labs, and the need for technological upgrades.

The revision and update of the National Medical Genetics Standard (including neonatal screening programmes) started in 2012. It envisages the expansion of the screening panel in accordance with the proposals of screening expert network in Bulgaria. National screening experts even propose to elaborate a separate regulation for neonatal screening, including the designation of the participating laboratories as “centres of expertise” on different sectors (diagnosis, clinical management, consultation, registries, training, etc.). The technological renovation of the centralised screening laboratories, as planned in the National Rare Diseases Plan, has not been carried out yet.

**Genetic testing**

National guidelines for performance of genetic testing in Bulgaria are regulated by the Health Law and the National Medical Genetics Standard. According to the Health Law (Section IV: Genetic health and genetic testing) genetic testing is performed by accredited genetic laboratories. Genetic tests for the diagnosis of rare disorders are provided mainly by the National Genetic Laboratory (NGL). NGL is a national reference laboratory that provides methodological guidance and control over the activities of the genetic laboratories and performs centralised some specialised tests. It was established more than 35 years ago, by initiating of biochemical analysis for some rare disorders and mass neonatal screening for PKU. At the moment NGL provides routine diagnosis with DNA analysis (including prenatal and evaluation of carrier status) for many disorders: cystic fibrosis, phenylketonuria, Wilson disease, neuromuscular disorders, Niemann–Pick (in target population), beta thalassemia, galactokinase deficiency (in target population), microdeletions and microduplications syndromes, inborn hypothyroidism and other. The NGL also has the capacity to perform routine enzymatic analysis and GS/MS analysis for diagnosis of many rare disorders (Krabé, Pompe, MPS). In 2010 the laboratory introduced MS/MS analysis for metabolic study of inherited disorders.

The government organises support of testing by financing diagnostic kits and consumables. Genetic testing abroad is possible for diseases for which the genetic test is not available in Bulgaria, after commission approval. Furthermore, clinical centres, as well as separate research teams have the opportunity through research funded projects by relevant universities and the Ministry of Education’s Research Fund to apply for routine implementation of molecular genetic diagnosis for certain rare diseases. In these cases, patients’ diagnosis is provided free of charge.

Diagnostic tests are registered as available in Bulgaria for 34 genes and an estimated 85 diseases in the Orphanet database.

**National alliances of patient organisations and patient representation**

The National Alliance of People with Rare Diseases (NAPRD) in Bulgaria is an umbrella organisation of around 30 rare disease patient associations and single members with rare diseases not represented by an association.

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8 Information extracted from the Orphanet database in December 2012.
It aims to create a link between the people with rare diseases and the representatives of the social and healthcare system. The Alliance works for the right to timely and equal medical care. The organisation also lobbies for the creation of adequate laws in the field of the protection of the rights of the people with rare diseases.

Public funding is available for nation-wide patient organisations in Bulgaria. Patient representatives are members of the management board of the National Health Insurance Fund, the committee for transparency at the Ministry of Health and the national consultative committee on rare diseases.

Sources of information on rare diseases and national help lines

**Orphanet activities in Bulgaria**

Since 2004 there is a dedicated Orphanet team in Bulgaria, currently hosted by the Information Centre for Rare Diseases and Orphan Drugs (ICRDOD). This team is in charge of collecting data on rare disease related services (specialised clinics, medical laboratories, ongoing research, registries, clinical trials and patient organisations) in their country for entry into the Orphanet database.

**Official information centre for rare diseases**

ICRDOD is a project and activity of the Bulgarian Association for Promotion of Education and Science (BAPES) – a non-government non-profit organisation, registered under the Bulgarian law on non-profit legal in 2003. ICRDOD is a free educational and informative service in Bulgarian and English, providing personalised replies to requests from patients, families and medical professionals. It operates a multilingual website (www.raredis.org) and a rare disease help line - (+359) 32 57 57 97. The ICRDOD also provides a bi-monthly newsletter ("Rare Diseases & Orphan Drugs"): every issue contains a cover story with an interview, news and announcements and a rare diseases reading list. It is electronic and distributed free of charge. The newsletter is published in two versions – Bulgarian (ISSN 1314-3581) and English (ISSN 1314-359X). The ICRDOD also provides an online registry of rare diseases patients, and a Rare Diseases Library in Bulgarian.

In 2010 ICRDOD started publishing reviews on rare diseases topics. These papers’ objective is to summarise important information on particular topics in the field and to present it in a reader-friendly format.

ICRDOD published in September 2012 an updated review of the access to orphan medicinal products for rare diseases in Bulgaria: the report contains 4 sections: orphan medicinal product designation and marketing authorisation; pricing, inclusion in the Positive drug list and reimbursement; mechanisms for accelerated access to innovative medicines; conclusions. There are 2 annexes, attached to the review: list of orphan medicinal products in EU and Bulgaria, which contains information about the trade name, ATC code, active substance, indication(s), marketing authorisation holder and date of marketing authorisation for each item (additionally, it is indicated whether the drug is present in the Positive drug list of Bulgaria and if it is reimbursed by public funds); and a list of references.

The Bulgarian Information Centre for Rare Diseases and Orphan Drugs (ICRDOD) released a new report in 2012 listing the epidemiological registries for rare diseases in Bulgaria: the report aimed to provide up-to-date and reliable information on the epidemiological registries for rare diseases in the country.

The ICRDOD site was substantially renewed and upgraded at the end of 2011. Besides a new look and new layout of content, the site offers several new features for its users, such a subscription for its newsletter

**Help line**

ICRDOD provides a rare disease help line - (+359) 32 57 57 97 providing personalised replies to requests from patients, families and medical professionals. ICRDOD is a member of EURORDIS-led European Network of Rare Diseases Help Lines and took part in the Network’s Caller Profile Analysis 2011 and 2012.

**Other sources of information**

Departments of Medical Genetics at all University Hospitals and the National Genetic Laboratory also provide information.

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9 [http://www.raredis.org/]
10 [http://www.raredis.org/?page_id=2147&mel=8&smel=81&lang=en]
11 [http://www.raredis.org/?page_id=2311&mel=7&smel=71&lang=en]
Good practice guidelines
Several national best practice guidelines are available in Bulgaria, for example the guidelines prepared, adopted and published by the Bulgarian Cancer Society for oncological diseases, including rare tumours, clinical guidelines for Gaucher disease, neuromuscular diseases, thalassemia.

In 2012, rare diseases experts from the University Pediatric Hospital in Sofia took part in the elaboration of “Growth and Endocrine Disorders in Thalassemia: The International Network of Pediatric Endocrinologists in Thalassemia (I-CET) position statement and guidelines”.

Training and education initiatives
The first Eastern European Rare Diseases Summer School14 designed for Russian health authorities and legislative institutions was held on 11-18 September 2011. The second edition of this initiative was once again organised in 201215. The Summer School was a joint initiative of BAPES, the National Association of Rare Diseases Patient Organisations “Genetics” (Russia) and the Italian National Centre for Rare Diseases (CNMR). The event gave the Russian policy and decision makers an opportunity to learn more about rare disease topics and to understand the significant added-value that rare diseases actions and measures bring to the table. The participants came from a wide range of public fields – federal and regional legislative bodies and health authorities, leading medical institutions, academia, patient organisations and the pharmaceutical industry. The week-long event, which covered a variety of topics including centres of expertise, orphan medicinal products, registries, and national plans, was considered a success by attendees, who reported feeling much better informed on the needs of rare disease patients by the end of the training.

Furthermore, BAPES organises each year a two-day rare disease training seminar for medical students. Medical students have the chance to get acknowledged with the main rare diseases concepts, such definition, major problems, important initiatives, etc. The information and education services, offered by ICRDOD and Orphanet were also presented as high quality and reliable source of information, that medical students could use anytime now during their training or further in their future professional practice. Patient representatives took part in the event too, giving personal testimonials and their own point of view on rare diseases issues.

The Screening Laboratory at the University Paediatric Hospital in Sofia also organises a 3-day training workshop for medical professionals each year.

National rare disease events in 2012
In Bulgaria the Rare Disease Day was celebrated through events in Sofia, Plovdiv, Stara Zagora and Pleven, including a release of balloons in solidarity with rare disease patients in the capital. In Sofia and Varna training sessions were held for general practitioners concerning how to relate to people with rare diseases.

The Third Bulgarian National Conference for Rare Diseases and Orphan Drugs16 was held on 14-15 September 2012. 163 participants, including officials from the Parliamentary Health Committee, the Ministry of Health and the National Health Insurance Fund attended the conference. Centres of expertise for rare diseases were the principle focus: a panel of experts focused on the EUCERD quality criteria for centres of expertise for rare diseases, with the goal of adopting a set of designation criteria that could be proposed to the Ministry of Health. Requirements such as multidisciplinarity, recognised expertise and reputation, networking with other similar national and European structures, cooperation with patient organisations were unanimously supported as criteria for designation of these centres. On the other hand, not all experts believed that the criteria for scientific contribution and participation in clinical trials can be fully met, mainly due to the insufficient human and material resources for these activities in the hospitals here. Apart from better care and follow-up of patients, the designation of a medical structure as a centre of expertise for rare diseases will provide new opportunities for the development of the institution, such as increasing knowledge and experience of professionals, and attracting additional external funding through participation in European reference networks and research projects. Logically, both doctors and patients are united behind the need for the status of these centres in Bulgaria to be as fast as possible officialised by the Ministry of Health. A proposal for the development of appropriate regulations will be submitted to the Minister of Health. European and international experience has clearly demonstrated the importance and benefits of such public health institutions: better quality of treatment and care, better organisation of medical services, and more efficient use of funds for rare diseases. The designation of centres of expertise at national level is an important topic considering the steps towards the implementation of the Cross-Border Healthcare Directive.

14 http://raredis.org/pub/Newsletter/Rare_Diseases_Summer_School_2011.pdf
16 http://conf2012.raredis.org/
ICRDOD and NAPRD organised for the first time in Bulgaria a workshop on health technology assessment for rare diseases on 1 November 2012 in Sofia. The event was under the auspices of and hosted by the Healthcare Commission at the 41st National Assembly of Bulgaria. The event aimed to advocate for a better comprehension, access and use of innovative health technologies, including orphan medicinal products.

Many different rare disease-specific training and scientific events were also organised by different stakeholders. For example, several workshops on thalassemia were held (2nd Workshop on thalassaemia major “Multimodal approach in therapy and follow-up”, Summer Academy “Quality of life of patients with β Thalassemia in Bulgaria – focus on endocrine complications”, etc).

Hosted rare disease events in 2012
The Bulgarian Association for Promotion of Education and Science launched and organised in 2005, 2006, 2008 and 2009, the annual "Eastern European Conference on Rare Diseases and Orphan Drugs". In 2010 for the very first time the event was hosted and co-organised outside Bulgaria, in Saint Petersburg, Russia, in conjunction with the first All-Russian Conference for Rare Diseases and Rarely Used Medical Technologies. It has proved to be an efficient strategy to foster rare diseases progress in Eastern Europe. In 2011, the sixth edition of this initiative was held and co-organised in Istanbul, Turkey. The Bulgarian National Alliance of People with Rare Diseases initiated and organised a Balkan patient meeting on 24 March 2012 in Sofia. Leading rare diseases experts and patients from Balkan countries took part in this event. A second edition is expected in 2013.

Research activities and E-Rare partnership

National research activities
In Bulgaria, there is no specific call for rare diseases at the national fund for research, although rare disease related projects can apply. The National Plan does not envisage any official policies to stimulate research on rare diseases; it only envisages encouraging partnerships.

Participation in European research projects
Bulgaria participates in European rare disease research projects, including: RAREBESTPRACTICES, EUROGLYCANET and TREAT-NMD.

E-Rare
Bulgaria is not currently a partner of E-Rare.

IRDiRC
Bulgarian funding agencies have not yet committed national funding to the IRDiRC.

Orphan medicinal products
ICRDOD issued an updated report in September 2012 reviewing access to medicines for rare diseases in Bulgaria. The report contains information on important orphan medicinal products activities and explained how they are set up in Bulgaria in 4 sections: orphan medicinal product designation and marketing authorisation; pricing, inclusion in the Positive Drug List (PDL) and reimbursement; mechanisms for accelerated access to innovative medicines; and conclusions.

Orphan medicinal product committee
There is currently no orphan medicinal products specialised committee in Bulgaria. Orphan medicinal products are subject as are all other medicinal products to the Commissions on the pricing of medicines and on the Positive drug list. In order to optimise these procedures, in 2011 the two commissions were merged into a single one under the Ministry of Health. Subsequently, the Parliament adopted in 2012 new legislation, by which a National Council for Pricing and Reimbursement of Medicinal Products would replace the Commission at the beginning of 2013. The new body would have a status of a State agency and would be responsible for both pricing and reimbursement of all medicinal products. The headquarters would be in Pleven, rather than Sofia. Medicinal products’ safety will continue to be monitored by the Executive Drugs Agency.

Orphan medicinal product incentives
No specific activity reported.

Orphan medicinal product market availability situation

Orphan drugs availability in Bulgaria and patients’ access to them is regulated on national level by two legal acts – Ordinance on the pricing and inclusion of medicinal products in the Positive Drug List (PDL) (adopted by Decree 340 of the Council of Ministers) and Ordinance 38 of the Minister of Health (on the list of diseases, whose outpatient treatment is covered by the National Health Insurance Fund, NHIF). Orphan drugs have a centralised market authorisation by EMA, so they are all formally registered in Bulgaria. However, to be accessible, they have to be priced and included in the PDL, as well as their indicated medical condition has to be in the NHIF list of diseases according to Ordinance 38.

By the end of 2012, 24 orphan medicinal products with EMA market authorisation are priced and included in the PDL. 15 of them are reimbursed by NHIF and 9 – by the respective hospital budget. These include: Atriance, Elaprase, Evoltra, Exjade, Fabrazyme, Glivec, Litak, Lysodren, Mozobil, Nexavar, Nplate, Revatio, Revolade, Somavert, Sprycel, Tasigna, Torisel, Tracleer, Tracleer, Trąvevis, Votubia, Xagrid, Yondelis, Zavesca.

Despite the recent increase of these figures, institutions dealing with planning and funding for treatment and rehabilitation of patients with rare diseases still do not have actual and reliable data on the number and distribution of patients in the country and information on the compliance and effectiveness of this expensive treatment. A recent cross-sectional observational study appearing in the journal *Health Policy* evaluated the conditions that impact orphan drug availability in Bulgaria and other Eastern European countries. The authors observe that in Bulgaria: “All the other orphan drugs, being not reimbursed, are practically inaccessible for the individual patients because of their high price. Despite the increasing number of orphan drugs, which are being reimbursed in Bulgaria, in reality there is no significant improvement of the accessibility of treatment for rare diseases. Neither a national epidemiological registry nor expert centres for rare diseases exist. Regulation of alternative access to orphan drugs (e.g., compassionate use, off-label use) is also missing. In these conditions many patients are left without correct diagnosis, adequate treatment, follow-up and rehabilitation.” The system for determining pricing is proving detrimental to orphan drug availability. Bulgaria uses a history-based budget through which funds for orphan drug treatments are allocated annually based on the previous year’s calculated needs. Thus if the population of newly diagnosed patients grows, medicinal treatment shortages and access limitations also increase. The inclusion of new drugs in the country’s reimbursement scheme results from improved awareness for rare diseases among physicians and patient associations, as well as the intentions of pharmaceutical manufacturers to market their products in Bulgaria. The authors outline the flaws with the current EU pricing system: “In Bulgaria, the mechanism of smallest value from a set of international reference prices reduces the opportunities for flexible solutions. The inclusion of reference countries with floating euro exchange rate makes the pricing of orphan drugs dependent on the macroeconomic indicators in these countries and thereby allowing serious fluctuations of the price. This is a specific reason for the reluctance of some companies to register prices of their orphan drugs in the small Eastern European non-Eurozone countries. Financial difficulties in one country may lead to lower prices in all referring ones. It should be also underlined that Member State authorities have little negotiating leverage since these medicines have no therapeutic alternative”. Another important element that negatively impacts small countries is a lack of clinical data around the cost-effectiveness of rare disease medicinal products. Epidemiological rare disease registries are needed to improve knowledge in this area. Thus the authors call for a reformed orphan drug policy-making process that is transparent, and based on a convergence of medical, economic, ethical and social elements.

Orphan medicinal product pricing policy

There is no specific orphan medicinal product pricing policy and orphan medicinal products are subject to the general conditions as any other medicaments. The negotiation of price and level of reimbursement of orphan medicinal products in Bulgaria is determined by the Ordinance on the pricing and inclusion of medicinal products in the Positive Drug List (PDL) (adopted by Decree 340 of the Council of Ministers), and it is based on reference pricing, using data from Romania, France, Estonia, Greece, Slovakia, Lithuania, Portugal, Italy, Finland, Denmark, Slovenia, Spain, Belgium, Czech Republic, Poland, Latvia and Hungary.

Orphan medicinal product reimbursement policy

There is no specific orphan medicinal product reimbursement policy and orphan medicinal products are subject to the general conditions as any other medicaments. Medicinal products’ reimbursement is regulated on national level by two legal acts – Ordinance on the pricing and inclusion of medicinal products in the Positive

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Drug List (PDL) (adopted by Decree 340 of the Council of Ministers) and Ordinance 38 of the Minister of Health (on the list of diseases, whose outpatient treatment is covered by the National Health Insurance Fund, NHIF). To be accessible, orphan drugs have to be priced and included in PDL, as well as their indicated medical condition has to be in the NHIF list of diseases according to Ordinance 38.

**Other initiatives to improve access to orphan medicinal products**
No information reported.

**Other therapies for rare diseases**
No information reported.

**Orphan devices**
No information reported.

**Specialised social services**
Respite care services and therapeutic recreational programmes are provided in certain medical centres in Bulgaria and are partially reimbursed by the National Health Insurance Fund.

In Bulgaria, there are currently no specialised programmes for people for rare diseases: these patients are forced to seek alternatives in the existing general schemes for the rehabilitation and integration of people with disabilities which do not often meet European standards and recommendations in the area. They are unevenly distributed across the country and public awareness of these services is low. In addition, rare disease patients may be denied access as the Territorial Expert Medical Commission’s legislation is not adapted to the specificities of rare disease. As such, one of the priorities of the National Plan is to work on an integrative approach and specialised programmes for physical and social rehabilitation of rare disease patients, however no progress has been made to date, and patients feel that specialised services for rare diseases should not be separated or be in opposition to current programmes for people with disabilities.

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**DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2012 IN BULGARIA**

**National plan/strategy for rare diseases and related actions**
On 27 November 2008, the Bulgarian Council of Ministers approved the National Plan for Rare Diseases – genetic disorders, congenital malformations and nonhereditary diseases (2009 – 2013). The Bulgarian National Plan for Rare Diseases started on 1 January 2009 and will last for 5 years.

The National Plan’s implementation was greatly disrupted by the country’s general economic difficulties and pending healthcare reforms. Nevertheless, individual efforts from medical professionals, university clinics, patient groups and association have greatly contributed for the overall progress of rare diseases issues in Bulgaria. Many new rare diseases activities have been launched by different stakeholders, an excellent rare disease network has been established within the country and rare diseases awareness has been significantly improved. Funded by the National Plan, national newborn screening programmes have been stabilised and improved, which has positively affected the start age of therapy in newborns picked up by mass and selective screening programs, diagnosis and therapy have also moved forward. There is now more mutual confidence and close collaboration among different groups, as well as more active “crosstalk” regarding rare diseases policy (especially rare diseases centres of expertise). Annual rare disease conferences continue to be organised so as to discuss with stakeholders the provisions and advancements of the national plan.

Two major developments concerning the NCCRD and the Bulgarian National Plan took place in 2012. The first one was the Annual National conference for rare diseases in September 2012, which gathered more than 160 national rare diseases stakeholders to discuss the official designation of rare diseases centres of excellence.

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20 Brief report of the 3rd National Conference for Rare Diseases and stakeholders feedback in “Rare Diseases & Orphan Drugs” (October 2012, in both Bulgarian and English) http://raredis.org/pub/Newsletter/Newsletter_12_EN.pdf
expertise in Bulgaria. The participation of the Chair of Committee on Healthcare at the Bulgarian Parliament (Dr. Daniela Daritkova), the Director of the National Health Insurance Fund (Dr. Plamen Tsekov) and leading experts from the Ministry of Health was an encouraging sign for all local rare diseases stakeholders. The second one was that NCCRD board was significantly renewed at the beginning of 2013. Its members are now supposed to take more active role in the EU Cross-Border Healthcare Directive transposition in Bulgaria, especially on the matter of rare diseases and centres of expertise.

Centres of expertise
The national plan was supposed to carry out a feasibility study on the necessity, possibility and criteria for the creation of a centre of expertise for rare diseases. However, by the end 2012 no such steps have been undertaken.

Nevertheless, rare diseases centres of expertise for rare diseases were the principle focus of the Bulgarian Third National Conference for Rare Diseases and Orphan Drugs (14-15 September 2012): a panel of experts focused on the EUCERD quality criteria for centres of expertise for rare diseases, with the goal of adopting a set of designation criteria that could be proposed to the Ministry of Health. Given the wide range of rare diseases and problems associated with them, stakeholders agreed that it is difficult to approach uniformly all these issues. However, requirements such as multidisciplinarity, recognised expertise and reputation, networking with other similar national and European structures, cooperation with patient organisations were unanimously supported as criteria for designation of these centres. On the other hand, not all Bulgarian experts believe that the criteria for scientific contribution and participation in clinical trials can be fully met, mainly due to the insufficient human and material resources for these activities in the hospitals here.

Registries
The first priority in the National Plan was to provide epidemiological data on rare diseases in Bulgaria through the establishment of a National Registry. By the end 2012 no specific steps for the implementation of this task have been undertaken.

The Bulgarian Information Centre for Rare Diseases and Orphan Drugs (ICRDOD) released a new report21 in 2012 listing the epidemiological registries for rare diseases in Bulgaria: the report aimed to provide up-to-date and reliable information on the epidemiological registries for rare diseases in the country. Eight nation-wide epidemiological registries concerning rare diseases have been identified by the ICRDOD: the National registry of Patients with Phenylketonuria, the National Registry of Patients with Primary Immunodeficiencies (PID), the National Registry of Patients with Thalassemia Major, the National Registry of Chronic Myeloid Leukaemia Patients, the National Registry of Crohn Disease Patients, the National Registry of Wilson Disease Patients, the National Registry of Gaucher Disease Patients, the National Registry of Mucopolysaccharidosis type 2 Patients. However, this data are not complete and only include registries whose coordinators have provided feedback. The survey will be organised once again in 2013 in order to catch the missing information, as well as the recent developments in this field.

On 28 October 2009, was In 2012 epidemiological data for the national thalassemia, chronic myeloid leukemia and Crohn disease registries have been updated by BAPES (Bulgarian Association for the Promotion of Education and Science), as well as pilot epidemiological studies for myelofibrosis and neuroendocrine tumors have been started.

The Screening Laboratory of the University Pediatric Hospital in Sofia has also created and is currently maintaining several databases, which are primary sources for analysis and evaluation of neonatal screening programmes in Bulgaria. Several modules are implemented (with no government funding): mass neonatal screening registry, primary congenital hypothyroidism, congenital adrenocortical hyperplasia, hyposomatotropism and Turner syndrome patient registries. There were talks with the Ministry of Health to link the Ministry of Health’s birth registry with the mass neonatal screening registry and to co-work in real time. All the University Pediatric Hospital registries have proved to be very efficient for the respective rare diseases patients’ follow up, as well as medical professional training.

Neonatal screening policies
The revision and update of the National Medical Genetics Standard (including neonatal screening programmes) started in 2012. It envisages the expansion of the screening panel in accordance with the proposals of screening expert network in Bulgaria. National screening experts even propose to elaborate a separate regulation for neonatal screening, including the designation of the participating laboratories as “centres of

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expertise” on different sectors (diagnosis, clinical management, consultation, registries, training, etc.). The technological renovation of the centralised screening laboratories, as planned in the National Rare Diseases Plan, has not been carried out yet.

Sources of information on rare diseases and national help lines

Official information centre for rare diseases
ICRDOO published in September 2012 an updated review of the access to orphan medicinal products for rare diseases in Bulgaria22: the report contains 4 sections: orphan medicinal product designation and marketing authorisation; pricing, inclusion in the Positive drug list and reimbursement; mechanisms for accelerated access to innovative medicines; conclusions. There are 2 annexes, attached to the review: list of orphan medicinal products in EU and Bulgaria, which contains information about the trade name, ATC code, active substance, indication(s), marketing authorisation holder and date of marketing authorisation for each item (additionally, it is indicated whether the drug is present in the Positive drug list of Bulgaria and if it is reimbursed by public funds); and a list of references.

The Bulgarian Information Centre for Rare Diseases and Orphan Drugs (ICRDOO) released a new report in 201223 listing the epidemiological registries for rare diseases in Bulgaria: the report aimed to provide up-to-date and reliable information on the epidemiological registries for rare diseases in the country.

Help line
ICRDOO is a member of EURORDIS-led European Network of Rare Diseases Help Lines and took part in the Network’s Caller Profile Analysis in 2012.

Good practice guidelines
In 2012, rare diseases experts from the University Pediatric Hospital in Sofia took part in the elaboration of “Growth and Endocrine Disorders in Thalassemia: The International Network of Pediatric Endocrinologists in Thalassemia (I-CET) position statement and guidelines”.

Training and education initiatives
The second edition of the Eastern European Rare Diseases Summer School24 designed for Russian health authorities and legislative institutions was in 201225. The Summer School was a joint initiative of BAPES, the National Association of Rare Diseases Patient Organisations “Genetics” (Russia) and the Italian National Centre for Rare Diseases (CNMR). The event gave the Russian policy and decision makers an opportunity to learn more about rare disease topics and to understand the significant added-value that rare diseases actions and measures bring to the table. The participants came from a wide range of public fields – federal and regional legislative bodies and health authorities, leading medical institutions, academia, patient organisations and the pharmaceutical industry. The week-long event, which covered a variety of topics including centres of expertise, orphan medicinal products, registries, and national plans, was considered a success by attendees, who reported feeling much better informed on the needs of rare disease patients by the end of the training.

Furthermore, BAPES organises each year a two-day rare disease training seminar for medical students. Medical students have the chance to get acknowledged with the main rare diseases concepts, such definition, major problems, important initiatives, etc. The information and education services, offered by ICRDOO and Orphanet were also presented as high quality and reliable source of information, that medical students could use anytime now during their training or further in their future professional practice. Patient representatives took part in the event too, giving personal testimonials and their own point of view on rare diseases issues.

The Screening Laboratory at the University Pediatric Hospital in Sofia also organises a 3-day training workshop for medical professionals each year.

National rare disease events in 2012
In Bulgaria the Rare Disease Day was celebrated through events in Sofia, Plovdiv, Stara Zagora and Pleven, including a release of balloons in solidarity with rare disease patients in the capital. In Sofia and Varna training sessions were held for general practitioners concerning how to relate to people with rare diseases.

The Third Bulgarian National Conference for Rare Diseases and Orphan Drugs was held on 14-15 September 2012. 163 participants, including officials from the Parliamentary Health Committee, the Ministry of Health and the National Health Insurance Fund attended the conference. Centres of expertise for rare diseases were the principle focus: a panel of experts focused on the EUCERD quality criteria for centres of expertise for rare diseases, with the goal of adopting a set of designation criteria that could be proposed to the Ministry of Health. Requirements such as multidisciplinarity, recognised expertise and reputation, networking with other similar national and European structures, cooperation with patient organisations were unanimously supported as criteria for designation of these centres. On the other hand, not all experts believed that the criteria for scientific contribution and participation in clinical trials can be fully met, mainly due to the insufficient human and material resources for these activities in the hospitals here. Apart from better care and follow-up of patients, the designation of a medical structure as a centre of expertise for rare diseases will provide new opportunities for the development of the institution, such as increasing knowledge and experience of professionals, and attracting additional external funding through participation in European reference networks and research projects. Logically, both doctors and patients are united behind the need for the status of these centres in Bulgaria to be as fast as possible officialised by the Ministry of Health. A proposal for the development of appropriate regulations will be submitted to the Minister of Health. European and international experience has clearly demonstrated the importance and benefits of such public health institutions: better quality of treatment and care, better organisation of medical services, and more efficient use of funds for rare diseases. The designation of centres of expertise at national level is an important topic considering the steps towards the implementation of the Cross-Border Healthcare Directive.

ICRDOD and NAPRD organised for the first time in Bulgaria a workshop on health technology assessment for rare diseases on 1 November 2012 in Sofia. The event was under the auspices of and hosted by the Healthcare Commission at the 41st National Assembly of Bulgaria. The event aimed to advocate for a better comprehension, access and use of innovative health technologies, including orphan medicinal products. Many different rare disease-specific training and scientific events were also organised by different stakeholders. For example, several workshops on thalassemia were held (2nd Workshop on thalassemia major “Multimodal approach in therapy and follow-up”, Summer Academy “Quality of life of patients with β Thalassemia in Bulgaria – focus on endocrine complications”, etc).

Hosted rare disease events in 2012

The Bulgarian Association for Promotion of Education and Science launched and organised in 2005, 2006, 2008 and 2009, the annual “Eastern European Conference on Rare Diseases and Orphan Drugs”. In 2010 for the very first time the event was hosted and co-organised outside Bulgaria, in Saint Petersburg, Russia, in conjunction with the first All-Russian Conference for Rare Diseases and Rarely Used Medical Technologies. It has proved to be an efficient strategy to foster rare diseases progress in Eastern Europe. In 2011, the sixth edition of this initiative was held and co-organised in Istanbul, Turkey. The Bulgarian National Alliance of People with Rare Diseases initiated and organised a Balkan patient meeting on 24 March 2012 in Sofia. Leading rare diseases experts and patients from Balkan countries took part in this event. A second edition is expected in 2013.

Orphan medicinal products

ICRDOD issued an updated report in September 2012 reviewing access to medicines for rare diseases in Bulgaria. The report contains information on important orphan medicinal products activities and explained how they are set up in Bulgaria in 4 sections: orphan medicinal product designation and marketing authorisation; pricing, inclusion in the Positive Drug List (PDL) and reimbursement; mechanisms for accelerated access to innovative medicines; and conclusions.

Orphan medicinal product committee

Orphan medicinal products are subject as are all other medicinal products to the Commissions on the pricing of medicines and on the Positive drug list. In order to optimise these procedures, in 2011 the two commissions were merged into a single one under the Ministry of Health. Subsequently, the Parliament adopted in 2012 new legislation, by which a National Council for Pricing and Reimbursement of Medicinal Products would replace the Commission at the beginning of 2013. The new body would have a status of a State agency and would be responsible for both pricing and reimbursement of all medicinal products. The headquarters would
be in Pleven, rather than Sofia. Medicinal products’ safety will continue to be monitored by the Executive Drugs Agency.

**Orphan medicinal product market availability situation**

By the end of 2012, 24 orphan medicinal products with EMA market authorisation are priced and included in the PDL. 15 of them are reimbursed by NHIF and 9 – by the respective hospital budget. These include: Atriance, Elaprase, Evoltra, Exjade, Fabrazyme, Glivec, Litak, Lysodren, Mozobil, Nexavar, Nplate, Revatio, Revolade, Somavert, Sprycel, Tasigna, Torisel, Tracleer, Ventavis, Volibris, Votubia, Xagrip, Yondelis, Zavesca.
LIST OF CONTRIBUTIONS

Contributions in 2010
Rumen Stefanov and Ralitza Jordanova (Orphanet Bulgaria, Bulgarian Association for Promotion of Education and Science BAPES – Information Centre for Rare Diseases and Orphan Drugs)
Tsonka Miteva (Bulgarian Association for Promotion of Education and Science BAPES – Information Centre for Rare Diseases and Orphan Drugs)
Vladimir Tomov (President of the National Alliance of People with Rare Diseases)

Contributions in 2011
Georgi Iskrov, Tsonka Miteva (Bulgarian Association for Promotion of Education and Science BAPES – Information Centre for Rare Diseases and Orphan Drugs)
Rumen Stefanov (Orphanet Bulgaria, Bulgarian Association for Promotion of Education and Science BAPES – Information Centre for Rare Diseases and Orphan Drugs)
Radka Tincheva (EUCERD Representative Bulgaria, Chair of the National Advisory Council on Rare Diseases, University Paediatric Hospital Sofia)
Alexey Savov (University Hospital of Obstetrics and Gynecology, Sofia)
Iva Stoeva (University Pediatric Hospital – Endocrine screening programme)

Contributions in 2012
Radka Tincheva (EUCERD Representative Bulgaria, Chair of the National Advisory Council on Rare Diseases, Coordinator of the National Plan for Rare Diseases, University Paediatric Hospital Sofia)
Rumen Stefanov (Orphanet Bulgaria, Bulgarian Association for Promotion of Education and Science BAPES – Information Centre for Rare Diseases and Orphan Drugs)
Vladimir Tomov (President of the National Alliance of People with Rare Diseases)
Alexey Savov (University Hospital of Obstetrics and Gynecology, Sofia)
Iva Stoeva (University Pediatric Hospital – Endocrine screening programme)
Georgi Iskrov, Tsonka Miteva (Bulgarian Association for Promotion of Education and Science BAPES – Information Centre for Rare Diseases and Orphan Drugs)

Contributions in 2013
Radka Tincheva (Chair of the National Consulting Council on Rare Diseases, University Paediatric Hospital Sofia)
Rumen Stefanov (IRDRC Interdisciplinary Committee Member, Orphanet Bulgaria, Dean of the Faculty of Public Health, Medical University of Plovdiv, Bulgarian Association for Promotion of Education and Science BAPES – Information Centre for Rare Diseases and Orphan Drugs)
Iva Stoeva (University Pediatric Hospital – Endocrine screening programme)
Irena Bradinova (EMA Committee for Orphan Medicinal Products Representative Bulgaria, National Genetic Laboratory)
Georgi Iskrov (Department of Social Medicine and Public Health, Medical University of Plovdiv, Bulgarian Association for Promotion of Education and Science BAPES – Information Centre for Rare Diseases and Orphan Drugs)
Tsonka Miteva-Katrandzhieva (Department of Social Medicine and Public Health, Medical University of Plovdiv; Bulgarian Association for Promotion of Education and Science BAPES – Information Centre for Rare Diseases and Orphan Drugs)

Validated by: Radka Tincheva (EUCERD Representative for Bulgaria, Chair of the National Consulting Council on Rare Diseases, University Paediatric Hospital Sofia)

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29 The contributors and validators of the report have contributed information which is accurate to the best of their knowledge. However, readers should take note that the contents of this report are illustrative and not exhaustive.

30 All websites and documents were last accessed in May 2013.
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