



RARE DISEASES ORPHAN DRUGS

Official Newsletter of the Bulgarian Association for Promotion of Education and Science (BAPES)

Dear friends and colleagues,

We are pleased to present you the first issue of our official newsletter. Another piece of our joint cause – the rare diseases.

For almost nine years we, the BAPES team, have been systematic and purposefully working through the Information Centre for Rare Diseases and Orphan Drugs (ICRDOD) and the Specialized Medical Centre “RareDis” for implementation of available, affordable and quality information, prevention, medical treatment, rehabilitation and social integration of people with rare diseases in Bulgaria and the region, realization of research projects and European cooperation, strengthening the NGO sector and assuring dialogue between all the field stakeholders.

Information is a powerful weapon. Particularly for the rare diseases field the lack of awareness and responsibility is a serious obstacle to solving the accumulated problems. That's why we work and will be working even harder to make reliable and adequate information available to all, because only informed decisions can help to move forward!

TOPIC OF THE ISSUE:

RARE DISEASES IN BULGARIA – ACTUAL STATE, POLICIES AND TENDENCIES

Despite initial skepticism of many people, we have come a long way. Our success is not only measured by the number of won and implemented projects, but also by the confidence and support that patients and their families, medical professionals and students, representatives of health authorities and pharmaceutical industries have given to us. Thank you!





ON YESTERDAY, TODAY AND TOMORROW

Information Centre for Rare Diseases and Orphan Drugs is pleased to present the book "On Yesterday, Today and Tomorrow". It has been funded by ICRDOD and contains the stories of 9 Bulgarian patients with rare diseases and their families. They describe their fears and hopes, their daily struggle to change the status quo. ICRDOD would like to thank the co-authors Maria Kancheva, Yordanka Petkova, Svetlana Atanasova, Elena Dimova, Desislava Hristakeva, Aneta Feodorova, Albena Dimitrova, Elena Eneva, Tsvetelina Yurukova.

Read more on page 4

DONATION FROM MC "RAREDIS"

Medical Center "RareDis" has made a donation of 12 000 BGN to the National Alliance of People with Rare Diseases (NAPRD) during the charity campaign "Be in Solidarity, Give Happiness!". The donation contains 10 individual vouchers, non-cash usable by members of NAPRD at Medical Center "RareDis".



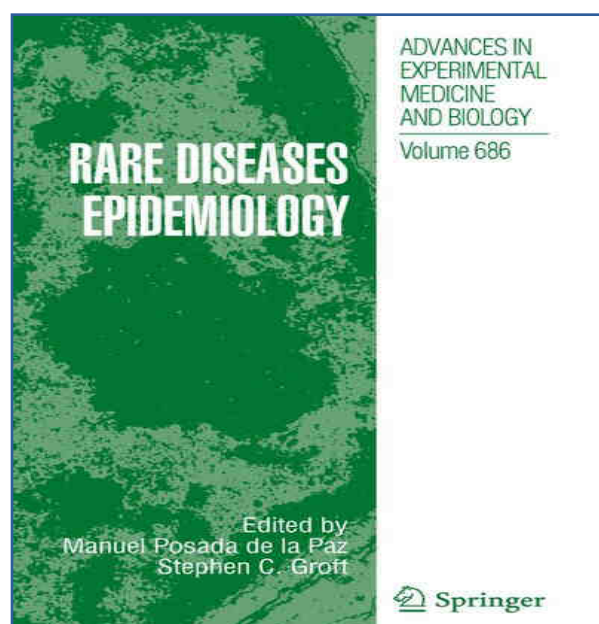
ROLE OF PATIENT ASSOCIATIONS

Roundtable discussions on the patient organizations' role as a factor in health policy making were held on 29 October in Plovdiv at the District Administration Office.

It was organized by the National Alliance of People with Rare Diseases, Bulgarian Association for Promotion of Education and Science and Italian-Bulgarian Foundation – Plovdiv. Hon. Mariella Bocciardo was a special guest to the event. Ms. Bocciardo is a member of the Italian Parliament and has a long experience in promoting patient empowerment.

RARE DISEASES EPIDEMIOLOGY

The newly published book offers numerous methods and approaches from the collective experiences of established research investigators who address significant issues, such as development of patient registries; collection, storage and selected distribution of bio-specimens from bio-banking activities; validation and utilization of genetic testing and newborn screening procedures; presentation of issues related to the importance of case reports to increase knowledge of rare diseases; challenges and models for population-based surveillance studies for rare congenital and inherited disorders; statistical methods for the geographical analyses of rare diseases; value and need for clinical trials and comparative effective studies; meeting the requirements of regulatory agencies. An expert from ICRDOD has taken part in the preparation of Chapter 26 of the book, "National Plans and Strategies on Rare Diseases in Europe".





ANETA AND SVETLANA –

NOMINATED FOR PERSON OF THE YEAR AWARD

Aneta Feodorova and Svetlana Atanasova from the Bulgarian Cystic Fibrosis Association have been nominated for the Person of the Year Award, Bulgarian Helsinki Committee's annual award for contribution to the human rights. For years, Aneta and Svetla have been fighting for the right of people with cystic fibrosis to adequate and quality treatment, physical rehabilitation and social integration. More than 1 200 people have supported their nomination, including Cystic Fibrosis Europe President – Ms. Karleen De Rijcke.

TRAINING SEMINAR FOR PEOPLE WITH RARE DISEASES

National alliance of people with rare diseases is organizing a training workshop "Psychological Methods - Way of Self-help to Improve Quality of Life of People with Rare Diseases and Their Families". Lecturers will be the prominent psychotherapist Ms. Madeleine Algafari and the psychologist Elenko Angelov. Participants will have the opportunity to take valuable advices, to discover the possibilities of the positive thinking and how best to use it. The seminar will take place on 12 February 2011 in Plovdiv. Participation is free.

I KNOW, I CAN, I SUCCEED

A training workshop for people with rare diseases will be held on 1-7 May 2011 in Veliko Tarnovo, Bulgaria. "I know, I can, I succeed" seminar will be focused on personal development, looking for a job and successful presentation in front of employers. Professional psychologists, including Mr. Laurent Gounelle from France will be lecturers. The event is funded by the European Grundtvig Workshops Programme and all the participants' expenses will be covered under it.

SECOND NATIONAL CONFERENCE FOR RARE DISEASES

The Second National Conference on Rare Diseases and Orphan Drugs will be held on 9-11 September 2011 in Plovdiv, Bulgaria. It will take place at the Congress centre of NOVOTEL – Plovdiv. The forum will include presentations by leading medical experts, workshops and discussions for all rare diseases stakeholders, poster session. Assessment of the Bulgarian National Programme for Rare Diseases implementation by EUROPLAN indicators and recommendations and analysis of the results from the epidemiological registries for rare diseases in Bulgaria will be among conference highlights.

MORE ON PERSONALIZED HEALTHCARE

A "FAQ on Personalised Healthcare" brochure has been added to ICRDOD's website F.A.Q. section. The material has been prepared by the European Genetic Alliances Network (EGAN) and translated in Bulgarian by the ICRDOD team.

As research advances, it is clear that a standard approach to treatment of cancer, heart diseases and other common physical and psychological disorders will not yield the results patients need. Increasingly therapies are being selected based on an understanding of the underlying genetic components of a condition and how these interact. Personalised healthcare is a conception, which when properly applied and adjunct to the clinical skills of doctors, will open up new opportunities for preventing, treating or curing many currently intractable diseases. It is an area where there has been much hype (both positive and negative) about possibilities for progress.



ON YESTERDAY, TODAY AND TOMORROW – 9 AUTHENTIC STORIES

At the end of October ICRDOD together with 9 patients with rare diseases prepared a booklet, called "On Yesterday, Today and Tomorrow". Despite being small in size and volume, it is bearing the huge emotional charge not only for the nine authors and their families, but also for all people with rare diseases in Bulgaria!

Although each unhappy family is unhappy in its own way, the fates of people with rare diseases are so similar! These are the heroes of our time, because of the dignity with which they are carrying their misery and the giant will, with which they are trying to overcome what seems impossible. These people deserve great respect!

The fact that nobody knows, nobody speaks and nobody deals with them has been always describing the rare diseases' field. Not the lack of funds, but the apathy and indifference are the main problem. We sincerely hope that this booklet will help for the huge mind change which our society so much needs!



Here, we present some selected texts from the stories. The book "On Yesterday, Today and Tomorrow" is distributed free of charge and you could download it from our website (www.raredis.org).

"... All these facts have determined the direction of my life – half went in hospitals and the other in non-stop fear and stress. This reflected not only on me but also on the others. Our society is deeply negative to the disabled people. Most people daily reaffirmed my sense of guilt that I was an obstacle in their lives and I should be removed. We want to be among you and live out here with dignity and head held high against the fate."

"... Silence and ignorance are the most scary. Sparse words like "I hope it is not that", "until the results come out, do not worry", and others. And you know nothing. What is that "this"? Examination results begin to arrive one after another. Reading, but understand nothing."

"...When the diagnosis was already confirmed, they gave me a videotape and sent me to the oncology ward upstairs to watch it alone. What irony! Perhaps they wanted to say that there were other sick children who were also suffering? Maybe by lying on somebody's pain, you will accept it easier? I do not know! A videotape that said such terrible things that I could not even cry anymore. I stood like frozen there in that room and I could not move. Then went back down to my little baby, taking a camera and starting to shoot, shoot, because I thought it would be the only thing that will remain behind. And in the same time I was saying that it is not true, I deny it and I won't accept it."

"... But I smile and keep telling, because my grown-up child is up to me and absorbs every word. And I reassure him that it can be handled with the problems, insist on inhalation and monitor the drugs. Sometimes it's hard, but I know this is the only way and I should not sit back."

"... By the third day after birth our little boy already had growing blisters on the elbows from the diaper. There were wounds in the mouth. Worried that he could not suck, I called the national consultant for EB, whose number a friend of mine had found. The first thing she said was: "You sound very calm for a mother of a child with such a diagnosis."



"... The hospital staff got familiar with us, they helped me with bandages because it was already impossible for one person to handle. They tried to provide what was in their power, were nice to me, answered my questions, discussed what could be done. I think they learned a lot from the condition of my child, saw some advanced materials that are not normally used in hospitals, understood that the parent is 24 hours with the child and his opinion must also be taken into consideration in order to make the treatment individualized and the most friendly as possible."

"... And although he can not move his hands, although he spends every day in different therapies, although he grows up in hospitals and have more time with doctors than with his peers, his face is always smiling! Yes, he is so joyful and positive! This is exactly what gives me strength to continue to fight and not to listen to remarks of doctors such as: "Well, you're young, you can have another child!". It is severe for a parent to accept and live with the thought that his child is different and will never be like the others! It is difficult to survive in our society where there is no place for sick and different people!"

"... If you meet me on the street I will just seem strange, but you will never understand. If we work together, you'll notice some particularities of my character, but you'll never assume. If we have fun together, you will find my very black humor, but you'll never see through. Because despite everything, I'm just one like you. Plain, young woman, dreaming to be a princess and have a personal hero. But I believe more in the goodness of ordinary people and that nothing is impossible!"

"...We waited the response three months, during which he was three times admitted to the intensive care ward. Finally we were told that because there is no clinical pathway for this rare disease in Bulgaria, we can not be sent for treatment abroad. Absolute paragraph 22 – because they do not treat here EB, they will not let us go where EB is treated. Several days later, our young son died.

I am grateful to the hundreds of Bulgarians who supported and helped us. I am grateful to the medical staff who did whatever was in their power. For the State the problems of my child did not exist any more."

"... I want to believe that someday our society will wake up and overpower the "little" things in life - houses, cars and luxuries, and will focus on really important issues. And eventually it will give hand to the patients with rare diseases."

RARE DISEASES IN BULGARIA: ACTUAL STATE, POLICIES AND TENDANCIES

ICRDOD announces the publication of the next of its reviews, now on the actual situation and tendencies in the rare diseases field in Bulgaria. It contains 5 main chapters: methodology and governance of the Bulgarian National plan for rare diseases (2009 – 2013); definition, codification and inventorying of rare diseases; research on rare diseases; centres of expertise, reference networks and access to orphan drugs; patient empowerment.

Our analysis is based on three main documents – EU Council recommendation on action in the field of rare diseases, EUROPLAN recommendations and the Bulgarian National plan for rare diseases. The comments and suggestions, which came from the participants of the EUROPLAN Bulgarian National Conference for Rare Diseases, have been had in mind when completing the explored indicators.

The full version of the review with detailed explanations and tables with indicators is available on ICRDOD website REGISTRIES AND STATISTICS section.

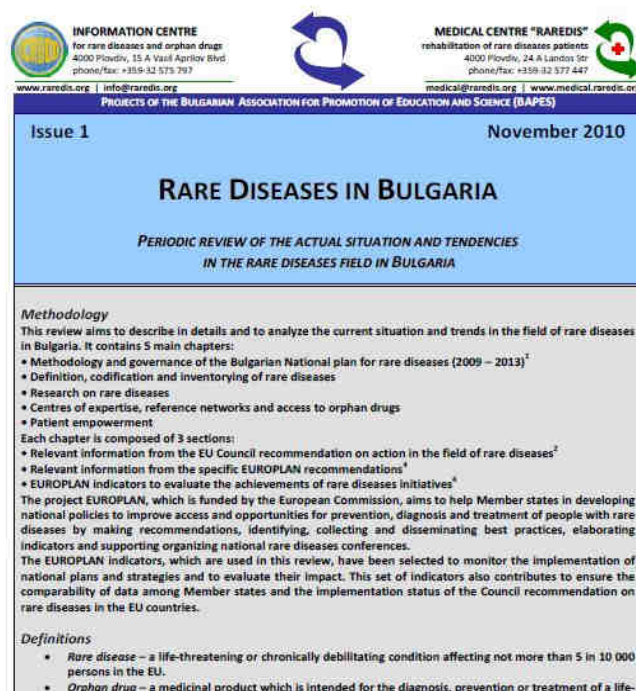


Rare diseases are a threat to the health of EU citizens as they are life-threatening or chronically debilitating diseases with a low prevalence and a high level of complexity.

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

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

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



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

However, nearly two years after its launch, people with rare diseases and their families still face daily discrimination, lack access to adequate treatment, barely any opportunities for rehabilitation and integration. The National Program for Rare Diseases was a glimmer of hope for these people, but its implementation is constantly held back by bureaucratic obstacles, lack of funding (although allocated such one) and not least, personal ambitions and interests.

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

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

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





Bulgarian Association for Promotion of Education and Science (BAPES) is a non-profit non-governmental organization registered under the Bulgarian law on legal persons with non-profit purposes in 2003.

The **main objectives** of the association are:

- to stimulate research;
- to encourage development of technologies and their practical application;
- to support all areas of education and science in medicine.



In 2004 BAPES launched the **Information Centre for Rare Diseases and Orphan Drugs (ICRDOD)** – the first and only Eastern European educational and information service dedicated to patients, medical professionals and associations, interested in rare diseases and orphan drugs.

The main functions of ICRDOD are providing free information on rare diseases, specialized clinics, laboratories in Europe and the world, bringing together leading experts, physicians and patients, creating and maintaining multilingual website for informational and educational purposes (www.raredis.org); organizing workshops and conferences; establishing contacts with scientific and patient organizations from Bulgaria and Europe.

ICRDOD is identified as a major and reliable source of information about rare diseases in Bulgaria on the website of the European Commission (DG SANCO) – http://ec.europa.eu/health/ph_threats/non_com/rare_9_en.htm.

BAPES together with other European partners **has successfully prepared, won and implemented 5 major European projects, funded by the European Executive Agency for Health and Consumers (EAHC)** – ORPHANET (2007-2010), EUROPLAN (2009-2011), RD PORTAL 2 (2010-2011), BURQOL-RD (2010-2013) and EPIRARE (2011-2014) with a total funding of over € 6,000,000. Additionally, **BAPES has won and implemented one own project** (4 EEC RDOD, 2009), which was co-funded by the EAHC.



In 2009, guided by the idea of enlarging and integrating its activities, BAPES started the **Medical Center "RareDis"** - a specialized medical centre for rehabilitation and training of patients with rare diseases and their families. **Multidisciplinary team of specialists is currently preparing intensely modern protocols for medical and social rehabilitation as well as projects for medical research.**

More information about us:

www.raredis.org

Editorial Box

This newsletter is prepared and published by the Bulgarian Association for Promotion of Education and Science (BAPES). The Information Centre for Rare Diseases and Orphan Drugs (ICRDOD) and Medical Centre "RareDis" are projects of BAPES (www.raredis.org).

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The information in this newsletter is provided free for educational purposes and should not be used for self-diagnosis or self-treatment. In case of a health problem you should contact a medical professional!

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