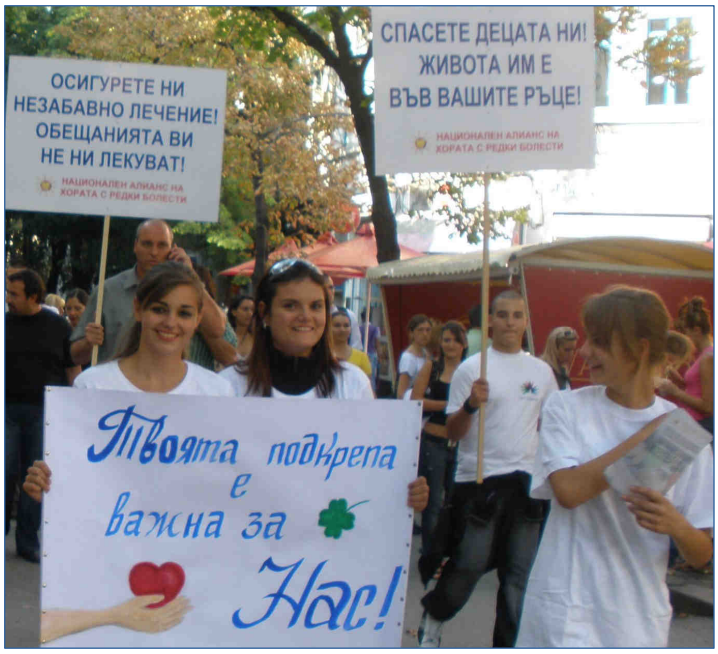
www.raredis.org February 2011 (Issue 1, Year II)

RARE DISEASES ORPHAN DRUGS

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

Dear readers,

We are happy to present you the second issue of BAPES official newsletter. Our fears that it will be difficult to gather enough information about the new issue in the last two months have failed. The pace at which rare diseases events take place in Bulgaria and across Europe is increasing and no wonder if the size and the frequency of this newsletter get bigger in the near future.



For many years people with rare diseases and their families were invisible for the society. Information among doctors was also extremely scarce. However, this situation has changed. The accession to the European Union has played a major role in it by creating opportunities for free exchange of knowledge and experience. But the principal change was in rare diseases patients' mind. They have understood the importance of unity and proactive stance on issues that directly concern their life and prosperity. Patients with rare diseases in Bulgaria have realized that they are equal citizens and along equal obligations they have also equal right to access to quality and adequate treatment. An outcome of this change is the creation of the National Alliance of People with Rare Diseases, which has briefly become a strong and active organization with many successful initiatives.

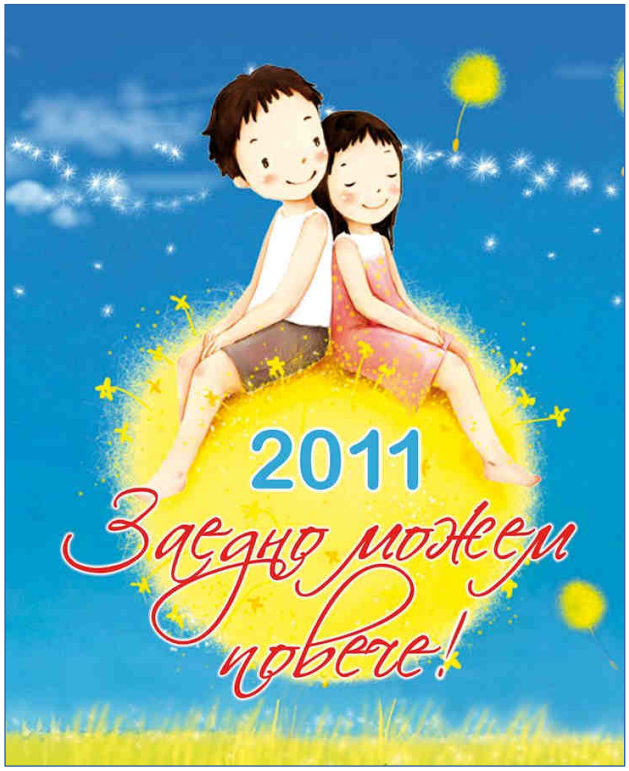
TOPIC OF THE ISSUE:

EPIDEMIOLOGICAL REGISTRIES – RARE DISEASES COME IN THE LIGHT

In this issue we would like to introduce you to two of the most important approaches to "get rare diseases in the light" and to inform the society on this problem's significance – the patient organisations' activities and the creation of the first epidemiological registries for rare diseases in Bulgaria.

We are also launching a new section in the newsletter – consultations on specific medical questions from RAREDIS medical professionals and ICRDOD consultants. Within each issue they will present the most updated information on a specific rare disease's prevention, diagnosis, therapy and rehabilitation, protocols and guidelines for social and psychological support and integration.

Enjoy reading ☺



SECOND NATIONAL CONFERENCE FOR RARE DISEASES AND ORPHAN DRUGS

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 of leading medical experts, workshops and discussions for all rare diseases stakeholders, poster session. The main highlights will include:

- Epidemiological registries for rare diseases
- Best practices and guidelines for prevention, diagnosis, treatment and rehabilitation of rare diseases
- Rare diseases health policy and legislation
- Availability and access to orphan drugs in Bulgaria
- Outcomes of the Bulgarian National plan for rare diseases
- European and international projects in the rare diseases' field

PATIENTS' RIGHTS IN CROSS-BORDER HEALTHCARE

On 19 January 2011 the European Parliament adopted a EU law clarifying patients' rights to access safe and good quality treatment across EU borders and be reimbursed for it. Patients travelling to another EU country for medical care will enjoy equal treatment with the citizens of the country in which they are treated. In a statement the European Commissioner for Health and Consumer Policy John Dalli says that the new legislation will help patients who need specialised treatment, for example those who are seeking diagnosis or treatment for a rare disease. It supports the development of "European Reference Networks" bringing together, on a voluntary basis, specialised centres of expertise already recognised in Europe. Health experts across Europe will be able to share best practices on healthcare and provide standards of excellence. This newly adopted directive sets all this out clearly in EU law and provides a uniform and coherent framework for all citizens in Europe.

At national level, Member States need to ensure that administrative procedures on the use of cross-border healthcare and on reimbursement of costs are in place, including complaint procedures as well as mechanisms to calculate costs. National governments have 30 months to integrate these measures into national legislation.

Credit ©European Union 2011

The Commission will set up networks to foster EU cooperation on health technology assessment and eHealth. It will also help facilitate the recognition of cross-border prescriptions.

RARE DISEASE DAY 2011

For fourth consecutive year the Rare Disease Day will be marked in more than 40 countries all over the world. This year event's slogan is "Rare but Equal" and focuses on the health inequalities for rare diseases patients and their families.

To get detailed information on Rare Disease Day 2011 events in Bulgaria and around the world, follow regularly ICRDOD and Rare Disease Day websites.

DEBRA BULGARIA

Starting from 2011 Association Epidermolysis Bullosa Bulgaria has officially a new name – **DEBRA Bulgaria**, showing the affiliation to the European and worldwide family of similar patient groups – DEBRA Europe and DEBRA International. The new chairperson of the association is **Mrs. Desislava Abadzhieva**.

For further information on Epidermolysis Bullosa and DEBRA Bulgaria, please visit the Internet sites of ICRDOD and DEBRA Bulgaria.

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". The prominent psychotherapist Madeleine Algafari and the psychologist Elenko Angelov will be lecturers. 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.

To read the seminar programme, please visit ICRDOD website.

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.

WE SHOULD NOT STOP IN ANY CASE IF WE REALLY WANT TO ACHIEVE GOOD OUTCOMES

The patient organizations' activities for protecting and defending the rights and interests of patients has long been regarded as an integral factor on the health policy making and public health management. Primarily this is a sign of a mature and active civil society.

During last four years, rare diseases patients in Bulgaria have undoubtedly increased their public activities. Besides the purely quantitative change (nearly 20 rare diseases patient associations, united by National Alliance – NAPRD), many patient groups are already actively involved in joint projects with medical and public institutions. Times, when the personal survival was on the top of the agenda, are gone and today people with rare diseases are determined as never before to change the public attitude and make the society aware of and engaged with the rare diseases' cause in Bulgaria.

In this issue we would like to present you Ms. Yordanka Petkova, chairperson of the Bulgarian association of patients with hereditary angioedema and coordinator of NAPRD for Plovdiv region. Ms. Petkova has been the driving force of the recent series of successful information and charity campaigns and charity of NAPRD.

How do the society reacts towards the problems of people with rare diseases? Do you find a real support?

My personal experience shows that the Bulgarian society is compassionate. If you succeed in reaching people in personal aspect, then they are ready to help you. The institutions are the biggest problem – these which are responsible to make the important decisions and to undertake the cardinal measures. Unfortunately, the real support there is very little.

Many of the rare diseases problems are due to the fact that people know very little about them, people work very little on them and especially – people talk very little about them. How can be raised the public awareness on these questions?

Yes, these difficulties are great, because any change requires effort, time and money. The 20-year transition of the country, the economic crisis and the general indifference of the Bulgarians keep the status quo. The change depends on all us – in both personal and public aspect. Raising-awareness campaigns are so useful. We should not stop in any case if we really want to achieve good outcomes.

You are organizing a training seminar with the famous psychotherapist Madeleine Algafari for a third time. How can the psychology and psychotherapy help rare diseases patients?

Psychology and psychotherapy are useful for all disorders. But the rare diseases need them most, since the lack of cure often damages much more in mental than in physical plan. That is why the hidden internal resources could be a good helper if they are properly used. Every patient with a rare disease need a psychotherapy but it is not always available and affordable.

Rare Disease Day is a very important date for the rare diseases' stakeholders on international level. What about Bulgaria?

It is a very important day in Bulgaria too. This is a great opportunity to bring together rare diseases patients and to seek adequate actions from the society on the problems of rare diseases.

Tell us more the upcoming events, organized by the National Alliance of People with Rare Diseases? You are planning a big charity event just before Easter.

Yes, together with Plovdiv Municipality authorities we are organizing a charity auction of items, donated by Bulgarian celebrities and VIP persons. The main idea of the initiative is to joint the voices of these people to our, rare diseases patients' appeal to the Bulgarian society and institutions. This is a part of the general rare diseases raising-awareness campaign in Bulgaria. I am fascinated by the responsiveness and the sympathy of country's prominent individuals who are ready to support us.

THE EPIDEMIOLOGICAL REGISTRIES – RARE DISEASES COME OUT IN THE LIGHT

A patient registry is defined as an organized program for the collection, storage, retrieval, and dissemination of a clearly defined set of data collected on identifiable individuals for a specific and specified purpose, as well as the collected data.

Patient registries are essential tools for public health surveillance and research inquiry and are a particularly important resource for understanding rare diseases. Registries provide consistent data for defined populations and can support the study of the distribution and determinants of various diseases. One advantage of registries is the ability to observe caseload and population characteristics over time, which might facilitate the evaluation of disease incidence, disease etiology, planning, operation and evaluation of services, evaluation of treatment patterns, and diagnostic classification.

Any registry program must collect high quality data to be useful for its stated purpose. Registries can be developed for many different needs and caution should be taken in interpreting registry data, which has inherent biases.

What is the legislative base in Bulgaria for the establishment of epidemiological register?

At this point there is no law that clearly and accurately treats the question of the establishment and functioning of epidemiological registries for rare diseases. Health Law and the Personal Data Privacy Law provide guidance on who has the right to collect health information of individuals and how that process must be organized.

Under the Health Law, "health record" means all forms of registration and storage of health information. HL states:

Art. 27. (1) Health information is all the personal data, concerning health, physical and mental development of individuals, as well as any other information which is presented in medical prescriptions, records, certificates and other medical documentation.

(2) Medical and health institutions, regional health inspection centres, doctors, dentists, pharmacists and other health professionals, as well as non-medical specialists, working in the national health system, collect, process, use and store health information.

Art. 28. (1) Health information can be provided to third parties when:

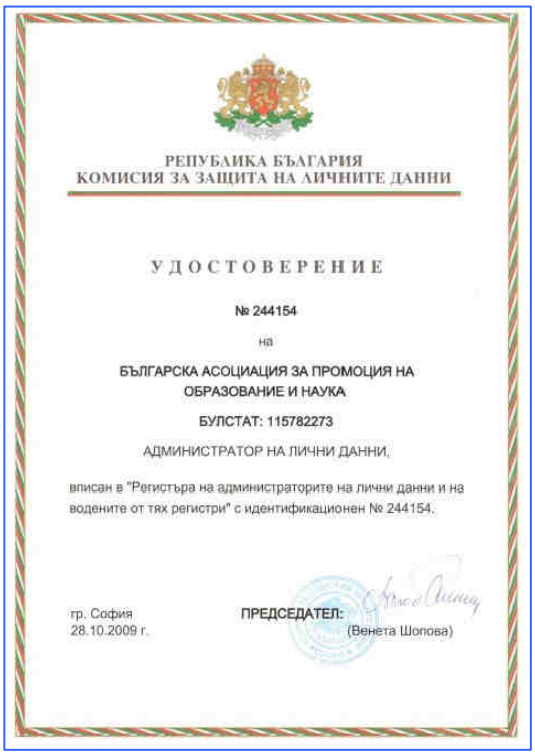
6. it is necessary for the purposes of the medical statistics or medical research after the data, identifying the patient, has been withdrawn;

7. (Amended – State Gazette, issue 98/2010, in force from 01.01.2011) it is necessary for purposes of Ministry of Health, National Centre for Health Information, National Health Insurance Fund, regional health inspection centres and National Institute of Statistics.

(3) Persons under Art. 27, paragraph 2 are required to ensure the protection and the privacy of stored health information against unauthorized access.

(2) Patient may authorize in writing another person to examine his medical records and make copies of them.

On the other hand, Personal Data Privacy Law clearly indicates that all information, related to an individual's health, may be collected only by authorized by the individual personal data administrator, who processes the personal data by self or by assigning another person. Article 4 of the Law states that processing of personal data is permitted only in cases where the individual, to whom the data is related, has given informed consent. Moreover, the administrator must ensure adequate protection of the stored personal data. This is certificated by the State Commission for Personal Data Privacy's mandatory inspection on all requirements for the appropriate level of protection.



РЕПУБЛИКА БЪЛГАРИЯ
КОМИСИЯ ЗА ЗАЩИТА НА ЛИЧНИТЕ ДАННИ

УДОСТОВЕРЕНИЕ

№ 244154

на

БЪЛГАРСКА АСОЦИАЦИЯ ЗА ПРОМОЦИЯ НА ОБРАЗОВАНИЕ И НАУКА

БУЛСТАТ: 115782273

АДМИНИСТРАТОР НА ЛИЧНИ ДАННИ,

вписан в "Регистъра на администраторите на лични данни и на водените от тях регистри" с идентификационен № 244154.

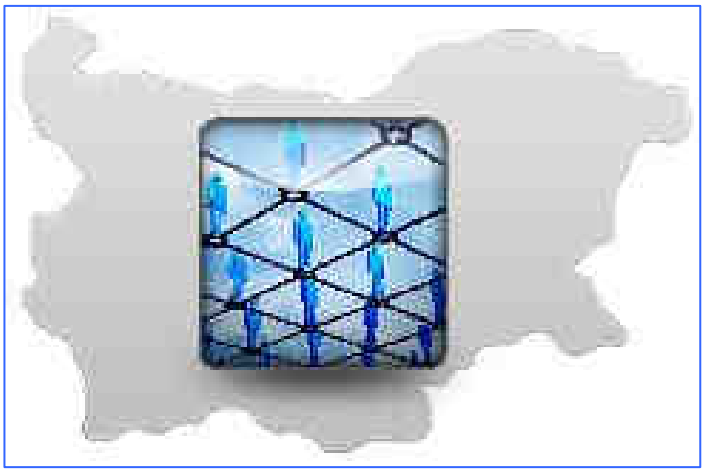
гр. София
28.10.2009 г.

ПРЕДСЕДАТЕЛ:
(Венета Шопова)

It is specifically stated that the personal data processing is permitted when it is necessary for the purposes of the preventive medicine, medical diagnosis, provision and management of health services and provided that the data is processed by a medical professional, legally required to observe professional secrecy, or by another person, subject to such obligation of secrecy.

According to this logical framework, there are currently 3 active epidemiological registries for rare diseases in Bulgaria – *thalassemia major, chronic myeloid leukemia* and *Crohn disease*. *Wilson disease* epidemiological registry is in final stage before starting, *Gaucher disease* and *mucopolysaccharidosis* registries are relatively clear in terms of size and content.

The second phase of the project "National registry of patients with thalassemia major in Bulgaria" was successfully completed in October 2010. The aim was to update the information on patients who were registered during the first phase of the project and to register newly diagnosed and not yet registered patients with thalassemia major. A total of 241 questionnaires (18 for primary epidemiological information about newly diagnosed patients and 223 for data update) were collected with the active assistance of medical specialists from blood transfusion centres and thalassemia patient association. After statistical processing of the submitted data, it was found that the number of men is 126 (52.28%) and 115 (47.72%) for women respectively. The average age of patients with thalassemia major is 19.5 ± 11.9 years. Their treatment is conducted in 12 hospitals in the country and chelation therapy takes place in the blood transfusion specialized centres (Sofia - three centres, Plovdiv - two centres, Varna, Stara Zagora, Pleven, Burgas, Ruse, Yambol and Silistra). These results were discussed and adopted as official for the country at a workshop of the Expert group on thalassemia, held in Varna in November. A subsequent update and collection of new epidemiological data in will be organized in March-April 2011.



The Bulgarian National chronic myeloid leukemia (CML) patient registry was started in November 2010 by concluding the first epidemiological study of CML patients. This project is implemented jointly with the Bulgarian Scientific Society of Clinical and Transfusion Hematology and Medical Center "RareDis". After processing the information a total of 248 CML patients were registered. The proportion of men is 50.40% (125 patients), while that of women is 49.60% (123 patients). Standardized annual prevalence is 3.27 per 100 000 people. The average age of registered patients is 53.86 ± 15.33 years. At the time of the study, patients were treated and followed up in eight hospitals in the country, primarily in university hospitals: 2 centres in Sofia – National Hematology Hospital with 78 (31.50%) patients and "Alexandrovska" University Hospital with registered 29 (11.60%) patients, Plovdiv - 56 patients (22.60%), Varna - 48 patients (19.40%), Pleven - 27 patients (10.90%), Ruse - 5 patients (2.00%), Haskovo - 4 patients (1.60%) and Vidin - 1 patient (0.60%). Patients from Ruse, Vidin and Haskovo receive medicinal therapy at the university clinics and are followed up by a hematologist in their home town. An update of the information on registered patients with CML, as well as registration of new cases will be held in 2011.

At the end of 2010 BAPES together with the Bulgarian Society of Gastroenterology, Gastrointestinal Endoscopy and Abdominal Echography concluded the first collection of epidemiological information on Crohn disease in Bulgaria. The initial data on Crohn disease prevalence, number of patients and their distribution, Montreal classification symptoms have been already processed. The detailed results will be published after official review by the Bulgarian Society of Gastroenterology.

ROLE OF THE REHABILITATION IN PRADER-WILLI SYNDROME MANAGEMENT

SHORT DESCRIPTION OF THE CONDITION

DEFINITION

Prader-Willi syndrome (PWS) is a complex genetic disorder caused by deletion or mutation of genes in chromosome 15 or maternal disomy of chromosome 15. Commonly associated characteristics of this disorder include decrased fetal activity, obesity, hypotonia, cognitive disabilities, problematic behaviour, short stature, hypogonadotropic hypogonadism, strabismus and small hands and feet.

PREVALENCE

It is estimated that one in 12,000 to 15,000 people has PWS. Although considered a "rare" disorder, Prader-Willi syndrome is one of the most common conditions seen in genetic clinics and is the most common genetic cause of obesity that has been identified. PWS is found in people of both sexes and all races.

DIAGNOSIS

The diagnosis is usually proved clinically first, and then confirmed by specialized genetic testing on a blood sample.

TREATMENT GUIDELINES

- *Special feeding techniques may be necessary for the first weeks to months of life.*
- *Early intervention in children under three years, particularly physical therapy, may improve muscle strength and encourage achievement of developmental milestones. When hyperphagia begins or weight is increasing (often age two to four years), a program of a well-balanced, low-calorie diet, regular exercises, and close supervision to minimize food stealing should be instituted to prevent obesity and its consequences.*
- *Growth hormone treatment normalizes height, increases lean body mass, decreases fat mass, and increases mobility, which are beneficial to weight management.*
- *Appropriate educational programming in children: speech therapy for language delay and articulation abnormalities in infancy and childhood; special educational, social skills training. Behavioural disturbance should be addressed with behavioural management programs.*
- *No medications are known to aid in controlling hyperphagia.*

Due to the lack of adequate drug therapy, attention should be focused entirely on the rehabilitation as an appropriate method for maximum use of the available functional capacities of children and decent complex of logopegical and pedagogical support.

In most European countries, depending on the type of health system and social policy of the country, specific programs or clinical protocols are developed in order to provide the most comprehensive help to these patients and their families. Recently, several publications came out, which study in retrospective aspect the most effective combination of rehabilitation activities for this disease.

Because of the difficulties in providing health care for these patients at home, coming from the constant hunger and outbursts of anger and aggression, most authors propose to undergo a rehabilitation program outside the home, which lasts at least 4 weeks, four times a year. The aim is not only to break away from the daily routine of the already difficult for socializing patients, but also to introduce a new dynamic stereotype of life. A strictly regulated diet of a reduced daily intake of 1500 kcal/day is one of programme's main features. The rehabilitation plan includes a vast number of specialists – psychologist, neurologist, physical therapist, geneticist, nutritionist, music therapist and others as needed.

Group therapy is preferred because of disease-caused difficult social contacts. It is recommended that physical exercises take average of 6.5 hours, divided into two parts – in the morning and in the afternoon. The variety of activities keeps the interest of patients, so it is recommended that apparatus exercises last no more than 30 minutes and nature walks and field treatment are included in order to diversify the rehabilitation. Since obesity is one of the main problems, the diet is very important and must be frequent – 5 times per day, consisting of two meals (lunch and dinner) and three snacks. Water intake is controlled to be only at mealtimes and no more than 1.5-2.5 litres per day.

Most authors propose that the psychomotor activities and the music therapy should occupy a significant place in the daily regime (2-4 hours) and must be almost equally divided during the day. They stimulate skills, coordination, concentration, social contacts and memory functions. This clinical protocol has demonstrated its effectiveness for many affected patients so far and is fully applicable in Bulgaria, having in mind the availability of good specialists, many resorts and suitable climatic conditions for tourism in most of the year. The program is feasible with outpatient reception within a day hospital.

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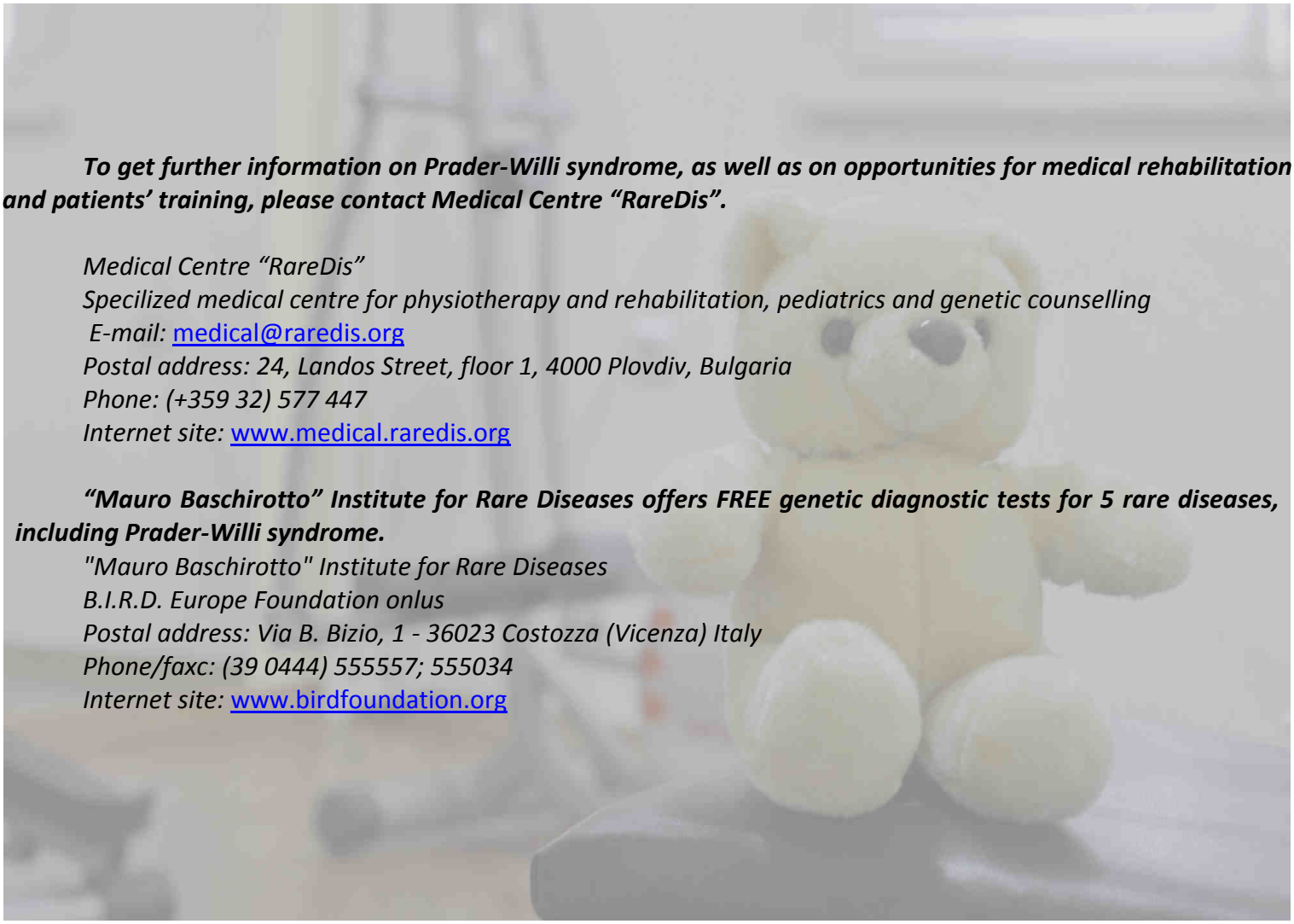

To get further information on Prader-Willi syndrome, as well as on opportunities for medical rehabilitation and patients' training, please contact Medical Centre "RareDis".

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"Mauro Baschirotto" Institute for Rare Diseases offers FREE genetic diagnostic tests for 5 rare diseases, including Prader-Willi syndrome.

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Bulgarian Association for Promotion of Education and Science (BAPES) is a non-profit non-governmental organization registered under 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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ISSN 1314-359X | © BAPES 2010

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