



BULGARIAN ASSOCIATION FOR PROMOTION OF EDUCATION AND SCIENCE

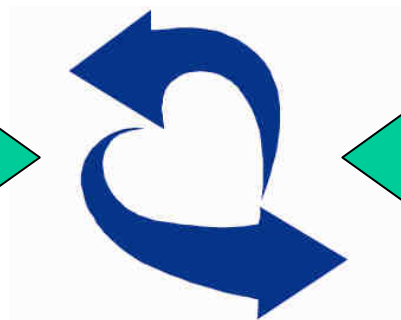
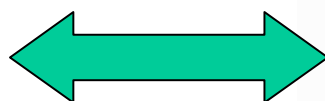
ANNUAL REPORT 2010

WHO WE ARE



ICRDOD

2004



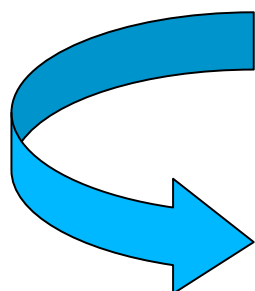
BAPES

2003

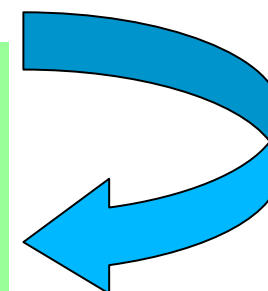


RAREDIS

2009



**INFORMATION AND
CLINICAL RESEARCH PLATFORM
FOR RARE DISEASES**

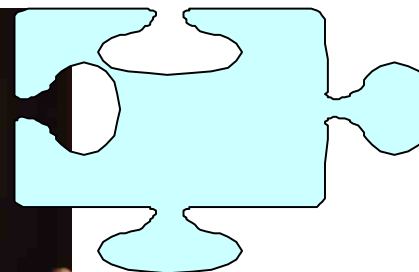




HOW WE WORK

- **Acting SYSTEMATICALLY AND PURPOSEFULLY on the problems of rare diseases patients**
- **Initiating and mediating for search of EFFECTIVE solutions**
- **Relying on ALL THE STAKEHOLDERS' opinion**
- **Following BEST EUROPEAN practices**

EDUCATION AND TRAINING



First National Conference for Rare Diseases and Orphan Drugs



- **More than 300 participants, 2 plenary sessions, 9 satellite workshops, 3 patient seminars!**
- **Part of EU-funded project EUROPLAN**
- **Detailed presentation and active discussion of the Bulgarian National Programme for Rare Diseases**

First National Conference for Rare Diseases and Orphan Drugs

All Bulgarian rare diseases stakeholders gathered at one place!

- **Medical specialists**
- **Patients and patient associations**
- **Academia**
- **Industry**
- **Media**
- **Special guests from EURORDIS, Thalassaemia International Federation, Cystic Fibrosis Europe**



Conference Memorandum

- **Full support of the priorities set out in the EU Council Recommendation on rare diseases actions, adopted on June 9, 2009**
- **Implementation of the Bulgarian National Programme for Rare Diseases with the appropriate funds, previewed in its budget framework**
- **Urgent legislative initiatives to protect the rights of people with rare diseases and ensure adequate prevention, treatment, rehabilitation and social cares**
- **Encouraging the epidemiological registries for rare diseases in Bulgaria**
- **Integrated approach to people with rare diseases and their families**
- **Organizing a public campaign aimed to fund and to stimulate research on rare diseases in Bulgaria**



Workshop

“Rare Diseases Patients and Medical Students – Ideas for Common Projects and Collaboration”



- **Sharing personal stories**
- **Looking for possibilities for:**
 - **patient empowerment**
 - **raising awareness among medical community, as well as the whole society**
 - **better professional qualification of medical students**

Update of WWW.RAREDIS.ORG – the Internet Portal for Rare Diseases

- New sections
- Analyses and results from own investigations
- Rare diseases encyclopedia in Bulgarian language
- Information brochures for patients and their families
- Newsletter

INFORMATION CENTRE FOR RARE DISEASES AND ORPHAN DRUGS
ИНФОРМАЦИОНЕН ЦЕНТЪР ЗА РЕДКИ БОЛЕСТИ И ЛЕКАРСТВА СИРАЦИ

EGAN
European Genetic Alliance Network

Често задавани въпроси за персонализираната медицина

Информационен Център за редки болести и лекарства сираци
ул. "Митко Митров" 2/а, София 1000
телефон (02) 575 7971
www.raredis.org | info@raredis.org

Медицински Център „РедДис“
реабилитационен център за редки болести
ул. "Никола Петков" 2/а, София 1000
телефон (02) 575 7971
www.raredis.org | info@raredis.org

Проекти на Българската Асоциация за Промощия на Образованието и Научата (BAPES)

Брой 1 **ноември 2010**

РЕДКИТЕ БОЛЕСТИ В БЪЛГАРИЯ

ПЕРИОДИЧЕН ОБЗОР НА АКТУАЛНОТО СЪСТОЯНИЕ И ТЕНДЕНЦИИ
В ОБЛАСТТА НА РЕДКИТЕ БОЛЕСТИ В БЪЛГАРИЯ

Методология
Настоящият обзор има за цел да опише подробно и да анализира актуалното състояние и тенденциите в областта на редките болести в България. Обзорът съдържа 5 основни раздела:

- Методология и управление на Националната програма за редки болести (2009-2013)¹
- Дефиниция, кодиране и квалификация на редките болести
- Научноизследователска дейност в областта на редките болести
- Експертни центрове, референтни мрежи и достъп до лекарства сираци
- Пациентски организации

Всички от тези раздели са съставени от 3 секции:

- Информация от Препоръките на Съвета на ЕС за действие в областта на редките болести²
- Информация от Препоръките по проект EUROPLAN³
- Индикатори за оценка, съгласно Препоръките по проект EUROPLAN⁴

Проектът EUROPLAN, финансиран от Европейската комисия, има за цел да помогне страните-членки при разработването на национални политики за подобряване достъпа и възможността за превенция, диагностика и лечение на хората с редки болести чрез изготвяне на препоръки, идентифициране

SCIENCE AND RESEARCH



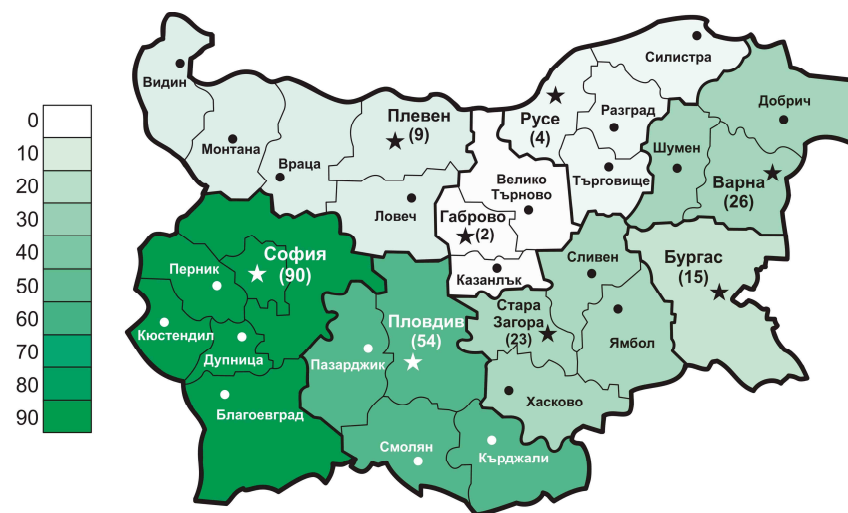
Epidemiological Registries for Rare Diseases

- **Epidemiological data for rare diseases prevalence and incidence**
- **Enrichment of medical science**
- **Establishment and update of protocols and standards for treatment and rehabilitation**
- **Data to health authorities for planning and supporting healthcare for rare diseases patients in Bulgaria**
- **Collaboration and partnership with familiar activities on European level**



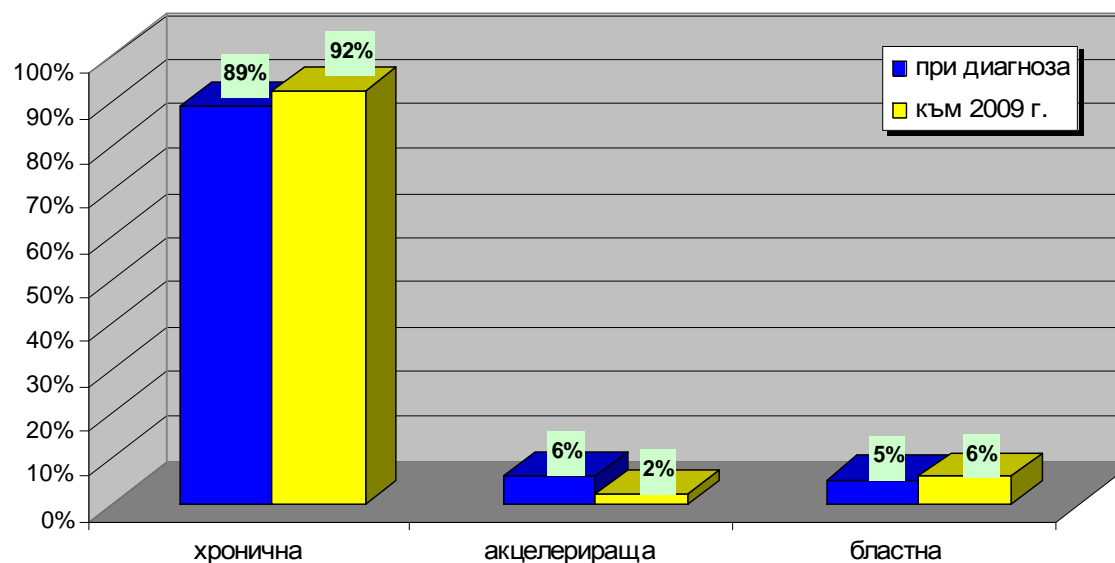
National Thalassemia Major Registry

- First one in Southeastern Europe
- Systematic data since 2009
- Example for successful collaboration between BAPES, Bulgarian Scientific Society for Clinical and Transfusion Hematology, patient associations and university hematology clinics
- Role model for future epidemiological registries

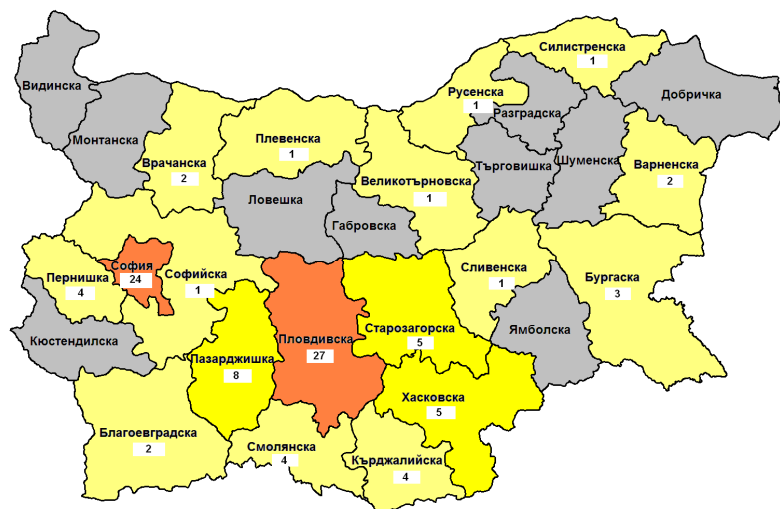


National Chronic Myeloid Leukemia Registry

- **Pilot survey conducted in 2009**
- **Starting epidemiological registry in 2010**
- **Example for successful collaboration between BAPES, Bulgarian Scientific Society for Clinical and Transfusion Hematology and university hematology clinics**



National Crohn Disease Registry



- Pilot survey for epidemiological data
- Collaboration between BAPES and the Bulgarian Scientific Society for Gastroenterology, Gastrointestinal Endoscopy and Abdominal Echography
- Information about Crohn diseases, number of patients and their distribution across the country, symptomatics of Crohn disease according to the Montréal Classification

In Process of Launching

Wilson Diseases

Mucopolysaccharidosis

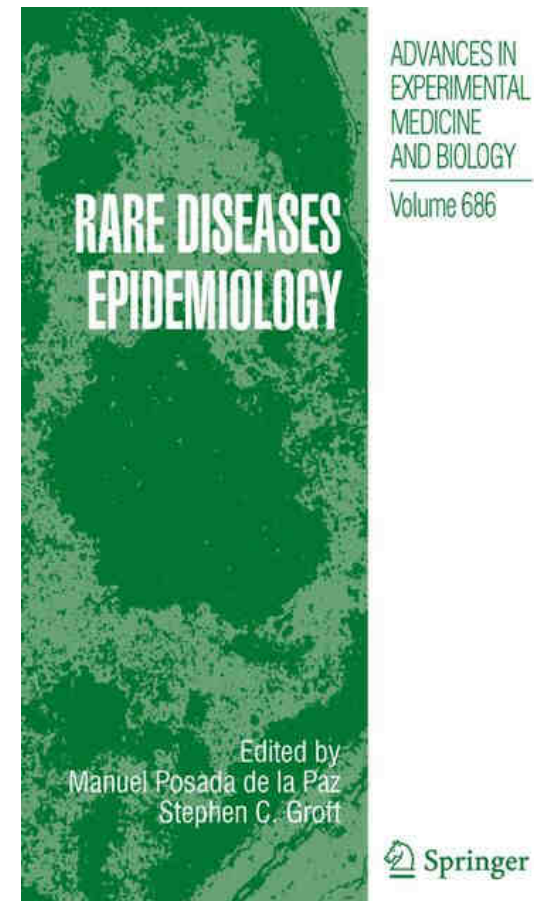
Gaucher Disease

Hereditary Angioedema



“Rare Diseases Epidemiology”

- **BAPES expert is a co-author of “Rare Diseases Epidemiology”**
- **Offering numerous approaches to increase the knowledge base of rare diseases and to facilitate the development and dissemination of prevention, diagnosis or treatment**
- **Presenting methods from the collective experiences of established research investigators who address these significant issues of:**
 - **patient registries**
 - **bio-banking activities**
 - **genetic testing and newborn screening**
 - **population-based surveillance studies**
 - **clinical trials and comparative effective studies**
 - **requirements of regulatory agencies**



Periodical Reviews on Rare Diseases Issues

Select language

Site translation

Powered by Google Translate

Menu

- News
- Rare Diseases
- Registries & Statistics**
- Patient Associations
- F.A.Q.
- Our Activities
- Links
- Forums
- About Us
- Consultants
- Contact Us

User Menu

- View Account
- Edit Account
- Notifications
- Logout
- Inbox (8)**
- Administration Menu

Supported by

Медицински център "Раредис"
www.medical.raredis.org

REGISTRIES & STATISTICS

PREVALENCE OF RARE DISEASES:

- **National registry of patients with thalassemia major in Bulgaria**
 - Information at the beginning of the project (August 2008)
 - Certification of BAPES as personal data administrator by the State Agency for Data Privacy Protection (October 2009)
 - Preliminary results (January 2010)
 - Phase I official results (April 2010)
 - Phase II official results (October 2010) **NEW!**
- **National registry of patients with chronic myeloid leukemia (CML) in Bulgaria**
 - Phase I official results (November 2010) **NEW!**
- Pilot epidemiological study of chronic myeloid leukemia (CML) (click here for more information)

RARE DISEASES WITHIN THE BULGARIAN PUBLIC HEALTH SYSTEM:

- **Review of the actual situation and tendencies in the rare diseases field in Bulgaria**
 - Issue 1 / November 2010 (click here to read the PDF file, 0.98 MB) **NEW!**

ORPHAN DRUGS:

- **Review of the access to medicines for rare diseases in Bulgaria**
 - Issue 1 / June 2010 (click here to read the PDF file, 130 KB)

For the relevant information on EU level, please visit Orphanet website.

Search

Search

Advanced Search

Our Conferences

- 2010
Първа Национална Конференция за Редки Болести
- 2010
Rare Diseases in the Focus of Personalized Medicine
- 2009
TOGETHER FOR INTEGRATIVE APPROACH TO RARE DISEASES
- 2008
RARE DISEASES – PREVENTION, DIAGNOSIS, TREATMENT
- 2007
First National Patient Conference of People with Rare Diseases
- 2006
Fostering Research on Rare Diseases in Eastern European Countries
- 2005
Promotion of Healthcare and Research on Rare Diseases

Available Online!

COLLABORATION AND INTEGRATION



EUROPLAN

Objective: *Recommendations for Development of National Plans for Rare Diseases*

BAPES Contribution:

- **Associated partner**
- **Monitoring on rare diseases issues in Bulgaria**
- **Assessing the Bulgarian rare diseases stakeholders' need and local conditions in prospect of the EUROPLAN recommendations**
- **Co-organizing the Bulgarian national EUROPLAN conference**



WWW.EUROPLANPROJECT.EU

RD PORTAL 2 (ORPHANET)

Objective: *Encyclopedia of rare diseases*

BAPES Contribution:

- **Associated partner**
- **Co-ordinator for Bulgaria**
- **Adding and updating specific rare diseases information about Bulgaria to the Orphanet database**
- **Promoting Orphanet among the Bulgarian rare diseases stakeholders by organizing workshops and discussions**

WWW.ORPHA.NET



The portal for rare
diseases and orphan
drugs

BURQOL-RD

Objective: *Measuring the socio-economic burden and health related quality of life of rare diseases*

BAPES Contribution:

- **Associated partner**
- **Selecting rare diseases to surveyed upon the project**
- **Project referent point for Bulgarian patients and patient associations**



WWW.BURQOL-RD.COM

EPIRARE

- **Defining European platform for rare diseases epidemiological registries and their optimal use**
- **Premises for European wide legal base and management guidelines**
- **Defining minimal data set to be included for epidemiological survey in prospect of health policy making, research, treatment and rehabilitation**
- **BAPES would be associated partner**
- **Partners from China would take part for the first time in European public health project**

Starting in 2011!

European Network of Rare Diseases Help Lines

- **ICRDOD is a member since 2010**
- **Training help lines staff**
- **Exchange of experience and good practices**
- **Better visibility for rare diseases information services**
- **Popularization of information sources in different languages**



**Help Lines
for Rare
Diseases**

Fifth Eastern European Conference for Rare Diseases and Orphan Drugs



BAPES Contribution:

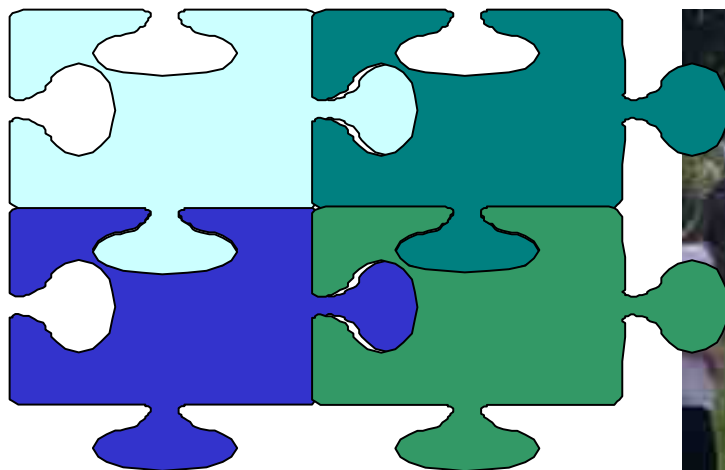
- **Granting the rights of the event**
- **Participation at both organizing and scientific committees**
- **Speaker and representatives from BAPES at the plenary sessions and satellite workshops**

Fifth Eastern European Conference for Rare Diseases and Orphan Drugs

- **For the first time outside Bulgaria – in St. Petersburg, Russia**
- **More than 400 participants from all over the world**
- **12 panel sessions and satellite workshops**
- **The biggest and most important Eastern European forum for rare diseases in 2010**



SUPPORT



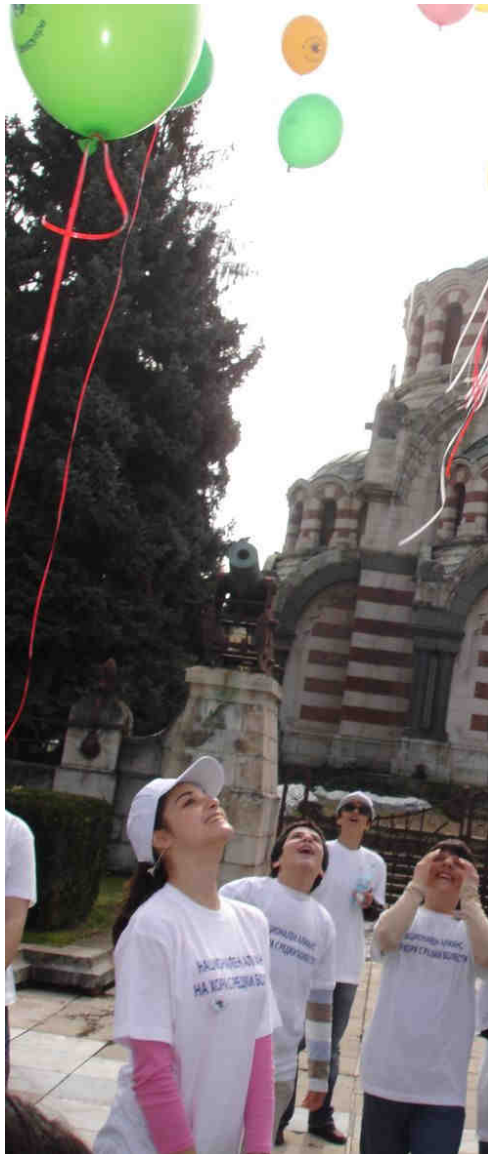
Rare Diseases Day



Rare Disease Day®

- **For third consecutive year BAPES has supported the National Alliance of People with Rare Diseases for the organization of Rare Diseases Day events in Bulgaria**
- **in 46 countries of all continents**
- **Official 2010 Slogan – Patients and Research, Partners for Life!**
- **Official patron of Rare Disease Day events in Bulgaria – Mrs. Zorka Parvanova, Bulgaria's First Lady**

BULGARIAN ASSOCIATION FOR PROMOTION OF EDUCATION AND SCIENCE (BAPES)
ANNUAL REPORT 2010



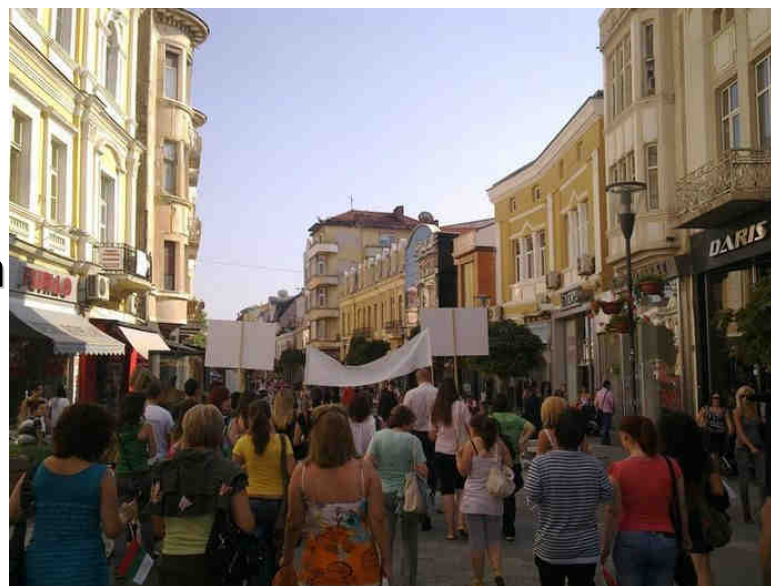
Rare Diseases Day

- Events in all biggest cities – Sofia, Plovdiv, Varna, Burgas, Pleven...
- Big charity concert in Plovdiv on 28 February



March “For a fair chance to our children”

On the start of the school year in Bulgaria (15 September) patient associations from the National Alliance of People with Rare Diseases organised a march under the motto "For a fair chance to our children" in order to draw again the attention of the society and to provoke actions from the state to eliminate the discrimination on rare diseases patients' medical treatment. ICRDOD supported and took part at the march.



Patient Organizations Role in Health Policy Making



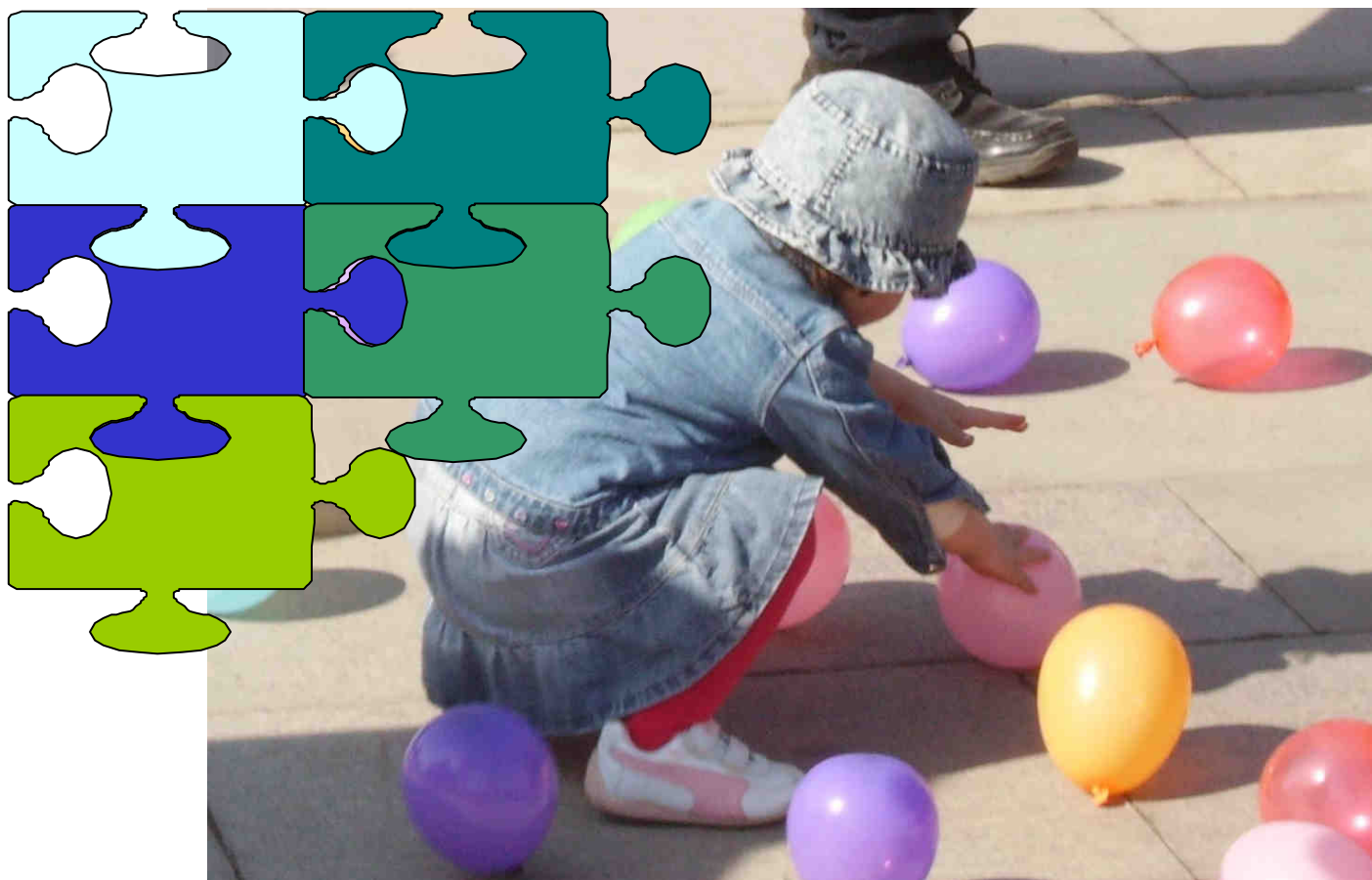
Roundtable discussions on the patient organizations role as a factor in health policy making were organized at Plovdiv District Administration Office by BAPES and its partners from the National Alliance of People with Rare Diseases and Italian-Bulgarian Foundation – Plovdiv. Special guest and speaker was Mariella Bocciardo. Mrs. Bocciardo is a member of the Italian Parliament. A patient herself, she has a long experience in promoting patient empowerment and research activities.

“On Yesterday, Today and Tomorrow”

ICRDOD organized and supported the publication of the book “On Yesterday, Today and Tomorrow”, containing stories of 9 Bulgarian patients with rare diseases. In this way we hope to contribute for the mind change, which our society needs not only for the rare diseases, but for all valueble and truly important things in life!



CARE AND FOLLOW UP



Medical Centre “RareDis”



- **Preparation and publication of specific protocols and programmes for physical, psychological and social rehabilitation of people with rare diseases**
- **Training of parents of children with rare diseases on quality everyday health cares**
- **Active collaboration with patient associations to access best innovative practices**

Donation from Medical Centre “RareDis”

Medical Center "RareDis" made a donation of 12 000 BGN to the National Alliance of People with Rare Diseases during the charity campaign "Be in Solidarity, Give Happiness!".

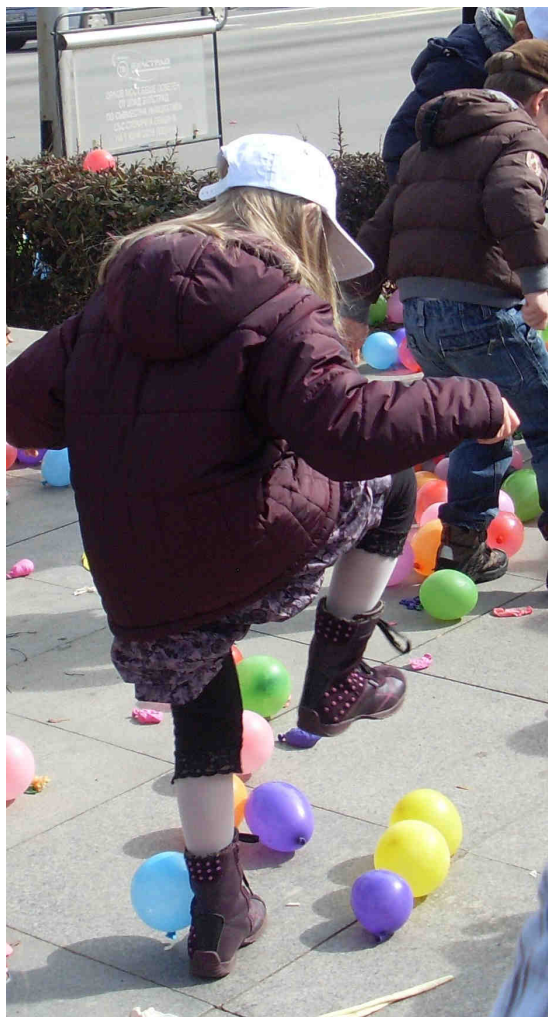


2011 Priorities

- **New epidemiological registries and research on rare diseases**
- **Territorial unification of ICRDOD and RAREDIS**
- **New design and structure of WWW.RAREDIS.ORG**
- **Research on quality of life of people with rare diseases**
- **Specialized protocols for complex rehabilitation and integration of people with rare diseases**



2011 Events



- **Second National Conference for Rare Diseases – 9-11 September 2011, Plovdiv**
- **Sixth Eastern European Conference for Rare Diseases and Orphan Drugs – October 2011, Istanbul (Turkey)**
- **European projects EUROPLAN, RD PORTAL 2, BURQOL-RD and EPIRARE**
- **Rare Diseases Day – 29 February 2011**

**TOGETHER WE
COULD DO MORE!**

