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RARE DISEASES ORPHAN DRUGS

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

Dear Friends,

The first edition of "Rare Diseases & Orphan Drugs" for 2012 is now in front of you. Besides our special message to you on the occasion of Rare Disease Day, we have gathered many interesting and useful news from the world of rare diseases.

As you have probably noticed, our Information Centre for Rare Diseases and Orphan Drugs (ICRDOD) has a new website. What's to find on it is described on page 2.

FOCUS ON:

RARE DISEASE DAY – SOLIDARITY IN ACTION!

News about important rare diseases events and activities in Bulgaria and Europe are on pages 3, 4 and 12. We present you the preliminary results of the Bulgarian rare disease patients' participation in the BURQOL-RD study, as well as some European novelties about rare diseases registries topics. Rare Diseases Library features the description of the rare disorder blepharospasm (pages 10-11).

February is always a special month for people whose life is somehow connected with rare diseases issues. The last day of this period, February 29 comes to make us think about how precious life is and how even at the beginning of XXI century for thousands of rare diseases and millions of people there are no cure, no care, no attention and no comprehension.

Rare Disease Day is not only to inform us once again on these facts, but it also tries to change something in us. This is an initiative that shows that anything is possible when everyone is together, standing behind a fair cause. The changes occur neither fast nor easy, but they need us all to take place. On 29 February everyone should think what it means solidarity. No man can change the course of events on his own, but together, weak and strong, small and big, patients and physicians, we are capable of doing great things. Today, not only people with rare diseases and their families, we all need this synergy around us.

SOLIDARITY IN ACTION!

INFORMATION CENTRE FOR RARE DISEASES AND ORPHAN DRUGS WITH A NEW WEBSITE

New information and education services for rare diseases

Since the beginning of 2012 ICRDOD has a new, upgraded website. Besides a new look and new layout of content, the site offers several new features for its users:

❖ **Subscribe for "Rare Diseases & Orphan Drugs"**

You can now subscribe to this newsletter and automatically receive each new issue via email. Please enter your e-mail in the form on the right of the homepage and follow the instructions. In this way you will always be informed of the developments in the field of rare diseases and orphan drugs – anytime and anywhere.

❖ **Registry of rare diseases patients**

This is a pilot initiative on our website. ICRDOD has always popularised the topic of registries for rare diseases and praised the benefits, that they produce to patients, physicians and institutions. To date ICRDOD maintains a number of national epidemiological registries for selected rare diseases. Help us create a unified registry of people with rare diseases in Bulgaria. Take a minute of your time and fill out the registration card (http://www.raredis.org/?page_id=2516) on our site. In this way you will help to collect more accurate and actual data on the number of people with rare diseases in the country. Your personal information will be stored in accordance with all the provisions of the personal data protection legislation. Moreover, that could help ICRDOD to inform you promptly on innovation projects and events, related to your rare disease. We hope it would help us achieving better communication and feedback between you and us.

❖ **Rare Diseases Library**

Rare Diseases Library has a new framework and updated contents. There you will find descriptions of over 100 rare diseases, which are prepared by the medical consultants of ICRDOD. Each description contains synonyms of the disease, ICD10 and Orphacode numbers, brief definition, etiology, clinical picture, genetic counseling, treatment, orphan drugs, patient associations, specialised clinics, links to further information resources. The descriptions are sorted alphabetically by the most used name of the disease. If you can not find a description of your disorder, do not hesitate to contact us. Medical consultant from our team will prepare a detailed one and we will send it back to you. We would like to remind you that all this information is provided free of charge, but it should not be used for self-diagnosis and/or self-treatment. If you have a health problem, please contact your medical professional.

❖ **Interaction with ICRDOD team**

Do you have further questions? You need to contact a medical specialist or a hospital in Bulgaria and the European Union? You want to get additional information about a project or an initiative for rare diseases? In all these and other cases, feel free to contact us (http://www.raredis.org/?page_id=1375). ICRDOD will reply and give you up to date and reliable information on rare diseases and orphan drugs. Looking forward to helping you!

SOCIAL-ECONOMIC COST AND HEALTH-RELATED QUALITY OF LIFE IN RARE DISEASES

A strong interest of the Bulgarian patients with rare diseases and their families in BURQOL-RD study

BURQOL-RD is a new European project on rare diseases, whose active part started in December 2010. Its survey is addressing the socio-economic cost and health-related quality of life of patients with rare diseases and their caregivers. It is a global European project, coordinated by the Fundación Canaria de Investigación y Salud (FUNCIS), Spain and co-funded by the European Executive Agency for Health and Consumers (EAHC).

Up to now, 132 Bulgarian patients with rare diseases have completed the online survey questionnaires! This is an extremely good result and a further evidence of the excellent comprehension and cooperation between doctors and patients in Bulgaria on the rare diseases issues.

Why is so important to take part in BURQOL-RD? By filling your rare disease questionnaire, you will help to give a greater and stronger voice to your patient association, a clearer picture of the daily difficulties that you face. The results of BURQOL-RD will be a fundamental basis for planning and development of future European policies on rare diseases. Bulgarian rare diseases patients and their caregivers could take part and fill the questionnaires until mid-March. "Rare diseases & Orphan drugs" invites the rare diseases patients to spend 10 minutes of their time to show an active position and to contribute to the cause of people with rare diseases in Bulgaria and Europe.

What are these questionnaires and how to take part in BURQOL-RD?

The survey is completely anonymous. Response records do not contain any identifying information and all the data will be treated in accordance with the Law on Personal Data Protection. The questionnaire consists of two parts – for the patient and for his/her main caregiver. The patient section is in two versions – for children (under 18) and adults.

The study is targeting 10 different rare diseases:

Cystic fibrosis	**Prader-Willi syndrome**	**Fragile X syndrome**
Hemophilia	**Epidermolysis bullosa**	**Duchenne muscular dystrophy**
Scleroderma	**Mucopolysaccharidosis**	**Juvenile idiopathic arthritis**
	Histiocytosis	

The survey is being conducted entirely online on HTTP://BURQOL-RD.EU/BG.HTML.

Take part in the study. In this way you are not only helping yourself, your family and your patient association, but you are also helping the Bulgarian society and the whole of Europe!

ICRDOD team would like to express special thanks to the Bulgarian Cystic Fibrosis Association, Bulgarian Hemophilia Association and DEBRA Bulgaria for their help in promoting this study. Thanks also to the medical professionals from the university hospitals throughout the country who are also involved, presenting the project to their patients.

EUROPEAN APPROACHES TO RARE DISEASES REGISTRIES ORGANISATION AND MANAGEMENT

Two events in the end of 2011 have returned again the focus of attention to the rare diseases registries, one of the rare diseases recent hot topics.

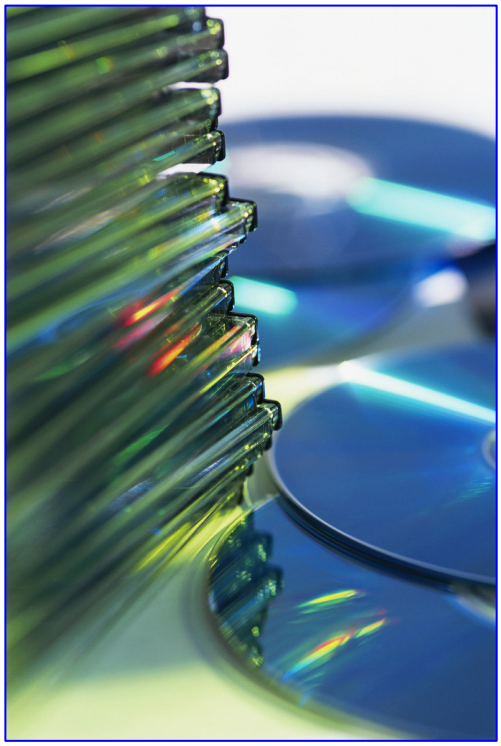
Preliminary results of EPIRARE online study

EPIRARE Project has published the preliminary results of its online survey on rare diseases registries. The study intends to explore the functioning, resources, problems, needs and expectations of existing rare diseases registries not only in the EU, but anywhere all over the world. The survey covers both active and expired rare diseases registries. The final aim is to develop tools and services in support of existing registries and to favour the creation of new ones where needed. If you are somehow involved in registry activities, please fill out the online survey. Any participation is of paramount importance as only through joint efforts great advances can be contributed to the rare disease field.

Some interesting outcomes so far:

- over 60% of studied registries have started after 2000
- almost 40% represent autonomous initiatives of clinicians, patient associations, etc.
- more than a quarter are set up with funding coming from national authorities

Source: *EPIRARE (http://www.epirare.eu/)*



Joint EUCERD/EMA workshop on the public-private partnership for registries in the field of rare diseases

In early October 2011 the EU Committee of Experts on Rare Diseases (EUCERD) and European Medicines Agency (EMA) organised a workshop at which more than 60 participants discussed the issue of public-private partnership concerning the rare diseases registries.

The present main focus is on organising registries by disease and not longer by product, in order to better deal with the requests of regulators and payers to access data for the assessment of the clinical utility of new drugs for which registries are an excellent source.

Currently, a consensus is established on several key aspects of rare diseases registries:

- fragmentation of data sources should be avoided
- public/private partnership should be considered as a reasonable option
- open-access to data should be promoted
- management by academia is identified as a solution to ensuring long-term sustainability

The outcomes of this workshop will serve as a basis for the elaboration of a EUCERD recommendation in this field. The full public report of this meeting can be found on EUCERD website:

http://www.eucerd.eu/upload/file/Meeting/EUCERD/EUCERDWorkshopRegistries2011.pdf

Source*: EUCERD (http://www.eucerd.eu/)*

29 FEBRUARY – INTERNATIONAL RARE DISEASE DAY

Changes take place slowly, but it is only up to us that they still happen

The last day of February gives us a formal occasion to think again about the rare diseases – it is the International Rare Disease Day. An initiative of the European organization of patients with rare diseases (EURORDIS), it started 5 years ago and it is gaining more and more followers all over the world. This year's Rare Disease Day theme is solidarity – the solidarity among patients, between patients and doctors, between patients and institutions, between patients and society, solidarity among people.

Solidarity is a very debatable concept. It means collectivity, common spirit, unity, cooperation, mutual support, reciprocity, community, consensus, togetherness. We will define it as a moral responsibility toward the others. Solidarity sounds great, people love to point it out as their own feature. But it is not shown in words, it is proved through actions. Solidarity people leave their activities speak for them, solidarity people help others without looking for a specific occasion or a reason.

Rare diseases are still a specific social problem. They are extremely severe and almost unknown to the unaffected members of the society. Misery and pain of people with rare diseases and their families are as so different and specific, as diverse are their disorders. One thing particularly unites them – the feeling of being overlooked by the others. Is there solidarity in a society, where in order to undertake actions some tragedy must happen first or some innocent children must be gone?

Solidarity is demonstrated and is not declared. Rare diseases problems do not need a specific day in the year when to recite speeches to the media. Rare diseases need appropriate action in all 365 days a year. Patients need care, follow up and rehabilitation, physicians need appropriate training, facilities and resources to do their job. Solidarity society is concerned about these issues, despite the fact that the patient with a rare disease may be a single one in the country. He may be our co-worker, our neighbour, our friend, our relative, this person also has its own dreams and potential to develop. The reasonable and solidarity people act on these issues rather than waiting for to become directly affected. Should we wait for our child, our friend or ourselves to be affected by rare diseases in order to realise the gravity of the problem?

The problem of rare diseases is not only the shortage of funds. It is much more a matter of respect, comprehension and adequate organisation. Situation, in which a patient with a rare disease, apart from anything else must pay an impossible price for a medicinal product just because he has a rare disease, he is the only patient in the country and institutions simply are not aware of his existence, is totally unacceptable. It is an absurd situation, in which a patient with a rare disease is denied access to treatment abroad, pointing out that here there is no clinical pathway for this disease. These examples are hundreds...

People with rare diseases in Bulgaria often say that it is not the lack of funds, but the indifference, which is killing them. On 29 February let's not jump to statements, exciting our solidarity with patients with rare diseases and our ful support to them. May on this day each of us remember that nobody is protected against problems as rare diseases, which could seem quite distant today, but tomorrow they may fall upon us in full force.

Meanwhile, people with rare diseases are not waiting for changes to come on their own. Transformation has to start from somewhere and patients have understood that they have to initiate and maintain this process. People with rare diseases in Bulgaria have realised that only complaining won't produce miracles. They have begun to organise their own campaigns – seminars for GPs and medical students, charity activities for repairing rooms for patients with rare diseases in major hospitals. They have known for themselves what it means solidarity – not to think about your own problem, but to act and help others by educating, informing and creating. In addition to raising awareness and lobbying for better health care, patients with rare diseases have started to teach their fellow citizens of solidarity.

You can too become a part of this change!

To our traditional blitz-interview we have invited representatives of two of the most active patient organisations of people with rare diseases in Bulgaria – Ms. Elena Eneva (DEBRA Bulgaria) and Mr. Todor Mangarov (Association Pulmonary Hypertension).

The team of "Rare Diseases & Orphan Drugs" would like once again to thank them for the time, spent to answer the questions!

Would you like to present yourself? What is the reason for your commitment to the rare diseases cause?

Elena Eneva (EE): My name is Elena Eneva. I am a member of the Board of DEBRA Bulgaria, patient organisation of people suffering from Epidermolysis bullosa. I first learned about this disease, when my youngest son was born with it. Fighting this rare disorder has changes the life of our family, it has required a lot of knowledge and resources and has showed me how important is people's support in a difficult situation. After the death of my son I donated all these materials to other children in need and so I met other people with rare diseases. Rare diseases patients and their families often have nobody to conctact and seek information, because few people are familiar with these problems. That's why rare diseases patients need to build their community in which to share experience and help each other. I am trying to help with whatever I can and I believe that people living with Epidermolysis bullosa in Bulgaria should be like a big family and feel the mutual care and support.

Todor Mangarov (TM): My name is Todor Mangarov. I am president of the patient association of pulmonary hypertension (APH). I live and work in Pleven. I am patient with the rare disease pulmonary arterial hypertension (PAH). The specific reason to get involved with rare diseases cause is personal – I want to live. I was diagnosed with PAH on 10 October 2006, when none of the drugs for the treatment of PAH was reimbursed. Currently this situation has changed dramatically. But there is still more work to do.

What is Rare Disease Day to you personally? What attitude, what hopes you imply in this event?

EE: Rare Disease Day provides an opportunity to talk about the problems of rare disease patients. It is also an excellent chance to inform the society about rare disease patients' specific needs.

TM: My personal opinion is that Rare Disease Day should be a major information campaign. People turn aside when someone talks about "diseases" and start to care only when it affects them personally. My experience shows that doctors, especially GPs, have high self-confidence, but the truth is quite different. A massive information campaign will help to enhance the awareness of both medical professionals and society.

How does this event contribute to the cause of patients with rare diseases in Bulgaria and rare diseases topics in generally?

EE: Rare Diseases Day is a good focal point, an occasion for organising events to draw public attention to the problems of people with rare diseases.

TM: Improving public awareness would also increase tolerance to different people. Better training of doctors would lead to a rapid and timely diagnosis, which means much more favorable prognosis for the patients.

Organising such an event is not an easy task, but attracting the public attention to it is the real challenge. How do you think society and ordinary people should be informed and involved in the ideas of this initiative?

EE: Information on rare diseases is very extensive, quite complex and generally concerns very few people. The personal stories are what attract the attention of the society. They always produce sympathy to a concrete patient and his family, but they do not usually cause large-scale actions to help the other affected people. I think it is the most important that public health authorities and medical professionals learn more about these issues because they are those who by their actions can contribute to a better life and treatment for all people with rare diseases.

TM: Personally, I am not a supporter of aggressive and intrusive methods of campaigning for sympathy. I prefer well-positioned promotion methods. For example, APH has previously organised a PAH week throughout the Bulgarian medical universities and major medical centers. We presented posters, we distributed brochures, introducing medical students, pulmonologists and cardiologists and patients with PAH features, as well as the activities of our association.

The theme of Rare Disease Day this year is solidarity – among the patients, between patients and doctors, patients and institutions, patients and society, solidarity among people. How do you see this concept in our country?

EE: People with rare diseases are a minority everywhere. For them it is very important to have solidarity with each other and mutual support. Everything that we achieved last year in Varna was a result of cooperation and solidarity between representatives of different patient groups. We set the beginning of good partner relations with the association of general practitioners, the governing body of “St. Marina” University Hospital and the students of the Medical University of Varna. The result of this multistakeholder partnership was the workshop for GPs “How to recognize and treat rare diseases”, “I breathe” and “Teddy bear hospital” events, as well as the project for repairing and furnishing separate rooms in the hospital for cystic fibrosis patients. A very beautiful symbol of our solidarity was the 2012 calendar with photos of children with different rare diseases. I am sure that now everyone is convinced that together we are capable of doing great things!

TM: There are recent developments on this issue, but there is still a lot to pursue. After the last Rare Disease Day we got connected with many new people with PAH. We have extended our contacts with physicians, institutions and industry. APH is now affirmed as the primary liaison between the different stakeholders. But the most important is that patients gained confidence that our organisation is able to help on specific problem very timely and properly.

In 2011 Varna and Pleven were the two most active cities in Rare Disease Day celebration. What do you plan for this year?

EE: As the first rare diseases seminar for GPs and medical students was greeted with huge interest, we are planning to hold again such an event, introducing some other rare diseases. I hope very much that it will coincide with the opening of the cystic fibrosis rooms, so we would have a double occasion to celebrate.

TM: To raise public awareness of rare disease we will organise stage productions of the paly "SubUrbia" by Eric Bogosian in Sofia, Plovdiv, Pleven and Stara Zagora. The theme touches on many actual problems of modern teenagers – drugs, alcohol, sex and money.

What is going to be the main accent this time?

EE: We will once again focus on diagnosis and treatment – the two most important things for people's survival and improvement of life.

TM: This year we will pay great attention to the future doctors – the present day medical students.

Rare diseases problems are literally endless, but from where to start in your opinion?

EE: Problems occur at every stage of healthcare for people with rare diseases – delayed diagnosis, difficult access to drugs, shortage of expert professionals, lack of rehabilitation, lack of support for social adaptation and integration. I believe that patient organisations can contribute with something in all these areas – by providing translated information about rare diseases and their treatment, lobbying government bodies for better care, organising training seminars for professionals, assisting individual patients, maintaining contacts with foreign associations, lobbying on European and worldwide level. Active patient organisations are the only chance for ensuring the rights of the smaller groups of suffering people.

TM: From the cradle of education – the university. This is a long-term investment in human capital.

The National Programme for Rare Diseases is now entering the fourth year of its action. Being proactive members of rare disease patient community, what are your impressions of it?

EE: The programme is a good plan for comprehensive actions to improve care for people with rare diseases, however so far it remains only on paper. Genetic services are the only activity, which has received funding from the programme. The goal is the prevention of genetic diseases, but unfortunately rare diseases are so numerous and unpredictable, so it is impossible to prevent rare diseases only through genetic testing. None of the programme activities, directed to the already born and adult patients, is being executed. It is time to stop declaring on different forums that Bulgaria has such a programme. We want to see concrete actions and outcomes.

TM: We have a national programme, that it is not working. In most EU countries there are no such programmes, but it is not problem for rare diseases patients to get high level treatment there.

What would you like to see in a year time when we would be once again marking this day?

EE: More generally, I would like to see people with rare diseases confident of receiving adequate treatment and using their energy to realise their dreams. And in particularly, I want to see the establishment of a center for rare diseases in Varna, an Epidermolysis bullosa center in Pleven, provision of bandages and medication for epidermolysis bullosa patients and gradual change in the society's attitude towards people with rare diseases.

TM: I should admit that during the past year PAH patients have seen a significant change in positive direction. I would be very happy if I could re-affirm it in a year time.

What will be your message to the people with rare diseases in Bulgaria on 29 February?

EE: Be tenacious and stand for your right to adequate care! Changes take place slowly, but it is only up to us that they still happen.

TM: Believe! There is always some hope!

And to the Bulgarian society?

EE: Rare diseases usually come unexpectedly and change the whole life of the patient and his family. Children are most affected. Be understanding and supportive to these people, so they can grow up and realise their potential!

TM: Patients with rare diseases are people just like you.

You would like to get involved with Rare Disease Day 2012?

"Rare but strong together" is the official motto of Rare Disease Day 2012. The Bulgarian National Alliance of People with Rare Diseases and Association Pulmonary Hypertension Bulgaria are organizing a charity performance of the theatre play "SubUrbia" in Plovdiv, Pleven, Stara Zagora and Sofia, respectively on 12, 18, 25 and 29 February 2012.

"SubUrbia" is a play of the American playwright Eric Bogosian. It features young artists from "Mask" Theatre Studio and it is directed by Lyubov Pavlova. The ticket price is 2 lv. For more information, please contact NAPRD coordinators in the corresponding cities.

Association "Cystic Fibrosis" continues its charity campaign to raise funds for renovation and equipment of a „cystic fibrosis" room at the children's ward of University Hospital "St. Marina". With the support of many friends and partners the association has prepared a new charity calendar for 2012, which is full of the smiles of children, suffering from rare diseases. If you want to help establishing a "cystic fibrosis" room, you can buy one of these smiling calendars. All proceeds from the sale will support this project, which will provide a better treatment of rare diseases kids. You could find how and where to buy this calendar on the website of the Association "Cystic Fibrosis".

NEW CONCEPTS OF BLEPHAROSPASM TREATMENT AND REHABILITATION

Our Rare Diseases Library expert and director of Medical Center "RareDis" – Dr. Radostina Simeonova, PhD gives an overview of the rare disease blepharospasm.

Definition

Blepharospasm is a localised form of muscular dystonia, characterized by involuntary clonic and tonic contractions of periocular muscles, causing complete or partial closure of the eyelids for several seconds to several minutes. The first symptoms are usually between 40 and 70 years of age. Women are affected more frequently than men. Male to female ratio is about 2:3.

Etiology

The exact cause of primary blepharospasm is unclear. It is most likely due to abnormal function of the basilar nuclei in the CNS, which play an important role in movement coordination – superior colliculus and substantia nigra of reticular formation. Blepharospasm may be also due to another underlying disease. In this case it is called secondary blepharospasm or reflexive spasm.

Genetic counseling

In most cases, there is no genetic disorder. However, familial forms have been recorded, especially in Meige syndrome. Family history is reported in 20% of cases.

Clinics

The manifestation of symptoms is usually gradual over several weeks or months. At the beginning it can be expressed as frequent blinking that later develops in classic muscle contractions. Symptoms are bilateral – both eyes are affected. Cramps are more defined when listening, walking, gazing, looking up. Conversely, they can significantly weaken while speaking, singing, looking down, coughing, and concentrating. Spasms are less intense in the morning and well expressed during the day.

Diagnosis

Diagnosis is based on typical clinical signs. The exact clinical form should be determined. It is important to distinguish primary from secondary blepharospasm, because their treatment is different.

Established therapy

Treatment of blepharospasm is symptomatic, unless it is secondary or due to another underlying disease (in this case, treatment is aimed at the primary disorder, which causes blepharospasm). Treatment could be conservative medication (it can be taken orally or by local injection), nonpharmacological, complementary and surgical techniques.

A. Conservative medication:

1. Medication, injected locally:
   - Injections of Botulinum toxin type A. It ensures a stable result for 3-6 months. In case of neutralising the action of toxin A by antibodies, it is possible to provide alternative treatment with Botulinum toxin B.
   - Alcoholic injections. The most common method until 1970.
2. Medication administered orally:
   - Benzodiazepines
   - Myorelaxants
   - Anticholinergics
3. Alternative treatment:
   - Psychotherapy, relaxation techniques (autogenic training), yoga, meditation, Feldenkrais method, hypnosis, controlled exercise such as Pilates and soft tissue techniques, acupuncture to relieve pain and spasms.

4. Physical therapy

The disorder primarily affects motor and facial muscles. That's why the applied physical methods are mostly symptomatic and have cosmetic effect. Teaching the patient to deal with muscle contractions and to use replacement movements and techniques of control is the task of the medical rehabilitation. Combinations that target all the patient complaints are often used and frequent rehabilitation courses should be sought – at least one in two months. Emphasis is placed on reflex methods in the neck, electrical and light treatment, kinesitherapy. The application of specialised kinesitherapy techniques significantly improves the patient's condition. Laser puncture is also one of the recommended approaches. Underwater gymnastics and underwater massage methods demonstrate a proven positive effect. It is very difficult to patients with blepharospasm to perform professional tasks due to limited eye sight and psychological barriers, posed by the aesthetic problem. Different variants of occupational therapy have double objective – a therapeutic and a re-training ones. Speech therapy, as well as consultations with psychologist may be also conducted.

B. Surgical treatment – it is recommended when the efficacy of botulinum toxin starts decreasing over time (due to the formation of anti-botulinum toxin antibodies) or it is ineffective from the very beginning (due to the absence of botulinum toxin receptors). Various surgical techniques are used.

Prognosis

Primary blepharospasm is not a life-threatening condition, but its progressive course strongly influences the patients in psychological and social aspects. The condition can be improved with prolonged conservative treatment and/or surgery.

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UPCOMING RARE DISEASES CONFERENCES, WORKSHOPS AND INITIATIVES

- **29 February 2012 – Rare Disease Day (more information on www.raredis.org, www.rare-bg.com and www.rarediseaseday.org)**
- **23-25 March 2012 – Balkan rare disease patients meeting, Sofia**
- **16 March 2012 – Annual workshop of ICRDOD medical consultants, Plovdiv**
- **31 March – 1 April 2012 – 3rd Rare diseases seminar for medical students, Plovdiv**
- **6-13 April 2012 – 2nd International rare diseases school for health authorities**
- **23-25 May 2012 – 6th European conference on rare diseases and orphan products, Bruxelles (www.rare-diseases.eu)**
- **21-23 June 2012 – 3rd All Russian conference for rare diseases and Kick-off meeting of the Euroasian rare diseases alliance, Moscow**
- **14-15 September 2012 – 3rd Bulgarian national conference for rare diseases and orphan drugs (detailed announcement in next issue)**

EUROPEAN LYMPHANGIOLEIOMYOMATOSIS FEDERATION

Lymphangioleiomyomatosis (LAM) is a degenerative and invariably fatal lung disease which affects women in their child bearing years. The disease significantly and progressively impairs respiratory function, leading to the formation of cysts in the lungs and subsequent destruction of healthy lung tissue. Difficult to pronounce and even more difficult to diagnose, LAM is often confused with other respiratory conditions, including asthma and bronchitis.

Following the first European LAM Research Conference in 2010, LAM patient and research groups within Europe aim to create a European LAM Patient Federation by December 2012. The newly formed federation will focus on supporting LAM research and will coordinate communication with existing LAM patient groups.

If you are further interested in LAM or you want to join the Federation, please visit http://www.europelamfederation.org/.

Editorial Box

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