www.raredis.org June 2011 (Issue 4, Year II)

RARE DISEASES ORPHAN DRUGS

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

FOCUS ON:
MEDICAL STUDENTS – PARTNERS FOR FUTURE

Dear friends,

Here comes the time for the new issue of our newsletter.

The month of June has been traditionally the time for planning – the summer leave, the remaining half a year, etc. So, we would like to use this opportunity to inform once again and to invite you at the Second National Conference for Rare Diseases and Orphan Drugs, which will be hold on 9-11 September 2011 at Novotel Plovdiv. Why is it so important to attend? First, you will have the opportunity to meet and listen to the most high-profile rare diseases stakeholders in Bulgaria. But not only that, you will have the chance that these people to have your feedback and comments on what's going on and to take account of them. That's because the overall interaction is the leading factor for success in the field of rare diseases. Of course, we have once again tried to invite interesting speakers who are leading experts in their field and to make a balanced program for all the groups of participants. So, do not hesitate - go register on the conference website – www.conf2011.raredis.org.

The main topic of this issue is medical students and their role and importance in the implementation of adequate strategies in the field of rare diseases. They not only form our so-called "bank" for future professionals in the field of healthcare, but they are also generators of new ideas resulting from the dynamic development of the younger generations. Properly trained and informed, they can be the key for solving long-standing problems. Moreover, as all young people, medical students are full of energy and ambition of self-proving and action. People just have to believe in them and give them a hand.

Have a nice time while reading the newsletter!

SECOND NATIONAL CONFERENCE FOR RARE DISEASES AND ORPHAN DRUGS

9-11 September 2011
Novotel Plovdiv

PRELIMINARY PROGRAMME

Session 1	**Health information and prevention of rare diseases**
Lecturers and moderators: Prof. Draga Toncheva, Prof. Rumen Stefanov, Dr. Tsonka Miteva **Main topics:** Best practices for prevention of rare diseases Epidemiological registries for rare diseases	
Session 2	**Best practices for diagnosis and treatment of rare diseases**
Lecturers and moderators: Prof. Lyudmila Mateva, Prof. Stefan Goranov, Prof. Asen Dudov, Prof. Valeria Kaleva, Prof. Iva Stoeva **Main topics:** Screening and functional endocrine diagnosis of rare diseases Therapeutic concepts for rare diseases management: Crohn disease, Hemophilia, Gaucher disease, Rare tumors, Thalassemia major, Chronic myeloid leukemia	
Session 3	**Rehabilitation and social integration of rare diseases patients**
Lecturers and moderators: Dr. Radostina Simeonova, Mariana Angelova **Main topics:** Role of the medical rehabilitation in rare diseases patients Role of the psychology in the integrative approach to rare diseases Specialized social services for rare diseases patients	
Session 4	**National programme for rare diseases**
Lecturers and moderators: Prof. Radka Tincheva, Vladimir Tomov	

For further information:

www.conf2011.raredis.org

WILLIAMS DISEASE SUPPORT GROUP

Support group for parents of children with Williams syndrome is being formed in Sofia. It is a proven fact that sharing of experience and support among other people having the same problem helps greatly in both psychological and practical terms to deal with daily cares for a child or a relative with a rare disease. It also builds stronger self-confidence and determination. That's why the group will aim to exchange experience and support among parents for better care for children with Williams disease, as well as information about progress in treatment and rehabilitation of patients with this disease. For more information, please contact Ms. Lucy Atanasova (lyusy.atanasova@abv.bg; +359 886 968 956).

EU CLINICAL TRIALS OFFICIAL REGISTER

EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

The European register of clinical trials has recently been launched. It is an online register where users can find information on the design, the sponsor, the investigational medicinal products and therapeutic areas involved and the status of the clinical trial. It covers clinical trials in EU Member States and the European Economic Area and clinical trials which are conducted outside the EU/EEA if they form part of a pediatric investigation plan. The website is hosted by the European Medicines Agency (EMA). For further information, please visit www.clinicaltrialsregister.eu.

EPIRARE PROJECT

EPIRARE
European Platform for Rare Disease Registries

The EPIRARE Project, funded by the EC, officially started on 15 April 2011. It will focus on preparing the feasibility of a future common European platform for rare diseases patient registries and its long-term sustainability. That's why rare diseases patients, researchers, HTA experts, medical professionals and health authorities will collaborate to build consensus and synergies to address regulatory, ethical and technical issues, associated with the registration of rare diseases patients in EU. Particular points of interest will be the elaboration of possible policy scenarios and the evaluation of the feasibility of registration of minimum data set, common to all rare disease. The project is coordinated by the Italian National Institute of Health (ISS). The Information Centre for Rare Diseases and Orphan Drugs through BAPES is an associated partner in this project. To read more information, please, visit the project official website – www.epirare.eu.

MEDICAL STUDENTS IN BULGARIA – PARTNERS FOR FUTURE

The future is in the children and young people,
the future of rare diseases is in the hands and minds of tomorrow's doctors.

The problems in the field of rare diseases are complex, they have been accumulating for years and today their successful solution depends on the joint and synchronous actions of all stakeholders. Among them there is a special category, whose skills and attitude will determine the direction of development not only of rare diseases, but of the public health in general. These are today's medical students, our future doctors. This is the lead story in this issue of our newsletter.

Rare diseases as a separate academic discipline are not present in the higher medical education in Bulgaria. This is perhaps one of the reasons why even very good doctors are not familiar with the problem of rare diseases and more specific approaches that they require. It is the timely awareness and targeted training which are key factors for improving the professional skills of medical professionals and hence the quality of life of people with rare diseases in Bulgaria. It is not exaggerated to say that investing in quality education of young doctors and raising their awareness of rare diseases are of great social importance and a kind of guarantee for successful future actions in this area.

Realizing these facts from the very beginning of its activities, BAPES have been actively cooperating with the Association of Medical Students in Plovdiv (AMS-Plovdiv), as well as with other similar associations in Bulgaria. AMS-Plovdiv is an important partner in our mission to inform, educate and influence for rare diseases. A number of our ideas and joint activities such as the Training seminar for medical students and patients with rare diseases have become a tradition, the assistance and contribution of medical students for organizing events such as the National conference for rare diseases and Rare disease day are irreplaceable. Furthermore, in this way we get very responsible partners and also highly informed and skilled on rare diseases young people who are eager to show their knowledge and competence in practice and to work for the cause of rare diseases in Bulgaria.

In this issue we will introduce to you just a small fraction of the initiatives that we have undertaken together with AMS-Plovdiv to show you how enterprising and creative can be medical students when working in rare diseases field.

TRAINING SEMINAR FOR MEDICAL STUDENTS AND RARE DISEASES PATIENTS

The main idea of this joint initiative is to raise awareness of medical students on the existence of the rare diseases problem and to present them the challenges, that as future doctors they will have to face. Holding such meetings in an informal setting allows the participants not only to get informed but also to express opinions and to share ideas, to be encouraged and motivated to not remain indifferent to the problems of people with rare diseases, something which is perhaps the worst fate for patients.

Another highlight of these seminars is that students can learn about these issues from the rare diseases patients themselves, who are the most reliable and complete source of information about rare diseases. The future doctors can not only see the purely medical side of the disease, but also to feel its social, economic, psychological and most importantly – its human aspect.

On the other side, patients with rare diseases have a chance to participate in shaping tomorrow's doctors, influence their views on what should be healthcare and not at last place, to find partners in their face for current activities and projects. The participation in the seminars held so far of representatives of the National alliance of people with rare diseases, Organization of thalassemics in Bulgaria and Association "Cystic Fibrosis" has notably contributed to implement these ideas.

Thanks to initiatives of this kind and the members of AMSB-Plovdiv, today more and more students and young doctors are starting to think about the education they receive. They want up-to-date quality, relevant knowledge and opportunities to develop their ideas and initiatives. All our civil society and rare diseases patients in particular need people like that.

That's why the partnership with the medical students' community will remain an important priority for BAPES – to promote education and science among students and to invest in the professional training and personal qualities development of the future medical professionals.

SUMMER SCHOOL FOR RARE DISEASES

Inspired and enthusiastic by the series of successful training seminars, medical students consider to be extremely necessary to spread knowledge on rare diseases among their community. Being part of European and global networks of associations of medical students, ASM-Plovdiv knows that the problems of patients with rare diseases are practically unknown for the student organizations. IFMSA (International Federation of Medical Students Associations) and EMSA (European Medical Students Association) are umbrella structures of medical students, building up international communities that allow not only the exchange of a huge flow of information, but they also give its members the opportunity to undertake activities in large scale, thus attracting public attention and expressing position on important social issues on international level.

Each year EMSA invites one of its full members to organize a summer school under its auspices. The hosts have the right to choose the theme of the summer school based on their experience and appraisal.

In the summer of 2012, this initiative will take place in Bulgaria and will focus on rare diseases. The objectives and tasks of the summer school will be based on the already successful model of partnership between AMS-Plovdiv and BAPES, which has been multiply applied in practice through the joint seminars. Using the extensive information channels of EMSA, rare diseases issues can find a wide resonance not only among the school participants themselves, but down through all the membership network of each national association. This will undoubtedly contribute a lot for moving rare diseases and the difficulties of these patients and their families out of the darkness.

On the other hand, AMS-Plovdiv and BAPES estimate that by raising awareness and specialized training of medical students, the future society will have a much more enlightened medical community. It is precisely one of the main hopes of people with rare diseases to finally deal with this difficult social problem, so are rare diseases.

TEDDY BEAR HOSPITAL

It is a well known fact that most rare diseases are of genetic origin and the vast majority of patients with rare diseases in our country are actually children. This inevitably affects kids' fragile psyche. The life of these children does not go into games and fun as their peers do, it is not so carefree and easy. They spend much of their time walking from doctor to doctor, undergoing procedures, giving blood sample or taking another medicine.

So, in Plovdiv, for the first time in Bulgaria, an event was organized, which was aimed exactly at this particular problem for rare diseases – helping kids to overcome the fear from medical doctors. Teddy Bear Hospital Project is part of an international initiative, which is undertaken by medical students in countries like Great Britain, Norway, Italy and France. The idea is through fun and games to show and convince the kids that visits to the doctor were not so horrible, that the disease is a normal part of life and they should not be afraid of it.

At the same time, Teddy Bear Hospital helps also the medical students to get acquainted with pediatrics, with children's feelings of fear and excitement, and most of all – to try now to learn how to build the relationship doctor – patient (child), which should be based extremely on mutual trust and warmth. It is something that may seem insignificant, given the very serious other problems for people with rare diseases and their families, but on the other hand, exactly because of the chronic nature of rare diseases it is very crucial point, which could ease greatly the little patients and their parents in the course of treatment, create a friendly atmosphere in the visited hospital and not least – help the doctor himself in performing his professional duties.

Teddy Bear Hospital Project took place in November 2010 in Plovdiv and was organized by AMS-Plovdiv with the support of BAPES. The example was really catchy and was followed by similar "hospitals" in Varna and Stara Zagora in 2011, once again as joint activity between students and patients.

We would like to thank ASM-Plovdiv and their team for the collaboration and assistance in preparing these articles.

RARE DISEASES THROUGH THE EYES OF MEDICAL STUDENTS – AWARENESS AND ATTITUDES

Four years ago BAPES together with AMSB realized a survey among the medical students. It aimed to explore their awareness and opinion about rare diseases and problems, which doctors and patients are facing in getting successful diagnosis and treatment. The questionnaire contained some principal issues, such as rare diseases prevalence in Europe, estimated number of people with rare diseases in Bulgaria, what is "orphan drug", specific activities for prevention of rare diseases. It also asked the respondents for their personal opinion and attitude towards the problems of people with rare diseases in our country, the public awareness of them. Students could express their view whether the medical education should include more information about rare diseases, give their advices and recommendations how to improve knowledge and competence of medical professionals and quality of service to patients and to increase support among the society as whole.

The results showed that the level of awareness of medical students to the rare diseases problems requires significant improvement. However, the more important is the strong support and positive attitude among this target group towards the actions in the field of rare diseases. Medical students understand that sizable and sustainable progress on that issue could be achieved only through the efforts of all. It should also be pointed the almost unanimous interest in the potential inclusion of new optional course on rare diseases to the curriculum in medical schools. This would already give a significant boost for rare diseases to move on top of the public health's agenda and consequently to the society's one too. BAPES and AMS-Plovdiv are already working on it and it is a matter of time for this idea to be implemented in practice.

In preparing this newsletter's issue we decided to check again the awareness and attitude of medical students about rare diseases. A freshman from the Medical University of Plovdiv and a sophomore from the Faculty of Medical Sciences at University of Groningen (Netherlands) answered our questions. The Dutch student was in Bulgaria under international student exchange programme and during her stay she visited the Information centre for rare diseases and orphan drugs in Plovdiv. We have deliberately chosen students who are just starting their medical education and are still forming their professional and personal attitude. The aspect of the different nationalities is also purposely sought, as we wanted to see the views from Bulgaria and a foreign country. Of course, no general conclusions based on these answers, nor claims for representativeness could be made. However, it is interesting and useful to see how the big number of actions and events in the field of rare diseases in recent years are felt and perceived by future doctors at national and EU level.

What is a rare disease?

Simeon Yordanov (SY): It is a condition which affects from 1 of 1 500 people to 1 of 250 000 persons.

Tessa de Wid (TW): A serious disease with a low prevalence.

Were you familiar with the problem before visiting the Information centre for rare diseases and orphan drugs?

SY: Yes, I knew that there are diseases which are extremely rare, but I didn't realize their big number, as well as all the problems arising from the specifics of the rare disorders.

TW: Not really. Of course, we have learned about some rare diseases during our curriculum, but not combined as a subject "rare diseases".

Do you know people suffering from rare diseases?

SY: No, I don't know people with rare diseases.

TW: I don't know such patients.

Do you think that the society pays enough attention to people with rare diseases?

SY: I don't think that the society gives enough attention to people with rare diseases.

TW: In the Netherlands there are a lot of campaigns for major diseases, such as cardiovascular diseases and cancer, to raise money for more research, but we hardly hear anything about rare diseases.

What is the policy on rare diseases in your country?

SY: I know that there is a strategy of the Ministry of Health on reimbursing the treatment of some rare diseases. However as far as I am informed, the numbers of drugs which are covered under this way is very low, so is the list of diseases. There are no specialized centres for rare diseases management, as well as the necessary conditions for treatment of these disorders in our country.

TW: No idea actually.

Do you think that there is a disease which is so rare, that patients suffering from it are not important?

SY: No, I don't think so.

TW: The patients are still important, but if you only have the money for one research project, it benefits more people if your research is about a more common disease instead of a rare one.

What would you do if there is someone around you suffering from a rare disease?

SY: I would do anything possible to get adequate treatment and cares. I would help to contact another people, suffering from the same condition. I will also look for information about advanced treatment and expert centres.

TW: Try to help him as good as possible and try to find other people in the world with the same disease, so we can learn more about it and they can exchange their experience, and their doctors can also exchange information.

What would you do to protect yourself and your family from rare diseases?

SY: So, that's why genetic testing of pregnant women and the newborn screening are very important.

TW: I don't think it is possible to protect yourself, since a lot of rare diseases have a genetic component.

We would like to thank Tessa de Wid and Simeon Yordanov for the collaboration and assistance in preparing this interview.

MULTIPLE SCLEROSIS – GUIDELINES FOR TREATMENT AND REHABILITATION

DEFINITION

Multiple sclerosis (MS) is an autoimmune disorder with a suspected genetic predisposition, which in combination with certain environmental factors, leads to an autoimmune reaction, resulting in inflammatory demyelination in the central nervous system (CNS). The conduct of impulses in the CNS is damaged, leading to a neurological deficit.

CAUSES

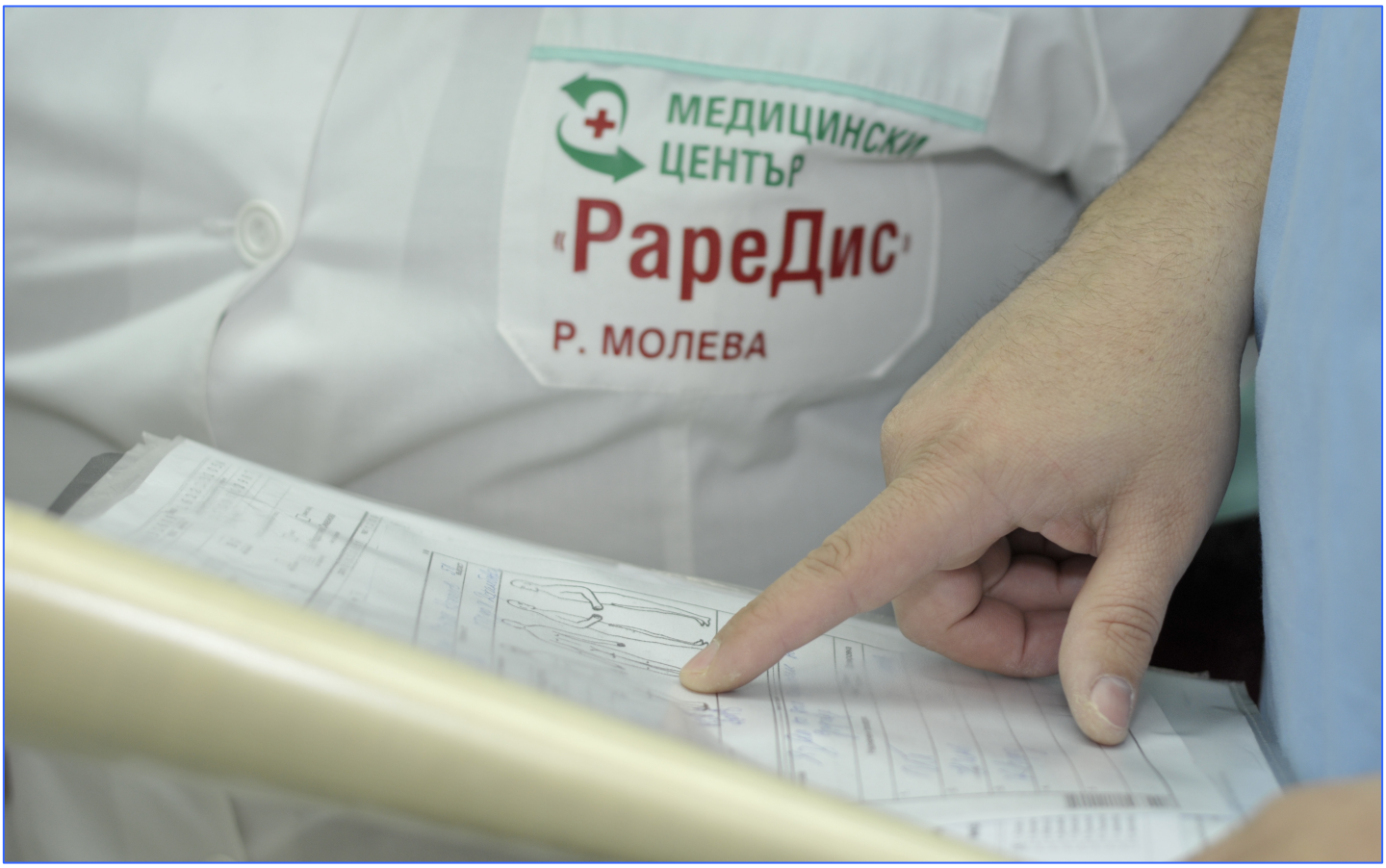
Activation mainly of T-lymphocytes and macrophages, inflammatory demyelination.

ESTABLISHED THERAPY

The therapy varies according to the form of the disease and whether MS is in relapse or remission.

The relapse treatment is to restore as far as possible the neurological symptoms which have occurred at this time and, of course, a large part of efforts is aimed at the rehabilitation process of the patient and his family.

There are several therapeutic approaches that are currently in use. Their indications are strictly specified. Depending on the format of the course of MS, betainterferon β-1a and β-1b, glatiramer acetate and the chemotherapeutic mitoxantrone are all used. Recently, a sphingosine 1-phosphate receptor modulator (SIPR), the first drug for per oral use with a unique mechanism of action has been approved. It acts as a functional antagonist of the receptor and prevents the release of certain subpopulations of lymphocytes from the lymph nodes, thereby reducing the autoreactive lymphocytes in the CNS.



REHABILITATION

The multidisciplinary team is of key role in the treatment of MS patients and the response to the psychological problems and social exclusion is of great importance the successful rehabilitation.

The neurorehabilitation stands on one of the top positions in the MS therapy. It aims to reduce the cognitive deficit, its impact on everyday life and to encourage the patient to successfully deal with the psychological effects of the disease – mood changes, depression and anxiety, reducing the stress and increasing the self-determination to manage the condition. It is only possible and efficient when an experienced psychologist is standing behind each patient and his family and competently consulting them. The comparative analyses show that patients receiving such therapy have better results when testing the memory functions and mobility.

MS is a disease that affects people of working age and their visual rehabilitation is a significant problem. Many patients lose their jobs, others retire due to illness. Training of these patients to use their residual visual capacity and appropriate professional prequalification would also help them revaluating their everyday lives. That's why the inclusion of specialized centres for visual rehabilitation and professional prequalification is not only therapeutic, but also a social commitment of the multidisciplinary team.

In most of the time patient recovers from the relapsing effects and the medical rehabilitation is a leading tool in this process. Various factors that can be applied and rehabilitation programme is strictly individualized according to the personal functional deficit. Few centres work efficiently and individually with these patients, because the needed time for daily procedures is about 3-4 hours and under the current healthcare regulations is very unprofitable and unattractive to the medical institutions in Bulgaria. A possible solution is to create specialized centres for rehabilitation of people with rare diseases, within the university hospitals with an adequate funding provided by the National Health Insurance Fund.

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To get further information on MS, as well as on the opportunities for medical rehabilitation and patients' training, please contact Medical Centre "RareDis".

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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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ISSN 1314-359X | © BAPES 2011

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