Editorial

*Patients’ preferred policy scenarios rely on you! Take action now*

Dear Readers,

Do you want to become involved in decisions on ethical issues that affect you and other rare disease patients and representatives? Is providing your views in opinion polls just not enough? Practice voicing your opinion and learn from those who may have opinions different from yours by participating in a new debate game called Play Decide developed by EURORDIS and its partner ECSITE, the European Network of Science Centres and Museums.

Take the opportunity to express your preferred policy scenario to address challenges of “Cross Border Healthcare”, propose answers to the question “Is there any upper limit on what we should spend on a single patient? - the case of Orphan Drugs”; make your position known on “Diagnosis, information of the patient, genetic counselling”, “Neonatal screening”, “Stem cell research” and “Pre-implantation genetic diagnosis”.

Debate games on these six hot topics for national and EU policies affecting people living with rare diseases have been developed based on a broad consultation of our members and scientific experts in 2009. Today the games are downloadable online in 22 languages at [www.playdecide.eu](http://www.playdecide.eu). For those interested in organising large-scale events and in need of support, contact Nick Perez at nick.perez@eurordis.org.

Sessions to play the games of ‘patient preferred policy scenarios’ have already been organised across Europe and the results are posted on the Play Decide website. Now is the time for you to plan your own sessions with your organisations’ members. Take this opportunity to organise an event on the occasion of your next membership meeting or awareness-raising event; set up sessions with your board and scientific committees or even local health professionals and policy makers. Take the time to develop opinion on these important policy issues and to share them with other rare disease patients across Europe by uploading the results of your discussion on [www.playdecide.eu](http://www.playdecide.eu).

Though the Play Decide exercise comes in the format of a game, it is a discussion tool on serious topics, designed for 4 to 8 participants lasting 1.5 hours. But time flies when you are having fun!

Yann Le Cam
"First International South Caucasian Conference for Rare Diseases & Orphan Drugs"

The rare disease movement spreading to the far end of Europe!

The First International South Caucasian Conference on Rare Diseases and Orphan Drugs (ISCORD) was held in Armenia in October and the First International Conference on Rare Diseases in Georgia was held in September. Both events could mark the beginning of a new era for rare diseases in this region of Europe...

Two pioneering rare disease conferences were held in Armenia and Georgia this fall. “These conferences are the result of years of work of a few dedicated health professionals and patient advocates. They are clear signs that the rare disease community is mobilising to include rare diseases in the health policy agenda, even in these remote parts of Europe where other health priorities are overwhelming,” says EURORDIS’ CEO, Yann Le Cam.

The First International Conference on Rare Diseases and Orphan Drugs took place in Yerevan, Armenia on October 7-8. A month before, another international conference on rare diseases had taken place on Sept 10-11 in Tbilisi, Georgia. The main aim of these meetings was to draw the attention of the medical, scientific, academic communities and government authorities to the field of rare diseases in these countries, and to help develop international and regional collaborations between researchers, doctors, patient organisations and the pharmaceutical industry.

The Georgian conference was organised by the Georgian Foundation for Genetic and Rare Diseases (GeRaD) and the Tbilisi State Medical University under the auspices of the First Lady of Georgia, Mrs Sandra Elisabeth Roelofs and her Charitable-Humanitarian Foundation SOCO. The Conference covered the following diseases: Osteogenesis Imperfecta, Cornelia de Lange syndrome, Shwachman Diamond syndrome, Epidermolysis bullosa, Phenylketonuria, Prader-Willi syndrome, beta-Thalassemia, Sjogren’s syndrome and Cystic fibrosis. It attracted international speakers from neighbouring Armenia, Bulgaria, Poland and Russia, and further afield from Canada, Germany and the Netherlands.
The Armenian event was organised by the Armenian Association for NeuroHereditary Diseases and co-organised by EURORDIS, under the auspices of the Centre of Medical Genetics of the Yerevan State Medical University. EURORDIS’ CEO Yann Le Cam was co-Chairman of the Conference and member of the Organising Committee. He presented an overview of rare diseases as public health issue in European policy. Successful experiences of rare disease patient mobilisation in neighbouring countries were presented by Dorica Dan, President of the Romanian National Alliance for Rare Diseases, Tamar Chigladze, President of the Georgian Foundation for Genetic and Rare Diseases (GeRaD) and Svetlana Karimova, head of Genetics, the Russian Information centre on rare diseases and orphan drugs. Prof Josep Torrent i Farnell, past-Chair of the COMP, presented an overview of the EU Orphan Drug regulation, its ten years of achievements, its benefits and challenges for patients in South Caucasus.

Scientific presentations focused on rare diseases such as MPS, rare inherited metabolic disorders, muscular dystrophy, mitochondrial diseases and phenylketonuria. Discussions focused on neo-natal screening, improving access to earlier and more reliable diagnosis, support for specialised centres in order to provide genetic counselling, care and access to medicines.

A special session co-organised by the International Prader-Willi Syndrome Association (IPWSO) was devoted to Prader-Willi syndrome featuring IPWSO’s activities and several scientific presentations on the state of the art for diagnosis and care of this disorder, as well as their experience in Armenia. A Prader Willi syndrome patient group will be established in Armenia following this conference.

In total, over twenty leading experts from 15 European countries and the USA participated in the meeting: scientists, doctors, public health officials and representatives of patient organisations from the EU, USA, Armenia, Georgia, and Russia.

The conference took place mainly through the efforts of Professor Albert Matevosyan MD, PhD, a dedicated physician and a familiar figure at EURORDIS, representing Armenia at EURORDIS’ Membership Meetings, European Rare Disease Conferences, Summer Schools and DIA Patient Fellowship Programmes for the last 8 years. Professor Matevosyan is the Head of the Republic’s Centre of Medical Genetics at the Yerevan State Medical University. The Centre includes the national laboratory of genetic diagnostics and clinical activities covering neuromuscular diseases and metabolic diseases, amongst others. He is also the President of the Armenian Association for NeuroHereditary Diseases, which he founded to increase the awareness of rare diseases amongst health care providers in Armenia, and to include rare diseases into the training program of medical students. In the last decade, the association has organised scientific conferences and training sessions on rare diseases for doctors. It has also promoted the inclusion of screening methods for phenylketonuria and hypothyroidism in the national plan for newborn screening. To organise this conference, Prof Matevosyan mobilised his students and
colleagues from the university hospital as well as friends from other hospitals and clinics.

“Great advances in the sphere of rare diseases in Armenia have been made possible due to collaboration and support from international partners like EURORDIS,” explains Prof Matevosyan. “In recent years we’ve been increasingly in contact with other actors in Europe. These encounters have helped break the isolation of health professionals and patient groups in this part of the world.”

The conference in Yerevan was also important because it brought the few existing concerned patient groups together (muscular dystrophy, haemophilia, cystic fibrosis and PWS): a core group which is planning to form an Armenian National Alliance of Rare Diseases.

“This conference is a starting point. It laid the foundations for future regional collaboration between Armenia, Georgia and possibly Azerbaijan, three countries that have cultural links and similar problems, as well as opportunities for addressing challenges in the field of rare diseases, despite the economic difficulties and the diplomatic tensions between the states. These collaborations are being developed in the framework of the EU policy on rare diseases and the growing dynamic of national strategies, while being inclusive of advocates and experts from other countries in Eastern Europe, such as Romania, Bulgaria and Russia, that share commonalities and can share their respective experiences” explains Yann Le Cam. “Even though these countries have many other health priorities to address, some activists are working hard to put rare diseases on the map at the far end of Europe!”

He adds “in Yerevan, Armenia, we agreed that the 2nd International South Caucasus Conference on Rare Diseases & Orphan Drugs will be held in 2011 in Tbilissi, Georgia. The conference will rotate every year between the different countries in the region”.

One of the organizers of the future conference, Executive Director of GeRaD, Dr Oleg Kvlividze said: “We would be very happy to welcome in Tbilisi people working in the field of rare diseases all over the world – representatives of patient-based organizations, health policymakers, medical doctors, researchers, representatives of international donor organisations and pharmaceutical companies. Their participation will enable us to adopt best practices in rare diseases and will be very helpful for the rare disease communities of South Caucasus countries. For our part, we will try our best to make their stay in our hospitable country full of most positive impressions.”

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**Rare Disease Day 2011: Focusing on health inequalities**
28th February 2011 will mark the fourth International Rare Disease Day coordinated by EURORDIS and organised with rare disease national alliances in 25 countries. On that day hundreds of patient organisations from more than 40 countries worldwide will organise awareness-raising activities and converge around the slogan “Rare but Equal”. If you would like to find out more about this year’s campaign, go to www.rarediseaseday.org. Get involved!

Patient organisations all over the world are starting to prepare already for one of the most important dates in the rare disease calendar: Rare Disease Day, 28th February 2011.

Rare Disease Day is intended to increase awareness of rare diseases amongst the general public, European, national and local health authorities and policy makers, health professionals, researchers, academics, the pharmaceutical and biotech industries, as well as the media.

Hundreds of patient groups and their partners, coordinated by national alliances at the country level, will organise a multitude of events to draw attention to rare diseases and the millions of people who are affected by them. Awareness-raising activities are planned across Europe, all the way to Russia, Georgia and Armenia, as well as in the US, Canada and as far as Australia, New Zealand, China and Japan!

This year, EURORDIS member national alliances in 25 countries will combine their efforts to put the spotlight on “Rare diseases and health inequalities” and converge around the slogan “Rare but Equal”.

More specifically, in 2011, Rare Disease Day will seek to draw attention to the:
• Gaps in health that exist for rare disease patients between and within countries in the EU;
• Gaps in health that exist for rare disease patients compared with other segments of society.

The campaign will serve to advocate for:
• Equal access for rare disease patients to health care and social services;
• Equal access to basic social rights: health, education, employment, housing;
• Equal access to orphan drugs and treatments.

“People living with rare diseases should be entitled to the same access and quality of care as any other patient. But today, the reality is far from that. The rarity of patients, medical experts, knowledge and resources aggravate the vulnerability of rare disease patients who are suffering from life threatening, debilitating, and chronic diseases. We are certainly not asking for more or better access and care than for other chronic diseases. On the contrary, we share the common cause of all chronic diseases. However, we believe that rare disease is one of the most dramatic cases of health inequality right now internationally and in Europe in particular,” argues EURORDIS’ CEO, Yann Le Cam.
This year the campaign will seek to drive home the message that it is important to level the playing field within and between countries in the area of rare diseases. At the European level, the immediate objective is to put rare diseases high on the public health agenda of each country involved in Rare Disease Day, and to promote the explicit inclusion of rare diseases in the third EU public health programme, which will decide policy and funding priorities for 2014-2020.

In order to contribute to this objective, EURORDIS is organising a one-day conference in Brussels, in partnership with DG Health & Consumers, to raise the issue of rare diseases and health inequalities and to discuss measures to mitigate them.

The conference will take place in the International Press Centre in Brussels. Participants will include patients and patient representatives, health professionals from rare disease centres of expertise, social researchers and academics working in the field of rare diseases, members of the EU Committee of Experts on Rare Diseases, industry representatives, as well as high-level officials from the European Medicines Agency and the European Commission’s Health and Social Affairs General Directorates.

The programme will be devoted to presenting the rare disease landscape in Europe and showing existing inequalities through case studies and surveys. The various surveys conducted by EURORDIS over the last 5 years, will be of particular use, especially EurordisCare 2 and 3 which investigated patients’ experiences and expectations regarding access to diagnosis and to medical and social services in 24 European countries, representing 18 different diseases. Other surveys, such as the ENserio Study conducted by the Spanish Alliance (FEDER) will also be used to illustrate the type of gaps in health care and social services that exist in a particular EU country.

Patients representing different countries and diseases will be invited to share their experience of access to health care and social services. Health professionals from selected centres of expertise will also explain difficulties of access their patients encounter on a daily basis.

In keeping with this year’s theme, EURORDIS is actively looking for stories of “rare disease health inequalities” that deserve to be showcased. A selection of stories will be posted on the website and included in the various communication tools prepared for Rare Disease Day. And, if the person submitting the story agrees, their testimony will be added to a bank of stories for the media.

“We are hoping to receive interesting and poignant testimonies to put a human face to health inequalities and to the policy we are advocating for,” explains Paloma Tejada, Communications Manager at EURORDIS. “We are also interested in showcasing surveys and studies (published or unpublished) that reveal health inequalities for specific rare diseases in specific countries.”
Patients, caregivers, social workers or doctors who have a story they would like to share are encouraged to contact the Rare Disease Day organisers at rarediseaseday@eurordis.org.

EURORDIS’ Council of National Alliances will meet in a week’s time to coordinate and plan the last details of this important event. If you would like to organise an awareness-raising activity you can still do so by contacting your national alliance or country organiser. To obtain their contact details go to www.rarediseaseday.org/country/finder.

If you are not a patient organisation or cannot see your country in the country page of the website, and would like to organise an activity or simply help relay the message to others, you can sign up as Friend of Rare Disease Day and add your name to a growing list of sympathisers on the website.

You can also join the more than 10,000 fans who have already signed up to Rare Disease Day Facebook, and send your video or photo (with commentary in your own language) to the RDD Flickr site and YouTube channel especially created for this day.

As you can see there are plenty of ways to get involved! Let’s combine our efforts to give hope to rare disease patients all over the world!

If you would like to find out more about this year’s campaign, go to www.rarediseaseday.org.

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**Chromosome 18 disorders: a European parent group to lean on**

**Acting at European level to gather families, networks, provide mutual support and stimulate research.**

The Chromosome 18 Registry and Research Society (Europe) is a charity that has been set up to bring together European families affected by any Chromosome 18 abnormality to share information and experiences. It held the first European-wide parent support conference on Chromosome 18 disorders in Glasgow, Scotland in July 2010.

There are five major disorders involving chromosome 18 (18q-, 18p-, ring 18, tetrasomy 18p and inversions of chromosome 18). Each of these conditions has a wide variety of characteristics and to complicate things, each of the conditions can vary in severity.
“My wife Judy and I, as grandparents of a grandchild with a Chromosome 18 q-disorder, attended the annual conference of the Chromosome 18 Registry and Research Society in 2007 held that year in Plymouth, Massachusetts. In Plymouth we met Bonnie McKerracher and her daughter Kathryn (also 18q-) who, like us, live in Scotland. Bonnie and Kathryn had attended many conferences. It struck all of us that the comfort and help given to us by the conference should be extended to Scotland, and Bonnie and I went home resolved to try and do this,” says Chris Wilkinson, Convenor of the Chromosome 18 Registry and Research Society (Europe).

Bonnie McKerracher’s daughter Kathryn was born with 18q- 30 years ago. She was not properly diagnosed until almost 7 years old. “She was born with a clubfoot and a huge umbilical hernia; she began missing milestones and was delayed in both speech and development. She was hearing impaired and had low muscle tone. More physical problems were discovered as she grew older” describes Bonnie, the society’s Secretary.

The Chromosome 18 Registry and Research Society (Europe) was set up in November 2008. “Bonnie and our son are members of Unique, the British charity for all chromosome disorders, and Unique allowed us to hold a meeting at the end of their annual conference where our constitution was formally adopted and the society set up,” remembers Chris. Twenty members were present at the first meeting and the society counts 83 members to date.

“Many parents are given a very pessimistic prognosis when their child is first diagnosed: the doctors who make the diagnosis have rarely seen other patients who are affected by the same disorder, and are unsure what outcomes they should expect. This leaves families confused and scared. Often the healthcare professionals are unaware of all the challenges, physical and developmental, that our children can face. And so parents chase around from clinic to clinic, getting eyes, ears, muscle tone, and bones checked, and those are the lucky ones whose children don’t have feeding and heart issues. Later, they may be slow to speak, and another round of specialists and therapists begins,” says Bonnie.

The Chromosome 18 Registry and Research Society (Europe) aims to make the difficult path from diagnosis to good care and living with the disease easier. The aims of the society are raising awareness amongst healthcare and education professionals about the challenges children and families face every day; and helping parents on a daily basis, making sure they are correctly supported, allowing parents to contact each other (through a 'Parents Networks' registry) and last but not least, informing parents that with accurate and consistent intervention, their child can grow up to experience a full life with all the possibilities that other children enjoy.

More details about the conference: www.chromosome18eur.org
News in Brief

EU-US bilateral workshop
A European Union – USA bilateral workshop on Rare Diseases and Orphan Products entitled “Fostering Transatlantic Cooperation on Research into Rare Diseases” was held in Reykjavik, Iceland on 27-28 October 2010. The work meeting co-organised by the European Commission and the National Institutes of Health, brought together 50 high-level scientists from academia and industry, regulatory authorities, and patient representatives, to develop recommendations for future coordinated activities between the USA and the EU. EURORDIS was invited to join in the discussions and address the issue of linking information resources for patients and families. The conference comes right after the release of an important report, commissioned by the NIH, which calls for a “coordinated national, and ideally global, effort to plan and begin systematically to implement new strategies for addressing the needs of patients with rare diseases is a timely consideration”.
Consult the Institute of Medicine report brief

Events

PatientPartner Final Workshop on Patients Partnering in Clinical Trials
7-8 December 2010
Brussels, Belgium
http://www.efgcp.be/Conference_details.asp?id=275&L1=10&L2=1&TimeRef=1

First Meeting of the European Union Committee of Experts on Rare Diseases
8-9 December 2010
Luxembourg
http://www.eucerd.eu/

4th International Workshop on AKU
10-11 January 2011
Cambridge, UK

4th International Meeting on the Congenital Disorders of Glycosylation and Related Disorders
13-14 January 2011
Leuven, Belgium
Announcements

Abstracts of posters and key presentations of ECRD 2010 in Krakow that were selected by the Programme Committee have now been published in the supplement of the Orphanet Journal of Rare Diseases. Read the selection of abstracts published on-line: www.ojrd.com

Orphan Drugs

Dated October 2010

Treatment of mantle cell lymphoma
(3S)-3-{4-[7-(aminocarbonyl)-2H-indazol-2-yl] phenyl}piperidine tosylate monohydrate salt

Treatment of hepatocellular carcinoma
(S-10-[(dimethylamino)methyl]-4-ethyl-9-hydroxy-4-O-[alpha-(2′″, 4′″, 5′″, 7′″-tetranitro-9′″-fluorenylideneaminoxy)propionyl]-1H-pyrano[3′, 4′, 6′,7′]indolizino[1,2-beta]-quinoline-3, 14-(4H), 12H)-dione, hydrochloride

Treatment of medulloblastoma
16-base single-stranded peptide nucleic acid oligonucleotide linked to a 7-amino acid peptide

Treatment of idiopathic pulmonary fibrosis
Ambrisentan
Treatment of moderate and severe closed traumatic brain injury
Ciclosporin

Treatment of small cell lung cancer
Maytansinoid-conjugated humanised monoclonal antibody against CD56

Treatment of Friedreich's ataxia
N-(6-(2-aminophenylamino)-6-oxohexyl)-4-methylbenzamide

Treatment of primary myelofibrosis
N-tert-butyl-3-[[5-methyl-2-[(4-(2-pyrrolidin-1-ylethoxy)phenyl]amino)pyrimidin-4-yl]amino] benzenesulfonamide dihydrochloride monohydrate

Treatment of Behçet's disease
Recombinant humanised anti-human interleukin-1 beta monoclonal antibody

Treatment of amyotrophic lateral sclerosis
Recombinant humanised monoclonal antibody to human Nogo-A protein of the IgG1/kappa class

Treatment of adrenocorticotropin-dependent Cushing’s syndrome
Synthetic double-stranded short interfering RNA oligonucleotide directed against proopiomelanocortin

Treatment of cowpox infection
Tecovirimat

New Marketing Authorisations

Vpriv
[velaglucerase alfa]
Gaucher disease
Shire Pharmaceuticals Ireland Ltd.
Ireland

What is Vpriv?
Vpriv is a powder that is made up into a solution for infusion (a drip into a vein). It contains the active substance velaglucerase alfa.

What is Vpriv used for?
Vpriv is used for the long-term treatment of patients with Gaucher disease. Gaucher disease is a rare inherited disorder, in which people do not have enough of an
enzyme called glucocerebrosidase, which normally breaks down a fat called glucocerebroside. Without the enzyme, glucocerebroside builds up in the body, typically in the liver, spleen and bone, which produces the symptoms of the disease: anaemia (low red blood cell counts), tiredness, easy bruising and a tendency to bleed, an enlarged spleen and liver, and bone pain and breaks.

Vpriv is used in patients who have type 1 Gaucher disease, the type that usually affects the liver, spleen and bones.

Because the number of patients with Gaucher disease is low, the disease is considered ‘rare’, and Vpriv was designated an ‘orphan medicine’ (a medicine used in rare diseases) on 9 June 2010.

The medicine can only be obtained with a prescription.