



December 2011  
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## EDITORIAL

Dear Readers,

Save the date: 23-25 May 2012! And before Christmas, don't miss early bird registrations for the European Conference on Rare Diseases and Orphan Products (ECRD 2012 Brussels) now open at [www.rare-diseases.eu](http://www.rare-diseases.eu)! Preliminary programme and all full conference details are already available.

ECRD 2012 Brussels is the unique platform across all rare diseases, all European countries, bringing together all stakeholders – academics, health care professionals, industry, investors, policy makers, healthcare managers and patients' representatives – providing the state-of-the-art of the rare disease environment and the most innovative initiatives. The forum is a mix of policy update, debates to shape future policies, discussions on actions and good practices. ECRD 2012 is conceived in synergy with national and regional conferences, enhancing efforts of all stakeholders, with a perspective on international collaboration. All presentations are prepared specifically for ECRD 2012.

The programme is organised around 7 themes covering concrete actions for national plans, guidance and practices for Centres of Expertise and strategy for their European Reference Networks. New policy and infrastructures for translational research within a more international strategy, current regulatory affairs in orphan drugs, issues of access to orphan products and innovative approaches to address them will be covered, as well as new information services and education programmes, new challenges in patient empowerment and specialised social services. A Call for Poster Abstracts has been launched and we encourage all stakeholders to submit poster abstracts that fall into any of the above themes. Full information is available by [clicking here](#).

ECRD 2012 is conceived as a European and international rare disease agora with lots of opportunities for networking, learning from each other, mixing stakeholders, dedicated poster sessions and oral presentations of posters, tutorials for industry and academia, capacity building workshops for patient advocates and academic satellite meetings. Interpretation is provided in English, French, German, Spanish, Dutch and Russian for the plenary and other selected sessions.

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ECRD 2012 Brussels is the 6th European Conference on Rare Diseases. Information on our previous conferences can be found by [clicking here](#). The scope of the conference covers both research and public health of rare diseases as well as all facets of orphan drugs. EURORDIS will co-organise ECRD 2012 Brussels for the first time with the Drug Information Association (DIA) Europe (the international non-profit professional forum organiser) and is partnering with the EU Committee of Experts on Rare Diseases (EUCERD), the European Medicines Agency (EMA and COMP in particular), ORPHANET, the European Society of Human Genetics (ESHG), industry, represented by EBE/EFPIA-EuropaBio, and with the US National Organisation for Rare Diseases (NORD).

This new format is brought to you thanks to a new partnership between EURORDIS and NORD with the DIA with the objective of synchronizing efforts on both sides of the Atlantic in order to assemble, educate, advocate and share experiences. The European Conference takes place in May, the US Conference takes place in October. Read more about the first event triggered by this partnership: the [US Conference on Rare Diseases and Orphan Products 2011 Washington](#), [highlights](#) of the event and my [blog post](#).

ECRD 2012 Brussels is made possible thanks to the financial support of the European Commission, AFM-Telethon, registration fees and exhibit tables by health companies and service providers.

Last but not least: ECRD will take place in Brussels in 2012 because this is the year in which the 3<sup>rd</sup> EU Health Programme 2014 – 2020 and 8<sup>th</sup> EU Research Framework Programme 2014-2020 will be finalised and budget decisions made, hence the presence of ECRD 2012 in Brussels is politically important as well as your presence for the plenary on 24<sup>th</sup> May.

I wish you a happy Holiday Season with your relatives and friends!

Yann Le Cam  
Chief Executive Officer, EURORDIS

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## Rare Disease Day: The countdown starts now!

Only 3 months to go till Rare Disease Day. Now is the time to get ready and start implementing your plan to raise awareness of rare diseases. Read on to find out everything you need to know about this year's campaign...

Rare Disease Day will be particularly special this year. 2012 is a leap year so the day will fall on a very "rare" day: **February 29**. On and around this date, the rare disease global community is invited to join forces to put the spotlight on rare diseases and the millions of people affected by them.

### *The theme: Solidarity!*

In its fifth year, Rare Disease Day will seek to draw attention specifically to the importance of SOLIDARITY in the field of rare diseases. Solidarity has been selected as the theme of the 2012 campaign to focus on the need for collaboration and mutual support amongst diseases, stakeholders and across countries.

The **slogan** of the 2012 campaign is **"Rare but Strong Together"**

The slogan is a reminder that acting together makes particular sense in the field of rare disease because patients are rare and expertise is scarce. Rare diseases are few and far between, but collectively they are many and people living with them all face similar challenges - so they will be stronger if they join forces.

### *The website: check it regularly!*

The [Rare Disease Day 2012 website](#) has been launched. It features a new design, in line with this year's theme and visual identity. The face of this year's campaign are three siblings from Lithuania living with a rare genetic condition known as MPS IV.

The website provides general information about the campaign and the theme, gives ideas of how to get involved, provides common tools for download and allows patients to share their story by uploading photos and videos. It will also include a calendar of national and local events that will be expanded as more countries sign up closer to the day. (To see the list of participating National Alliances [click here](#))

### *The materials: ready and easy to use!*

The logo, poster, banner and an information pack are now available in the '[Downloads](#)' section of the website. The communication materials are easily adaptable and should be disseminated as widely as possible. You can add your own logos and translate the slogan as needed.

### *Friends of Rare Disease Day: your hub to join!*

The website will display the people and organisations who have signed up as a "[Friend of Rare Disease Day](#)". This display will be updated on a daily basis in the weeks preceding the day, to show the growing list of individuals and sympathisers from industry, public authorities, research and others.

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### ***Patient stories: real life!***

The website offers the possibility to everyone to [upload](#) a video or a photo and post a short testimony in their own language. The Rare Disease Day **Photo Wall** and the collection of Rare Disease **Videos** that are shown on the website help create awareness and build the community.

### ***Social media: join the vibrant networks!***

The Rare Disease Day [Twitter](#) page, [Flickr](#) gallery, [YouTube](#) channel and [Facebook](#) group - with more than 18,000 followers to date - are all excellent channels to share information, make contacts, expand the rare disease community and create a buzz around Rare Disease Day!

We encourage you to use Rare Disease Day social media and to invite your members, friends and contacts to do the same. The more people join, the more effective we will be at spreading the messages of Rare Disease Day and raising awareness of rare diseases.

### ***Video viral campaign: it's safe to pass it on!***

EURORDIS is preparing a short promotional video for Rare Disease Day 2012. The video will give general information about rare diseases and seek to create awareness. Not intended to promote one disease or organisation in particular, but rather to promote Rare Disease Day, the date, the theme and the website. The idea is to disseminate it as widely as possible through a viral approach.

### ***Reaching out to the media: you are the voice!***

The Rare Disease Day website will make a selection of documents, videos and publications available that can be used to inform and engage the media. A changing selection of press articles will be displayed on the website's homepage to monitor media attention.

### ***Staying in touch***

Questions? Comments? Send us an email at [rarediseaseday@eurordis.org](mailto:rarediseaseday@eurordis.org)

If you want updates on these features and the general campaign, sign up to the Rare Disease Day [mailing list](#). Visit us now on [www.rarediseaseday.org](http://www.rarediseaseday.org)!

## Collecting real life experience from Centres of Expertise

### EURORDIS conducts a field study to evaluate centres of expertise and make recommendations

Centres of expertise for rare diseases are relatively new and vary in size and organisational model. EURORDIS, and its partners in the POLKA project, have come up with an original approach to evaluate them from the patients' perspective. Read on to find out more about this field study, which questioned over 200 patients and healthcare professionals from 25 centres of expertise in Denmark, England and France and issued 16 recommendations to improve the quality of their services...

"I find the current procedures complex, with long waiting times, which costs a lot and is not practical because the

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rheumatologist is not familiar with the disease, and I need to be followed by different specialists (dermatologist, psychologist etc.) I would rather go to a place where everyone comes together and consults.”

This was a quote from a rare disease patient taking part in a field study to evaluate the satisfaction of rare disease patients and their doctors relating to Centres of Expertise. The main objective of this study was to give a forum for patients to express their views and compare them with those of the healthcare professionals in order to evaluate the interaction between patients and specialised centres.

To meet this objective, EURORDIS and its partners decided to conduct a survey using a version of the technique known as the Delphi method. The process consisted of three rounds: a questionnaire, face-to-face meetings, and a validation to rate the recommendations drawn from the results of the questionnaire and the face-to-face meetings.

Overall, there was strong convergence on the ideal centre of expertise: one that provides diagnosis, treatment and care, as well as training and research. The importance of multidisciplinary care was high in the minds of participants. They also emphasised the importance of linking with outreach clinics to ensure continuity of care and minimise travel time. Help in navigating social care systems was also deemed important; as was the smooth transition of patients and patient records from paediatric to adult care.

Another key finding is that patients want to be involved in the evaluation and governance of their centres. “43% of patients did not know there was a satisfaction survey, where 82% of health care professionals knew one existed. 71% of patients and 46% of healthcare professionals did not know whether patients were represented on the governing bodies of their centres. More than three-quarters of participants said that there should be meetings between patients and professionals at the centres at least once a year!” highlights François Houyez, Health Policy Director at EURORDIS. “All this denotes lack of communication between patients and their centres and the need for this type of study to become more systematic: Centres of Expertise have responsibilities above and beyond providing high quality care and should communicate better all aspects of their activities (epidemiological research, clinical research, training etc).”

Another striking finding is that, although more than half of respondents said cross-border agreements should be established in order to allow European patients to access other European centres when needed, 60% of health professionals reported that their centre rarely referred their patients to seek expertise in other centres in Europe when necessary.

The study was carried out in Denmark, England and France, three EU countries that have long-term experience of specialised centres for rare diseases. In total 155 patients and 52 health care professionals agreed to participate. They were recruited through eight participating centres (1 in Denmark and 7 in England). The French centres of expertise were unable to participate in the face-to-face meetings. However 58 French patients completed the questionnaire.

“This pilot shows that it is possible to collect feedback of real life experience from centres of expertise to improve the services provided,” says EURORDIS Chief Executive Officer Yann Le Cam. “The same questionnaire could be used across Europe and the pilot could possibly be expanded in the future.”

The 16 recommendations that came out of this study have fed the [EUCERD Recommendations "Quality Criteria on Centres of Expertise in Europe"](#) which were adopted in October. The results will continue to be disseminated throughout the European rare disease community. They will also be integrated into [EURORDIS Guidelines of Good Practices for Relationship between Patient Groups and Centres of Expertise](#).

The Polka project was funded by the European Commission DG Sanco, Public Health Programme and also by CSL Behring, Novartis, and Sigma Tau.

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[Read the full Polka report on Centres of Expertise](#)

## RareConnect: On Line Rare Disease Communities

[www.rarediseasecommunities.org](http://www.rarediseasecommunities.org)

Isolation and a lack of access to quality information are two of the most common problems facing patients and families living with rare diseases. Thanks to the internet and more particularly Google and social networks such as Facebook patients have been able to break down those barriers but challenges still remain.

A simple Google search can deliver a bewildering amount of information that is not always of the highest quality, nor is it always relevant. Conversation and sharing of experiences may also be taking place in a language that patients may not understand. [RareConnect](#), the Rare Disease Communities project, is a patient-led social network that sets out to address some of these challenges by placing patients and patient groups at the heart of the process.

The project began when in 2010, EURORDIS and NORD formed a transatlantic, patient-driven strategic alliance for rare diseases, to bring patient advocates from Europe and the United States together to promote rare diseases as an international public health priority. The partnership's first actions have included RareConnect and encouraging cooperation amongst member organisations.

RareConnect, the Rare Disease Communities project, began with a community for a little known disease group called CAPS. The 5-language platform now covers **14 rare disease specific communities**, most recently in November 2011, Waldenstrom Macroglobulinemia, a rare form of cancer.

**RareConnect is patient-driven.** Communities are created by dedicated patient organisations, and members know that moderators represent people with real experience with their disease. They also know that the online community is sustainable; it cannot be deleted at the whim of a creator. Sensitive patient data is owned and secured by EURORDIS and NORD, while all advertising is banned. Communities are based on a [Charter for Patients' Discussion Lists](#).

**103 Patient Organisations from over 22** countries have joined the global conversation and ensured quality information is transmitted to patients through moderation of forums. The forums feature a hugely interesting feature which is an on-demand **human translation** service allowing visitors to the site to request translations across all possible combinations of the French, Spanish, Italian, English, and German languages. The translation is completed within 3-5 hours on average and automatically published in the forum whereupon an alert is sent to the visitor who requested it. "As a result of this we are finally beginning to see the language barrier break down," says Denis Costello, Web Communications Officer at EURORDIS, "now a French patient can speak to an Italian, or a German to a Spaniard in their own language".

These international patient groups not only create and maintain the community for their disease, they also nominate **volunteer moderators** like Quarda Fareh of SIMBA Behcet's Disease Italian patient group.

"I noticed that even patients from North Africa were writing and asking for information. This allowed me to reach out and highlight **trusted sources of information** to answer some questions while offering further support from the patient group," Quarda explains.

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Even when no formal patient group exists, volunteers like Nancy Morrison, an FMF patient advocate from the United States, share their research on recently updated treatment information from trusted sources and upcoming clinical trials with an international audience.

Throughout the year, EURORDIS and NORD reach out to patient organisations via meetings and workshops on social media and RareConnect. At the end of October, 23 patient group representatives from 20 rare disease organisations met in Paris for a 1-day workshop. Disease-specific communities are most often created in partnership with the NORD and EURORDIS membership organisations that are most active and expert for given disease areas.

Plans for the future include continuing to create new communities, while maintaining a commitment to offering a place where patients and families can safely exchange with others and find information.

The RareConnect philosophy can be summed up in the words of a patient who posted their story on one of the communities...

"I try to remain positive about life, and it can be hard sometimes, but finding a place like this website and reading the stories of others has given me some hope, and it really helps to know **I'm not alone with my struggles.**"

The numbers also tell the story:

- Over 7000 unique visitors per month from 108 countries on average
- 198 articles and stories added by patient groups and individual site members

## Huntington disease: In sickness and in health, two husbands become heroes

Huntington disease is a genetic disorder that affects parts of the brain. Roger Picard and Denis Ryan's lives were changed when their wives were diagnosed with this condition...

The onset of a rare disease brings huge changes in its wake, not only for the person diagnosed but also for their family. Denis Ryan and Roger Picard, two men leading different lives, share a common bond in the Huntington disease (HD) that has affected both their wives.

Denis Ryan lives in Ireland with his wife, Anne, who was diagnosed with Huntington's in 1996. Anne's younger sister had already been diagnosed and her older sister was also showing symptoms. Denis had suspected for some time that his wife had HD but knowing for sure, he explained, "is a life-changing moment. You realise that things will never be the same again."

Before HD, life was fairly normal. Anne looked after the children and Denis worked as an engineer.

"While the children were growing up life goes by in a bit of a 'blur'. When they have left home the parents often wonder 'what now'? Well HD certainly answered that question..." says Denis.

[Huntington disease](#) is a genetic disorder causing degeneration in parts of the brain, leading to physical, mental and emotional

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changes varying greatly from person to person but including anxiety, irritability, paranoia, psychosis, dementia, mobility, speaking and swallowing difficulties. Occurrence is estimated at 1/10,000-1/20,000.

Denis and Anne took advantage of the slow development of the condition to do some of the things they'd planned, like travelling.

"These things are not possible now but she still 'boasts' about seeing the Grand Canyon on our 40th wedding anniversary!" says Denis. "Although Anne is quite disadvantaged now, we try not to let it get in the way of enjoying our lives as much as possible."

The difference is that Denis has to make all the decisions alone. "While Anne became a different person, because of the effects of HD, I also had to learn to become a different person. This loss of your partner is one of the big changes."

Roger Picard's wife was diagnosed with HD in 1993 and, like Anne, she had been showing symptoms for some time before. After 20 years of family life together in France everything changed dramatically. Between 1992 and 1994 Denise made nine suicide attempts, the last involved setting fire to their home.

"The disease and the burning of the house completely changed this quiet life. I had to take care of everything. It became impossible for me to continue my work so I filed for bankruptcy."

Both men have had to learn to become carers, with all the complications of managing everyday life, organizing medical care, finding external support for their wives and for themselves. There is no uniformity of care across Europe and, as Denis explains, in Ireland alone, "Services provided seem to differ from area to area, it is not possible to get a 'check list' of all the people you need to talk with. Also, service providers differ in the amount of interest they show."

Denis turned to the [Huntington's Disease Association of Ireland \(HDAI\)](#), of which he is now chairperson. "Through the pooled resources of the members we learned how others dealt with particular problems and what services they had obtained."

Roger feels they were fortunate in that Denise was referred to the Neurology Department of Creteil hospital that, in 2004, became the Reference Centre for Huntington disease. However, problems relating to the social care/support of patients exist. "Once diagnosed, they are left to themselves with all the consequences of the disease. As a carer we are not recognised or considered. If I do not do everything myself there is nothing."

For this reason Roger set up the [Fondation Denise Picard](#), of which he is president. Its primary aim is to set up and manage facilities for neuro-degenerative diseases.

Both men stress the importance of taking care of themselves in order to take care of their wives but it isn't always easy to get the respite they need. Their biggest fear, however, is for their children, HD is an inherited condition and your chances of getting the disease if one of your parents has it are 50/50.

As Denis says, "probably the worst aspect of all is the knowledge that your offspring are fast approaching the age when symptoms are expected to appear."

"Living with someone with HD can be a living nightmare. It is not easy to see the person you knew and loved become a person that you no longer know (and may no longer know you!). It is like a long term mourning."

Nevertheless both Roger and Denis try to focus on the positive aspects of their situation, the surprising kindness of people, the new friends, their work and achievements with HD associations, and both continue to hope that a cure will be found, if not

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for their wives, then for their children.

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## EURORDIS New Position Paper expresses Patients' Priorities in Rare Disease Research

EURORDIS' position paper outlines the [needs and priorities for rare disease research](#) identified by patients for the decade ahead. The paper is complementary to another position paper on the importance of investing in rare disease research, released by EURORDIS a year ago. While the first paper seeks to answer the question: [Why research on rare diseases?](#) This latest paper presents an overall strategy based on "What" are the research priorities and "How" to achieve them. The strategic areas identified for priority funding are: registries and other infrastructures; understanding the natural history of rare diseases; translating research into therapies for patients; designing broad strategy trials and research in social sciences.

This paper is particularly timely as EURORDIS urges public decision-makers to take a stance in advancing rare disease research on the eve of the 8th EU Research Framework Programme 2014–2020 and in the midst of discussions about National Plans or Strategies on Rare Diseases. The paper was also circulated at the 3rd meeting of the International Consortium on Rare Disease Research (IRDiRC) that took place in Montreal, Canada at the beginning of October.

Read [position paper](#)

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## EVENTS

### [International Congress on Tuberous Sclerosis Complex](#)

6-9 September 2012

Naples, Italy

### [3rd TIF Pan-European Conference](#)

25-26 October 2012

Limassol, Cyprus

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## ANNOUNCEMENTS

### EURORDIS adds a 7<sup>th</sup> language

The first Russian issue of the EURORDIS newsletter appeared in November 2011. We are pleased to announce that the EURORDIS website has a new section in Russian. Click [here](#) to view

### AFM Téléthon

AFM Téléthon France takes place on 2-3 December. Check out the work the AFM Téléthon has achieved in the field of Rare Diseases in the last 25 years [here](#) (in French). You can make a donation [here](#).

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*New désignations dated October 2011***Treatment of acute myeloid leukaemia**

1-(4-{4-amino-7-[1-(2-hydroxyethyl)-1H-pyrazol-4-yl] thieno[3,2-c]pyridin-3-yl}phenyl)-3-(3-fluorophenyl) urea

**Treatment of glioma**

2-hydroxyoleic acid

**Treatment of mucopolysaccharidosis type III B (Sanfilippo B syndrome)**

Adeno-associated viral vector containing the human alpha-Nacetylglucosaminidase gene

**Treatment of hepatocellular carcinoma**

Brivanib alaninate

**Prevention of oral mucositis in head and neck cancer patients undergoing radiation therapy**

Clonidine hydrochloride

**Diagnosis of gastro-entero-pancreatic neuroendocrine tumours**

Gallium (<sup>68</sup>Ga)-pasireotide tetraxetan

**Treatment of glycogen storage disease type II (Pompe's disease)**

Glycosylation independent lysosomal targeting tagged recombinant human acid alpha glucosidase

**Treatment of glioma**

L-cysteine, L-leucyl-L-alpha-glutamyl-L-alpha-glutamyl-L-lysyl-L-lysylglycyl-L-asparaginy-L-tyrosyl-L-valyl-L-valyl-L-threonyl-L-alphaaspartyl-L-histidyl-S-[1-[(4-carboxycyclohexyl)methyl]-2,5-dioxo-3-pyrrolidinyl]-, complex with keyhole limpet haemocyanin

**Treatment of mantle cell lymphoma**

Lenalidomide

**Treatment of hypercortisolism (Cushing's syndrome) of endogenous origin**

Mifepristone

**Treatment of primary membranoproliferative glomerulonephritis**

Recombinant human minibody against complement component C5

**Treatment of cystic fibrosis**

Sinapultide, dipalmitoylphosphatidylcholine, palmitoyl-oleoyl phosphatidylglycerol, sodium salt and palmitic acid

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