Organiser: Anja Helm
Chair: Yann le Cam

22 participants from 16 National RD Alliances in 15 countries met in Paris for a 1.5 day workshop (list of participants) to discuss the following topics.

**National Plans for Rare Diseases**

Contacts: Valentina Bottarelli, Ariane Weinman

1) Overview of National Plans – where we stand (tour de table)

Each alliance (NA) shortly presented the National Plan situation in their country. There is one common problem that most NAs are facing: the lack of funding. The only country that does have a Plan and funding for it in place is France, but there is a problem with access to off label drugs which are not reimbursed anymore. The Europlan conferences have helped in countries where they took place to get different stakeholders to work together, draft strategies and in general advance towards a National Plan.

2) Guidance to organise EUROPLAN II national conferences

Presentation available on www.eurordis.org/content/national-alliances-rare-diseases

EUROPLAN II is part of a broader picture:
The EUCERD Joint Action on Rare Diseases (EJA): March 2012 – August 2015
Within this EJA, as a continuation of EUROPLAN I, EURORDIS will support and coordinate 19 National Conferences on RDs to be organised by National Alliances/ patient groups from
EU. One additional EU country is being recruited. EURORDIS will also support with its own core funding 4 additional National Conferences in Russia, Georgia, Ukraine and Serbia.

Countries organising a 1\textsuperscript{st} EUROPLAN Conference are: Belgium, Cyprus, Finland, Luxembourg, Portugal, Slovakia, Georgia, Russia, Ukraine, Serbia.

Countries organising a 2\textsuperscript{nd} EUROPLAN Conference are: Croatia, Denmark, France, Greece, Hungary, Italy, Ireland, Netherlands, Poland, Romania, Spain, Sweden, United Kingdom.

10 Advisors have been assigned the following countries:
Avril Daly Ireland
Christel Nourissier France
Dorica Dan Hungary, Romania, Slovakia
Lene Jensen Belgium, Denmark, Portugal
Lily Cannon Cyprus, Luxembourg
Maria Gardsåter Finland, Sweden
Oleg Kvlividze Georgia, Russia, Ukraine
Simona Bellagambi Greece, Italy, Spain
Stephen Nutt The Netherlands, United Kingdom
Vlasta Zmazek Croatia, Poland, Serbia

The advisors work with the EURORDIS staff to develop common format and content guidelines for all EUROPLAN conferences; Act as a liaison between NAs and EURORDIS staff; and advise one or two National Alliances in addition to their own NA in preparing the EUROPLAN National Conference.

EURORDIS is producing a set of tools to support NAs in the organisation of EUROPLAN Conferences including:
- Common Conference Format;
- Content guidelines reflecting the thematic structure of the EU main documents (Council Recommendation, EUROPLAN Recommendations and Indicators, etc.);
- Guidelines for Chairs and Rapporteurs;
- Presentations for the thematic Workshops aligned with the main European documents.

The Common Format for national conference has been presented, as well as the Content Guidelines for the 6 thematic workshops of each conference:
1. Methodology and Governance of a National Plan
2. Definition and Inventorying of RD + Information and Training on RD
3. Research on RD
4. Care - Centres of Expertise and European Reference Networks
5. Orphan Medicinal Products
6. Specialised Social Services for Rare Diseases

Based on the Common Conference Format and the Content Guidelines, each conference organiser with the support of their Advisors will establish a conference programme that will be then discussed with EURORDIS.

The complete toolkit can be found here: [http://www.eurordis.org/europlan-support](http://www.eurordis.org/europlan-support)

Europlan 2 means to continue to support the momentum across Member States for National Plans, which should be implemented by 2013.
In countries where a National Plan has already been adopted, the conferences will help assess the plan in place and to prepare for a next plan.
National initiatives to support the 116 number application for RD help lines
Presentation available on www.eurordis.org/content/national-alliances-rare-diseases

Contact: François Houÿez

EURORDIS, on behalf of the European Network of RD Help Lines, has asked the European Commission, DG Connect, to reserve a 116 number for RD help lines. If it is accepted, citizens from all Member States will have one single number to call wherever they are in Europe any time they need RD information, whether they are travelling or in their home country.

Rare Disease Help Lines are an important part of National Plans for Rare Diseases. The proposal to create a 116 number for help lines is supported by the health programme of the European Commission (cf Commission Communication & Council Recommendation)

National Alliances are invited to advocate at national level, as nowadays the European Commission requests 14 MS to support the establishment of a 116 number. So far, the following countries are supporting the 116: France, Italy, Portugal, Romania and Spain. 9 more MS are therefore needed to support the proposal.

To advocate for the 116 number, NA can make sure that the creation or development of national help lines are included in the National Plan. The national conferences can be a good moment to advocate for RD help lines and the 116 number.

Rare Disease Day 2013

Presentation available on www.eurordis.org/content/national-alliances-rare-diseases
Info Pack available on: www.eurordis.org/content/national-alliances-rare-diseases

Contact: Paloma Tejada

The theme of Rare Disease Day 2013 is solidarity. The slogan 2013 is: Rare Disorders without Borders
Of course, this slogan, which sounds great in English, can be adapted in the national language if the direct translation doesn't sound good or doesn't make sense. The message that we want to convey is: Fighting diseases which affect only a few people in each country and for which expertise is scarce and scattered requires cross-border cooperation.
The common features amongst National Alliances and at the international level will be the “Joining hands” activity and the official video that EURORDIS is currently producing with an Italian filmmaker who has made a film documentary about a rare disease (XP). It will be a mix of real filming with patients and stop-motion animation. We will have approx. 2 min and 30" version for TV use. The casting was organised thanks to Simona Bellagambi (from the Italian NA UNIAMO). The official video will be promoted via email blasts, the RDD website, our members and partners websites, RDD social media and via YouTube.
NAs are invited to organise photo shoots in their countries, ideally in front of a recognisable landmark and to upload it (before or on the day), on http://www.rarediseaseday.org/article/joining-hands-around-world. You can also send it with a comment to rarediseaseday@eurordis.org

A digital awareness campaign powered by a Facebook application will also encourage anyone across the world to share their picture joining hands. The digital campaign will feature a short video clip with real patients joining hands to encourage others to do the same and share the message via FB. The video clip is being produced free of charge by a group of students from the University of Porto in Portugal. The photos uploaded will be registered into an interactive map that will be displayed on the website. This will give a visual representation of the movement of solidarity with rare disease patients throughout the world.

EURORDIS will launch the 2013 version of http://www.rarediseaseday.org/ in November. Participants will be able to download material (Info Pack, Logo, Banner, Flyer, E-mail signature). The poster will be available beginning of December in pdf and photoshop versions for NAs to adapt as needed.

NAs are requested to fill in their Country Pages as soon as they can announce their events and to report back to EURORDIS, on the website via the country pages and also via the e-mail rarediseaseday@eurordis.org before and after the event by sending short highlights, press clippings, photos, videos and any other relevant information to show the outcomes of the campaign at local and national level.

EURORDIS has been approached by a publishing company called Media Planet, which has run rare disease supplements for NAs in the UK, Canada and the US. They, and other similar companies that ran similar operations, are interested in approaching patient organisation to supply them with content and sometimes to put them in touch with potential advertisers from the pharma and biotech industry. A list of Media Planet contacts per country was distributed at the meeting for those NAs that would like to pursue this partnership further and ran a rare disease supplement in a big newspaper in their country on Feb 28. In addition, EURORDIS contacted a paper called Metro that is distributed in most European cities free of charge at metro stations. Running a Rare Disease Day banner and/or editorial has a cost that varies depending on country and circulation. A list of prices and contacts per country was also distributed at the meeting.

EURORDIS will organise its yearly advocacy Rare Disease Day event in Brussels. In 2013, the event will be a public Hearing that will take place at the European Parliament on February 26, hosted by MEP Antoynia Parvanova, the rapporteur of the EU Directive on Transparency that will be voted the next day. This Directive means to accelerate the process to get orphan drugs on the market and to speed up decision-making on pricing & reimbursement.

A small working group of volunteers is in charge of the strategy of the Disease Day. Current members are: Mirjam Mann, ACHSE, Germany, Claudia Delgado, FEDER, Spain, Simona Bellagambi, UNIAMO, Italy, Mary Dunkle, NORD, USA, Marie Roinet-Tournay, Alliance Maladies Rares and Avril Daly, GRDO, Ireland
More volunteers are very welcome to join this working group and are invited to inform Lara Chappell by e-mail to lara.chappell@eurordis.org. Lara will be replacing Paloma during her maternity leave in 2013.

NAs have been distributed the Info Pack of RDD 2013 and can send in their comments and suggestions until Nov 7. After that it will be considered approved by the CNA and uploaded on to rarediseaseday.org

Working together within Rare Connect
Presentation available on www.eurordis.org/content/national-alliances-rare-diseases

Contact: Denis Costello, Rob Pleticha

RareConnect www.rareconnect.org responds to rare disease patients’ need for information and connection. A joint initiative of EURORDIS and NORD, the National Organization for Rare Disorders, RareConnect was introduced as a pilot in 2009. Today, 32 disease-specific communities are now active on the site. RareConnect is endorsed by more than 200 patient organisations and visited by approximately 16,000 unique visitors from 132 countries each month. At www.rareconnect.org, rare disease patients and their caregivers share their stories, discuss common issues, find information vetted by patient organisations, and post or learn about new events and articles related to their disease. The site is published in and translated by humans among five languages: English, French, German, Italian and Spanish. Each disease-specific community is a secure environment, moderated by trusted patient representatives, where patients can connect, share their experiences, and learn about their disease. Activities are organized around three sections: Understand, Meet and Learn. NAs are invited to promote this tool and to endorse it possibly with putting their logo on the website.

More information: Denis Costello, Senior Web Communications Manager and RareConnect Project Leader (EURORDIS) denis.costello@eurordis.org, Robert Pleticha, Online Communities Manager robert.pleticha@eurordis.org, or tel. 34.6.71.88.22.18.
Promoting the European Rare Disease blog
http://www.rarediseaseblogs.net/

Contact: Denis Costello

RareDiseaseBlogs.Net is a joint EURORDIS/NORD project whose aim it is to bring together key international opinion leaders in the fields of rare diseases & orphan drugs in order to promote debate and foster interaction with members of the public.

Bloggers are drawn from a broad range of stakeholders and interests the rare disease community including patient advocates, rare disease national and international patient federations, regulators, public policy decision makers, national health authorities and research.

NAs are invited to participate in the debates on the blog and to promote it on their websites.

Information on Mutual Exchange Programme “Learning from Each Other”
Guidelines, Application & report form available on www.eurordis.org/content/national-alliances-rare-diseases
Contact: Anja Helm

In September 2012 EURORDIS launched a new Exchange Program for National Alliances in the form of Short Term Fellowships to enable more direct exchange, transfer of knowledge and collaboration between one National Alliance with another and to offer means of mutual support and capacity building. The programme is designed to provide short term, quick and flexible financial support to enhance the current efforts of the National Alliance, it shouldn't be considered as a source of significant or recurrent financial support.

Financial support is limited to a total of 600 € per Fellowship and a maximum of 2 fellowship per year and per National Alliance.

This program will be continued in 2013, the relevant documents will be sent out to all NAs at the beginning of 2013.

Information on EPIRARE Survey
Presentation available on www.eurordis.org/content/national-alliances-rare-diseases

Link to survey in English: https://www.surveymonkey.com/s/3KBY96V
For other languages go to: http://www.eurordis.org/news/eurordis-conducts-survey-rare-disease-registries

Contact: Monica Ensini

There is robust evidence that when a registry exists for a certain rare disease:
•quality of care and life expectancy for patients improves dramatically
• higher level of networking between medical experts
• more research activities on-going
• more therapies under development

The EPIRARE project objectives are:

• To elaborate propositions for EU policies on RD patient registries
• To propose the framework of a future EU registry platform/hub for RD (legal basis, governance framework, option for sustainability, etc…).
• To explore the definition of a minimum data set common for all RD

The EPIRARE survey conducted by EURORDIS is specifically targeted to patients in order to gather their perspectives and expectations on RD patient registries. The survey is available in the following languages:
English, French, German, Italian, Portuguese, Spanish, Greek (GR), Czech (CZ) and Romanian (RO), and will soon be available in Danish (DK) and Hungarian (HU).

As of today: more than 3000 patients have accessed the survey.
The preliminary data show that:
• Patients expect registries to also fulfil health/social services planning for RD
• There seems to be quite a dominant diffidence towards industry
• Patients express interest and engagement with the core registry governance issues
• Patients mainly expect the EC to provide long term funding for RD registries
• Patients wish for a uniform EU legislation and for a EU reference platform/hub for RD registries

EURODIS, NORD and CORD, have recently agreed of 10 Key Principles for Rare Disease Patient Registries that can be found http://www.eurordis.org/en/content/eurordis-nord-cord-release-joint-declaration-10-key-principles-rare-disease-patient-registri

**Creation of an Interest Group for translational research**

*Translational research is scientific research that facilitates the translation of findings from basic science to practical applications that enhance human health and well-being. (From bench to bedside)*

Contact: John Dart

John Dart, COO Debra International and EURORDIS Deputy General Secretary proposed the creation of an Interest Group for translational research. Many of the European Federations and National Alliances already have experience in research projects and are reaching the point of translation of potential new therapies from the laboratory to the clinic; it would be very interesting to pool that knowledge across the groups and diseases to discuss ways of approaching and promoting Rare Disease research, particularly around preparation for clinical trials, intellectual property issues, strategic planning and attracting and working with external partners.

EURODIS will create a mailing list for this group that the representatives of Federations and Alliances can sign up to and use to mutualise and spread knowledge.

A specific e-mail will be sent shortly to all groups.
**Specialised social services**
Presentation available on www.eurordis.org/content/national-alliances-rare-diseases

**Contact: Raquel Castro**

Across EU there is a common need to integrate Rare Diseases into social policies and to guarantee PLWRD access adequate Specialised Social Services. The EUCERD joint Action (EJA; 2012-2015) includes a Work Package on Specialised Social Services addressing the 3 following main subjects:

1) Mapping of services  
2) Training of staff and volunteers  
3) Integration of RD into Social Policies

Specialised Social Services for RD include:
- Respite Care Services  
- Therapeutic Recreation Programmes  
- Adapted Housing and other habilitation services  
- Resource Centres for Rare Diseases

A literature review paper has been developed to facilitate advocacy actions for the promotion of Specialised Social Services for PLWRD, to be made available in December 2012. Currently, services are being mapped in different countries and within the different categories, in order to prepare the European map of Specialised Social Services for PLWRD. The EURORDIS website section on Specialised Social Services – including the map of services in Europe, facts/guidelines and testimonies – is to be made available in December 2013 with on-going update.

Two country visits have been performed in 2012: Frambu Resource Centre, Norway and Agrenska Respite Care Service, Sweden. Data collection of these visits will be disseminated in 2013 in the format of “case study”.

A workshop on “Guiding Principles for Specialised Social Services” will take place on the 6-7th December, in Romania, involving EUCERD representatives, Specialised Social Services, Europlan Advisors, some academia and other relevant authorities. The outcome of the workshop discussion shall be disseminated in 2013 in the form of report.
Access to treatments in the context of the crisis

Contact: Yann le Cam
More information: EUCERD Recommendation for a CAVOMP information flow

Equitable and timely access to market-authorised orphan medicinal products is an issue for thousands of rare disease patients and their families across the European Union, as reported by many patients and patient groups as well as robustly shown in the EURORDIS Surveys on Patients Access to Orphan Drugs. Large disparities in access exist between and even within the European Member State countries.

Increasing cooperation between EU-level authorities and the Member States (MS) has been identified as a means of improving access to Orphan Medicinal Products (OMP) and reducing inequities. EURORDIS has promoted with several Position Papers the concept of a mechanism to improve and evaluate the Clinical Added Value of Orphan Drugs (CAVOD) together with our EURORDIS Round Table of Companies and the industry platform of EuropaBio-EBE-EFPIA Task Force. The auditing firm Ernst & Young was mandated by the European Commission to identify and assess the possible options for the creation of a mechanism for the exchange of knowledge between the MS and European authorities on the scientific assessment of the relative effectiveness of orphan medicines and issued a report in December 2011. Consequently, the European Commission asked the European Union Committee of Experts of Rare Diseases (EUCERD) to make recommendations in the area of facilitating the exchange of scientific information on orphan medicinal products that would support the clinical assessment processes undertaken by the MS. This “EUCERD Recommendation on the Clinical Added Value of Orphan Medicinal Products Information Flow (acronym: CAVOMP) to the European Union and Member States” has been adopted by the EUCERD in June 2012 by consensus of all its members (not unanimity because Austria abstained). We are now working on its implementation from 2013. But it is of major importance than the key measures adopted for Member States be introduced in the current MSs policies or upcoming national plans.

Also, EURORDIS represent its members in the EU Corporate Social Responsibility Forum piloted by EC DG Enterprise for the Working Group “Mechanism of Coordinated Access to
Orphan Medicinal Products” (acronym: MOCA). After two years of work, the final report will be released in December 2012. The main measures, of great importance and usefulness for NAs, are: first, the adoption of a Common European Transparent Value Framework for exchange of information between Member States on the value of an orphan drug to have national “well informed” decisions based this European exchanged; second, pilots from 2013 of negotiation of prices of orphan drugs coordinated at European level based on this Value and on Volume (the targeted population of patients to be treated) as well as on commitment for post-marketing studies on long term safety, efficacy and effectiveness; third, joint procurement contract through which several Member States could coordinate their purchase of orphan drugs, particularly relevant for ultra-orphan diseases and for small or medium size countries.

Besides, EURORDIS is active on supporting its members when issues are arising in their country such as national measures to reduce prices or access due to the crisis (ex: Germany or Greece or Spain or Poland or France), or HTA review of value of orphan drugs (ex: Netherlands CVZ or Sweden TLV), or on crisis situation (ex: access to Enzyme Replacement Therapy in Lithuania). We will continue to do so increasingly; François Houyez new position on “Information & Access to treatments” has been created to address this growing issues as well as to expand our work on compassionate use, off-label use of medicines, patients reported outcomes etc and making the link with our webservices and helplines.

EURORDIS will continue to inform patient representatives of the developments and will issue a EURORDIS Policy Fact Sheet on Improving Access to Orphan Medicinal Products. The issue will as well be discussed at the EURORDIS Membership Meeting (EMM) 2012 Dubrovnik.

Yann invites all participants to look at the relevant documents they have previously received and to send their information on access to medication problems in their country to the CNA/CEF.

The EURORDIS blog can as well be used to exchange views or alert on specific problems.

**Results of the off label use survey**

Presentation available on [www.eurordis.org/content/national-alliances-rare-diseases](http://www.eurordis.org/content/national-alliances-rare-diseases)

**Contacts: François Houÿez, Rob Camp**

Off label use of medicines is very frequent amongst patients with rare diseases. Orphan medicines with a specific indication with a rare condition are in fact the exception. The objective of the Off-label use (OL) study launched by EURORDIS was to:

- Document patients’ experience with OL use
- Learn about the information patients receive
- Create database of off-label uses in rare diseases
- Explore how patients handle Adverse Drug Reactions (ADRs) ADRs with OL use
- Define future actions on OL use in rare diseases

The Off label use survey, consisting in a questionnaire available on the internet in 5 languages during summer 2012, has been developed by the DITA Task Force. So far, 255 responses have been analysed, the process is ongoing.
Cross Border Health Care and European Reference Networks
Contact: Yann le Cam, Flaminia Macchia

The national transposition of the CBHC Directive is now in the hands of the national Ministries. The directive, in article 13, specifically mentions Rare Diseases and the specific issues in Health Care at EU level.

Patient organisations can impact the process, via the EUCERD and National Alliances, notably on the aspects of mobility and patient's rights. The EUCERD will advise MS to involve patient organisations in the transposition process. Currently, the working groups responsible for the national transposition do not have to involve patient organisations. It is important that patient representatives get involved at the national level. A workshop at the EMM 2013 Dubrovnik will be dedicated to this subject. Hopefully Ms Nathalie Chaze of the EC will participate in this workshop. She is currently going to each MS to meet the people in charge of the national transposition.

Most CNA participants do not know who sits in their national working group; It is important that they identify these people and try to get in contact with them to make their voice heard. The list of national representatives in the EUCERD can be found on: http://ec.europa.eu/health/rare_diseases/docs/list_members_eucerd_en.pdf

M. Margetidis of the EC underlines that each MS will have to put in place a contact point that will give answers to all questions concerning prior authorisation, reimbursement etc. The question of whether patients should travel to experts or experts to patients is discussed. The Directive says clearly that expertise should travel first, but there can of course be cases where patients should travel to an expert centre if needed, in an organised way and not on an ad hoc basis as before the directive. It is important that bio samples travel to the right laboratory for diagnosis, biobanking etc.

European Reference Networks (ERN) will gather the knowledge and expertise. The development of centres of expertise and European Reference Networks in the field of rare diseases is encouraged in the Council Recommendation on an Action in the Field of Rare Diseases and CBHC Directive as a means of organising care for the thousands of rare conditions affecting patients across Europe. In order to share knowledge and expertise more efficiently, the EUCERD recommendations seek to introduce harmonious standards of quality practices by elaborating criteria for the Member States to incorporate into their process to designate centres of expertise, especially in the context of national plans/strategies for rare diseases which the Council has urged all Member States to elaborate by 2013.

Next CNA Meeting
The 15th workshop of the Council of National Alliances will take place May 30 in Dubrovnik, Croatia, as a satellite meeting of the EURORDIS Membership Meeting EMM 2013 Dubrovnik (30 May-1st June)