

# *Report*

## *5th Workshop of the Council of European Federations*

### *Paris, 30 & 31 October 2012*

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Representatives of 19 European Rare Disease Federations (participants list attached) met in Paris on October 30 and 31. For the first time, part of the CEF meeting was organised jointly with the Council of National Alliances, to discuss topics of common interest and learn from each other. October 31<sup>st</sup> a training on the Promotion of Research took place.

This workshop was organised by Anja Helm and chaired by Yann le Cam, EURORDIS.



CEF meeting Paris 2012

## 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 EURORIDS 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.

EURORDIS 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: <http://www.eurordis.org/content/european-disease-specific-federations>

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 Centers 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 ongoing 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: <http://www.eurordis.org/content/european-disease-specific-federations>

*Contacts: François Houyez, 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](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 too 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.*

### Support to European RD Federations 2013

Terms of References 2012 available on <http://www.eurordis.org/content/european-disease-specific-federations>

Contact: Anja Helm

In September 2012 EURORDIS launched a new Exchange Program for European Federation in the form of Short Term Fellowships to enable more direct exchange, transfer of knowledge and collaboration between one Federation with another and to offer means of mutual support and capacity building:

*European Federations' Exchange Program- Learning from Each Other*

All European Federations member of EURORDIS can apply for this support. The program covers the travel expenses (main travel and hotel) of one representative of a European Federation to visit another Federation for a short to collaborate on a specific pre-defined topic.

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

This program will be continued in 2013.

The other support program, entitled "Support to European Federations" will be continued as well.

Once the EURORDIS Board of Directors has approved the budget 2013, all member federations will receive the relevant documents and application form.

Whereas the *Support to EF* aims at helping federations to organize meetings ( board meetings, conference, meetings with researchers, network meetings...) the *Learning from each other* program means to enhance

the exchange between individual federations, who can benefit greatly from each other's experience in order to pool and mutualize knowledge and best practices.

## **RareConnect for European RD Federations**

Presentation available on <http://www.eurordis.org/content/european-disease-specific-federations>

Contacts: Denis Costello, Rob Pleticha, Marta Campadal

RareConnect [www.rareconnect.org](http://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](http://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*.

Rare Connect communities are not only a place for patients and caregivers to exchange, but can be as well a shared project that acts as a catalyst for starting and maintaining a vibrant European federation. Some European federations have already successfully launched their community, such as: EDRIC/Dysnet, HSP Europe, Waldenstrom Europe, AKU and others.

One of the questions that comes back often is the added value of Rare connect versus Facebook:

- Human translation
- Moderator support and training
- Ongoing technical improvements
- Non profit model
- Allows for disease conversations and Facebook conversations to be separate
- 3 full time Community Managers
- Your conversations and stories are there to stay

According to representatives who already have an active community, there is huge added value as well compared to national websites, RareConnect acts like an extension of the national website, the translations allowing for discussions across Europe or the world.

## **October 31st: CEF training: "Research coordination & promotion"**

Most European Federations have been or are actively involved in research activities, but only a few (Debra International, SIOPE, CF Europe) have or are participating in EU funded projects, be it DG SANCO or DG Research. All federations are very interested in projects funded by the EC but writing proposals is difficult. A few have tried and failed.

Angelika Klucken of Hoffnungsbaum, Germany and Kay Parkinson, Alstrom UK, are participating in EU funded projects, with very different experiences.

## TIRCON - Collaborative Research for a very rare disease as a global approach under FP7 Experiences of the German NBIA patient organization "Hoffnungsbaum e.V." as TIRCON Partner,

Angelika Klucken

Presentation available on <http://www.eurordis.org/content/european-disease-specific-federations>

Hoffnungsbaum is the German NBIA organisation, which provides support to families, information and networking between patients, clinicians and researchers, notably via the bi-annual family conferences. Hoffnungsbaum supports NBIA research financially and by national and international networking.

TIRCON (Treat Iron Related Childhood Onset Neurodegeneration) is a research consortium comprising 13 partners from 8 countries, funded by the EU under FP7. 2 NBIA patient organisations are partners in TIRCON: Hoffnungsbaum e.V. (Germany) and the NBIA Disorders Association (USA). TIRCON's concept is to bring together the existing outstanding but scattered expertise in NBIA research and care through Europe and internationally. The project aims to set up a structured network to improve diagnosis and treatment of NBIA. The scientific workpackages include a patient-registry, biobank, biomarker-studies, a clinical trial and further preclinical studies. The other TIRCON- workpackages are ethics, management and dissemination.

Hoffnungsbaum helped, to bring researchers from Europe and the USA together, which was the start of the TIRCON project, born from a spirit of cooperation.

In TIRCON, both patient organisations are mainly in charge of the Work Package "Dissemination" with Hoffnungsbaum e.V. as WP-leader. Hoffnungsbaum is member of the Scientific Steering Committee.

TIRCON Dissemination tasks (not only of the patient groups but of all TIRCON-partners) include:

- ✓ Networking and communication plan to raise knowledge and awareness of PKAN and other NBIA
- dissemination strategy
- set-up of TIRCON website [www.tircon.eu](http://www.tircon.eu) with patient information in several languages,
- development of target-group specific information packages
- networking with key stakeholders in health care systems and Rare Diseases policy
- the use of the advocacies' usual media like websites, newsletters, leaflets
- development of new services like a healthcare notebook for patient families to track medical history
- ✓ "doctor's telegram" as online newsletter and patient-friendly care-guidelines for PKAN, scientifically elaborated by TIRCON-partner OHSU
- ✓ NBIA Network in Europe and USA to harmonize care standards
- development of a clinical NBIA network, currently the TIRCON clinical centers
- development of an international network of NBIA patient advocacies, the NBIA ALLIANCE
- ✓ Dissemination of research results
- Publications (scientific and non-scientific journals), Website
- International scientific conferences (e.g. neurology, neuropediatrics, human genetics )
- ✓ Teaching PKAN/NBIA
- Specific educational training of the early-career scientists and clinicians
- Creation of an international research prize for early-career scientists

Hoffnungsbaum and NBIA Disorders Association USA are as well partner in 3 other Work Packages: Clinical Trial, patient registry and Ethics.

The experience as partner in an EU project has been positive, the integration of patient organisations as partners a success because they can contribute specific competences like networking abilities, expertise with PR and act as a bridge between scientists and patients. It is important that the patients' perspective is brought into science.

To make the partnership work, patient organisations need to stick to the rules of the Consortium Agreement and respect Responsibility Confidentiality, Reliability



John Dart, Kay Parkinson

### The EURO-WABB Project (DG Health), an instrument to increase knowledge, improve the lives of affected people through better management, and to develop clinical research

Kay Parkinson, Alström Europe & Partner EU WABB

Presentation available on <http://www.eurordis.org/content/european-disease-specific-federations>

Alström Syndrome UK was created by Kay Parkinson in 1998 with three key aims:

- to help people with Alström Syndrome to provide support for them, their carers and the professionals who are working with them
- to raise awareness amongst both the public and medical professions of Alström Syndrome
- to raise funds to promote research into Alström Syndrome

One of the key achievements of Alström UK is the development of patient led NHS funded multi disciplinary clinic for Alström Syndrome.

EURO-WABB is an EU Rare Diseases Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes. The EURO-WABB Project is a collaboration of doctors, scientists and patient support groups from all over Europe. It is supported by the EU Directorate General for Health and Consumers (DG-SANCO) via its Executive Agency for Health and Consumers. The overall aim is for this register to be a key instrument to increase knowledge on these rare diseases, improve the lives of affected people through better management, and to develop clinical research.

Alström Syndrome UK was invited to join the project, on EURORDIS recommendation, to lead the work package Dissemination. There was no patient input when the project was drafted.

The Work Package Dissemination includes:

- Identifying and engaging with stakeholders
- Awareness Raising
- Associate Partner & Member of the Project Management
- Committee contributing to the delivery of Euro-WABB
- Leading analysis of the learning and information needs of patients and families
- Contributing to the development of guidelines

The Plans for continuation are:

- EU health providers: network of experts in WABB diseases
- Industry: new indications for existing drugs
- Disease specific charities



Future developments:

- Alström Europe and later possibly European Wolfram and BBS Federations

There are many challenges for EURO WABB as the conditions are all extremely rare; and there are no patient organisations for these diseases across Europe, apart from the newly formed Alström Europe. Recruitment, therefore of patients to participate in the registries is proving difficult for some of the conditions in the group.

Medical consultants with databases of these diseases are often reluctant to share their information, having collected the information over many years; some degree of “ownership” is seen and quite a few are reluctant to join the EURO WABB project. As the conditions are already rare this means that for a condition like Alström Syndrome for example, there are currently three separate databases in place.

Patient groups are usually more likely to share data as patient benefit is paramount for them- unfortunately two of the conditions involved in EURO WABB do not have patient groups ready to form European Associations.

It is difficult when one country which has evolved specialist care for very rare conditions tries to expand this knowledge EU wide before consulting all patient groups involved. There is no doubt patients EU wide could and should benefit from the advances in one country, however, reaching these patients before the patient group networks are in place via medical professionals is proving to be a barrier because of the reasons stated above.

EURORDIS advice from the start for EU projects which include very rare conditions could lead to better patient outcomes.

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### Wrap up:

It is important that patient organisations be included from the start in research projects, whether DG Research or DG Health funded. It's equally important that the patient organisations are included as equal partners and with an adequate budget. The upcoming EU Framework Programme for Research and Innovation is called Horizon 2020. It will represent the financial instrument implementing the Innovation Union, a Europe 2020 flagship initiative aimed at securing Europe's global competitiveness. Running from 2014 to 2020 with an €80 billion budget, the EU's new programme for research and innovation is part of the drive to create new growth and jobs in Europe.

Patient organisations can be involved in other Work Packages than Dissemination.

EURORDIS for example only gets involved in projects where participation in the Steering Committee is assured. The patient organisations need to make sure that the project they are invited to participate in fits into their strategy, that the partners involved are the right ones and that the PO can actually add real value to the project.

It is therefore very important that the PO sees the full outline of the project.

EURORDIS will organise capacity building workshops for patient organisations to increase their knowledge of EU funded research projects. The CEF meeting in November 2013 could be an occasion to have this workshop. Patient advocates need to get involved in clinical work and to increase their knowledge by participating in, for example, the EURORDIS Summer school and/or specific webinars that are organised on the topic.

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