

EUROPEAN  
ORGANISATION  
FOR RARE DISEASES

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# ACTIVITY



EURORDIS  
Rare Diseases Europe

# REPORT

2014

&  
WORKPLAN 2015

“ The Voice of Rare Disease Patients in Europe ”



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# FOREWORD

## BY THE PRESIDENT AND THE CHIEF EXECUTIVE OFFICER

**I** EURORDIS has reached **678 members in 63 countries** (26 EU countries)\*, including 36 national alliances and 46 European federations. In 2014 alone, **50 new members** joined EURORDIS. A membership re-assessment process to examine whether members still fulfil our strict criteria is also now in place. These progressions further enhance EURORDIS' mission to build a strong pan-European community of patient organisations and the 30 million people living with a rare disease in Europe, in order to be their voice at the European level and to fight against the impact of rare diseases on their lives.



Yann Le Cam / Chief Executive Officer

The **European Network of National Alliances** was further strengthened with new national rare disease alliances created in Latvia and Ukraine. Over the course of 2014, 20 national alliances signed the **Common Goals & Mutual Commitments between EURORDIS & National Alliances in Europe**, an initiative that aims to promote strategic synergies between national alliances and EURORDIS, as much as between national alliances themselves. This initiative further structures and sustains the Rare

Diseases Europe movement and enhances the collaboration and capacities of patient advocacy groups for the achievement of common results.

The **EURORDIS website**, with over 350 000 visits per year, was further developed in 2014. The website includes a dynamic homepage driven by the eNews, an increased visibility to our members' events, a more prominent presence of social media and rare disease video content through EURORDIS TV. New website sections included the Compassionate Use page and a Volunteer section, while other sections such as Rare Disease Policy, What we do and Photo Contest were redesigned and updated. Further sections will be launched in 2015.

**Rare Disease Day** continues to grow internationally with **84 countries participating in 2014**. The media outreach was strong, with hundreds of news articles collected and over 1000 events organised all over the world. A **new Rare Disease Day website** was launched in 2014 to improve user friendliness and accessibility and a **mobile site** for smartphone users was developed for launch in 2015. Rare Disease Day has now truly become an international event and an essential force to raise public awareness and promote national rare disease policies.

The EURORDIS online patients' community network **RareConnect** experienced further growth in 2014. By the end of 2014, 70 disease-specific online communities had been successfully created from international partnerships with **560 different patient organisations and the engagement of 246 volunteer moderators**. This helped to build cross-border disease knowledge and reach isolated people living with a rare disease. **EURORDIS is planning to rebrand the RareConnect platform in 2015** with a new logo, visual identity and new features such as discussion groups. Further partnerships between RareConnect and national alliances will be explored.

In 2010, EURORDIS made a long-term commitment to the development of **national rare disease plans/strategies throughout Europe and beyond** and this remained one of our main and foremost key advocacy priorities for 2014. EURORDIS and the national alliances continued to organise **national conferences in EU Member States and across Europe** in order to **promote good national measures and dialogue between all relevant stakeholders**. **26 national rare disease conferences** were organised by the end of 2014.

As these plans develop, **advocacy activities need to become more technical/ focused and should increasingly be based on an exchange of best measures across Europe**. EURORDIS' active participation in the **European Commission Expert Group on Rare Diseases** (through its **EURORDIS Policy Action Group** composed of volunteers and staff) is essential to reach that goal. The key priorities of the Commission Expert Group in 2015 are: European Reference Networks, cross-border healthcare, registries, access to orphan drugs, implementation of rare disease coding in healthcare systems, social policy/ services and national plan indicators.

EURORDIS has contributed substantially to positioning the voice of patients at the heart of the development of **European Reference Networks (ERNs)**. Throughout 2014, EURORDIS continued paving the way for the next steps in ERN implementation to take place in 2015. Support of the implementation of the EU policy on European Reference Networks, the optimal uptake of this policy for rare diseases, the orientation of ERNs toward patients' health outcomes, and the preparedness of our members and stakeholders to be involved in ERN development are all other current top priorities. Finally, it is also priority to support to the development of an **EU platform on rare disease registries** and participate in new **long-term infrastructure projects**.

In 2014, EURORDIS increased activities in the field of **rare cancers**, with the formation of a dedicated Policy Action Group on Rare Cancers and the nomination of 2 patient representatives to the newly formed Commission Expert Group on Rare Cancers. Through this group, we are analysing the commonalities between all rare diseases, with a particular focus on rare cancers, and advocating for synergy in policies.

The **European Conference for Rare Diseases & Orphan Products (ECRD) 2014 Berlin** was a milestone for EURORDIS. ECRD 2014 was also part of a broader strategy to push toward concrete implementation of a new EU rare disease policy framework, both at European and national levels. A multi-stakeholder event, it attracted 768 attendees (patient representatives, academics, healthcare professionals, industry, payers, regulators, and policymakers) from 43 countries. In 2015, we are already preparing the ECRD 2016 Edinburgh to be held on 26–28 May 2016, which will focus on the theme of 'Game Changers in Rare Diseases'.

Lack of equitable access to orphan medicines across Europe remains a high concern aggravated by the impact of the financial and economic crisis. In 2014, EURORDIS sustained its advocacy work in favour of European-level initiatives aimed at improving access for patients, including: the medicines Adaptive Pathways project, the Patient Progressive Access, the Mechanism of Coordinated Access to Orphan Medicinal Products (MOCA), parallel scientific advice by EMA and HTA agencies and future common HTA assessment reports within the SEED Project. **Earlier, wider, equitable, sustainable access to medicines continues to be a key theme for 2015.** EURORDIS will continue to promote European collaboration between Member states on pricing that is based on value, volume and post-marketing authorisation data generation, also exploring managed entry agreement and equitable pricing mechanisms for improving access. The EURORDIS **Access Campaign** was launched in 2014 to assess difficulties that patients currently experience in accessing medicines and medical care. The campaign will be sustained throughout 2015 and its results will be analysed and disseminated for the purpose of promoting targeted solutions.

In 2014 **EURORDIS continued its active support of the participation of patient representatives in the committees and working parties of the European Medicines Agency** (COMP, PDCO, CAT, PCWP, SAWP, CHMP), culminating in 166 days of meetings and 456 scientific dossiers examined for scientific advice/protocol assistance, paediatric studies and overall clinical development up to marketing authorisation. 2014 also brought the first participation of patients in a **Committee for Medicinal Products for Human Use (CHMP)** oral explanation to discuss the marketing authorisation of a new rare disease treatments. This was the result of years of EURORDIS advocacy actions that called for the involvement of patients in the benefit/risk evaluation of medicines.

In 2014, EURORDIS continue to grow its activities in **health technology assessment (HTA)**. EURORDIS is one of four patient representative organisations at the EUnetHTA Stakeholders Forum and is contributing to a vision for the future of HTA in EU. EURORDIS is promoting the involvement of patients in HTA procedures and is coordinating the identification and involvement of patients in the SEED project.

Rare diseases are an international public health challenge and international policy convergence and collaboration can enhance European activities in research, health care organisation, information and therapy development. In 2014, EURORDIS amplified its work towards launching **Rare Diseases International (RDI), a global network of rare disease patient organisations**, by finalising the Joint Declaration: Rare Diseases as an International Public Health Priority and by holding a first RDI meeting in Shenzhen, China that was attended by key international patient organisations. RDI will be officially launched in Madrid in May 2015.

EURORDIS was able to grow in 2014 in spite of a challenging economic environment thanks to actions to diversify its public funding and new initiatives in fundraising. We believe **our organic growth reflects the positive resonance our actions have within the rare disease community.** However, funding of our core activities remains a critical issue when it comes to sustaining our development. **EURORDIS will continue to diversify and consolidate its resources in 2015.**

In 2014, EURORDIS was proud to rely on **320 volunteers** including **72 volunteer patient advocates, 2 office support volunteers and 246 volunteer RareConnect moderators.** The EURORDIS volunteers have unique insight into the complexity of different rare diseases across Europe and reinforce EURORDIS as a grassroots movement.

**Our community is governed by common values and modes of action.** We believe in solidarity and equity, social justice and fairness. We work through patients' empowerment and capacity building of patient advocates, exchange and mutual support, gathering of experience, networking and partnership with all stakeholders. We are daring and innovative. We proactively collect facts and data to support the promotion of patient-centred policy. We trust that facts and words shape policy and actions resulting in better outcomes. We understand what patients need and firmly believe that a patient-driven agenda has the capacity to act as a catalyst for positive change.



Terkel Andersen / President

\*Correct at time of printing

# EURORDIS IN BRIEF

EURORDIS, is a non-governmental patient-driven alliance of patient organisations and individuals active in the field of rare diseases, dedicated to improving the quality of life of all people living with rare diseases in Europe.

EURORDIS was founded in 1997 by four patient groups from different therapeutic fields: the Association Française contre les Myopathies (AFM), Vaincre la Mucoviscidose, Ligue nationale contre le Cancer (LNCC), and AIDES Fédération.

Today it is supported by its members and by the Association Française contre les Myopathies, AFM – Téléthon, the European Commission, corporate foundations and the health industry.

EURORDIS is the voice of 30 million people affected by rare diseases throughout Europe.

## KEY FIGURES 2014



## EURORDIS' MISSION

To build a strong pan-European community of patient organisations and people living with a rare disease, to be their voice at the European level, and – directly or indirectly – to fight against the impact of rare diseases on their lives.





# STRATEGIC APPROACH 2010–2015

The EURORDIS Strategy 2010–2015 was adopted at the 2009 Annual General Assembly in Athens. The Strategy was reviewed end of 2012 and revisions were adopted by the EURORDIS Board of Directors in March 2013.

1

**EURORDIS in 2015 has consolidated its position as the organisation of reference for rare diseases in Europe and is recognised as an actor in worldwide processes having impacts on patients living with rare diseases in Europe**

- Rare Diseases as a Public Health priority in Europe (8th EU Framework Programme for Research & Technology, 3rd EU Public Health Programme, other policy legislations and programmes)
- Being the voice of all rare diseases, genetic or not, including rare cancers, and very rare diseases, open to Europe at large (48 countries)
- Raising rare disease awareness amongst general public (in particular the International Rare Disease Day and European Year of Rare Diseases campaign)
- Rare diseases become an international movement and gain visibility and influence in international instances (Council of Europe, WHO & UN)
- Production, sharing and accessibility of patient-generated knowledge

2

**EURORDIS in 2015 is facilitating the effective implementation of European regulations and strategies at national levels in more policy areas for the benefit of patients and families**

- A Public Health priority in European countries (Member States and beyond)
- National Plans in each Member State with patient-centered approaches incl. Centres of Expertise, research, medicines, registries, information, social services. Monitor their implementation including indicators.
- Development / Consolidation of European Networks integrative of European and National levels:
  - European Reference Networks of Centres of Expertise
  - European Research Networks and European Research Infrastructure for rare diseases
- European Network of Information Help Lines
- Adjust actions on the basis of feedback from PO members on the effective implementation of rare disease regulations and policies (evaluation process) and remaining unmet needs (research budget, Centres of Expertise, Best Clinical Practice on Diagnostics and Care, Quality of Life)
- A special focus on research. The role of EURORDIS shall aim at:
  - A higher public awareness in support of rare disease research
  - A more favourable research policy framework for rare diseases

- A development of EURORDIS' interactions with the research community and learned societies
- A promotion of the development of European Research Networks and European Research Infrastructure for rare diseases
- A promotion of the participation of patients in research and therapeutic developments – which enhances capacity building of patient representatives
- A promotion of the participation of patient representatives in ethical committees in clinical research and human genetics
- A special focus on information and quality of life. The role of EURORDIS shall aim at being a direct operator in the following fields:
  - Rare disease-specific help lines in national languages linked in a European Network
  - Patient-based knowledge, generated and shared in care, cope and quality of life in a holistic approach
  - Educational information on the management of specific symptoms which are common to across different rare diseases (ex. hyperactivity, sleep disorders, etc.)
- Educational information on managing the impact of rare diseases on family life (effect on parents, siblings, integration at school, empowerment of young adults, aging etc.)
- A special focus on information and quality of life. The role of EURORDIS shall aim at enhancing and catalysing actions in the following fields by the means of partnerships:
  - Production and availability of educational material and courses for health care professionals, social workers, etc
  - Production and availability of validated and updated Information on Respite Care Services (RCS), Therapeutic Recreation Programmes (TRP), Resource Centres (RC) and other rehabilitation services such as Adapted Housing (AH).
  - Promotion of research on quality of life in EU framework programmes in research and public health



### **EURORDIS in 2015 has developed enriched and more supportive capacity building relationships with its members and empowerment of volunteers**

- Maintain a high level of legitimacy and credibility by maintaining a high level of consent amongst EURORDIS' members
- An integrative (in main areas of activities) and supportive volunteer programme well recognised inside and outside
- Intensify capacity-building and networking with and between the National Rare Diseases Alliances and European Rare Diseases Federations for improved efficacy and decentralization
- Member patient organisations as key relays to their families and patients to generate and access relevant customised information
- Capacity building networking, sharing experience and common tools, giving an easy access to good practices to empower patient advocates



### **EURORDIS in 2015 is more sustainable in terms of human, financial and organisational resources and governance**

- Values and governance: Well established shared Values and governance processes
- Leadership sustainability of the Board
- Decreased and better-balanced workload inside the staff, more high level volunteers, efficient balance of workload between staff / volunteers
- Web communications central in strategy / organisation / work process
- Financial sustainability: Attract more public funding, diversify sources of revenues (Corporate other than pharmaceutical or medical device companies, Foundations), generate own unrestricted resources (Gala dinner, donations). Consolidate administrative process and budget control.
- Integrative IT infrastructure, database and tools



# COMMUNITY STRUCTURE OVERVIEW

- | National Alliances on Rare Diseases
- | European Rare Diseases Federations or Networks
- | Patient Organisations' Members other than above
- | Patient Organisations and Patient Outreach in All of Europe
- | RareConnect
- | Committees, Task Forces, Group

# STRATEGIC PARTNERSHIPS

- | AFM–Téléthon: Association Française Contre les Myopathies
- | NORD: US National Organization for Rare Disorders
- | CORD: Canadian Organization for Rare Disorders
- | ORPHANET: The web server of medical experts generated and validated information
- | JPA: Japan Patients' Association
- | RPU: Russia Patients' Union
- | RVA: Rare Voices Australia
- | INDUSTRY through EURORDIS Round Table of Companies and other initiatives
- | ACADEMIA for education & capacity building and for social & policy research



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# HIGHLIGHTS 2014

- | EURORDIS reached **646 member patient organisations**, located throughout 60 different countries, including 526 full members in 26 EU countries.
- | The **EURORDIS Membership Meeting (EMM 2014 Berlin)** was held in Berlin in May back to back with the **European Conference on Rare Diseases (ECRD 2014 Berlin)**. EMM 2014 Berlin attracted 180 participants including 34 patient representatives that received fellowships from EURORDIS. ECRD 2014 Berlin was attended by 768 people from 43 countries representing all stakeholders.
- | **Rare Disease Day 2014** was organised in collaboration with partners in 84 countries worldwide, with over 1000 related events taking place. A Rare Disease Day European Policy Event was organised in Brussels. A brand new website was launched for Rare Disease Day 2014 and a mobile version for smartphone users was developed for Rare Disease Day 2015.
- | **RareConnect** continued to expand with 20 new online patient communities launched in 2014 reaching 70 global communities in collaboration with 560 patient organisations and 246 volunteer moderators and over 13.000 members.
- | Since 2010, EURORDIS alongside National Alliances has facilitated the organisation of **26 EUROPLAN National Conferences** whose main goal is to promote the design, the adoption and the implementation of patient-centered national plans/strategies for rare diseases. The EUROPLAN National Conferences have been currently embedded in the European Joint Action on Rare Diseases.
- | EURORDIS continued its work in the field of **Rare Cancers** reaching 64 member rare cancer patient organisations and rare disease organisations concerned by cancers in 2014. In addition, a EURORDIS Policy Action Group on Rare Cancers was set up with 6 patient representatives, 2 of which were appointed as EURORDIS patient representatives to the new **Commission Expert Group on Rare Cancers**.
- | The **7th EURORDIS Summer School** for patient advocates was held in Barcelona in collaboration with ECRIN-IA, for the third year in a row. 2014 gathered 37 participants representing 16 countries and 26 different diseases. EURORDIS also continued its strong involvement with the European Patients' Academy on Therapeutic Innovation project (EUPATI). Since 2008, 227 patient advocates have been trained so far.
- | **456 dossiers on orphan drugs**, advanced therapies and paediatric investigation plans were reviewed as part of participation in the European Medicine's Agency Scientific Committees. These include: 259 dossiers for orphan drug designations, 28 classification or certifications by the CAT and 172 Paediatric Investigation Plans by the PDCO.
- | EURORDIS continued its activities on **Health Technology Assessment**, working on the vision for the future of HTA in EU, advocating for engagement of patient representatives in HTA, and for adaptive approaches to rare diseases and orphan medicinal products.
- | The "**Common Goals & Mutual Commitments between EURORDIS & National Alliances in Europe: An Agenda between 2014 & 2020**" were adopted and promoted with 20 National Alliances having signed them by end of 2014. This structured collaborative framework aims at promoting greater convergence between National Alliances and EURORDIS as much as between National Alliances Themselves. The **16th Workshop of the Council of National Alliances** (32 National Alliances for Rare Diseases) was held in conjunction with the **7th Workshop of the Council of European Federations** (44 rare diseases-specific networks).
- | EURORDIS continued the work towards launching **Rare Diseases International** as an informal global network of rare disease patient organisations. A first RDI face-to-face meeting was held in November 2014 in Shenzhen China, attended by several international patient organisations and a pre-formation group, was formed, which will comprise the informal governance structure of the initiative until its official launch in May 2015.
- | EURORDIS further consolidated its **international partnerships with patient organisations** in 2014. EURORDIS has signed partnerships (MoUs) with the US National Organization for Rare Disorders (NORD), the Canadian Organization for Rare Disorders (CORD), the Japan Patients Association (JPA), Rare Voices Australia (RVA) and has MoUs under development with the Russian Patients Union and the Chinese Organisation for Rare Disorders (CORD).
- | EURORDIS has long called for the involvement of patients in benefit/risk evaluation of medicines. In 2014, this advocacy activity culminated with the first participation of patients in a **Committee for Medicinal Products for Human Use (CHMP)** oral explanation to discuss the marketing authorisation of a new treatment for a rare disease.
- | EURORDIS has contributed substantially to positioning the voice of patients at the heart of the development of **European Reference Networks** within the framework of both the EU Directive on Patients' Rights in Cross-Border Healthcare and the reflection process on Centres of Expertise and ERNs. Throughout 2014, EURORDIS continued paving the way for the next steps in ERN implementation to take place in 2015.
- | EURORDIS was proud to rely on **320 volunteers** in 2014 with 72 dedicated volunteer patient advocates, 2 office support volunteers and 246 volunteer RareConnect moderators, from a wide variety of diseases and countries, and all adhering to the EURORDIS Charter for Volunteer.
- | EURORDIS celebrated the 10 years of the **EURORDIS Round Table of Companies (ERTC)** with its 45 member companies in February 2014 with an ERTC workshop on "Unlocking Europe's Potential in Rare Disease Therapies" between now and 2020. Over the past ten years, the twice yearly round tables have regularly tackled the key issues that can unblock bottlenecks and advance real solutions for more, safe, efficient, effective, affordable, accessible therapy development.



# ADVOCACY

## Sustaining Rare Diseases as an EU Public Health Priority

### COMMISSION EXPERT GROUP ON RARE DISEASES

*The Commission Expert Group on Rare Diseases was set up by a Commission Decision of July 2013, replacing the former European Union Committee of Experts on Rare Diseases (EUCERD). The members and their representatives to the Commission Expert Group on Rare Diseases were appointed in January 2014 following a call for expression of interest. To support EU policy on rare diseases, the Expert Group may assist the Commission in the drawing up of legal instruments and policy document; advise the Commission in the implementation of Union actions and suggest improvements to the measures taken; advise the Commission in the monitoring, evaluation and dissemination of the results of measures taken at Union and national level; advise the Commission on international cooperation; provide an overview on Union and national policies; and foster exchanges of relevant experience, policies and practices between the Member States and the various parties involved.*

The work of the Commission Expert Group on Rare Diseases builds on the achievements of the EUCERD (2010–2013), including the 5 Recommendations adopted by this committee on centres of expertise, European Reference Networks, registries, indicators for national plans and improving informed decisions based on the clinical added value of orphan medicinal products ([www.eucerd.eu](http://www.eucerd.eu)).

The eight seats (four full members and four alternates) for patients on the Expert Group are held by EURORDIS members, mostly volunteer patient advocates, selected based on their expertise, advocacy track records, representation of large disease groups and geographic outreach. The four full members (and their alternates) each represent the following organisations: EURORDIS, the

European Network of National Alliances for Rare Diseases, the Network of European Federations of Rare Diseases and EGAN (the Patients Network for Medical Research and Health). Out of the current eight representatives, four members were already on the EUCERD.

This group of patient representatives is supported in their work by two observers from EURORDIS. Altogether, they form the EURORDIS Policy Action Group (PAG).

Throughout 2014, the patient representatives participated in the following CERD meetings:

- 1st meeting, 11–12 February 2014, EC, Luxembourg
- 2nd meeting, 03–04 July 2014, EC, Luxembourg
- 3rd meeting, 12–13 November 2014, EC, Luxembourg

The most important achievements of the Commission Expert Group on Rare Diseases in 2014 were the implementation report on the Commission Communication on Rare Diseases: Europe's challenges and Council Recommendation of 8 June 2009 on an Action in the Field of Rare Diseases (5 September 2014) and the Recommendation on Ways to Improve Codification for Rare Diseases in Health Information Systems (12–13 November 2014).

Major topics discussed in 2014 include: the ongoing work of the European Joint Action for Rare Diseases; the adoption and implementation of national plans for rare diseases; the design and implementation of future European Reference Networks for Rare Diseases; registries of rare diseases; activities of the International Rare Diseases Research Consortium (IRDIRC); Social Services for rare diseases.

All reports and recommendations produced by the Expert Group in 2014 are available on the new website:

[http://ec.europa.eu/health/rare\\_diseases/expert\\_group/index\\_en.htm](http://ec.europa.eu/health/rare_diseases/expert_group/index_en.htm)



## COMMISSION EXPERT GROUP ON CANCER CONTROL

*On 3rd June 2014, the European Commission adopted a decision which established a Commission Expert Group on Cancer Control. The members of this Expert Group were appointed following an open call of expression of interest. The Expert Group assists with drawing up legal instruments, policy documents, guidelines and recommendations on cancer control, as well as help prepare guidelines on cancer data. In addition, the Expert Group will fulfil an advisory role by providing the Commission with guidance on different issues. It will also advise the Commission in the implementation of Union actions and suggest improvements to the measures taken.*

Two volunteer representatives of EURORDIS have been nominated to represent patients affected by rare cancers in the Commission Expert Group on Cancer Control. One of them also sits on the European Commission Expert Group on Rare Diseases. EURORDIS considers that it is important to create a bridge between the two groups as several health policy issues related to rare cancers are common to these two groups: national rare disease / cancer plans, registries, European Reference Networks, quality criteria for Centres of Excellence, research policy, creating evidence and data in small populations, research policy and regulation, genetic testing and counselling, etc.

The patient representatives attended the first meeting of the CEG-CC on 23–24 September 2014 in Milan, Italy.

The most important achievement of the CEG-CC in 2014 was the Adoption of the 4th version of the European Code Against Cancer while major topics discussed in 2014 comprised:

- Ongoing European Joint Action CANCON (European Guide on Quality Improvement in Comprehensive Cancer Control).
- Rare Cancers
- Research, palliative care
- Cancer information systems

The work of the two EURORDIS volunteer representatives is supported by the EURORDIS Policy Action Group on Rare Cancers (PAG-RC).



## RARE CANCERS

EURORDIS' mission is to be the voice of all rare disease patients, including patients affected with rare cancers. As of end of 2014, EURORDIS counted amongst its members 64 rare cancer patient organisations and rare disease patient organisations concerned by cancers.

Activities pertaining to rare cancers throughout the year included:

- The nomination of two EURORDIS patient representatives on 29 August to the newly established European Commission Expert Group on Cancer Control. They both voice the rare cancer patients' perspectives and needs.

- The establishment of the Policy Action Group on Rare Cancers (PAG-RC) which supports the work of our volunteer patient representatives on the Commission Expert Group on Cancer Control. The PAG-RC is composed of six patient representatives from different rare cancer organisations nominated by the Board on 2 July and one member of staff.

- An initial work on documenting the similarities and differences between rare cancers and rare diseases which was initiated by the PAG-RC in order to highlight the benefits of joining forces for the purpose of improving access to treatment. This work will be further elaborated within 2015.

## THE EU PUBLIC HEALTH PROGRAMME 2014–2020: "HEALTH FOR GROWTH"

*On 26 February 2014, the European Parliament adopted the Commission proposal for the establishment of the third Health Programme 2014–2020: "Health for Growth".*

Rare diseases are recognised as a priority. Since 2010, EURORDIS has conducted intense advocacy activities and in 2012 worked with MEPs to table amendments to enhance the rare diseases priority.

## THE EU RESEARCH PROGRAMME 2014–2020: “HORIZON 2020”

*In December 2013, the first Work Programmes of Horizon 2020, the new EU Research Framework Programme for 2014–2020, were published. The topics of the Work Programme 2014–2015 for Health, Demographic Change and Wellbeing are characterised by an overarching, non-disease specific approach. However, a special focus on rare diseases is maintained, in line with IRDiRC priorities and the Regulation establishing Horizon 2020, following the amendments adopted by the European Parliament in 2013 that EURORDIS brought forward.*

A rare disease specific topic focuses on the development of “New therapies for rare diseases”, with a budget of 62 million EUR and between 10 and 15 proposals to be funded. However, 421 proposals were submitted for this topic, thus highlighting the interest in research for new rare disease therapies and the lack of sufficient EC funding to support them all.

Another rare disease specific topic addressed the creation of an “ERA-NET: Rare Disease research implementing IRDiRC objectives”. EURORDIS will collaborate with the

consortium to deliver an innovative funding scheme between patient organisations, members of EURORDIS, and national research funding agencies.

In 2014, EURORDIS submitted its contribution to the European Commission public consultations on the priorities for the next Work Programme for Health, Demographic Change and Wellbeing 2016–2017, as well as for the Work Programme for Science in and for Society 2016–2017. EURORDIS also shared rare disease research priorities with patient representatives at the Horizon 2020 Advisory Group for Health, assisting the European Commission in the development of Horizon 2020 Work Programmes.

In view of the adoption of the forthcoming Work Programmes, EURORDIS was in contact with the European Commission, DG Research and Innovation, and other stakeholders to have insights on the next calls for proposals and to spell out rare diseases research priorities.

## RARE DISEASE DAY EURORDIS POLICY EVENT

*Since 2008, EURORDIS has organised an annual Policy Event in Brussels to mark the occasion of Rare Disease Day. The aim of the Policy Event is to raise awareness and moves forward key policies and initiatives benefiting people living a rare disease.*

Keeping with this tradition, EURORDIS organised a Policy Event in Brussels for Rare Disease Day 2014 on 23 February. The Policy Event was on the topic ‘Improving Access to Rare Disease Care: The Vision of Patients’. Patients offered their vision on how to improve access on the following topics (1) Empowering People with Rare Diseases & Their Families (2) Addressing Unmet Medical Needs (3) Improving Patient’s Access to Quality Care and Innovative Treatments. The event was filmed and added to EURORDIS TV and remains available for public viewing.



# Support to National Plans for Rare Diseases in Europe: a 2013–2014 milestone!

## WHAT A EUROPLAN NATIONAL CONFERENCE MEANS FOR PATIENTS LIVING WITH RARE DISEASES?

*Since 2010, EURORDIS together with the National Alliances for Rare Diseases, or a patient organisation when a NA is not yet in place in a country, have organised a series of EUROPLAN National Conferences in 26 European countries: Belgium, Bulgaria, Croatia, Cyprus, Denmark, Finland, France, Georgia, Germany, Greece, Hungary, Ireland, Italy, Lithuania, Luxemburg, the Netherlands, Poland, Portugal, Romania, Russia, Serbia, Slovakia, Spain, Sweden, Ukraine, United Kingdom.*





The main goal of these conferences is to promote the design, the adoption and the implementation of patient-centered national plans/strategies for rare diseases, based on the European Commission Communication on Rare Diseases and EU Council Recommendations of 2009 in order to ultimately improve the life of the patients with rare diseases and their families.

The EUROPLAN National Conferences have been embedded in the EU co-funded project EUROPLAN (2008–2011) and the current European Joint Action on Rare Diseases (2012–2015) and co-funded by EURORDIS itself.

The **strength and success** of the EUROPLAN National Conferences lie in the philosophy of one **common approach**

**and format** for all countries, applying the following principles so that conferences are **patient-centred**, involve **varied stakeholders**, follow a **common format and content guidelines** on main key policy areas from research to health and social care, and facilitate the **integration of EU measures and recommendations** into national health systems (e.g. quality criteria for RD Centres of Expertise).

The patients' organisations are playing an instrumental role in shaping rare disease policy and advocating for patients' rights at national and European levels, an effort that often generates results in the long term. To date, 19 EU Member States have a rare disease national plan/strategy as compared to only 4 in 2008.

National Rare Disease Alliances all agree that EUROPLAN Conferences have been fundamental in enabling discussion around EU rare disease policy and planning, as well as in providing a great opportunity to bring various stakeholders together. You will find below the testimonies of several of them.

### BELGIUM – RADIORG, BELGIUM ALLIANCE FOR RARE DISEASES – 28 FEBRUARY 2014

The EUROPLAN conference has helped strengthen relationships and pave the way for further collaboration between Radiorg and the Belgian competent authorities. Even if the cooperation within the EUROPLAN project was mainly financial, we do consider this as recognition of Radiorg's important role in representing patients living with rare disease in Belgium. EUROPLAN did not play a specific role in the development of the Belgium National Plan because it was already officially published on the date of the conference; however, all the participating stakeholders were ready to further discuss the Plan's implementation.

### CROATIA – CROATIAN ALLIANCE FOR RARE DISEASES

17–19 SEPTEMBER 2010 et 27–28 FEBRUARY 2014

The Croatian EUROPLAN Conferences have served to bring together relevant stakeholders. During the Conferences, it was established that there was a need for improved collaboration and communication between all stakeholders. The involvement of patients in the organisation of the Conferences and in policy activities that followed has improved directly as a result of EUROPLAN. Having tackled many challenges, Croatia has now approved a national rare disease plan for 2015 – 2020.

### CYPRUS – CARD, CYPRUS ALLIANCE FOR RARE DISORDERS – 14–15 NOVEMBER 2013

The EUROPLAN Conference held in Cyprus has impacted the Cyprus Alliance for Rare Disorders' work and voice in the following ways:

- More stakeholders, in addition to the Ministry of Health, have taken an interest in rare diseases (for example, other patient organisations, the Ministry of Labour and

Social Insurance, the Ministry of Finance and political leaders).

- Several meetings have been held to highlight issues with the Parliamentary Health Committee, leaders of political parties, pharmaceutical services and the Ministry of Health.
- The Ministry of Health has seriously taken on board the designation of Centres of Expertise.

Unfortunately progress is hindered due to the economic crisis and changes in the healthcare system.

### DENMARK – RARE DISEASES DENMARK

19 NOVEMBER 2010 et 23 JANUARY 2015

Rare Diseases Denmark has strengthened its relations with key stakeholders through the organisation of EUROPLAN Conferences as they were notably involved in the EUROPLAN Steering Committee. These key stakeholders included government agencies on health care and social affairs, important rare disease health professionals and other key persons. New stakeholders have also appeared throughout the process, offering to contribute to future activities. The Conferences also contributed to a new level of awareness at a regional and local level, particularly of the special needs of people living with a rare disease who should have the same opportunities and rights as other citizens. However, the Danish strategy does not hold a concrete plan for budget and implementation; the effectiveness of the strategy remains to be seen.

### FRANCE – FRENCH ALLIANCE FOR RARE DISEASES – 30 SEPTEMBER 2010 et 13 JANUARY 2014

The second EUROPLAN Conference in 2014, organised by the French Rare Disease Alliance, was the opportunity to assess the outcomes of the 2nd national rare disease plan (2010 – 2014) and to maintain rare diseases as a national priority. Following two national rare disease national plans, this conference demonstrated the motivation of stakeholders to continue their commitment to the rare disease spectrum (from research to care). Finally, the role of the French Rare Disease Alliance has been strengthened through the organisation of the French EUROPLAN Conference. The second national plan has been extended by two year up to the end of 2016.

## GREECE – PESPA, GREEK ALLIANCE FOR RARE DISEASES – 26–27 NOVEMBER 2010 et 1ST DECEMBER 2012

The EUROPLAN Conferences were both a big success in terms of informing patients and stakeholders about rare diseases. PESPA had the opportunity to reach a large group of members of the general public. However, the Ministry of Health was unable to provide the level of support that PESPA had hoped for, perhaps as a result of the difficult economic and political situation in Greece at the time. For instance, between 2010 and 2014 there were 7 new Ministers of Health. Nevertheless, it was a chance for all stakeholders to get together and to form several partnerships with private health care institutions.



## HUNGARY – HUFERDIS, RARE DISEASES HUNGARY 15–16 OCTOBER 2010 et 25–26 OCTOBER 2013

The EUROPLAN Conferences catalysed a strong cooperation among all concerned stakeholders, resulting in the creation of the Hungarian rare diseases national plan. Now, we have practical, useful and progressive tools for the implementation of the national plan for rare diseases on a local level and in local institutions, following the EU recommendations. The EUROPLAN conferences contributed not only to strengthening the collaboration between stakeholders, but also to increasing the general knowledge and awareness of rare diseases in the Hungarian society.

## IRELAND – GRDO, GENETIC AND RARE DISORDERS ORGANISATION – JANUARY 20, 2011

The first EUROPLAN Conference in Ireland was facilitated by EURORDIS and organised by a committee of patient-led umbrella groups. In the weeks following the conference, the Minister for Health appointed a national steering committee to develop a national plan for rare diseases. This included four patient representatives, with a Rare Disease Task Force to support them. Following considerable work and consultation, the plan was published in July 2014 and implementation is now underway. The legacy of EUROPLAN1 can be measured by the structures that informed the development of Ireland's first National Plan for Rare Diseases and are now enabling its implementation. The Rare Disease Task Force remains in place today and is supporting the planning of the 2nd EUROPLAN Conference, which will take place in Ireland in June 2015. This will be an important event to ensure the implementation of the Irish NPRD.

## ITALY – UNIAMO, ITALIAN FEDERATION FOR RARE DISEASES

11–13 NOVEMBER 2010 et 27–28 JANUARY 2014

The EUROPLAN Conferences instigated the start of a new model of collaboration between national and regional institutions as well as with other stakeholders. This model was reproduced in other projects led by UNIAMO on different issues included in the national rare disease plan with successful outcomes:

- 1 – The use of telemedicine and teleconferences to gather and share expertise has recently been agreed by the regional technical boards that are tasked with optimising resources;
- 2 – Broader neonatal screening for 40 metabolic diseases will be implemented in all regions;
- 3 – Focus on transitional health care to meet shared needs in all regions and across diseases.

## LUXEMBURG – ALAN, RARE DISEASES LUXEMBURG – 19–20 NOVEMBER 2013

The EUROPLAN Conference in Luxembourg took place during a period when a new government was being established. The conference was a major milestone in the recognition of rare diseases; it raised awareness amongst the general public through good media coverage. In addition, the issue is now better understood by journalists. The discussion that took place between various stakeholders has increased communication and enhanced solidarity. On a national and international basis, networking has been greatly facilitated. The Health Minister, Lydia Mutsch, recently confirmed the Government's engagement to develop a national rare disease plan during her term in office and before spring 2018. A team will be set up at the Ministry of Health (probably in 2016) to better deal with all issues concerning rare diseases.



## NETHERLANDS – VSOP, THE DUTCH NATIONAL ALLIANCE FOR RARE AND GENETIC DISEASES

19 NOVEMBER 2010 et 14–15 NOVEMBER 2013

The second EUROPLAN conference entitled “**Quality for rare**”, with 150 participants, addressed two key issues: 1) Standards of care and 2) Centres of Expertise (CoE) for RDs. VSOP is the major developer of standards of care for RDs in the Netherlands. During the conference, VSOP and other patient organisations inspired participating stakeholders to develop (inter)national standards of care for a large number of RDs. Regarding CoE, as a result of both the first EUROPLAN Conference and the Dutch National Plan for Rare Diseases (October 2013), patient organisations, together with scientific assessors from Orphanet-NL, have an equal voice in the accreditation of several hundreds of candidate CoEs. The accreditation process (facilitated by the Netherlands Federation of University Medical Centers and VSOP) was evaluated during the conference. Further discussion took place on the important role of CoEs in creating networks with other healthcare providers, patient organisations, pharmaceutical companies, health insurance companies and European Reference Networks. In conclusion, the Conferences paved the way towards a sustainable healthcare infrastructure for providing integrated care for RD patients in the Netherlands.

## POLAND – ORPHAN, POLISH NATIONAL FORUM ON THE TREATMENT OF ORPHAN DISEASES

22 OCTOBER 2010 et 27–28 SEPTEMBER 2013

The Polish EUROPLAN Conference of 2013 attracted a great level of interest from patients, doctors and policy actors. Attended by over 160 people, it helped to put pressure on the Ministry of Health to adopt the national plan for rare diseases. During the conference there was a public debate organised with patient organisations, national payers, academy, philosophers, ethicists and drug manufacturers. Its result was the publication of a list of recommendations on the necessary changes in the Polish health care system related to rare diseases treatment in Poland. The need for an egalitarian approach to these disorders has been widely promoted.

## PORTUGAL – APADR, PORTUGUESE ALLIANCE FOR RARE DISEASES – 28 FEBRUARY 2015

- Planning the EUROPLAN Conference in Portugal strengthened cooperation with the national Orphanet team.
- Health professionals became even more eager to cooperate with rare disease patients. General awareness regarding both APADR and rare diseases in general improved greatly. A new strategy for rare diseases was announced on Day one of the conference, not launched in connection with the conference, nor as detailed as the previous national plan for rare diseases, but a fresh document to start working from.

## ROMANIA – RONARD, ROMANIAN ALLIANCE FOR RARE DISEASES

18–19 JUNE 2010 et 24–25 MAY 2013

The EUROPLAN Conferences have improved relations between all stakeholders including the Ministry of Health and helped create a sense of belonging, of being part of Europe. In the spirit of collaboration, all stakeholders concentrated on common problems and solutions rather than on differences. Rare Diseases have become one of Romania's health priorities and the National Plan for Rare Diseases is imbedded in the national strategy for health.



## SPAIN – FEDER, SPANISH FEDERATION FOR RARE DISEASES

5–6 NOVEMBER 2010 et 20–21 NOVEMBER 2014

The EUROPLAN Conferences have improved relations among stakeholders and centralised needs of rare disease patients. The 2014 Conference provided an invaluable opportunity to reflect on FEDER's priorities and to clarify the organisation's main goals, the foundation of FEDER's strategy. The main conclusions of the Conference have been used to write a top-ten list of priority rare disease policies, which has been shared with the main political parties and public administrations for their approval.

## UK – RDUK, RARE DISEASE UK

16 NOVEMBER 2010 et 24 JUNE 2014

The last EUROPLAN Conference facilitated joint working between the four nations of the UK (England, Scotland, Wales and Northern Ireland) by bringing together health officials from each nation and identifying a number of key areas for interaction and collaboration. The Conference highlighted the progress towards national implementation plans and put pressure on the nations that had fallen behind on their commitment to implementing the UK Strategy for RDs. Ahead of the General Election, RDUK with the support of its members continues to raise awareness of rare diseases and push each health department to deliver the 51 commitments included in the UK Strategy for RDs.

## GEORGIA – GERAD, GEORGIAN FOUNDATION FOR GENETIC AND RARE DISEASES – 17 APRIL 2013

The EUROPLAN Conference was instrumental in raising awareness of rare diseases and advocating for creating a political framework that would support the development of a national strategy/plan for rare diseases that will tackle key issues: including RDs in the list of main health care priorities, improving access to diagnostics based on the implementation of everyday practice of genetic and molecular testing, approving a series of RD guidelines for better treatments, fostering the development of effective registries, creating residential homes and other educational programmes for rare disease patients and strengthening Georgia's integration into the international RD community. Now, health authorities as well as relevant stakeholders have started to give more attention and allocate more human and financial resources to rare diseases.

## SERBIA – NORBS, NATIONAL ORGANISATION FOR RARE DISEASES – 6-7 DECEMBER 2013

The EUROPLAN Conference on rare diseases organised in Serbia created the first opportunity to bring together all stakeholders to enter into open dialogue about a national rare disease strategy. After the conference, NORBS held numerous meetings with the Ministry of Health and the national health insurance fund so as to encourage dialogue on the different aspects of the lives of patients. The Ministry of Health has now decided to start working on the National Strategy for Rare Diseases 2015-2020 and has formed a working group to draft this Strategy. The NORBS President and Executive Director have both been appointed to this working group as full members. The first draft of the document might be ready for public review mid-2015. NORBS is very proud to now be recognised as an equal partner among policy makers.

## UKRAINE – CSMA, KHARKIV CHARITABLE FOUNDATION – CHILDREN WITH SPINAL MUSCULAR ATROPHY – 27 MARCH 2013

While the health care system in Ukraine was neither ready nor motivated to recognise rare diseases as a health priority at the State level, the EUROPLAN Conference of March 2013 served as a catalytic event: the patients' voice has been heard and rare diseases have received more attention from policy makers. In April 2013, the first Ukrainian law for Orphan Diseases was adopted. The next step is to develop a national strategy and improve the implementation of the law.

**As a conclusion**, National Alliances and rare disease patient organisations all over Europe need to continue shaping national strategies and participate in national and European decision-making processes. Significant progress has been made in recent years in terms of recognising rare diseases as a health priority but there is still a long way to go to ensuring equal access to care and social services for rare disease patients. **At the present time, securing the implementation, furthering the EU integration and enhancing the sustainability of national rare disease plans appear to be the main challenges across the EU.** Patient representatives play a decisive role in securing that momentum is not lost, that all parties maintain their engagement and commitments are respected.

A final challenge is to ensure that solid monitoring and evaluation processes take place that help refocus actions and pave the way for the next generation of rare disease policy planning.

EURORDIS wishes to warmly thank all the EURORDIS-EUROPLAN Advisors who have played a major role over these last years to accompany and advise National Alliances for organising successful conferences having a significant political impact.

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- Vlasta Zmazek, Croatian Alliance for Rare Diseases, Croatia





# Rare Diseases: An International Public Health Priority

## RARE DISEASES INTERNATIONAL

*Rare Diseases International (RDI) is an initiative which aims at creating an informal network of patient organisations for the purpose of expanding the movement of rare diseases at an international level, providing mutual support between patient organisations and being able to speak with one voice.*



**RARE  
DISEASES  
INTERNATIONAL**

A EURORDIS INITIATIVE

In consultation with international partners such as CORD and IAPO, EURORDIS developed a survey that was disseminated to over 100 contacts/patient organisations internationally. The survey received over 60 responses from 37 countries around the world. The results were analysed in early 2014 and respondents were overwhelmingly in favour of the creation of a Rare Diseases International initiative (97.62% replied that they would be interested in joining).

Throughout 2014, contact was made with a number of prominent international rare disease organisations in order to follow up on their interest in creating this international network. In addition, the Joint Declaration: Rare Diseases as an International Public Health Priority, which has been a work-in-progress for several years, was updated and finalised using information received through the international survey. The Joint Declaration was presented at a first RDI face-to-face meeting which was held on 7 November 2014 in Shenzhen China, back to back with the International Rare Disease Research Consortium (IRDiRC) conference. 12 representatives of large scale international patient organisations were present at the meeting and agree to the creation of an RDI Pre-Formation Group. This group, which is composed of



international organisations that EURORDIS works closely with and/or has signed MoUs with, will act as the initial governance structure of the RDI initiative and will work on finalising the RDI structure, documents and logo until its official launch in May 2015.

## INTERNATIONAL PARTNERSHIPS WITH PATIENT ORGANISATIONS

In recognition of the need to unite, expand and reinforce the rare disease movement of patient organisations and patient advocates around the world, EURORDIS has established several partnerships with common objectives. EURORDIS signed Memorandums of Understanding (MoUs) with the **US National Organization for Rare Disorders (NORD)** in 2009 which was renewed in 2014, the **Canadian Organization for Rare Disorders (CORD)** in 2012, the Japan Patients Association (JPA) in 2013 and **Rare Voices Australia (RVA)** in 2015.

The MoUs are part of the ongoing effort to stress the international dimension of the rare disease movement and the global benefits to be gained by collaboration and enhanced cooperation with partners. The MoUs specify the common objective to strengthen the united

international voice of rare disease patients and promotes a global approach through participation in the annual awareness-raising event Rare Disease Day, international patient forum RareConnect, Rare Diseases International, and other possible joint actions such as common position papers and collaboration in diverse areas including research, diagnostics, information, treatment, and quality of life.

The partnerships reiterate the willingness of national umbrellas of rare disease patient organisations to collaborate globally in order to gain mutual benefits as much as to promote rare diseases as an international health policy priority.

Currently, there is an ongoing development of partnerships with the **Russian Patients Union (RPU)** and the **Chinese Organisation for Rare Diseases (CORD)** which are both planned for 2015.

## EURORDIS–NORD STRATEGIC PARTNERSHIP

*For the past 15 years, EURORDIS and the US National Organization for Rare Disorders (NORD) have joined efforts to improve the lives of rare disease patients on both sides of the Atlantic by providing mutual support to the development of a series of activities.*

In 2009, a five-year strategic partnership, aimed at converging strategies between the two organisations, was signed by both organisations. This strategic partnership came to an end in July 2014 and was renewed with a new five year MoU for 2014–2019.

The two organisations continued to play a key role in promoting rare diseases as a public health priority in 2014 with notable areas of collaboration being: Rare Disease Day 2014 (NORD is EURORDIS' official partner for RDD in the USA); joining the Rare Diseases International preformation group and the European Conference on Rare Diseases and Orphan Medicinal Products 2014 Berlin, where NORD was an official conference partner. EURORDIS also participated in NORD's Regional Summit in Alexandria, Virginia, USA, on 20 October and attended the NORD Rare Diseases and Orphan Products Breakthrough Summit on 21–22 October.



## EURORDIS–CORD PARTNERSHIP

*EURORDIS and the Canadian Organization for Rare Disorders (CORD) have been collaborating together for many years. CORD is a member of the EURORDIS Council of National Alliances. In 2012, an official five-year partnership was established between CORD and EURORDIS to further enhance collaboration on international advocacy activities, especially in the areas of Rare Disease Day, Rare Diseases International and collaborations on conferences.*

In 2014, CORD and EURORDIS continued their close collaboration: CORD actively participated in Rare Disease Day 2014 (CORD is EURORDIS' official partner for RDD in Canada); joined the Rare Diseases International preformation group; and the European Conference on Rare Diseases and Orphan Products (CORD was an official conference partner and also actively participated in the event).



## EURORDIS–JPA PARTNERSHIP

*EURORDIS and the Japan Patients Association (JPA) signed a Memorandum of Understanding in 2013, bringing patient advocates from Europe and Japan together to promote rare diseases as an international health priority.*

In 2014, JPA and EURORDIS continued their collaboration. JPA attended the European Conference on Rare Diseases and Orphan Medicinal Products 2014 Berlin, joined the RDI Pre-formation Group and attended a face-to-face meeting on RDI organised and led by EURORDIS in Shenzhen, China on 7 November.





# Improving Access to Orphan Medicines

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*Disparities in access to market-authorised orphan medicines exist between as well as within the EU Member States. This situation translates into varying delays from the marketing authorisation date to the pricing and reimbursement decisions amongst Member States. Different EU initiatives on how to improve **access to treatments based on patients' health outcomes are currently being explored and how to coordinate these at the European level is being examined. To this end, EURORDIS continues to join efforts with representatives of industry and regulators in addition to HTA, payers and the EMA.***

In 2014, EURORDIS sustained its advocacy work in favour of the definition, adoption and implementation phases of these European-level initiatives aimed at improving access for patients, including the Mechanism of Coordinated Access to Orphan Medicinal Products (MOCA), Parallel Scientific Advice by EMA and HTA agencies and future common HTA assessment reports within the SEED project.

## MECHANISM OF COORDINATED ACCESS TO ORPHAN MEDICINAL PRODUCTS (MOCA)

*EURORDIS has participated in the Mechanism of Coordinated Access to Orphan Medicinal Products (MoCA) of the Platform on Access to Medicines in Europe since its creation in 2010. This platform is one of the three work areas of the Process on Corporate Responsibility in the field of Pharmaceuticals launched by the European Commission (DG ENTR).*

In 2014, EURORDIS followed up the implementation phase and launch of first pilots through the MEDEV and has been advocating for coordinated mechanism towards EU decision makers as well as encouraging industry to submit pilots.

## DIFFERENTIAL PRICING

EURORDIS participated in the meetings of the Group on "Equity of Access and Sustainable Pricing Approaches for Pharmaceuticals in Europe", organised by FIPRA International, and chaired by former MEP John Bowis and the Head of the Institut national d'assurance maladie-invalidité (the Belgian National Health & Disability Insurance), Jo De Cock. Meetings attended include those of October 2014 on "Equity of access and sustainable approaches to financing of pharmaceuticals" in Brussels and the Debrief from Italian Presidency Informal Council in September 2014. EURORDIS has been liaising with different stakeholders on how to tackle inequalities of access to medicinal products, within and beyond Europe and has regularly underlined the need to have all stakeholders at the table, especially the most reluctant ones: payers and HTA bodies.

This Group will be focusing on access to precision medicines in the beginning of 2015.

## PATIENT PROGRESSIVE ACCESS

*Patient progressive access, also referred to as Medicine Adaptive Pathways to Patients (MAPPS), has continuously been promoted by EURORDIS since 2012. EURORDIS position is to optimise the use of current EU Regulatory Framework – Conditional Approval, Exceptional Circumstances, Risk and Benefit Management Plans – to provide earlier access to rare disease therapies in the absence of alternative therapies or when highly innovative treatments may translate in patients' medical benefits. EURORDIS has been, and will continue to be in the coming years, instrumental in elaborating the concept and piloting of adaptive pathways as part of the overall strategy and efforts aimed at improving access to orphan medicinal products. EURORDIS has regularly liaised with the European Medicines Agency (EMA), HTA bodies and European Commission on this initiative. EURORDIS has had a fundamental role in promoting dialogue between all major stakeholders involved in improving access to patients with particular focus on getting HTA bodies and Payers engaged into different platforms and mechanisms such as the MAPPS and the MOCA as elaborated above.*

EURORDIS participated as the organisation representing patients at the first meeting on Accelerated Access to Medicines to Patients co-hosted by the EMA, IMI, EFPIA, and MIT NEWDIGS (NEW Drug Development Paradigm) in London in April 2014. This meeting laid down the basis of stakeholder dialogue on the MAPPS mechanisms. In October 2014, EURORDIS was invited to represent rare disease patients at a senior conference at the Royal College of Physicians about new trial pathways and better patient data, which pointed out the challenges around payers' engagement into MAPPS. This conference was also organised by EFPIA, IMI, and MIT NEWDIGS.

Advocacy work carried out in the second half of 2014 included sending to the IMI team a paper on the vision of rare disease patients on ways to optimise patient engagement in the medicine development process with particular emphasis on the adaptive pathways mechanism.

The area of medicine development is rapidly evolving and challenging society faced with national health budgets pressure. While the landscape is rapidly changing, the opportunities of innovation are growing. One key area of change is the engagement of patients all along the life cycle of a product, at the time of development with academia and industry, at the time of assessment with regulatory or HTA bodies and payers.

## PATIENTS' INVOLVEMENT IN THE BENEFIT / RISK EVALUATION OF MEDICINES

EURORDIS has long called for the involvement of patients in benefit/risk evaluation of medicines. Since 2011, EURORDIS has played a frontline advocacy role in promoting such involvement by working closely with the Patient and Consumer Working Party of the Committee

for Medicinal Products for Human Use (CHMP)<sup>2</sup>, starting patient involvement in several scientific advisory groups of the CHMP and training patients for such activities.

In 2014, this advocacy activity has culminated with the first participation of patients in a CHMP oral explanation with the marketing authorisation applicant to discuss the marketing authorisation of a new treatment for a rare disease.

# Improving Access to Quality Care

## FOLLOWING-UP THE IMPLEMENTATION OF THE DIRECTIVE ON PATIENTS' RIGHTS IN CROSS-BORDER HEALTHCARE

*Directive 2011/24/EU on patients' rights in cross-border healthcare clarifies the rules on access to healthcare in another EU country, including reimbursement issues. EURORDIS has been instrumental in placing the focus of the Directive on patients living with a rare disease and on the specificities of rare diseases which require mobility of experts and expertise, of data and of patients at some crucial moments such as diagnosis and reassessment of therapy. The three main elements of EURORDIS advocacy activity have been reflected in the Articles relating to: 1. Rare Diseases; 2. The European Reference Networks for Rare Diseases; and 3. Cooperation between Member States on Health Technology Assessment.*

the Directive through contributing to the development of European Reference Networks and being involved in the EUnetHTA Stakeholders Forum, an EU initiative to improve coordination and harmonisation of the assessment tools used by the main HTA agencies in Europe.

## EUROPEAN REFERENCE NETWORKS (ERN)

*Within the framework of both the EU Directive on Patients' Rights in Cross-Border Healthcare and the reflection process on Centres of Expertise and ERN that started in 2005, EURORDIS has contributed substantially to the development of ERNs from the very beginning through to the finalisation of the EUCERD Recommendation on ERN.*

EURORDIS has put forward a patient-centred vision of the overall system to be achieved at an EU level, aimed at establishing a limited number of ERN that cover all rare diseases and that are gathered according to therapeutic area. These ERN will pragmatically stem from existing networks of experts and patient groups and will widen in a step-wise approach to the most mature centres wishing to establish a reference network at the European level. EURORDIS advocates that the overarching goal is to reach a global system where no rare disease patient will be left out, so that each patient can 'find a home'. This should also include undiagnosed patients by the end of the process.

To this effect, EURORDIS' actions in 2014 included:

- Raising awareness and capacity building on ERNs among EURORDIS' member patient organisations, national alliances and European Federations. This was done through internal consultation and annual conferences to prepare for the first call for tender for ERN applications in 2015;
- Building awareness and capacity with patients and patient organisations through participating in regional conferences lead by the European Patients Forum in northern, central and eastern European countries;
- Engaging with international consortia for undiagnosed conditions to raise awareness of ERN and stimulate discussions on models for undiagnosed patients in ERNs;
- Completing a comparison review of the EUCERD



EURORDIS has engaged in extensive advocacy work with respect to the cross-border Directive from the very beginning of the process. Following the approval of the Directive, EURORDIS has accompanied the transposition phase of the Directive into National Laws encouraging National Alliances, throughout 2013, to engage into discussion with their health authorities for the transposition of the Directive at national level. EURORDIS is currently following the implementation of

<sup>2</sup> Committee for Medicinal Products for Human Use



Recommendation on European Reference Networks for Rare Diseases, the Delegated Decision and the Implementation Decision;

- Contributing to the engagement of EUCERD through a Joint Action workshop with Member State leads to review the specificities of rare diseases that should be considered in the development of ERN and the assessment manual and technical toolbox that will be used for healthcare provider ERN applications;
- Reviewing funding opportunities for ERNs through European funding mechanisms, such as Structural Funds;

■ Broadly disseminating EUCERD Recommendations, Delegated Acts and information on ERNs to rare disease patient organisations;

■ Including ERNs as part of the European Conference on Rare Diseases, Berlin 2014;

■ Implementing content guidelines, which are based on EUCERD recommendations, for national conferences on national plans; and

■ Disseminating information through Eurordis.org and the EURORDIS eNews.

## Other issues

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### ADVOCACY TOWARDS OPTIMAL ENVIRONMENT FOR CLINICAL TRIALS IN THE RD FIELD

EURORDIS continues to encourage better cooperation between patient representatives and sponsors of clinical trials in the rare disease field. EURORDIS has been following the new EMA policy in publications of clinical trial data and is heavily involved in the EMA policy around this issues. EURORDIS also continues to advocate for the review of statistical methods for clinical trials in small populations, surrogate endpoints in clinical trials for rare diseases and alternatives to animal models.

EURORDIS attended an event during which we expressed our overall support to the revised EMA policy in this field. The event was hosted by the European Ombudsman at the end of December 2014 at the European Parliament on "Transparency and Public Health: How accessible is scientific data".

In September, EURORDIS co-chaired an event, organised by the European Forum for Good Clinical Practice on "How to ensure optimal ethical review within the new clinical trials regulation". At the workshop issues touched upon were: the independence of ethical review committees; how can the patients voice be heard; how should governmental support for patient participation in ethics committees be organised; how should the ethics review process be organised.

### DATA PROTECTION

*The Revision of the Data Protection Directive has been identified as a key issue involving rare disease specificities. EURORDIS has been carrying out an internal reflection process on data protection issues, in particular in relation to patient registries and mobility of data for research purposes while protecting patients' privacy at the same time.*

The 21st EURORDIS Round Table of Companies workshop in September 2014 was dedicated to "Sharing Rare Disease Patient Data: Translating Principles into Action". The workshop presented the opportunity to review and discuss issues around collecting, registering and sharing



patient data in the drug development process upstream in order to foster the development of treatments for rare diseases downstream. The need to harmonise and share patient data so that researchers are able to make the best use of results was highlighted. Patients expressed both willingness and caution regarding data sharing, particularly genetic data which provides new benefits for research but also increases the risks of discrimination. Rare disease patients request active representation in data collection programmes and need to trust the data-sharing network. Finally, this ERTC workshop explored the feasibility of establishing public-private partnerships in patient data collection. It was highlighted how researchers, companies and patient groups have already progressed well in building consensus around guiding and aspiring principles for collecting and sharing patient data to maximise opportunities for improved diagnosis and treatment development.



## TRANSPARENCY DIRECTIVE

The Council failed to agree on a way forward towards the adoption of a European Transparency Directive in the area of pricing of medicinal products. The Commission proposal is currently frozen.

## EMA CONSULTATIONS WITH PATIENTS IN 2014

Among other important EMA consultations, EURORDIS contributed to the EMA consultation for a procedure for safety public hearings. The first public hearing could take place in 2015 as a pilot and a learning process for the agency and stakeholders.

Another important reflection relates to the EMA Access Policy and Clinical Trials Transparency. This policy has now been adopted by the EMA Management Board, and EURORDIS emphasised the need to ensure greater transparency, whilst properly managing access to data by third parties so that the informed consent can be respected. This will help to avoid counter-productive controversies when third party findings differ from the clinical trial results.

EURORDIS also commented on the EMA Communication on Medication Errors, the EU Framework for Patient Registries, the draft functional specifications for the EU

Portal and EU Database of Clinical Trials created by the new Clinical Trial Regulation.

Regarding off-label use of medicines, EURORDIS commented on the Terms of Reference for an off-label research project mandated by the European Commission.

## MEMBERSHIP OF THE INTERNATIONAL SOCIETY FOR PHARMACOECONOMICS – ISPOR

Outcome research is rapidly evolving and major changes are already being implemented. One common feature is the agreed consensus on the importance to obtain the opinion of the patients or their representatives in all processes, and also sometimes to collect information from them. The measurement of the impact of medicines in the patient's life is a key component of the reflection on how to best assess the value of medicines for society.

In 2014, several EURORDIS members renewed their membership of ISPOR.

A second meeting of the Patient Centred Special Interest Group (PC-SIG) took place in 2014. Actions include a survey to ISPOR members regarding how they engage with patients in their work and the drafting of an ISPOR recommendation on how and why to engage with patients.

In 2014 EURORDIS participated in the ISPOR 17th Annual European Congress session 'Patient-Reported Outcomes and Patient Preference Research issues'.

## PARTNERSHIP WITH THE EUROPEAN FEDERATION OF INTERNAL MEDICINE (EFIM) & THE FOUNDATION FOR THE DEVELOPMENT OF INTERNAL MEDICINE IN EUROPE (FDIME)

The European Federation of Internal Medicine (EFIM) is a scientific organisation founded in 1996 to provide a scientific organisation of internal medicine on a European level. EFIM was formed by bringing together the national societies of internal medicine in European countries, both inside and outside the European Union. EFIM currently comprises 33 member Societies representing over 30,000 internists.

- IN 2013, EURORDIS, EFIM and FDIME signed an MoU agreement to work together to promote or develop: awareness of rare diseases, initiatives aimed at improving access to information, quality diagnosis, treatment and multidisciplinary care and also rare disease research.
- In 2014, EFIM was an official partner of the European Conference of Rare Diseases and Orphan Medicinal Products Berlin.
- EFIM organised a session on rare diseases at their European congress in June 2014 in Geneva, inviting EURORDIS to present its actions.
- EFIM invited EURORDIS to its annual seminar for young internists in July in Paris, to present the views of patients vis à vis ethical aspects in clinical research.



## INFORMATION & NETWORKING

### Membership Base – 646 members

50 new members joined EURORDIS in 2014 (23 full members and 27 associate members). New countries represented: Japan, Kazakhstan, Lebanon, Singapore, Uruguay.



### EURORDIS Membership Meeting – Berlin 2014

*Every year EURORDIS organises its Membership Meeting (EMM) in a different European city. This is an occasion for patient representatives to gather and learn from each other. The EURORDIS Membership Meeting comprises the annual General Assembly, a conference and several workshops. The majority of participants are EURORDIS member organisations and other patient organisations, while about 25% of participants represent policy makers, industry and academia. Each EMM has a specific focus on issues of high importance.*

The EURORDIS Membership Meeting (EMM 2014 Berlin) was held in Berlin in May back to back with the European Conference on Rare Diseases (ECRD 2014 Berlin). The event attracted 180 participants and comprised 8 'learning from each other' and capacity building workshops on topics including "Crowdfunding", "Views on ethical, legal and social issues in research" and "Patient advocacy to improve access to OMPs".

180 participants attended EMM 2014, including 34 patient representatives that received fellowships distributed by EURORDIS.





## Council of National Alliances

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National rare disease alliances serve to bring together the many rare disease organisations in a particular country. The CNA (Council of National Rare Disease Alliances), established by EURORDIS, allows national representatives of rare disease patients to work together on common European actions.

EURORDIS supports a network of 36 national alliances, 32 of which constitute the CNA. Two new national alliances joined the CNA in 2014, representing Latvia and Ukraine.

The CNA's main activities in 2014 were:

- a) The work on national plans for rare diseases (notably in the framework of the EUCERD Joint Action on Rare Diseases) by interacting with the ten EURORDIS EUROPLAN Advisors and beyond through regular and in-depth exchange regarding their experiences and the latest situation of national plans across Europe.
- b) The preparation and coordination of Rare Disease Day 2015.
- c) The finalisation and adoption of the Common Goals and Mutual Commitments between EURORDIS & National Alliances in Europe.

In 2014, a two-day CNA workshop took place in Paris in October, which was again held alongside the Council of European Federations (CEF) meeting in order to allow cross-cutting discussions on common issues.

The first day of the workshop gathered 33 participants focused on the following topics: Rare Disease Day 2015; an update on mutual commitments; early results & next steps for the Access Campaign survey; an update on CERD, the Joint Action and the Rare Cancer Expert Group; national rare disease plans; structural funds; European Reference Networks; and a presentation of the current projects of the French National Rare Disease Alliance.

On the second day the meeting was held together with the CEF and gathered 45 participants to focus on: RareConnect; Rare Diseases International; and a debrief on the EUCERD Joint-Action Workshop 'Guiding Principles for Social Care in Rare Diseases' and next steps in the drafting of CERD recommendations in the social field.

### COMMON GOALS & MUTUAL COMMITMENTS BETWEEN EURORDIS & NATIONAL ALLIANCES IN EUROPE

The Common Goals & Mutual Commitments between EURORDIS & National Alliances in Europe is an initiative that aims to promote greater convergence and collaboration between national alliances, and between national alliances and EURORDIS, for an optimal synergy based on a set of common goals. The initiative is a shared process for the purpose of accelerating positive change for people living with rare diseases by 2020, notably

by further building a strong pan-European community of patient organisations and people living with rare diseases, by being their voice at the national, European and international level and by directly or indirectly fighting against the impact of rare diseases on their lives.

The elaboration of these Common Goals was a long process that was developed and refined between the CNA and the EURORDIS Board of Directors.

By the end of 2014, 20 national alliances within Europe had signed the Common Goals document.



## Council of European Federations & Networks

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*European Federations aim to federate national rare disease-specific patient organisations at the European level. The CEF (Council of European Federations), established by EURORDIS, allows European Federations to work together on common European actions.*

Representatives of European rare disease federations gathered in Paris in October 2014 to discuss issues that are important across Europe and across diseases. For the third time, and as detailed above, part of the meeting took place in conjunction with the CNA.

The second part of the meeting was dedicated to the topics 'Organisation of Centers of Expertise & European

Reference Networks' and 'New procedures at the CHMP/EMA involving patient representatives'. The next day representatives participated in a one-day capacity-building session on registries with trainer Anil Mehta, Dundee University.

EURORDIS continued the program "Support to European Rare Disease Federations" for the fifth year in a row; the smallest and/or youngest organisations often have great difficulties in financing their network meetings (board meetings, network meetings, conferences etc.) – EURORDIS has granted a total of 14 402,52 € to 11 European rare disease federations to help them organise their various meetings.

## European Network of Rare Disease Help Lines

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*The European Network of Help Lines for Rare Diseases aims at better serving the needs of the callers by sharing resources, best practices, common tools and knowledge base. It was created in September 2006 and is coordinated by EURORDIS. The network aims at increasing awareness, efficiency and best practice standards for its members.*

In 2014, 12 help lines from 8 countries participated in the activities: Bulgaria (ICRDOD), Croatia (National Help Line Zagreb), France (Maladies Rares Info Services, AFM-Téléthon), Italy (Coordinating Centre for Rare Diseases Veneto Region and Telefono Verde Malattia Rare), Portugal (LinhaRara), Romania (NORO, Myastenia Gravis Romania), Spain (SIO-Feder and ENERCA) and Switzerland (Info Maladies Rares, a new help line)

In addition to its annual meeting, the network conducted its 7th Caller Profile Analysis based on all enquiries received in October, a report for which is available. An article was published in the Interactive Journal of Medical Research: "A European Network of Email and Telephone Help Lines Providing Information and Support on Rare Diseases: Results From a 1-Month Activity Survey".

The various help lines participated in the EURORDIS Access Campaign, the questionnaire for which was translated in 19 languages. Help line respondents were invited to fill in the questionnaire when enquirers phone regarding difficulties they have accessing any type of care, or to propose the enquirers fill the survey in themselves.

## Rare!Together

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Rare!Together aims at helping in the creation, operation and management of European rare disease federations, in particular through the website [Raretogether.eurordis.org](http://Raretogether.eurordis.org). This website a good practice reference handbook and toolkit for existing European Federations and for patient organisations planning to set up their European Federation.

# RareConnect – Rare Disease Online Communities

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Launched in 2010 by EURORDIS in partnership with the US National Organization for Rare Disorders (NORD), RareConnect.org, the online patient community portal, has helped patients and families connect through an online social network in order to support each other and share vital experiences on aspects of living with a rare disease. Organised into disease-specific communities, this platform also provides links to quality information and involves patient associations in the creation, governance and growth of each community. Additionally, it offers a translation service whereby patients can request a human translation of any forum post into any one of the available languages.

RareConnect experienced further growth in 2014. By the end of 2014, 70 disease-specific online communities had been successfully created (22 new ones in 2014). These communities saw international partnerships emerge from 560 different patient organisations that are committed to international networking, building cross-border disease knowledge and reaching isolated people living with the disease.

During 2014, the RareConnect website received over 80,000 visits per month from 22 countries. Subscribed members reached a total of 13,438, amounting to a 70% increase compared to 2013.



RareConnect is a multilingual platform in five languages (English, French, German, Italian, and Spanish). During 2014, RareConnect translated approximately 1.2 million words across twenty language pairs.

The RareConnect network is also composed of over 250 moderators who volunteer their efforts and expertise in managing diseases and finding quality information online. Many of these moderators come from EURORDIS member organisations and play a key role in requesting, creating, moderating and maintaining the communities with the goal of enhanced global cooperation and increased knowledge on daily living with a rare disease. Moderators are supported daily via a series of tools which include individual attention from one of the team's 2 community managers, a peer-driven mailing-list to exchange experiences, a best-practises blog available at moderators.rareconnect.org and a training guide.

In May, a EURORDIS Membership Meeting workshop in Berlin was dedicated to the RareConnect project and social media use.



# EURORDIS Website

The EURORDIS website provides information relating to the role of patient organisations in the development of rare disease and orphan medicines policy translated into 7 languages (English, French, German, Italian, Portuguese, Russian and Spanish), while also outlining the activities of EURORDIS. The total number of website visits over the year amounted to 367 208, a 19% increase compared to 2013.

Based on user feedback, several improvements were introduced to the website in order to enhance overall navigation and to give greater visibility to EURORDIS projects and volunteers. New website sections include

those on compassionate use, volunteers, the European Year for Rare Diseases 2019, information on the EURORDIS eNews (and a new sign-up form). Several other webpages were redesigned and updated, such as those on rare disease policy, what EURORDIS does and the EURORDIS Photo Contest.

The development of a 'related documents' tool allows users to have a direct link from the page they are consulting to a list of EURORDIS related documents when possible, and thus go deeper into details of the current subject.



www.eurordis.org

# EURORDIS TV

EURORDIS TV was launched in April 2013. It provides a dedicated, EURORDIS branded, video portal available within Eurordis.org that aggregates quality rare disease video content and promotes videos produced by EURORDIS.

Through EURORDIS TV, video footage is provided of testimonies, speeches & events, conference presentations, training or capacity building activities and Rare Disease Day events. In addition, EURORDIS TV promotes in-house recordings presenting key policy issues, papers, the EURORDIS Summer School and EURORDIS Membership Meeting testimonies. One 'video of the week' is selected every week and promoted on Eurordis.org and in the EURORDIS eNews, connecting with our audience in a new interactive way. In 2014, 84 videos were added to EURORDIS.



# EURORDIS InfoHub

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The internet is one of the first places people living with a rare disease turn to in search of information. Without wishing to duplicate the resources which already exist on the web, EURORDIS has developed the EURORDIS InfoHub for the purpose of meeting three basic needs: to provide a guide to patients on how to find quality rare disease information on the internet; to provide a place where people can find a quality-driven selection of rare disease resources; to comprise a specific tool which gives access to search results based on an index of affiliated patient organisation websites.



# EURORDIS Photo Contest 2014

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*Initiated in 2006, the EURORDIS Photo Contest is an annual event that raises awareness of rare diseases among members of the general public and members of the rare disease community. It helps to build the EURORDIS network of people living with a rare disease and their families and helps us to collect photos that can be used in EURORDIS communication materials.*

The EURORDIS Photo Contest 2014 was open to public voting via social media. The photo contest was a great success with 410 photos received from 44 countries. Three separate awards were given out: the Public Award selected by the public at large (180,000 votes were received through Facebook); the Staff Award, voted for by the EURORDIS Staff; and the Expert Award, selected by professional fashion photographer, Rick Guidotti.

1. The winning photo for the Public Award entitled 'My hero' is of 19-month-old Karlo from Croatia who lives with Niemann-Pick disease type C.
2. The Expert's Choice photo is of 2-year-old Daniel from Russia who has Williams syndrome.
3. The EURORDIS Favourite prize is awarded to the photo of Lola entitled "The world through her eyes". Lola lives with diploid-triploid mosaic syndrome.



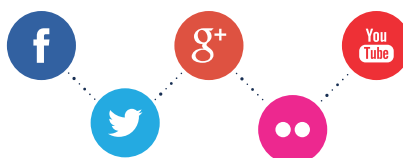
# EURORDIS eNews

The EURORDIS eNews is a weekly news report that communicates breaking news of interest to patient advocates and policy makers. Each eNews features a lead article (devoted to important news in the rare disease community or EURORDIS activity) in addition to short news sections on topics including new RareConnect communities, member events and EURORDIS TV content. Content is also made available via EURORDIS Facebook and Twitter.

The eNews is entirely written, constructed and disseminated in-house by EURORDIS staff or automatic programs created by EURORDIS staff. The only element that is outsourced is the weekly translations; the eNews is available in all 7 languages of the website (English, French, German, Italian, Portuguese, Russian and Spanish). In 2014, 45 eNews issues were written, translated and produced/ distributed via email to over 8,000 subscribers.



# EURORDIS Social Media



EURORDIS has its own Facebook page ([facebook.com/eurordis](https://facebook.com/eurordis)), Twitter account ([twitter.com/eurordis](https://twitter.com/eurordis)), Flickr account ([flickr.com/photos/eurordis](https://flickr.com/photos/eurordis)), YouTube channel ([youtube.com/eurordis](https://youtube.com/eurordis)), and Google +.

EURORDIS continued to leverage existing social media tools in order to communicate and interact with the rare disease community in 2014.

In 2014, EURORDIS Facebook posts and updates received 474,000 views and generated 27,000 interactions with Facebook subscribers. The EURORDIS Facebook page reached a total of 25,354 likes by the end of the year, receiving 6,205 likes alone during the Photo Contest period. EURORDIS Twitter was also particularly active in 2014 with a first interactive twitter-chat run for the EURORDIS Policy Event held in Brussels on Rare Disease Day 2014. A new feature was also introduced for ECRD 2014 – a screen displaying live tweets under with #ECRDBerlin. By end of 2014, Twitter followers had reached 7,417.



# EURORDIS Policy Fact Sheets

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*EURORDIS has created specific policy fact sheets to help rare disease patients and organisations better advocate issues of importance with national and/or European policy decision makers. The EURORDIS policy fact sheets provide comprehensive, validated information on specific topics relevant to rare diseases. All these documents are available in printed form and online on Eurordis.org in English and are widely distributed to members at relevant events.*



## Rare Disease Day 2014

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*Rare Disease Day is an annual, awareness-raising event co-ordinated by EURORDIS at the international level and by national alliances and patient organisations on the national level. The main objective of Rare Disease Day is to raise awareness amongst the general public and decision-makers about rare diseases and their impact on patients' lives.*

28 February 2014 marked the 7th edition of the Rare Disease Day Campaign with the slogan '**Join Together for Better Care**'. The campaign saw active participation in 84 countries, 36 of which are European countries and 27 EU countries. EURORDIS and 29 rare disease national alliances, together with patient groups acting as country organisers, mobilised thousands of patient organisations across 5 continents. New national alliances from Finland, Slovakia and the Czech Republic joined in 2014.



Rare Disease Day

Around the world over 1000 unique events were staged on or around Rare Disease Day by patient organisations and other partners.

EURORDIS organised a Policy Event in Brussels entitled 'Improving Access to Rare Disease Care: The Vision of Patients' (for further information refer to the 'Rare Disease Day Policy Event' in the Advocacy section of this report).

Rare Disease Day 2014 saw the launch of a new website including easier navigation for users, more visible links to social media and a completely revamped backend, facilitating organisations to upload pictures, videos and logos. The website includes: a downloadable toolkit with an information pack, logo, poster and other materials accessible to organisers; press releases for journalists; links to social media networks; and a section for patients to upload photos and videos. In January and February the website received over 150,000 unique visits, with 50,000 visits on 28 February alone (over double the amount of visits compared to 2013). Rare Disease Day 2014 received over 40 000 likes on Facebook and there were 28,000 tweets on the subject of Rare Disease Day, breaking down to 17 tweets per minute.

For the 3rd year in a row EURORDIS proceeded with the production of a Rare Disease Day video, which was again a great success. EURORDIS worked with TheGloryDays, a production company based in Barcelona and several people living with a rare disease were actors in the video, which showed different scenes of care such as the home, a hospital and therapy. The video was translated into 21 languages including Russian, Arabic, Hebrew and Mandarin.

Hundreds of people responded to our call to 'Raise & Join Hands' to show solidarity with rare disease patients on 28 February. People could submit their photos of people joining hands via the website and these can be viewed on the Rare Disease Day Flickr gallery.

Finally, a Rare Disease Day mobile site for smartphone users was developed at end of 2014 for a launch prior to Rare Disease Day 2015.



# EURORDIS Black Pearl Gala Dinner

The third EURORDIS Black Pearl Gala Dinner, entitled 'Solidarity & Hope for Rare Disease Patients throughout Europe' and held on the occasion of Rare Disease Day 2014, took place in Brussels on 25 February 2014.

Over two hundred people attended the event – Brussels community members, national and international officials, researchers, professionals striving to make treatments available to rare disease patients, individuals living with a rare disease and their families and friends. They joined together to recognise the rare disease cause and the recipients of the EURORDIS Awards 2014.

The net proceeds of more than 238,000 euros collected from the Gala support community-building initiatives aimed at breaking the isolation of rare disease patients and their families in Europe and at empowering leaders of the rare disease community through training, capacity-building activities and exchange to stimulate research and increase public awareness about rare diseases.



"The EURORDIS Gala Dinner will go a long way towards raising awareness of the issues affecting the millions of European citizens living with rare diseases while securing much needed funds to support the continuation of the vital work undertaken by EURORDIS."

**AVRIL DALY, RARE DISEASES IRELAND**

"It is a paradox that rare diseases are common. Nor do rare diseases mean few patients. Some 33 million people in Europe live with one of the 6,000 rare diseases. 70% of these are neurological and 75% of such diseases affect children. That is why we need to put rare diseases higher up the health and research agenda and that is why the excellent EURORDIS is so crucial to this, and deserves our support."

**JOHN BOWIS OBE, FORMER MP, MEP & HEALTH MINISTER, UK (GALA COMMITTEE CHAIR)**

"Je ne savais pas qu'il existait tant de maladies rares touchant des millions d'enfants et d'adultes. Puis, j'ai connu EURORDIS, grâce à qui aujourd'hui, les malades ainsi que leurs familles, les soignants, les chercheurs reçoivent l'attention dont ils ont tant besoin. J'ai été impressionnée par le professionnalisme et la profondeur d'EURORDIS, par sa vision holistique, ainsi que par le travail quotidien de ses membres. Ils engendrent l'espoir. C'est pour moi un honneur et une joie d'apporter mon soutien au gala 2014."

**PRINCESS ANNE DE LIGNE, BELGIUM**

"I believe that the Black Pearl Gala is a wonderful initiative to raise public awareness of people suffering from rare diseases and to help promote their cause"

**DR. ECKART CUNTZ, AMBASSADOR OF THE FEDERAL REPUBLIC OF GERMANY TO BELGIUM**

"C'est un honneur de pouvoir apporter mon soutien au dîner de gala d'EURORDIS pour attirer encore davantage l'attention sur les maladies rares au profit des personnes qui en sont atteintes"

**JEAN-JACQUES WELFRING, AMBASSADOR OF LUXEMBOURG TO BELGIUM**

"I applaud EURORDIS' outstanding contributions to ongoing efforts to improve the lives of those facing the challenges of rare diseases."

**DAVID PLUNKETT, AMBASSADOR OF CANADA TO THE EUROPEAN UNION**

"I strongly support the mission of EURORDIS to increase public awareness of rare diseases, advanced therapy research, rare disease treatment and care, and to maintain rare diseases as a European Union priority in public health and research."

**MRS. MAJA IVANOVA, FIRST LADY OF THE REPUBLIC OF MACEDONIA**

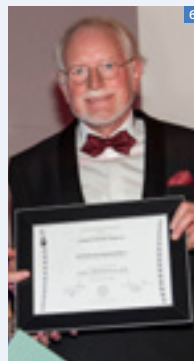


# EURORDIS Awards

*The EURORDIS Awards are designed to recognise the outstanding commitment and achievements of patients' advocacy groups, volunteers, scientists, companies, media and policy makers who have contributed – directly or indirectly – to reducing the impact of rare diseases on people's lives. These prestigious awards are judged by the EURORDIS Board of Directors based on over 130 nominations received from EURORDIS members, non-member patient groups, volunteers, staff and the general*

*public with the aim of promoting leadership and the highest achievements in favour of people living with rare diseases.*

2014 marked the third EURORDIS Awards for outstanding accomplishments in the field of rare diseases, which were presented in Brussels on the occasion of Rare Disease Day. The 2014 EURORDIS honorees were as follows:



## ■ LIFETIME ACHIEVEMENT AWARD 2014

**Marlene Haffner, MD, MPH** <sup>1</sup>

For her immeasurable impact upon the development of orphan drug therapies.

## ■ EUROPEAN POLICY MAKER AWARD 2014

**Dr. Antonyia Parvanova** <sup>2</sup>

For her dedication and commitment in addressing the needs of people with rare diseases as a Member of European Parliament.

## ■ EUROPEAN LEADERSHIP AWARD 2014

**Professor Guido Rasi, Executive Director of the European Medicines Agency (EMA), Professor Luca**

**Pani, Director General of the Italian Medicines' Agency (AIFA), and Paola Testori Coggi, Director General for Health and Consumer Protection at the European Commission.** <sup>3</sup>

To recognise their leadership and support for rare diseases and their capacity for far-reaching, innovative policies.

## ■ PATIENT ORGANISATION AWARD 2014

**Allianz Chronischer Seltener Erkrankungen (ACHSE)** <sup>4</sup>

For their instrumental role in turning Germany into one of the most committed Member States in the European Union for rare diseases in the fields of research, information, healthcare organisation and drug development.



#### | VOLUNTEER AWARD 2014

##### Lise Murphy <sup>5</sup>

For her outstanding contribution in closing the gap between patients, health care professionals and pharmaceutical agencies, demonstrating the importance of patient dialogue; and for using her experience and patient expertise generously for the benefit of the entire rare disease community.

#### | SCIENTIFIC AWARD 2014

##### Professor Hans Hilger Ropers, Director at the Max-Planck-Institute for Molecular Genetics in Berlin <sup>6</sup>

A clinical geneticist, Professor Ropers has made many contributions to the molecular elucidation of monogenic disorders with a focus on eye diseases, deafness and particularly intellectual deficit.

#### | MEDIA AWARD 2014

##### Rick Guidotti

For his efforts to raise public awareness for the beauty of difference via his beautiful photography and his non-profit organisation, Positive Exposure.

#### | COMPANY AWARDS 2014

##### Swedish Orphan Biovitrum (Sobi) <sup>7</sup>

For the excellence and consistency of their work in developing treatments for rare diseases and their track record of creating successful dialogue with patient communities.

##### Orphan Europe <sup>8</sup>

For their robust portfolio and strong pipeline of rare disease treatments, as well as for their committed engagement with patient organisations.



## Campaign for a European Year for Rare Diseases in 2019

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*Each year since 1983, the EU has chosen a specific topic for the European Year in order to encourage debate and dialogue within and between European countries. EURORDIS is spear-heading the campaign for a European Year for Rare Diseases in 2019 at the EU level with the support of rare disease national alliances, European Federations and patient groups.*

The aim of the European Year on rare diseases would be: to communicate on all rare diseases to raise public awareness; to find solutions to the challenges faced by patients, families and their care-givers both in the public and political spheres; to encourage researchers to focus on these rare, mostly unknown, seriously debilitating and often life-threatening diseases, which affect children and adults; and to create the conditions

for better health, medical and social care for all. 2019 has been chosen because it is an important year for the rare disease community – it marks the 20 year anniversary of the adoption of the EU Regulation on Orphan Medicinal Products and 10 year anniversary of the Commission Communication and Council Recommendation on rare Diseases.

Following the adoption of the Call for a European Year for Rare Diseases by EURORDIS' members at the General Assembly 2013 Dubrovnik, several actions have taken place in 2014. A dedicated webpage was launched in 2014, containing information about the campaign, a form to sign up and a counter widget and a media campaign will be further developed throughout 2015.



Please sign up to support the Campaign for a European Year for Rare Diseases at [eurordis.org](http://eurordis.org)!

# HEALTH POLICY & HEALTH CARE SERVICES

## European Conference on Rare Diseases & Orphan Drugs (ECRD 2014 Berlin)

*The European Conference on Rare Diseases & Orphan Products is the unique platform/forum across all rare diseases and all European countries, bringing together all stakeholders – patient representatives, academics, health care professionals, industry, payers, regulators and policy makers. It is a biennial event, providing the state of the art of the rare disease environment, monitoring and benchmarking initiatives. It covers research, development of new treatments, health care, social care, information, public health and support at European, national and*

*regional levels. It complements national and regional conferences, enhancing efforts of all stakeholders. There is no competition with them, but efforts fully respect initiatives of all, aiming at integrating EU and national policies and actions.*

ECRD 2014 Berlin took place at Andel's Hotel from 8 – 10 May 2014. 768 people attended from 43 countries representing all stakeholders.

### PARTICIPANTS



768

attendees

138

session chairs  
/speakers

38

sessions

+200

posters

43

countries

### HIGHLIGHTS



#### **Pertinent conference programme**

Discussion of hot topics: organisation of healthcare, research, development and availability of orphan medicinal products in Europe.



#### **High-level session chairs and speakers**

Diversity and complementarity of topics



**Case studies from:** Germany, Denmark, France, Belgium, Austria, UK, Portugal, Netherlands, Spain, Italy, Canada, USA, Sweden, Finland, Romania, Ireland, Norway and Hungary



#### **Involving a diversity of stakeholders:**

patient representatives, academics, healthcare professionals, industry, payers, regulators, and policymakers



#### **Highest participation since the first ECRD**

**2001:** 768 participants from 26 EU/EEA countries & 15 Eastern European countries



## Specialised Social Services and Social Policies

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In 2014 EURORDIS continued its focus on specialised social services and on social policies, mainly through its involvement in Work Package 6 of the EUCERD Joint Action Working for Rare Diseases (EJA).

The definition of these specialised social services continued to be disseminated via the concept paper entitled *Rare Diseases: Addressing the Need for Specialised Social Services and Social Policies*, through policy fact sheets and via the document *Guiding Principles for Specialised Social Services*.

The documents *Guiding Principles on Training for Social Services Providers* and *on Examples of Training Programmes for Social Services Providers* were finalised and distributed to the CEGRD, to EURORDIS members and made available online via the EURORDIS and the EUCERD websites.

EMM 2014 Berlin provided the occasion for EURORDIS members to learn from each other on education and employment initiatives. A capacity building workshop on 'Integration of Rare Diseases into Social Policies' was also organised at EMM 2015 Berlin, focused on the identification of social challenges of people living with a rare disease and on training for social services providers.

Social challenges and social policy were the main subjects of the Theme 6 of ECRD 2014 Berlin, entitled 'Beyond Medical Care'. The sessions focused on social challenges in rare diseases and on different approaches to overcome those. Presentations included outcomes of the EJA, social policy issues, case studies of service provision and testimonials of patients who have succeeded to live independently in spite of being confronted with significant social challenges. The poster on 'Integration of Rare Diseases into Social Services', developed based on the outcomes of EURORDIS' work within the EJA, received a

ECRD 2014 Berlin poster award and was disseminated via the *Orphanet Journal of Rare Diseases* later in the year.

EURORDIS continued to map specialised social services in cooperation with national alliances and European federations, reaching over 90 services from 22 countries by the end of 2014. The map of these services, in addition to definitions, fact sheets and testimonies of beneficiaries and volunteers can be found on the EURORDIS website in the designated section entitled 'Services to Patients: Specialised Social Services'.

Country visits to the CREER Resource Centre and to the Respite Care Service of Hendaye Hospital were also carried out in 2014.

In October, a workshop on 'Guiding Principles for Social Care in Rare Diseases' was organised to discuss care pathways and social care provision at Frambu Resource Centre, Norway, with 20 participants from 13 countries. The workshop was organised in the scope of the preparation of the CEGRD recommendations in the social field, for which EURORDIS is responsible as leader of the EJA Work Package 6.

In parallel to these actions, EURORDIS continued to distribute all collected information on specialised social services and social policies for people living with a rare disease to EURORDIS members, patient advocates, policy makers, authorities and the general public through the eNews and EURORDIS social networks.

During 2014, EURORDIS connected with several partners working on social policy in order to exchange important information and good practices: Orphanet Disability Projects; the International Federation of Social Workers; the Social Platform; and the European Social Network.





# RESEARCH, DRUGS & THERAPIES

EURORDIS intensified its direct role in advancing research in the field of rare diseases by expanding its efforts to strengthen ties with the research community, so as to improve diagnostic and therapeutic possibilities for rare disease patients. More specifically, EURORDIS continued promoting the development of sustainable, harmonised and integrated registry, biobanking and genetic data infrastructures and networks that reflect the patient's best interests. This was achieved by supporting the

involvement of patients and their representatives at both the governance and clinical research levels of several international projects and consortia.

In 2014, EURORDIS activities aimed to promote rare disease research by consolidation of patients' and EURORDIS' role in IRDiRC activities through contributions to meetings, governance, policy papers and recommendations; involvement in research projects like ASTERIX and InSPIRe; and participation in E-RARE.

## Shaping and Supporting Research Policy

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### INTERNATIONAL RARE DISEASE RESEARCH CONSORTIUM – IRDiRC



*The International Rare Disease Research Consortium (IRDiRC) teams up researchers and organisations investing in rare disease research in order to achieve two main objectives by the year 2020, namely to deliver 200 new therapies for rare diseases and means to diagnose most rare diseases.*

The International Rare Disease Research Consortium (IRDiRC) teams up researchers and organisations investing in rare disease research in order to achieve two main objectives by the year 2020, namely to deliver 200

new therapies for rare diseases and means to diagnose most rare diseases.

EURORDIS continued its involvement in IRDiRC with its presence in the Executive Committee (Béatrice de Montleau, EURORDIS Board member and Yann Le Cam, Chief Executive Officer of EURORDIS, Chair of IRDiRC Therapies Scientific Committee). This involved the preparation of and attendance at meetings in Berlin and at the second IRDiRC conference in Shenzhen, China; participation in conference calls; and the provision of specific contributions to the update of the governance and policy paper. Specifically, EURORDIS promoted a better dialogue and interactions between the three Scientific Committee Chairs, the Chair of the Executive Committee

and the Scientific Secretariat of the Consortium. Moreover, EURORDIS promoted a clear and tight agenda for the achievement of the overarching IRDiRC objective of having 200 more medicinal products for rare diseases on the market in 2020.

EURORDIS also continues to be involved in other IRDiRC governing bodies. Yann Le Cam, is the Chair-Elect of the Therapies Scientific Committee (TSC) since October 2013. Maria Mavris, who was member of the TSC since 2012, left EURORDIS this year. Virginie Hivert was nominated to replace her.

Yann Le Cam and Virginie Hivert are respectively members of the Working Groups on Orphan Drug-Development and Regulatory Processes and on Chemically-Derived Products Including Repurposing. Anna Kole (EURORDIS staff) is member of the IRDiRC Working Group on Registries and Natural History, and Tsveta Schyns (volunteer) is a member of the Working Group on Ethics and Governance.

EURORDIS staff provided inputs and support to its representatives in IRDiRC, in particular by helping them prepare for meetings and by providing regular contributions to key policy documents. In order to ensure optimal involvement, conference calls are regularly organised between EURORDIS representatives involved in IRDiRC.

## PROJECT ASTERIX

*The ASTERIX (Advances in Small Trials dEsign for Regulatory Innovation and eXcellence – FP7 project) Project is specifically designed to optimise methodology for clinical trials in small populations to achieve more reliable and cost-efficient clinical development of treatments for rare diseases.*

The main objectives of the Project are to: develop and design analysis methods for single trials and series of trials in small populations; include patient-level information and perspectives in design and decision making throughout the clinical trial process; and validate new methods and propose improvements for regulatory purposes.

Tsveta Schyns is a member of the Advisory Board of project. The board will provide expert advice on key open project decisions, general philosophy and direction of ASTERIX. It will also be instrumental in dissemination and exploitation of project results, as well as for the quality of deliverables and overall project status.

In October 2014, Kerry Lesson-Beevers participated in the first ASTERIX Patient Thinktank in Utrecht (NL). The members of the Patient Thinktank are asked to participate in annual stakeholder workshops, held in parallel to annual meetings, and will also be consulted on an ad-hoc basis when issues arise in the project that require input from the perspective of patients.

## PROJECT INSPIRE

*Project InSPiRe (Innovative methodology for small populations research – FP7) focuses on the development of novel methods for the design and analysis of clinical trials in rare diseases or small populations defined, for example, by a rare genetic marker. Tsveta Schyns is a member of the Independent Scientific Advisory Committee of the project, which oversees its scientific direction and progress of the project. This comprises international clinical and non-clinical experts and patient group representatives.*

## E-RARE

*EURORDIS is also a member of the External Advisory Board of E-Rare, the Era-Net project that has been established to develop synergies between eight public national research programmes on rare diseases in the European Union and to set up a coordinated research policy on rare diseases in Europe.*

The E-Rare-2 project aims to deepen and extend cooperation among partners involved in E-Rare 1 and four new partners by: systematic exchange of information; yearly launching of joint calls; thorough assessment of the funding mechanisms and results of the funded research projects; and carrying out strategic activities aiming at a sustainable development and extension of the network. E-Rare-2 activities will thus further contribute to reducing fragmentation of research and resources through the enhanced coordination and transnational funding of excellent research on rare diseases, thereby shaping the European Research Area for rare diseases.



EURORDIS participated in the following E-Rare meeting in 2014: E-Rare 'Scientific Meeting & Strategic Workshop' that took place in January 2014 in Athens; the E-Rare Symposium in Rome in October 2014 on the involvement of patient organisations in research funding; and the ERA-Net Info Day in Bologna in December, during which information on current and future E-Rare consortia was provided.

The third E-Rare project was officially launched in December 2014 and has the same objectives as the previous E-Rare-1 and E-Rare-2 projects. Patient organisations from Europe (represented by EURORDIS) and beyond will be invited to contribute to collaborative efforts for research promotion and funding. The participation of EURORDIS in E-Rare-3 creates an opportunity for patient organisations to foster their engagement in funding of research on rare disease at the transnational level.

# Promoting the Development of Effective Rare Disease Research Infrastructures

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Rare disease research infrastructures such as patient registries, genomic databases and biobanks are key instruments for advancing clinical research in the field of rare diseases. EURORDIS intensified its direct role in advancing research in the field of rare diseases by maintaining strong ties with several key research infrastructures in the field specifically, promoting the development of sustainable, harmonised and integrated registry, biobanking and genetic data infrastructures and networks that reflect the patient's best interest.

This was achieved by supporting the involvement of patients and their representatives at both the governance

and clinical research levels of several international projects and consortia.

## TREAT-NMD

*As a founding member of TREAT-NMD (Translational Research in Europe – Assessment and Treatment of Neuromuscular Diseases), EURORDIS continued its service to the newly governed “TREAT-NMD Alliance” as a member of the Executive Management Committee Composed of academic and patient representatives, the Executive Management Committee governs the overall network.*

## Biobanks

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*Patient organisations are aware of the need for more rapid progress in rare disease research and of the difficulties that researchers and clinicians face in obtaining human biological material to conduct research on rare diseases, and EURORDIS has represented this patient-driven view.*

As the former coordinator of **EuroBiobank**, EURORDIS continued its strong commitment in promoting and actively participating in the governance of the first

network of biobanks dedicated to rare diseases. EURORDIS continued to raise the importance of supporting biobanks for rare disease research to European and national level policy makers, liaising with the **Biobanking and Biomolecular Resources Research Infrastructure (BBMRI) Consortium**, most recently joining the BBMRI Rare Disease Working Group.

## Genomics Data

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*Funded by the EU Seventh Research Framework Programme, RD-Connect will develop a global infrastructure for sharing outputs of research projects, starting with the EUREnOmics project focusing on the causes, diagnostics, biomarkers, and disease models for rare kidney disorders, and the Neuromics project that will use next generation whole exome sequencing to increase genetic knowledge of rare neurodegenerative and neuromuscular disorders. The RD-Connect project supports the overarching IRDiRC 2020 goals.*

EURORDIS joined as one of 27 full partners in **RD-Connect**, an integrated platform connecting databases, registries, biobanks, and clinical bioinformatics for rare disease research. EURORDIS is active in many facets of the project and thus has many opportunities to reflect the voice of patients throughout the various activities of the project. To structure the input of patient views in advising the governing board of the project, EURORDIS has established and continues to chair a 16-member Patient Advisory Council.

EURORDIS encouraged opportunities for patient involvement throughout each of the RD-Connect project work packages including the preparation and delivery of educational materials and the consultation of RD patients on issues related to the research on “-omics” and the linkage of patient data across different infrastructures and across borders. Finally, EURORDIS identified opportunities for interaction and coordination of the RD-Connect network with other initiatives within and beyond Europe and the dissemination of the RD-Connect project outcomes at the international level to be carried out from this point forward.





# A Platform for Sharing Best Practices for the Management of Rare Diseases

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In January 2013, EURORDIS joined, as one of 15 partners, the RARE-Bestpractices project, a 4 year (2013–2016) project funded by DG Research under FP7-HEALTH-2012 INNOVATION-1. This project will develop a sustainable platform for sharing best practices for the management of rare diseases, supporting the collection of standardised and validated data and the efficient exchange of knowledge and reliable information on rare diseases. The expected impact of the project is an improvement of health outcomes and quality of life for rare disease patients, through the reduction of inequality in rare disease care and an access to better diagnostics and therapeutics at EU level. RARE-Bestpractices is aimed at collecting, evaluating and disseminating best practices and agreeing on a methodology suitable to develop and update best practice guidelines. The project will also create a forum for exchanging information, sharing lessons learnt and facilitating collaborations.



EURORDIS acts as a transversal partner in the different work packages, ensuring the involvement and capacity-building of patient organisations across Europe regarding the project. EURORDIS is especially involved in the work packages that focus on dissemination, on scientific

coordination and networking with relevant activities in the field of rare diseases and on defining a common methodology for the production of guidelines on clinical management of rare diseases.

In the context of the RARE-BestPractices project, a workshop was organised during the EURORDIS membership meeting in Berlin in May 2014. Juliette Senecat of EURORDIS highlighted the importance of healthcare guidelines with a focus on rare diseases, as well as the role of EURORDIS in the project. Following a session on patients as guideline developers/ users presented by the coordinators of the project (Cristina Morciano and Paola Laricchiuta from the Istituto Superiore di Sanità, Italy), volunteers Avril Kennan, Debra Ireland and Eric Low, Myeloma Patients Europe illustrated the concepts with two case studies on Epidermolysis bullosa Myeloma. This helped the group to identify obstacles and ways to involve patients in healthcare guidelines development. In October 2014, Avril Kennan and Mathieu Boudes represented EURORDIS at a workshop in Edinburgh aiming to assess the use of a standardised tool to appraise existing clinical practice guidelines for the management of rare diseases.

## Promoting Patients' Involvement and Rare Diseases Health Technology Assessment (HTA)

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*In parallel to the marketing authorisation process, patients are now consulted when HTA bodies assess new health technologies (medicines, and also medical devices, connected devices, complex surgery, diagnostic tests etc.), at least at the European level. Since September 2014, patients have been invited to participate in early dialogues between HTA experts and industry (through the SEED<sup>3</sup> project), and as experts in the review of joint assessments led by EUnetHTA<sup>4</sup>.*

EURORDIS continued its activities on Health Technology Assessment, focusing on acquiring a knowledge base, training rare disease patient representatives, gathering their experience in HTA procedures and promoting the involvement of patients in HTA procedures across Europe.

### TECHNICAL AND SCIENTIFIC ACTIVITIES: THE EUROPEAN NETWORK FOR HEALTH TECHNOLOGY ASSESSMENT (EUNETHTA)

EURORDIS is one of four patient representative organisations at the **EUnetHTA Stakeholders Forum**. EUnetHTA is an EU initiative to improve coordination and harmonise the assessment tools used by the main HTA agencies in Europe. It is funded by the European Commission and Member States. EUnetHTA is a joint action between the European Commission and Member States; it is now in the second joint action (JA2).

The current main objectives of EUnetHTA are to promote the actual use of methods and guidelines by HTA agencies at the national level and see which barriers may remain to better harmonising the assessment of the value of medicines across the EU, in order to decrease the disparity that exists regarding reimbursement of medicines in Europe.



<sup>3</sup> Shaping European Early Dialogues (<http://www.earlydialogues.eu/has/>)

<sup>4</sup> European Network of HTA Agencies (<http://www.eunetha.eu/>)

EUnetHTA gathers 62 HTA agencies from the EU and beyond and progress is presented regularly to the Stakeholders Forum. Members of the Stakeholders Forum represent not only patients and consumers, but also the pharmaceutical industry, payers (e.g. private insurance companies) and healthcare providers (e.g. hospitals).

EURORDIS's Director of Treatment Information and Access, François Houyez, is the current co-chair of the Forum. Visit [Eunethhta.eu](http://Eunethhta.eu) to learn more.

### EARLY DIALOGUES BETWEEN DEVELOPERS OF NEW HEALTH TECHNOLOGIES AND HTA EXPERTS: THE SEED PROJECT<sup>5</sup>

The French HTA agency, Haute Autorité de Santé, is coordinating the SEED project that involves thirteen other

HTA bodies from eleven Member States. The SEED project will end mid-2015. It offers an opportunity for industry to meet with HTA experts at an early stage to discuss their development plans.

The objective of an early dialogue is "to reduce the risk of inadequate data when products are presented for evaluation in aim of reimbursement by national health insurance".

Twelve early dialogue meetings will have been conducted when the project ends, seven of which focus on rare diseases. Each meeting is a pilot from which experts and companies draw lessons to improve the next ones. The EMA sometimes participate through EMA-HTA parallel scientific advice.

Since September 2014, two patients can be invited to each procedure. EURORDIS is coordinating the identification and the involvement of patients in the SEED project.

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### POLITICAL AND STRATEGIC ACTIVITIES: THE EUROPEAN HTA NETWORK AT THE EUROPEAN COMMISSION REFLECTION ON LONG-TERM HTA COLLABORATION IN EUROPE

The HTA Network, composed of Member States representatives and the European Commission, adopted a long term strategy for HTA collaboration in Europe. This paper explains how long term collaboration could work, beyond current initiatives such as EUnetHTA.

EURORDIS, and its Drug Information Transparency and Access taskforce in particular, noted that 25 years has already passed since the first European projects started a debate on how to cooperate in HTA. It also acknowledged the necessity to foster the generation and dissemination of evidence-based practices in assessing the effectiveness and cost-effectiveness of medical technologies and organisation of care, as well as to

improve the transparency of reimbursement decisions so that they are understood by European citizens.

The patient community sometimes lacks the resources it needs to be fully equipped to play the desired role in HTA. EURORDIS calls on the European Commission to systematically conduct an impact assessment on the consequences of involving civil society, here patients and part of the public, when developing new legislation.

EURORDIS stated clearly that the EMA would be the most logical choice to host European HTA cooperation in the long-term. For citizens, it would be much more logical to have one single agency responsible for coordinating all aspects of medicines and other health technologies evaluation; the current situation, under which the EMA evaluates the benefit/risks of medicines but has no mandate to discuss price or reimbursement, is not understood by the public and therefore not acceptable.

### SUPPORT TO PATIENT REPRESENTATIVES AT THE EUROPEAN MEDICINES AGENCY

*EURORDIS is in the unique position of having patient representation in the following European Medicines Agency (EMA) Committees and Working Parties: the Committee for Orphan Medical Products (COMP); the Paediatric Committee (PDCO); the Committee for Advanced Therapies (CAT); and the Patients' and Consumers' Working Party (PCWP).*

Dedicated patient representative volunteers contributed to the examination of a total of 456 dossiers in 2014 through the work of the scientific committees they belong to. These include: 259 dossiers for orphan drug designations, 28 classification or certifications by the CAT and 172 paediatric investigation plans by the PDCO.

The Therapeutic Action Group (TAG), composed of the EURORDIS representatives in the above-mentioned scientific committees and working partly at the EMA, continued their work and maintained communication internally with monthly conference calls and emails. They also had their annual meeting with the EURORDIS Board of Directors to exchange views on future actions.

In addition to these permanent activities at the EMA, patient representatives are also invited on a sporadic basis to attend the scientific committees and the Scientific Advice Working Party (SAWP) as experts for their disease.

In 2014, 12 patient representatives attended meetings of



the SAWP for protocol assistance. Protocol assistance is a version of scientific advice specific to orphan medicinal products and is a way for the company developing the medicine to obtain scientific and regulatory advice on the manufacture of a medicine, as well as on pre-clinical and clinical tests being performed. Patients are involved in order to provide first-hand input on the most relevant outcome measures and endpoints for clinical trials. The process of scientific advice/protocol assistance is recommended to avoid major objections (regarding the design of clinical trials) during evaluation of the marketing authorisation application.

The inclusion of patient representatives at the EMA either as permanent members of the committees and working party or sporadically is considered an important contribution by the Agency.

The Patients' and Consumers' Working Party, of which

## THE DRUG INFORMATION, TRANSPARENCY AND ACCESS TASK FORCE (DITA)

*The Drug Information, Transparency and Access Task Force (DITA) represents a group of volunteers who are trained (via the EURORDIS Summer School) and active in issues concerning therapeutic development of medicines for rare diseases. The Task Force supports and/or advises the EURORDIS representatives who participate in EMA Scientific Committees and Working Parties, or in the European Network of Health Technology Assessment (EUnetHTA) and the HTA Network (DG Sante). It is consulted on papers prepared by EURORDIS.*

One DITA Task Force meeting was held in Warsaw in April 2014 (linked to the EUPATI Workshop).

DITA members exchanged information and elaborated contributions on among other issues:

- Health Technology Assessment contributing to the HTA Network Long Term Strategy paper and providing comments on the EUnetHTA recommendations on the implementation of a sustainable European network for HTA
- The EURORDIS Access Campaign which is an online survey on access difficulties for care/medicines used in rare diseases in the context of the economic crisis. The survey was conducted from May to September 2014 and collected 1,846 responses from 31 European countries.
- Questionnaire on off-label use of medicine. A new research project has been proposed to explore how patients are actually taking their medicines, how they understand the black symbol for additional monitoring which is printed on some package leaflets and how they see the use of off-label products. The questionnaire was adapted from a pilot version used in 2012. The research will be submitted to an Institutional Review Board (INSERM) before starting the field work.
- Participation in EMA meetings with DITA task force members such as the 8th Stakeholder Meeting on the implementation of the new Pharmacovigilance

EURORDIS is a member, is a unique forum where all scientific committees of the Agency meet with patients and consumers. It held four meetings in 2014 and organises the involvement of patients and consumers in all of the EMA activities (in 2013 a total of 551 patients/consumers were involved in one of the EMA activities). On the evaluation of benefit-risk for a marketing authorisation, even though patients are not yet members of the Committee for Human Medicinal Products (CHMP) at the EMA, the agency has put in place processes for the CHMP to consult with patients and their organisations when needed.

Applications were submitted for the extension of the mandate for the PDCO for Tsveta Schyns as member and Kerry Leeson-Beevers (Alstrom Syndrome association UK) as alternate. Both applications were successful and their mandate started on 1st August 2014.

legislation; 4 meetings of the Patients and Consumers' Working Party; EMA workshop on a mobile app for adverse drug reactions (Web-RADR); Committee for Advanced Therapies (CAT) – Interested Parties meeting – 11/12 (2 members)

- Other conferences such as the European Summit on Hospital Pharmacy organised by the EAHP (14–15 May, Brussels) and the Eurometing of the International Society of Pharmaco-economics and Outcomes Research 10–12/11, Amsterdam





# New European Projects in the Field of Pharmacovigilance

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## WEB-RADR

*In 2014, the IMI-Web-RADR project was launched. Its main objectives are to create data mining tools to analyse patients' discussion in relation to side effects on social media and to develop a mobile application to facilitate the spontaneous reporting of suspected adverse drug reactions.*

EURORDIS is a member of the WEB-RADR consortium and will help interview patients regarding their needs, but also barriers and obstacles vis-à-vis adverse drug reactions reporting tools. EURORDIS will also take part in the guidelines development, together with regulators, data protection supervisors, industry, patients and clinicians.

More on Web-RADR: [Web-radr.eu](http://Web-radr.eu)

## SCOPE

*In parallel to the launch of Web-RADR, EURORDIS continued to advise the SCOPE joint action (Strengthening Collaboration for Operating Pharmacovigilance in Europe).*

26 Member States work together to explore how they can best play their roles in pharmacovigilance: informing the public on risks, collecting spontaneous reports from patients and healthcare professionals, encouraging patients to report side effects more often, detecting and analysing new pharmacovigilance signal and auditing their systems.

More on SCOPE: [Scopejointaction.eu](http://Scopejointaction.eu)

# EURORDIS Capacity Building & Training for Patient Advocates

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*In 2014, EURORDIS continued its capacity-building & training activities for patient advocates by: holding the highly successful EURORDIS Summer School for the 7th consecutive year; expanding its online e-learning tools; and continuing involvement in the DIA Patient Fellowship Programme. In 2014, collaboration continued with two training projects: the European Patients' Academy on Therapeutic Innovation (EUPATI) and the European Clinical Research Infrastructure Network – Integrating Activity (ECRIN-IA).*

## EURORDIS SUMMER SCHOOL

*The EURORDIS Summer School was initiated in 2008 as part of our continued commitment to empowering people living with rare diseases. This four-day course provides*

*training in aspects of medicines development and EU regulatory processes where patient representatives can be involved.*

The 7th EURORDIS Summer School session was held in Barcelona in June 2014 and once again aimed at training patient representatives in clinical trials, medicines development and EU regulatory processes. As in previous years, this four-day training course gathered together a dynamic group, 37 participants representing 16 countries and 26 different diseases.

The 2014 Summer School provided participants with a fundamental understanding of the processes involved in drug development, the time required and the different stages of clinical trials. Coupled with formal presentations of the topics, the participants were divided into small groups and encouraged to share their experiences and



knowledge in the context of documents provided to complement the lectures.

In addition, practical 'hands-on' examples of the work performed by the representatives at the EMA Scientific Committees and working parties was presented using orphan drug designation dossiers from the Committee for Orphan Medicinal Products and an example of a waiver request from the Paediatric Committee.

For the third year in a row, non-rare disease patients were included as part of EURORDIS' collaboration with the European Clinical Research Infrastructure Network – Integrating Activity (ECRIN-IA), which develops e-services and education materials to train professionals and patient associations to communicate with users, patients, citizens and policymakers (WP3). It supports the structuring and connection to ECRIN of disease-, technology-, or product-oriented investigation networks and hubs focusing on specific areas: rare diseases (WP4), medical devices (WP5) and nutrition (WP6). Through this collaboration, nine participants representing nutrition and medical devices were also included in the EURORDIS Summer School. This will be continued for 3 more years in the context of this project.

## ONLINE LEARNING

*Based on the experience gained from the EURORDIS Summer School, a comprehensive e-learning tool covering topics such as clinical trial design, statistics and ethics has been designed and is freely available on the EURORDIS website. In addition, all presentations have been recorded, downloadable pdf versions of which are all available in the training section of the Eurordis.org.*

All of the above experiences will be further developed to include additional topics and to target different disease

groups through the European Patients' Academy on Therapeutic Innovation (EUPATI), a 5-year project funded by the Innovative Medicines Initiative (IMI). This patient-led academy will provide scientifically reliable, objective, comprehensive information to patients on pharmaceutical research and development.

EUPATI comprises a unique combination of pan-European patient, academic and not-for profit organisations, as well as EFPIA member companies. EURORDIS is involved in the development of content, in particular for the areas of drug safety and risk/benefit assessment of (novel and existing) medicines, design and objectives of clinical trials and in the area of deployment, where two face-to-face training sessions in 2015 and 2016 will be organised.

## DRUG INFORMATION ASSOCIATION (DIA) PATIENT FELLOWSHIP PROGRAMME

*The annual DIA Eurometing attracts over 3,000 representatives of the pharmaceutical industry, academia and public health authorities. Patient fellows are able to learn about regulatory affairs, clinical trials, latest drug developments and innovative therapies, and meet with many different stakeholders. Since 2008 patient fellows have had a dedicated booth supported by EURORDIS in the exhibition hall of the Eurometing, which has helped increase their visibility.*

At the annual DIA EuroMeeting 2014 in Vienna, 22 fellowships were granted to highly qualified patients' representatives. In addition, 14 speakers from patient organisations were invited to share their expertise in various sessions.

Since 2006, over 200 patient representatives across diseases and European countries received a fellowship; about half of them are members of EURORDIS.





## EURORDIS Round Table of Companies (ERTC) – 2014 Workshops

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*Initiated in 2004, the EURORDIS Round Table of Companies (ERTC) is a 'club' of pharmaceutical companies with a common interest in rare diseases and orphan drug development.*

In 2014, the ERTC comprised:

- 45 members
- 6 new members: Boehringer Ingelheim International GmbH, Cydan Development, Inc., Insmid Incorporated, NPS Pharma, Therakos, Inc., Vanda Pharmaceuticals, Inc.

As is the case every year, two workshops were held in 2014:

**'Unlocking Europe's Potential in Rare Disease Therapies' A 10 Year Anniversary Workshop, 26 February 2014, Brussels: 84 attendees from 12 countries**

The 20th ERTC workshop marked the 10th anniversary of the creation of the Round Table of Companies workshops. This workshop explored the ambitious overarching theme of 'Unlocking Europe's Potential in Rare Disease Therapies' between now and 2020, celebrating ten years of dialogue and collaborative efforts between patient advocates, industry, regulatory agencies and national competent authorities to move forward the development of treatments for rare diseases. Over the past ten years, the twice yearly round tables have regularly tackled the key issues that can unblock bottlenecks and advance real solutions for more, safe, efficient, effective, affordable and accessible therapy development.

**'Sharing Rare Disease Patient Data: Translating Principles into Action' 30 September 2014, Barcelona: 79 attendees from 12 countries**

The 21st ERTC workshop was the occasion to review and discuss issues around collecting, registering and sharing patient data in the drug development process upstream in order to foster the development of treatments for rare diseases downstream. The need to harmonise and share patient data in order for researchers to make the best use of results was highlighted. Patients expressed both willingness and caution regarding data sharing, particularly genetic data which heightens benefits of research but also the risks. They desire active representation in data collection programmes and need to trust the data-sharing network. Finally, this ERTC workshop explored the feasibility of establishing public-private partnerships in patient data collection. It was highlighted how researchers, companies and patient groups have already progressed well in building consensus around guiding and aspiring principles for collecting and sharing patient data to maximise opportunities for improved diagnosis and treatment development.



# GOVERNANCE, ORGANISATION & FUNDING

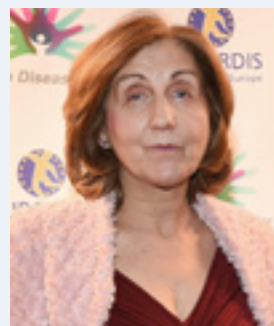
## Board of Directors

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During the General Assembly held in Berlin on 8 May 2014, EURORDIS full members voted on four vacant positions in the Board of Directors, re-electing the Greek Alliance for Rare Diseases PESPA, Greece, represented by Dimitrios Synodinos; Rare Diseases Denmark (Sjældne Diagnoser), Denmark, represented by Birthe Byskov Holm; the Romanian Prader Willi Association (RPWA), Romania, represented by Dorica Dan; and the Federación Española De Enfermedades Raras, Spain, with the new representative Gema Chicano Saura.

The Board of Officers, elected annually by the Board of Directors following the General Assembly, was voted as follows: President: Terkel Andersen, Denmark; Vice President: Avril Daly, Ireland; General Secretary: John Dart, UK; Treasurer: Dimitrios Synodinos, Greece; and Officer: Dorica Dan, Romania.

The Board of Directors extended a heartfelt thank you to outgoing Board member, Rosa Sanchez de Vega (FEDER) for her longstanding dedication to EURORDIS and the rare disease cause.



## Staff Organisation

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The team comprised 35 people, 28 full-time equivalent (FTE) as of December 2014. The team is composed of paid staff, one office volunteer, one consultant and trainees. Most staff members are based in the Paris office located in the Rare Disease Platform. EURORDIS' European Public Affairs Director and European Public Affairs Advisor are based in the Brussels office. The EURORDIS RareConnect team managing the online patient communities network is based in Barcelona. The Chief Executive Officer and the Research and Healthcare Director share their time between the Paris and Brussels offices.

The following are the main changes in human resources in 2014 :

- Paloma Tejada, Rare Diseases International Senior Manager, Geneva, has returned from parental leave
- Lenja Wiehe, Fundraising Manager, has joined the Paris Office
- Matt Johnson, Research and Healthcare Director, has joined the Paris Office
- Virginie Hivert, Therapeutic Development Director, has joined the Paris Office
- Maria Mavris, Therapeutic Development Director, has been seconded to EMA

- Louise Taylor, Communications and Development Writer, has left EURORDIS
- Eva Bearryman, Junior Communications Manager, has joined the Paris Office
- Juliette Sénecat, Health Research Projects Manager, has temporary left for a maternity / parental leave



# Volunteers

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Most EURORDIS volunteers are either parents of patients affected with a rare disease or patients themselves. Due to the rarity of their disease and lack of available information, they have consequently become experts of their disease and of their respective national health care system. Other volunteers, indirectly affected by rare diseases, have also become very knowledgeable on rare disease related issues and are all very committed to the cause.

These volunteers are called the EURORDIS volunteer patient advocates.

They are driven by the urgent need to improve access to adequate diagnosis and care for rare disease patients, as well as access to adapted social services. Raising awareness is their credo.

The specific role of volunteer patient advocates consists in sharing their knowledge and experience to advocate for better national and European public health policy measures in favour of rare diseases. They participate in committees, working groups, Task Forces (DITA and RD Connect), and speak at international conferences.



## OUR REPRESENTATIVES ON THE EMA SCIENTIFIC COMMITTEES

- EMA Committee for Orphan Medicinal Products (COMP):
  - Ms Birthe Byskov Holm, Danish Osteogenesis Imperfecta Society and Rare Disorders, Denmark, full member
  - Ms Lesley Greene, EURORDIS, UK, Vice-Chair of the COMP
  - Dr Maria Mavris/ Dr Virginie Hivert, EURORDIS Therapeutic Development Director, Permanent Observer
  - Mr Julian Isla, Dravet Syndrome Foundation, Spain, Observer
- EMA Paediatric Committee (PDCO):
  - Dr Tsvetana Liharska-Schyns, European Network for Research on Alternating Hemiplegia Association, Belgium, full member
  - Ms Kerry Leeson-Beevers, Alstrom Syndrome UK; United Kingdom, alternate
- EMA Committee for Advanced Therapies (CAT):
  - Dr Michele Lipucci di Paola, Associazione Veneta Lotta Talassemia, Italy, full member
  - Mr Chris Sotirellis, UK Thalassaemia Society, Observer
- EMA Patients' and Consumers' Working Party (PCWP):
  - Mr Richard West, Behçets Syndrome Society, UK, full member
  - Mr François Houÿez, Information & Access to Therapies Director, full member

The EURORDIS patients' representatives and staff on EMA scientific committees form the Therapeutic Action Group (TAG). The TAG holds monthly conference calls to update on the work of each EMA committee and take actions where necessary.

## OUR REPRESENTATIVES ON THE COMMISSION EXPERT GROUP ON RARE DISEASES

The 8 seats (four full members and four alternates) for patients are held by EURORDIS members, mostly volunteer patient advocates, selected based on their expertise, advocacy track records, representativeness of large disease groups and geographic outreach. The four full members (and their alternates) represent each the following organisations: EURORDIS, the European Network of National Alliances for Rare Diseases, the Network of European Federations of Rare Diseases and EGAN (the Patients Network for Medical Research and Health). Yann Le Cam was the only staff member; other representatives were volunteers:

- Ms Dorica Dan, Chair of RONARD, the Romanian National Alliance for Rare Diseases – European Network of National Alliances for Rare Diseases, full member
- Mr Jan Geissler, Vice President of the Leukemia Patient Advocates Foundation – Network of European Federations of Rare Diseases, full member
- Mr Alastair Kent, Director of the Genetic Alliance UK and Chair of Rare Disease UK & EGAN, full member
- Mr Yann Le Cam, Chief Executive Officer of EURORDIS, full member
- Ms Amanda Bok, Chief Executive Officer of the European Haemophilia Consortium – Network of European Federations of Rare Diseases, alternate

■ Ms Lene Jensen, Chief Executive Officer of Rare Disorders Denmark – European Network of National Alliances for Rare Diseases, alternate

■ Mr Flavio Minelli, co-founder and Board member of the Italian Ichthyosis Union (UNITI), member of UNIAMO, the Italian National Alliance for Rare diseases & EGAN, alternate

■ Mr Christopher Natchtigäller, President of ACHSE, the German Alliance for Rare Diseases and EURORDIS, alternate

Flaminia Macchia, European Public Affairs Director, and Ariane Weinman, European Public Affairs Manager, were both observers on the Expert Group and supported the group of volunteers.

Altogether, they form the EURORDIS Policy Action Group (PAG).

## OUR REPRESENTATIVES ON THE COMMISSION EXPERT GROUP ON CANCER CONTROL

Two volunteer representatives of EURORDIS have been

nominated to represent patients affected by rare cancers in the Commission Expert Group on Cancer Control. One of them also sits on the European Commission Expert Group on Rare Diseases. EURORDIS considers that it is important to create a bridge between the Commission Expert Group on Rare Diseases and the Commission Expert Group on Cancer Control as several health policy issues related to rare cancers are common to these two groups: national rare disease / cancer plans, registries, European Reference Networks, quality criteria for Centres of Excellence, research policy, creating evidence and data in small populations, research policy and regulation, genetic testing and counselling, etc.

Mr Jan Geissler, EURORDIS / Vice President of the Leukemia Patient Advocates Foundation, full member

Ms Kathy Oliver, EURORDIS / Chair and Co-Director of the International Brain Tumour Alliance (IBTA), alternate

The work of the two EURORDIS volunteer representatives is supported by the EURORDIS Policy Action Group on Rare Cancers (PAG-RC) composed of other four patient representatives from different rare cancer organisations and one staff.

# Finance and Support Services

Finance and support services' activities in 2014 included:

- Accounting and monthly financial reporting in a timely manner including cash flow and risk analysis detailed report.
- Monthly meetings with managers to update the budget and the year-end financial forecast.
- Operating Grant Steering Committees organised every third month to review progress on implementation, deliverables and budget.

- Management of human resources activities, such as recruitment.
- Management of office support: IT infrastructure, contact database, office supplies.
- Management of legal and fiscal matters related to contractual matters.

# Contract Grants

## NEW

ECRD2014, European Conference on Rare Diseases & Orphan Products 2014, DG Health and Consumers, 8 months, 2014.

Web-Radr, Recognising Adverse Drug Reactions, IMI-JU, 36 months, 2014–2017.

Genetic Clinic of the Future, a stepping stone approach towards the Genetics Clinic of the Future, DG Research, 30 months, 2014–2017.

## RENEWED

Advocacy and core activities, AFM-Téléthon, 48 months 2014–2017.

Operating Grant for year 2014 ("EURORDIS\_FY2014"), single beneficiary, DG Health and Consumers, 12 months.

eNews and Website in Russian, Association of International Pharmaceuticals Manufacturers (AIPM), 12 months.

## ONGOING

EJA, EUCERD Joint Action, DG Sanco, 42 months, 2012–15.

EUPATI European Patients' Academy on Therapeutic Innovation, Innovative Medicines Initiative – a DG Research / EFPIA Joint Undertaking, 60 months, 2012–16.

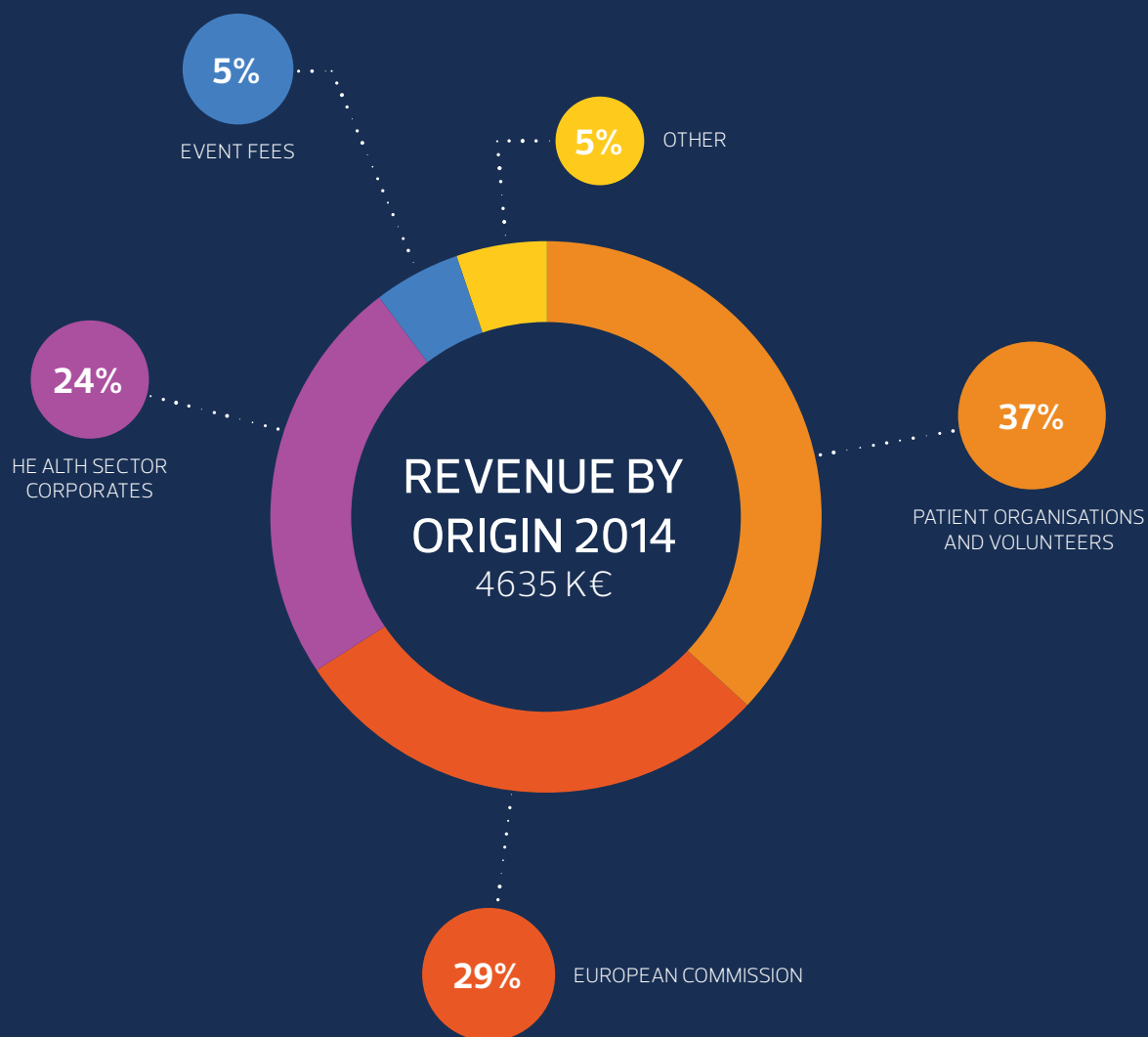
ECRIN-IA European Clinical Research Infrastructures Network – Integrating Activity, DG Research, 48 months, 2012–15.

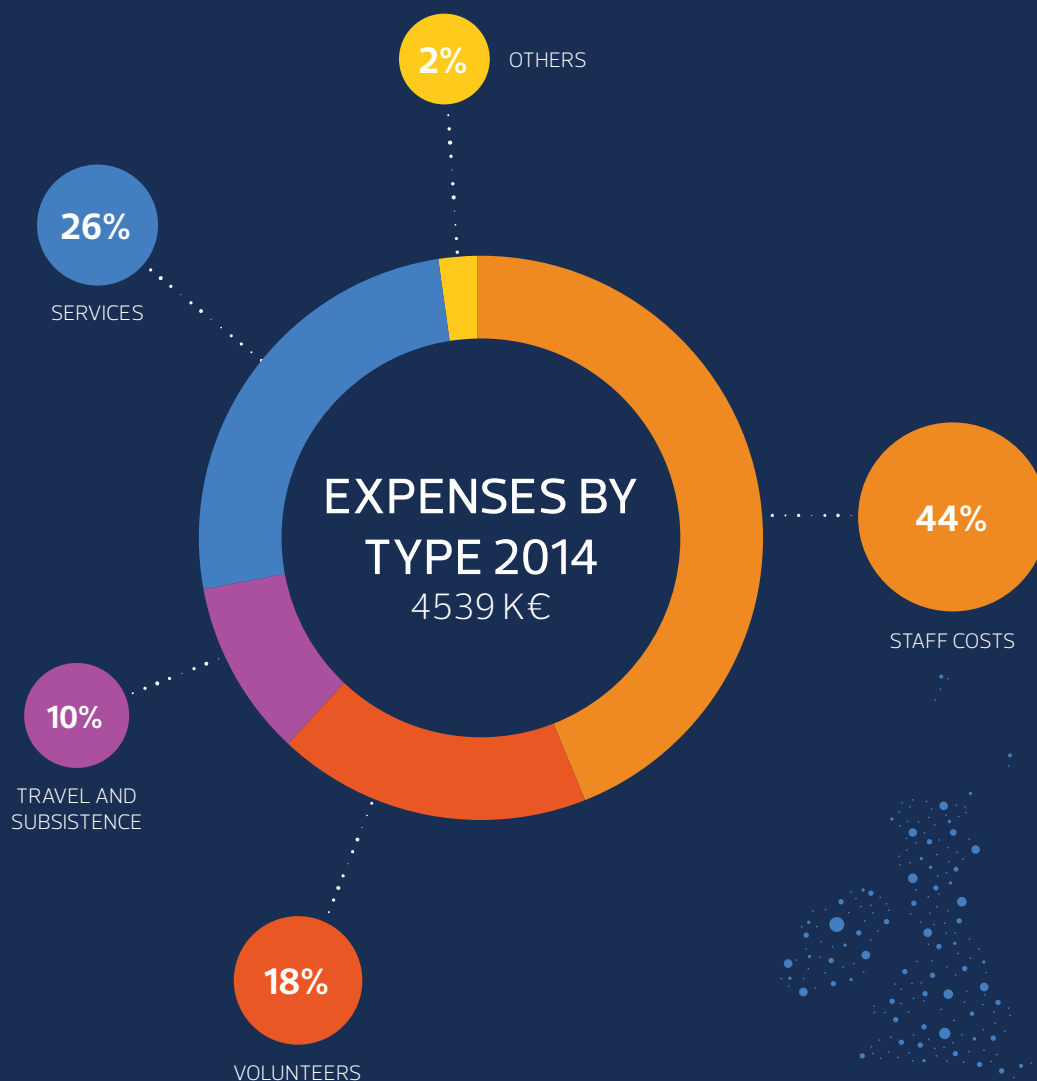
RD-Connect, an integrated platform connecting registries, biobanks and clinical bioinformatics for rare disease research, associated partner, DG Research, 72 months 2012–18.

RARE-Bestpractices, a platform for sharing best practices for the management of rare diseases, DG Health and Consumers, 48 months, 2013–2016.



# REVENUE & EXPENSES 2014







# BOARD OF DIRECTORS MAY 2014 – MAY 2015

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<b>Ms Vlasta Zmazek</b>	Croatian Alliance for Rare Diseases	Croatia





# CONFERENCES & WORKSHOPS 2014

## | EUCERD Joint Action workshop: "Cross-Border Genetic Testing for Rare Diseases", Newcastle, UK, 15–16 December

Kerry Leeson–Beevers, Birthe Holm and Dorica Dan represented EURORDIS

## | ERA–NET Info Day: "Opportunities and Strategies for Transnational Research Calls", Bologna, Italy, 15 December

Mathieu Boudes: Presentation of EURORDIS with an emphasis on research, patients' involvement in drug development and our participation in EMA committees

## | "Development Pathways for Advanced Therapy Medicinal Products" – organised by EBE (European Biopharmaceuticals Enterprises), EMA and the Scientific office of the Italian Embassy in the UK, London, UK, 15 December

Michele Lipucci di Paola represented EURORDIS

## | INFRAFRONTIER, Marseille, France, 9–10 December

Mathieu Boudes: "How Patient Groups are Involved in Research"

## | "Evidence Generation and Successful Knowledge Transfer in Public Health – Integrated care as a vehicle for innovation in health care delivery", organised by the European Commission, Glasgow, UK, 19 November

Matt Johnson: "Patient's Perspective on Added Value of Integrated Care for Rare Disease European Reference Networks"

## | Cilia 2014 Conference, Institut Pasteur, Paris, France, 18 November 2014

Mathieu Boudes: "EURORDIS, the voice of Rare Diseases patients in Europe"

## | ISPOR– International Society for Pharmacoeconomics and Outcomes Research – 17th Annual European Congress, Amsterdam, The Netherlands, 8–12 November 2014

François Houjiez: Panelist in the session "Patient–Reported Outcomes and Patient Preference Research issues"

Rob Camp and Russel Wheeler also represented EURORDIS

## | 2014 World Orphan Drug Congress in Europe, Brussels, Belgium, 12–14 November

Yann Le Cam: "Rethinking the Models for Orphan Drug Development, Access and Pricing"

## | IRDiRC 2nd International Conference, Shenzhen, China, 6–9 November

Yann Le Cam: "Therapies for rare diseases: achievements and challenges" – "Why adaptive pathways are necessary to bring new drugs to patients?"

Flaminia Macchia, also represented EURORDIS

## | II Ibero–American conference of Rare Diseases Patients' Organisations (Rarissimas, FEDER, ALIBER) Lisbon, Portugal, 4–7 November

Gema Chicano: Presentation of EURORDIS

**Rare Diseases Conference under Italian EU Presidency, Rome, Italy, 31 October**

**Yann Le Cam:** Presentation of EURORDIS and its Achievements

**Valentina Bottarelli,** Flaminia Macchia also represented EURORDIS

**HTA 2.0. Conference, Rome, Italy, 30–31 October**

**François Houÿez,** moderator of the panel "Getting an effective technology from the lab to the patient in Europe –challenges and opportunities"

EURORDIS' representatives: **Vesna Aleksovska, Claudie Baleyrier, Rob Camp, Yann Le Cam, Luc Matthysen, Danijela Szili, Tatiana Foltanova, Ellen Van Veldhuizen, Richard West, Russel Wheeler, Vlasta Zmazek**

**E-Rare Symposium: "Fostering Collaboration in Rare Diseases Research", Rome, Italy, 30 October**

**Yann Le Cam:** "Involvement of Patients' Organisations in Research Funding"

**Mathieu Boudes** also represented EURORDIS

**Informal Joint EMA CHMP – COMP – CAT Meeting, Rome, Italy, 28–30 October**

**Lesley Greene, Virginie Hivert, Birthe Holm, Michele Lipucci di Paola** represented the patients on EMA COMP and CAT

**Workshop on "European Reference Networks and Structural Funds within the European Joint Action on Rare Diseases", Rome, Italy, 28–29 October**

EURORDIS' representatives: **Yann Le Cam, Valentina Bottarelli, Matt Johnson, Flaminia Macchia and Ariane Weinman**

**"NORD's Rare Diseases and Orphan Products Breakthrough Summit", Alexandria, Virginia, USA, 21–22 October**

**Zoe Alahouzu** represented EURORDIS at the NORD Regional Summit

**"Better Science, Better Health: New Trial Pathways and Better Patient Data", Royal College of Physicians, London, UK, 21 October**

**Yann Le Cam** panellist in the session: "The Patient's Understanding of Benefit Risk"

**Flaminia Macchia** also represented EURORDIS

**3rd Angelman Syndrome International meeting, Paris, France, 17 October**

**Lara Chappell and Ariane Weinman** represented EURORDIS

**Rare–Best Practices project: Workshop organised by Health Improvement Scotland, Edinburgh, UK, 16 – 17 October**

**Mathieu Boudes** represented EURORDIS

**Information Meeting for Members of the French National Alliance for Rare Diseases, Paris, France, 16 October**

**Sharon Ashton:** "European Year for Rare Diseases – 2019"

**ICORD 2014, Ede, the Netherlands, 8–10 October**

**Yann Le Cam:** "EU Major Policies from the patients' perspective & International engagements"

**"The importance of using the European Structural and Investments Funds to drive sustainable healthcare systems" organised in the framework of EU Open Days, Brussels, Belgium, 7 October**

**Valentina Bottarelli** represented EURORDIS

**European Patients' Forum (EPF) Fourth Regional Cross Border Healthcare Conference (for Patient organisations' representatives from Denmark, Estonia, Finland, Latvia, Lithuania and Sweden), Tallinn, Estonia, 6–8 October**

**Matt Johnson:** "EU cooperation in Cross–Border Healthcare: European Reference Networks for Rare Diseases: The Patient's Perspective"

**European Commission workshop: "High Performance Computing in Health Research", Brussels, Belgium, 1 – 2 October**

**Yann Le Cam,** Panellist: "Future activities and implementation"

**Mathieu Boudes** also represented EURORDIS

**EFGCP / DIA / EMA Annual Conference 2014 on Better Medicines for Children: "Explore Ways to Enhance Collaboration Between Key Players", EMA headquarters, London, UK, 30 September – 1 October**

**Kerry Leeson–Beevers** represented EURORDIS and Alström Syndrome UK

**International Conference on Rare and Undiagnosed Diseases, Rome, Italy, 29–30 September**

**Matt Johnson:** Presentation of EURORDIS

**Annual meeting of EuroBioBank, Milan, Italy, 29 September**

**Mathieu Boudes** represented EURORDIS

**"CHAM" – 6th international Convention on Health Analysis and Management, Chamonix, France, 26–27 September**

**Yann Le Cam,** Panellist in the Round Table: "Patients: How to access to therapies?"

**EUSTM – European Society for Translational Medicines – Conference, Vienna, Austria, 22–25 September**

**Lesley Greene:** "New Horizons in Orphan Diseases Research and Development: A Collaborative Approach to encourage research and promote new treatments for orphan diseases"

**Pre–Health Council public conference: "System on Innovation – at the Service of Patients", Rome, Italy, 22 September**

**Yann Le Cam,** Panellist

**Conference on "Rare Diseases and Orphan Drug Development Initiatives in India", Hyderabad, India, 18–19 September**

**Yann Le Cam:** "Why Rare Disease Policies are needed? A European & International Perspective"

**| Behçet's International Conference – Paris 18,19 September**

**Marta Campabadal** "Connecting Rare Disease Patients Globally"

**| International Conference on Rare Diseases, Antalya, Turkey, 13–14 September**

**Yann Le Cam:** "Why Policies on Rare Diseases Matter? A European & International Perspective"

**| EFGCP Multi-Stakeholder Workshop: "How to Ensure Optimal Ethical Review within the New Clinical Trials Regulation?", Brussels, Belgium, 11 September**

**Flaminia Macchia,** Co-chair (together with Jozef Glasa), Session: "Identification of the Opportunities and Challenges for Ethical Review in Real Life for the Different Stakeholders"

**| "Integrating Personalised Medicine into the EU Strategy" organised by the European Alliance for Personalised Medicine (EAPM), Brussels, Belgium, 9–10 September**

**Flaminia Macchia:** "The Value of Personalised Medicine / what is Personalised Medicine?"

**Mathieu Boudes** also represented EURORDIS.

**| General Assembly of the Interstitial Cystitis Association, Rome, Italy September 6**

**Simona Bellagambi:** "The impulse comes from Europe: Concrete involvement of patients in the advancement of research, access to orphan drugs and treatments."

**| European Patients' Forum (EPF) Third Regional Cross Border Healthcare Conference (for Patient organisations' representatives from Austria, Czech Republic, Hungary, Slovakia and Slovenia), Ljubljana, Slovenia, 7–9 July**

**Matt Johnson:** "EU cooperation in Cross-Border Healthcare: European Reference Networks for Rare Diseases: The Patient's Perspective"

**| Technology for Associations Congress 2014, London, UK, 2–3 July**

**Sharon Ashton:** "Case Study EURORDIS: Setting clear objectives for your Mobile Event App"

**| First Symposium on New methodologies for clinical trials for small population groups, Vienna, Austria, 1–2 July**

**Tsveta Schyns** represented EURORDIS

**| Workshop on "EU Data Reforms and What They Mean for Research", Wellcome Trust and European Foundations Centre, Brussels, Belgium, 2 July**

**Jill Bonjean** represented EURORDIS

**| European Policy Centre high-level roundtable discussion: "Fostering Pharmaceutical Innovation – What role for regulation?", Brussels, Belgium, 25 June**

**Yann Le Cam & Flaminia Macchia** represented EURORDIS

**| Workshop on "European Reference Networks", organised by the European Commission – Directorate Health and Consumers, Brussels, Belgium, 23 June**

**Flaminia Macchia,** Chair of Session: "Quality, clinical criteria and performance assessment: Clinical and professional criteria"

**Matt Johnson and Juliette Sénécat** also represented EURORDIS

**| "Ad Augusta per Angusta" ("through difficulties to great things"), organised by NoRo Centre, Cluj Napoca, Romania, 19–20 June**

**Raquel Castro:** "Integration of Rare Diseases into Social Policies and Specialised Social Services in Europe – EUCERD Joint-Action"

**| International Glucogenosis Conference, Barcelona, Spain, 4–8 June**

**Marta Campabadal:** "Creation and management of European Rare Diseases Federations"

**| 55th Annual General Meeting of GIRP (The European Association of Pharmaceutical Full-line Wholesalers): "Innovative Solutions for Health – Shaping the Future of European Healthcare", Vienna, Austria, 2–3 June**

**Flaminia Macchia and Jill Bonjean** represented EURORDIS

**| "Health Inequalities in Europe: Monitoring and Directions for Action", Paris, France, 2 June**

**Ariane Weinman** represented EURORDIS

**| IMI (Innovative Medicines Initiative) Stakeholder Forum, Brussels, Belgium, 21 May 2014**

**Flaminia Macchia, Mathieu Boudes and François Houÿez** represented EURORDIS

**| IDIS – Insights on Access Forum, Camberley, U.K, 14–15 May**

**Jill Bonjean:** "Impact of early access on the rare disease community"

**| 2014 European E-Health Forum, Athens, Greece, 12–14 May**

**Denis Costello** represented EURORDIS

**| Working group on health systems – Friends of Europe – Brussels, Belgium, 6 May**

**Yann Le Cam and Flaminia Macchia** represented EURORDIS

**| ISCT (International Society for Cellular Therapy) Annual Global Meeting, Paris, France, 26 April**

**Dr. Sara Casati** represented EURORDIS

**| International Conference for Clinical Ethics Consultation (ICCEC 2014), Paris, France, 24–26 April**

**Yann Le Cam:** "L'empowerment des patients est-il un facteur de justice sociale ?" (Does patient's empowerment promote social justice?)

**| World Orphan Drug Congress USA, Washington D.C., USA, 23–25 April**

**Jill Bonjean** participated in the panel: "The role of patients in developing orphan medicinal products"



**| 1st Conference on Rare Diseases in Republika Srpska (the Bosnian Serb Republic), Banja Luka, 3 – 4 April**

**Vlasta Zmazek** represented EURORDIS

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**| “Global Updates on Accelerated Pathways for New Medicines to Patients in Need”, co-hosted by EMA, EFPIA, IMI & MIT NEWDIGS, London, UK, 2–3 April**

**Yann Le Cam – Flaminia Macchia:** Panel Discussion: Stakeholder Perspectives on Adaptive Licensing/Medicines Adaptive Pathways to Patients (“MAPPs”)

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**| EUPATI 2014 Workshop: “Reaching a Public Audience on Medicines Development”, Warsaw, Poland, 2 April**

**Maria Mavris:** “Presentation of the Rare Disease Day”

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**| 26th Annual DIA EuroMeeting, Vienna, Austria, 25–27 March**

**Maria Mavris:** “Empowering and Educating Patients in Medicines Research and Development”

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**| First “Central European Initiatives” Conference for building regional network of organizations for Rare Diseases, Skopje, Macedonia, 23 – 24 March**

**Vlasta Zmazek:** Presentation of the EUROPLAN project and EURORDIS

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**| RARE-Bestpractices General Assembly, Rome, Italy, 20–21 March**

**Juliette Sénécat and Mathieu Boudes** represented EURORDIS

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**| “International Forum on Rare Diseases and Orphan Drugs: Ensuring the access”? Sao Paulo, Brazil, 18 March**

**Yann Le Cam / Ariane Weinman:** “Access to Orphan Drugs in Europe”

**| RE(ACT), 2nd International Congress on Research of Rare and Orphan Diseases, Basel, Switzerland, 5–8 March**

**Yann Le Cam:** “European Rare Disease Research Agenda: The Patients’ Perspective”

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**| Journée des maladies rares 2014 (Rare Disease Day), Berne, Switzerland, 1 March**

**Denis Costello:** “RARECONNECT: “Créer des liens avec des personnes qui vous comprennent” (create links with people who understand you)

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**| Jornada Dia Mundial de les Malalties Minoritàries al CCCB, Barcelona, Spain, 28 February**

**Yann Le Cam:** “Empowerment of Patients”

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**| RD connect, EuRenOmics, NeurOmics Joint annual meeting, Heidelberg, Germany, 23– 26 February**

**Anna Kole:** “Contribution to a Patient-Centred Approach”

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**| United Parent Projects Muscular Dystrophy, Rome, Italy, 21 February**

**Maria Mavris:** “EURORDIS, EMA, Patients and Training”

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**| 2014 E-Rare Scientific Meeting, Athens, Greece, 20–22 January**

**Maria Mavris:** “EURORDIS, rare disease patients and research”

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# ACKNOWLEDGEMENTS

EURORDIS would like to thank the following organisations and companies for their financial support in 2014:

## Not-for-Profit Organisations and Public Entities

• • • • •

### AFM – Téléthon

The "Association Française contre les Myopathies", for the annual core activities grant and the office space they make available to the organisation free of charge.



### NORD

The National Organization for Rare Disorders for its contribution to the RareConnect web platform.



### CNSA

« Caisse Nationale de Solidarité pour l'Autonomie » (CNSA) for co-funding of the EUCERD Joint Action: Working for Rare Diseases (EJA), WP6 « Specialised social services »



### European Commission (DG Health and Food Safety)

- The Operating Grant for year 2014 (EURORDIS\_FY2014)
- The EUCERD Joint Action Working for Rare Diseases (EJA)
- The Conference Grant for the European Conference on Rare Diseases in Berlin (ECRD 2014)



Co-funded by  
the Health Programme  
of the European Union

### European Commission (DG Research and Innovation)

- RD-Connect Project to improve connections among different clinical datasets such as registries, biobanks and clinical bioinformatics for rare disease research.
- The European Clinical Research Infrastructures Network Integrated Activity (ECRIN-IA) project, which partners with and supports the EURORDIS Summer School
- The Innovative Medicines Initiative-Joint Undertaking (IMI-JU) project: European Patients' Academy on Therapeutic Innovation (EUPATI)
- RARE-Bestpractices – A platform for sharing best practices for the management of rare diseases
- The Innovative Medicines Initiative-Joint Undertaking (IMI-JU) project: Web-Radr, Recognising Adverse Drug Reactions
- Genetic Clinic of the Future, A stepping stone approach towards the Genetics Clinic of the Future



### AIPM

Association of International Pharmaceutical Manufacturers for co-funding the Russian version of EURORDIS Website and eNews



### EFPIA

For co-funding the Innovative Medicines Initiative-Joint Undertaking (IMI-JU) project: European Patients' Academy on Therapeutic Innovation (EUPATI)



# Pharmaceutical and Biotechnology Sector Companies

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EURORDIS has diversified its Pharmaceutical and Biotechnology Sector Companies' sponsorship from 41 to 43 different companies in 2014. EURORDIS believes that diversification of funding is a key success factor to minimise potential conflict of interest with donors. Companies have supported EURORDIS primarily through the EURORDIS Round Table of Companies<sup>1</sup>, the EURORDIS Membership Meeting 2014 Berlin, the RareConnect® Online Patients Communities<sup>2</sup> and the EURORDIS Gala Dinner<sup>3</sup> which funds actions to reduce isolation of people living with a rare disease, to increase awareness of rare diseases and to empower leaders of rare disease communities through training, capacity-building and exchange. The breakdown of each company's donations by project is detailed on the EURORDIS website on the "Corporate" tab of the "Financial Information" section.

## THE TOP FIVE DONORS

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SHIRE



GSK



CELGENE



CSL BEHRING



PFIZER



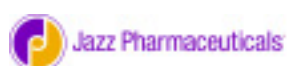
<sup>1</sup> <http://www.eurordis.org/content/ertc-members>

<sup>2</sup> <http://www.rareconnect.org>

<sup>3</sup> <http://galadinner.eurordis.org>










## OTHER PHARMACEUTICAL COMPANIES (LISTED ALPHABETICALLY)



## OTHER HEALTH SECTOR CORPORATES

<b>Cydan</b>	For EURORDIS Round Table of Companies membership fees	
<b>Idis</b>	For EURORDIS Round Table of Companies membership fees and Black Pearl Gala Dinner 2014 contribution	
<b>Marc Krueger &amp; Associates, Inc.</b>	For EURORDIS Black Pearl Gala Dinner 2014 contribution and support to one ERTC Workshop Welcome Dinner	
<b>Pharmaphorum media</b>	For in-kind contribution of communications services in the context of ECRD 2014	

## OTHER CONTRIBUTORS

<b>Arès Life Sciences</b>	For EURORDIS Black Pearl Gala Dinner 2014 contribution	
<b>Burson-Marsteller</b>	For on-going in-kind contribution of communications advice to build awareness of rare diseases	
<b>DLA Piper UK LLP</b>	For in-kind contribution of legal services	
<b>Multiburo</b>	For the discounted pricing of the office space made available to the Public Affairs team based in Brussels	
<b>DAREN Direction Artistique</b>	For in-kind contribution of communications services in the context of the European Year for Rare Diseases 2019	
<b>Medioplanet</b>	For in-kind contribution of communications services in the context of Rare Disease Day 2014	
<b>Carlo Hintermann and Moonchausen</b>	For the in-kind contribution of the creation and production of the Official Rare Disease Day 2015 Video	

# WORKPLAN 2015



**EURORDIS**  
Rare Diseases Europe

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# OUR 2015 ACTION PLAN

## Advocacy

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### ADVOCACY ISSUES

- Promoting rare diseases as a sustainable public health priority in the EU programmes beyond public health: research, enterprise, digital, social
- Making rare diseases a public health priority in all EU Member States
- Promoting rare diseases as a public health priority internationally
- Improving access to orphan medicinal products and treatments for rare diseases
- Improving quality and access to rare disease diagnosis, as well as to medical and social care
- Accessing cross-border healthcare and making possible patient mobility
- Promoting research and bridging patients and researcher activities
- Addressing the issues of genetic testing, genetic counselling & newborn screening

### ADVOCACY ACTIONS

- **Supporting implementation of the EU strategy on RDs adopted in 2008 “Commission Communication on Rare Diseases: Europe’s Challenges” and in 2009 with the “Council Recommendation on an Action in the Field of Rare Diseases” through participation in:**

- 1 The Commission Expert Group on Rare Diseases (CERD):

Participation of four EURORDIS patient representatives, their four alternates and two observers in the Commission Expert Group on Rare Diseases.

- 2 The Commission Expert Group on Cancer Control:

Participation of one EURORDIS patient representative, one alternate and one observer in the Commission Expert Group on Cancer Control.

- 3 The EURORDIS Policy Action Group (PAG):

The patient representatives in the Commission Expert Groups together with public affairs staff form the PAG

- The PAG teams up with other volunteers and EURORDIS staff on the issues of the two Commission Expert Groups:

- National strategies, national plans & indicators
- Centres of Expertise and European Reference Networks

- Cross-border healthcare
- Patient registration and data collection
- Codification and classification of rare diseases
- Guidelines on diagnostics and care
- Gene Testing and Counselling
- Newborn screening
- Information provision to patients and professionals
- Access to rare disease therapies and orphan medicinal products
- Integration of rare diseases into social policies and services

■ The PAG contributes to the Commission Expert Groups with concrete policy proposals in order to address patient needs when implementing the strategy foreseen in the EU framework. The PAG coordinates patients' views on policy on rare diseases and policy on cancer so to address the needs of people affected by rare cancers

■ The PAG intends to lead and support collaboration around the Commission Expert Group on Rare Diseases in the following specific areas: Rare Disease Day, European Conference on Rare Diseases and Orphan Products, information provision to patients and professionals, improving access to medicines, comprehensive approach to address the health and social needs of people living with rare diseases (PLWRD) and the promotion of international cooperation.

■ The PAG contributes actively to the 'State of the Art of Rare Disease Activities in Europe' through direct EURORDIS input and involvement of its national alliances.

#### 4 EUCERD Joint Action Working for Rare Diseases (March 2012–September 2015)

- EURORDIS supports the implementation of the EUCERD Joint Action as a full partner across all Work Packages and disseminates its outcomes (EURORDIS is the NGO representing patients in this Joint Action involving the EU Member States and the European Commission).
- EURORDIS is specifically involved in two Work Packages of the EUCERD Joint Action:
  - Leader of Work Package 6 on 'specialised social services and integration of rare diseases into social policies and services'.
  - Co-partner of Work Package 4 on the 'support for the implementation of plans or strategies for RDs at MS level' (continuation of EUROPLAN).

#### 5 2nd Joint Action on Rare Diseases (2015–2018)

Contribute to the development of the 2nd Joint Action on Rare Diseases so to address our advocacy issues and actions while promoting a multi-stakeholder dialogue

6 Disseminate EU policy outcomes, monitor progress, promote new strategies and innovative approaches, all to integrate all main stakeholders at EU and national levels, and share common objectives through EMM 2015 Madrid, internal dissemination, EURORDIS Website, eNews and social media

#### ■ Promote the sustainability of rare diseases as a policy and budget priority in the EU programmes for the period 2014–2020:

- As a public health priority in the 3rd EU Public Health Programme 'Health for Growth' & Annual Work Programmes and advocate on the rare disease community's priorities
- As a research priority in the 8th EU Research & Technology Framework Programme 'Horizon 2020' & biannual Work Programmes and in the Innovative Medicines Initiatives (IMI 2), and advocate on the rare disease community's priorities
- As a priority which can be relevantly articulated with the EU policy agenda for the next years on growth, science-based innovation, competitiveness, efficiency of healthcare systems and social services and digital economy

#### ■ Monitor the implementation of the EU Directive on Patient's Rights to Cross-border Healthcare adopted in 2011 providing a legal basis for patients' rights for accessing care abroad and for the establishment of European Reference Networks aimed at addressing specific needs of rare disease patients.

- Disseminate information on and promote access to national contact points
- Facilitate reporting of real life experience of patients

#### ■ Advocate to improve access to orphan medicinal products and promote a new business model sustainable for society

- Develop and adopt EURORDIS position paper on 'Improving Access to Orphan Medicinal Products in Europe and its sustainability for society'
- Promote and take an active part in the dialogue on Medicine Alternative Development Pathways with EFPIA and EMA
- Support the implementation of the objectives of the Clinical Added Value of Orphan Medicinal Products (CAVOMP) process based on the EUCERD Recommendation, in conjunction with EMA, EUnetHTA, MEDEV, and the EFPIA-EuropaBio Taskforce on Rare Diseases and Orphan Drugs
- Promote innovative approaches, policies and pilots: early dialogue/ scoping / de-Risking, EMA-HTA-Payers dialogue involving patient representatives and medical experts, progressive patient access, adaptive licencing, compassionate use and early access programmes, shortages, and off-label use of medicines in rare indications
- Contribute to new approaches to benefit/risk assessment, taking into consideration patient values & patients' preferred treatment options

- Contribute to the pilots of the Mechanism of Coordinated Access (MoCA) to Orphan Drugs, developed within the EU Stakeholders Forum on Corporate Responsibility in Pharmaceuticals, promoting pan-European collaboration between EU Member States on Orphan Drugs based on a common approach to the value of new medicines, pricing, volume and post-Marketing Authorisation data generation
- · Explore the wider use of Joint Procurement, Managed Entry Agreement and Differential Pricing
- · Contribute to developing at EU level adaptive approaches on methodology and criteria for the Health Technology Assessment of the Effectiveness or Relative Effectiveness as for evaluation of the value of Orphan Drugs
- · Promote these innovative approaches in Member States & national plans on rare diseases
- · Increase direct cooperation with leaders from rare disease therapies sector
- · Prepare and widely disseminate the annual EURORDIS Access Campaign report based on the ongoing survey
- · In emergency situations, support actions to defend patients' access to rare disease therapies

#### ■ **Promote rare diseases as an international public health priority through:**

- Establish Rare Diseases International as an informal network of rare disease patient organisations, and its subsequent development through: the creation of an RDI governance group, the Council of RDI; recruitment of members; development of an action plan; creation of a mailing list and communication tools with a distinct visual identity. RDI official launch will take place in Madrid in May 2015
- The adoption of a joint declaration 'Rare Diseases: an International Public Health Challenge' by major rare disease patient organisations
- Signing of MoUs with international patient organisations
- The international development of Rare Disease Day
- Continued active participation in the International Rare Disease Research Consortium
- Elaboration of a strategy to integrate into the UN System: ECOSOC, WHO, and Council of Europe

#### ■ **Advocate in support of rare disease research, a high priority in the 2010–2015 Strategy:**

- Contribute to the consolidation of rare disease priorities and activities in Horizon 2020, IMI 2, JRC, eRare, InnoRare and IRDiRC

- Engage EURORDIS in European research infrastructures, policy debates and projects.
- Promote IRDiRC objectives, governance and contribute to IRDiRC policy papers
- Empower patient advocates through policy fact sheets and capacity-building workshops at the EMM 2015 Madrid

#### ■ **Contribute to the revision of EU Regulation on Advanced Therapies / EURORDIS Statement**

#### ■ **Contribute to the assessment of emerging issues in biosimilars / EURORDIS Statement**

#### ■ **Contribute to the future Regulation on Data Protection and adopt EURORDIS statement**

#### ■ **Adopt a position paper on Integration of Rare Disease in Social Services**

#### ■ **Advocate on the impact of new knowledge on genetics through the:**

- Elaboration of EURORDIS positions on genetic testing, screening and Next Generation Sequencing
- Contribution to the reflection processes on stratified medicines and fragmentation of frequent diseases

#### ■ **Prepare for the revision of EU pharmaceutical legislation**

### PATIENT KNOWLEDGE-BASED ADVOCACY

#### ■ **Launch of EURORDIS Patient Voices**

- The EURORDIS Patient Voices programme to be launched in 2015 aims at the creation of a patient-derived knowledge survey programme. EURORDIS has a long-standing experience in collecting the patient perspective through surveys, debates, online communities, workshops etc. Examples of our activities centred around patient-derived knowledge are The voice of 10.000 patients, RareConnect Community Polls, Rare Diseases International Survey and the Access Campaign. The new programme will further develop, centralise and structure our efforts with the overall objective of becoming the go-to source for the perspective of the rare disease patient community on a number of transversal topics important to people living with rare diseases. Specific topics covered in 2015 will include data sharing and data protection, the future of genetic clinics and one additional topic (implementation of ERNs, social needs).



# Information & Networking

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## COMMUNITY BUILDING

### **Maintain the EURORDIS Membership over 650 members and ensure regular interaction**

### **Maintain implementation of process of regular membership reassessment, as established in 2013**

### **Recruitment of members at large, particularly in all EU member states, acceding and candidate EU Member States, rare cancer groups and European Federations**

### **Organise EURORDIS Membership Meeting 2015 Madrid on 28–30 May with capacity building workshops**

### **Build capacities of the European network of 33 national alliances through:**

- Promoting greater convergence and collaboration between national alliances, as well as between national alliances and EURORDIS, through implementation of the 'Common Goals & Mutual Commitments between National Alliances in Europe and EURORDIS: An agenda between 2014 & 2020'
- Sharing information, experience, guidance and common actions in national plans, Rare Disease Day, cross-border healthcare, access to medicines and evaluation of Centres of Expertise
- Organising two workshops of the Council of National Alliances
- Organising telephone or web-based conference calls on specific topics
- Maintaining direct interaction or visits between EURORDIS and national alliances
- Operating a EURORDIS "Learning from Each Other" Exchange Programme for national alliances to enable more direct exchange, mutual support and capacity building between national alliances
- Encouraging contact between patient organisations in same countries and the creation of new national rare disease alliances where there are none yet

### **Build capacities of the European network of 45 disease-specific European / International Federations & Networks through:**

- Sharing information, experience, good practices and guidance – integration of RareTogether! Website in Council of European Federations (CEF) section – between European Federations and common actions in European Reference Networks, Rare Disease Day, drug development & interaction with EMA & access to medicines & patients reporting of adverse events, cross-border healthcare, social policy & services, and Online Patient Communities
- Providing regular advice to European Federations & Networks

- Organising a two day workshop of the Council of European Federations, including a one day training course

- Expanding the EURORDIS Programme to Support European Federations & Networks with money for their governance meetings, membership meetings, first European conferences on their disease

- Operating a EURORDIS "Learning from Each Other" Mutual Exchange Programme for European federations or networks to enable more direct exchange, mutual support and capacity building between disease-specific networks or federations

### **Outreach to patient groups in Central & Eastern Europe, Balkans, Russia and Caucasia, support of their actions to raise public awareness and promote policy on rare diseases and promote the creation of national alliances:**

- Maintain and expand EURORDIS Website, eNews and main documents available in Russian; develop RareConnect in Russian
- Provide official EURORDIS endorsement/ promote and take part in national conferences across Central & Eastern Europe on request of our members
- Support Russian and Caucasian endeavours in raising awareness of rare diseases in their regions through support of their national conferences or EURORDIS–EUROPLAN conferences on national RD strategies as well as Rare Disease Day
- Provide access to EURORDIS Patient Advocate Fellowships Programmes for the EURORDIS Membership Meeting 2015 Madrid and to the EURORDIS Summer School 2015 Barcelona
- Maintain dissemination of Decide Topics available in 22 languages

### **Support EURORDIS Volunteers' involvement through:**

- Strengthening the identification, recruitment and support of volunteers to be increasingly involved in EURORDIS activities based on the EURORDIS Charter of Volunteers, participating in NGO partnerships and representing patients in the European Commission and EMA working groups and committees
- Creating four EURORDIS Thematic Working Groups of volunteers on the following issues: national plans, social services, registries, best practice clinical guidelines
- Briefing on key topics, access to shared reference documents and public presentations
- Maintaining and expanding the volunteer section on the EURORDIS website

### **Expand RareConnect, the global social network of online patients communities:**

- Implement the RareConnect Action Plan 2014–2016

and Strategy for 2020

- Adjust governance and management structure to new development phase with the participation of more national alliances
- Maintain & support the 70 existing online patient communities in conjunction with the over 570 patient groups and over 300 volunteer moderators involved
- Launch 20–25 new online patient communities, adapting the tool to the needs of each specific community in conjunction with relevant existing patient groups for these rare diseases
- Develop an evolved translation model for RareConnect based on developing a plan to seek investment in a volunteer translator management tool, along with a volunteer translator programme, which will include outreach to translation institutions, sourcing volunteer translators from other sources and the incorporation of part-time or volunteer coordinator into the team
- Add an additional two to three languages to RareConnect (Russian, Portuguese)
- Enhance the presentation of information on existing communities to bring conversations and patient testimonials to the forefront and improve the presentation and quality of medical information
- Launch ten RareConnect Discussion Groups. Discussion Groups will be lighter to set up and maintain than full communities and be purely conversation driven.
- Pilot RareConnect as a platform to perform surveys
- Invest in a mobile interface for RareConnect to enable visitors to participate on a more regular basis and accomplish priority tasks from their mobile device
- Develop partnerships with national alliances, but also where appropriate and where there is crossover (with topic groups for example) with learned societies
- Develop a communication plan for RareConnect based on online advertising (via in-kind Google Adwords and investment in Facebook advertising) and printed material (flyers, brochures and posters)
- Explore opportunities of partnership between RareConnect and researcher/patient exchange platforms in partnership with academic research teams
- Examine the feasibility of building on the Summer School to expand capacity building of rare disease patient advocates in social media & communications

## INFORMING & RAISING AWARENESS

- **Organise Rare Disease Day 2015: around the theme “Living with a Rare Disease” and with the slogan “Day-by-day, hand-in-hand” keeping the overarching theme of “Solidarity”**
  - Maintain the new user-friendly Rare Disease Day website with more features and information and create a mobile version for access from smartphones

- A promotional video in over 25 languages to be disseminated through a social media campaign and the EURORDIS websites
- Marketing & communication plan: info pack, poster & slogan, patient testimonies, social media
- Continue to expand the Join and Raise Hands campaign and tie it in to social media campaign
- Storify: A digest of social media and country pages information disseminated in the weeks leading up to 28 February

- **Organise a Policy Discussion Event in Brussels to mark the occasion of Rare Disease Day on 24 February 2015 on “Living with a Rare Disease”**

- **Plan Rare Disease Day 2016: marketing and communication plan, info Pack, video, poster & slogan, website, patient testimonies, social media**

- **Organise the EURORDIS Awards 2015, designed to recognise the outstanding commitment and achievements of patient organisations, volunteers, companies, scientists, media and policy makers who have contributed to reducing the impact of rare diseases**

- **Take actions towards a European Year for Rare Diseases (EYRD) in 2019:**

- Promote the EYRD campaign launched in 2014 to rare disease stakeholders and outreach more to the general public
- Update the common sign-up web page on the EURORDIS website to make it more appealing to a broader public
- Build the base of a patrons programme
- Promote a social media campaign in multiple languages to reach out to patient organisations, people living with a rare disease, policy makers and other stakeholders
- Identify and approach a political leader and form a group of MEPs to champion the campaign within the European Parliament

- **Maintain content quality and information architecture of EURORDIS Website:**

- Centered on target audiences: patient organisations & advocates, patient and families, other stakeholders (secondary)
- Maintain navigation and user-friendliness of website
- Maintain quality, updated information in seven languages (EN, FR, DE, ES, IT, PT, RU) in all sections
- Further develop content and facilitate access via three focal points: the core EURORDIS.org website, its satellites (eNews, Rare Disease Day, RareConnect, Help Lines) and EURORDIS Social media (Facebook, Twitter, Flickr, YouTube)
- Within core website, consolidate issues of strategic importance, adding dedicated sections on: Access Campaign, cross-border healthcare, European

Reference Networks & CoEs, 'Get Involved', 'International' and 'Donate'

- Better organise the publications section so that important documents such as Q&A, fact sheets are more visible and easily accessible
- Implement the social media strategy and internal organisation for more vibrant social media
- Develop strategy for promotion of EURORDIS website's main new services, sections, information and capacity-building tools for patients and patient advocates
- Enhance EURORDIS website software (development of Drupal)

#### ■ **Continue social media and patient empowerment webinar series to moderators and patient groups**

#### ■ **Maintain EURORDIS TV, which provides updated video footage of: news & current affairs, living with a rare disease stories, patient groups, research, health policy, orphan drugs & therapies, EURORDIS speeches & events, conference presentations, training or capacity building videos and Rare Disease Day events**

- Make use of pre-existing recorded material, edit and upload to EURORDIS TV
- Record videos – in-house and outsourced – to present key policy issues, papers, Summer School and EMM testimonies
- Rebrand EURORDIS TV and further promote it

#### ■ **Publish EURORDIS eNews in 7 languages (EN, FR, DE, SP, IT, PT, RU) every second week all year round (except August)**

#### ■ **Present Photo Contest 2014 at EMM 2015 Madrid**

#### ■ **Organise Photo Contest 2015 with an eventual presentation at EMM 2016 Edinburgh**

#### ■ **Conceive a deeper and long-term strategy on photo and video projects to promote people living with a rare disease in the public space**

### INFORMATION SERVICES TO PATIENTS

#### ■ **Improve access to and quality of information through rare disease help lines:**

- Develop the European Network of Rare Disease Help Lines:
  - Governance & business meeting, develop membership base
  - Explore new financial tools for the help lines (Structural Funds)
  - Submit and publish an article on the Caller Profile Analysis in a scientific journal
  - Organise the annual caller profile analysis
  - Engage and train help lines on reporting adverse events of medicines used in all rare disease treatments (whether orphan or not, and off-label) as part of the EURORDIS DIOD project (Daily Impact of Drugs in patients' lives) or of other activities
  - Link to national plans
  - Link to ERNs

#### ■ **Maintain and improve the EURORDIS InfoHub feature on the Eurordis.org website and RareConnect platform to improve access to quality information sources on the internet for patients, families and relatives as well as structured access and use of information for patient advocates**

## Health Policy & Healthcare Services

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### PROMOTING RARE DISEASE HEALTH POLICY DEVELOPMENT

Support the national alliances in their action in advancing national rare disease policy through the following activities:

#### ■ **EUCERD Joint Action / Work Package 4: "support for the implementation of plans or strategies for RDs at MS level" (continuation of EUROPLAN):**

- Support 20 national alliances in the European Union to facilitate the organisation of a EUROPLAN National Conference aimed at promoting the implementation of a national plan for Rare Diseases. This second

round of EUROPLAN National Conferences will finish in May 2015 when the Irish National Conference takes place

- The EUROPLAN National Conferences use the same format and methodological tools derived from EUROPLAN I (2008–2011) in addition to content guidelines that have been developed for six policy areas, including technical policy guidance, notably on EUCERD Recommendations. The organisers can adapt the layout of the Conferences to the most pressing needs of the rare disease community in their countries
- 10 EURORDIS Advisors, all from different national alliances, are in charge of advising one to three



national alliance(s) for the preparation of the EUROPLAN National Conference. The Advisors work with the EURORDIS public affairs staff

- The 10 EURORDIS Advisors participate in the EUROPLAN capacity-building workshops together with representatives of national authorities
- Synthesis on 5 key points of national plans and national conference final reports: governance, CoEs & ERNs, research policy, social policy, access to OMPs & rare disease therapies

#### ■ EURORDIS Membership Meeting 2015 in May in Madrid

- Information & experience-sharing on national planning across members of EURORDIS
- Capacity-building workshop for rare disease patient advocates: promoting national plans, proposing adequate measures, playing an active role in national plan steering committees, and being trained in new policy areas

#### ■ EURORDIS Policy Fact Sheets for Patient Advocates

- 2-page document enabling patient representatives to better advocate issues of specific importance with national and/or EU policy decision makers (e.g. on policy and services expected by patients and their families)
- Development of new policy fact sheets on: access to orphan medical products; orphaCode and its national potential use; EU structural funds for national plans; update of European Reference Networks; and integration of rare diseases in social services

#### ■ Targeted communication

- Promote exchange of information and sharing of experiences through the website by expanding more user-friendly EURORDIS web sections on EU and national policies, the eNews and participation in national conferences or other meetings
- Promote exchange of information, experience, good measures and concerns between national alliances, the 10 EURORDIS Advisors and the staff through means such as a national plan mailing list and the eNews

## PUTTING RARE DISEASE PATIENTS AT THE HEART OF THE HEALTHCARE SYSTEM

### ■ Support the implementation of the policy on European Reference Networks (ERN) and Centres of Expertise (CoE) toward a patient-centric approach aiming at clinical excellence and best possible patient health outcomes. Build the capacities of the EURORDIS membership and their readiness to support European Reference Networks

#### ■ Implement policy on ERN, CoE, expert networks & healthcare pathways on rare diseases:

- Promote the long-term EURORDIS strategic vision on ERN

- Disseminate the Declaration of Common Principles on CoE & ERN
- Take action to help prepare rare disease patient groups for the launch of the Call for Proposals for ERN
- Develop a proposal on grouping rare diseases so to take optimal advantage of the limited number of future ERNs while ensuring that on in the long term every rare disease patient has a home, a healthcare pathway
- Initiate (and debate with EURORDIS members) future possible organisation of ERN in Community Advisory Boards, to be structured per grouping of rare diseases. Initiate actions to support collaboration between European federations, patient organisations or online communities and ERNs in key areas such as: governance of ERNs, disease registries, biological repositories, clinical trials, treatment protocol trials, standards of diagnosis & care, information to patients and outreach to patients
- Organise a session during EMM 2015 Madrid dedicated to ERN & CoE
- Organise three follow-up webinars for all EURORDIS members, European federations and national alliances on the implementation of ERN, that will take place after key European conference leading up to the first call for tenders for ERN applications at the end of 2015.
- Publish an updated fact sheet on ERN
- Maintain a dedicated and regularly updated website section on CoE and ERN

### ■ Support the implementation of ERNs through lead and manage the Consortium for Partnership for the Assessment of European Reference Networks Clinical Excellence (PACE-ERN), formed by EURORDIS together with the European Organisations of Hospitals (HOPE) and the internationally renowned health technology assessment agency Accreditation Europe / Accreditation International.

The Consortium will support the Commission in the development of ERNs, through producing a technical proposal of the assessment manual for ERN application that the Commission will consult prior to the implementation of the assessment framework, including:

- Design the detailed application and assessment process, detailed operational and eligibility criteria
- Develop the manual & tool box to be used by ERN applicants and independent agencies responsible for their assessment

### ■ Participate in the development of new long-term projects on the methodologies for creation and review of best clinical practices:

- Partnering in the EU project RARE-BestPractices, a platform for sharing best practices for management of rare diseases, in order to:
  1. Build capacities of rare disease patient organisations and of people living with a rare disease on the importance, use and benefits of best practice guidelines. Do this through training activities targeted at patient advocates

and dissemination of information and project outcomes (WP1)

- 2. Establish a EURORDIS working group on best clinical practices
- 3. Contribute to the development and the implementation of a core methodology on best practice guidelines for rare diseases (WP3)
- 4. Ensure a strong interaction and coordination of the RARE-BestPractices project with other initiatives such as EUROPLAN II, RD-Connect and IRDIRC and contribute to the dissemination of the RARE-BestPractices project outcomes (WP1,6);

- How to better use scientific progress in biomedical research to translate results into tangible benefits for rare disease patients and to offer high quality information to patients and healthcare professionals;
- How to promote and support a consistent level of healthcare services for rare disease patients in the EU while implementing the EU Directive on Patients' Rights in Cross-border Healthcare

#### ■ Engage in, promote and develop policy on upcoming genetic developments:

- Create a dedicated website section and disseminate information, including better promotion of information available from EuroGentest, the European Society of Human Genetics, the International Society for Neo-Natal Screening and the Commission expert Group on Rare Diseases.
- Participate in the Genetic Clinic of the Future (GCF) project, led by the University of Utrecht, Netherlands. The GCF project's main objectives are:
  - To ensure that the future implementation of high-throughput genome technologies is relevant to the needs of patients and responsive to the interests and concerns of citizens and stakeholders;
  - To engage all relevant groups in constructive dialogue by enabling 'radically interdisciplinary' collaborations between genomics researchers, clinical geneticists and other medical specialists, bioinformaticians, patient representatives, policy makers and experts from ethics, social science and law;
  - To implement key Science with and for Society (SwafS) issues (ethics, patient and citizen involvement, education, communication and public engagement and policy development) in the GCF, ensuring that ethical reflection and stakeholder involvement do not occur in parallel, but are effectively integrated in the core of the project;
  - To establish a robust communication and implementation strategy that implements the project's outcomes and recommendations in research and clinical practices, as well as policy developments, outlining opportunities for a more responsive health research and innovation system
- Support an increase of capacities to analyse new

data resulting, from newborn screening for patients, and engage in recommendations with Commission expert Group on Rare Diseases and learned societies. Newborn screening is rapidly changing the access to and capacities for diagnostic of rare diseases and profiling for precision treatments

#### ■ Contribute to the transparency of clinical trials data (both global results and individual patient's data):

- Adopt a position on the EMA policy on access to clinical trials data

### INTEGRATING RARE DISEASES INTO SOCIAL POLICY AND SPECIALISED SERVICES TO PATIENTS

#### ■ Promote integration of the challenges faced by people living with a rare disease into social policy:

- Within the EUCERD Joint Action (EJA) (2012-2015), EURORDIS leads the work package on "Provision of Specialised Social Services and Integration of Rare Diseases into Social Policies and Services". This involves: mapping existing specialised social services; developing case studies on experienced services based on information collected during country visits; addressing issues related to the training of specialised social service providers and elaborating the draft CERD recommendations in the social field
- Also within the EJA, EURORDIS will coordinate with the work package on national plans: 1) ensuring the integration of the different issues concerning social challenges into of the remaining EUROPLAN National Conferences and 2) analysing the outcomes of EUROPLAN Conference reports and reviewing national plans in order to extract good practices and ideas from MS within social services and social policies
- Advocate for the promotion of EU and national policies for social research and quality of life studies
- Disseminate the EURORDIS position paper on the social challenges faced by people living with a rare disease/their relatives and the need to integrate rare diseases into existing social policies and services;
- Follow-up of social research projects and dissemination of relevant findings on the socio-economic difficulties faced by people living with a rare disease in Europe;
- Perform preliminary research and plan for a EURORDIS Patient Voices survey on the social and financial burden of rare diseases for patients and families in coordination with INNOV-Care (if approved)
- Promote patient-centred services, namely via the INNOV-Care project (if approved), which proposes the implementation, testing, evaluation of impact/ cost-benefit of a social care pathway. This will be organised in coordination with an European network of resource centres, national resource centres, regional case managers and local care professionals
- Networking and building relationships with other organisations and professional associations active in the social field at the EU level

**| Stimulate the development and the improvement of access to respite care services, therapeutic recreation programmes, adapted housing and resource centres for rare diseases:**

- Maintain and expand EURORDIS website sections for respite care services, therapeutic recreation programmes, adapted housing and resource centres to contain all relevant definitions, information and contacts collected
- Continue mapping of services and updating information (available online)
- Facilitate the exchange of experiences between services through: the dissemination of a document on consensual guiding principles for specialised social services; the dissemination of case studies on respite

care services, therapeutic recreation programmes, adapted housing and resource centres derived from country visits performed as part of the EJA; and the dissemination of a document on consensual principles for the training of social service providers

- Advocate for the creation of new respite care services, therapeutic recreation programmes, adapted housing services and resource centres through the policy fact sheets, presentations delivered at the European Conference on Rare Diseases (under Theme 6 – 'Beyond Medical Care') and the monitoring of measures adopted in national plans
- Encourage EURORDIS staff, volunteers and members to volunteer at summer camps

## Research, Drugs & Therapies

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### INVOLVEMENT WITH RESEARCH PUBLICATIONS

**| Member of the Editorial Board of OrphaNews Europe**

**| Member of the Editorial Board of the Journal Expert Opinion on Orphan Drugs**

**| Link with Orphanet Journal of Rare Diseases**

Reference Networks, as well as planning of healthcare services & social services, clinical practices, medicine development

- Website section on registries

**| Participate in the development of new long-term infrastructure projects on registries, bioBanks and -omics through:**

- Engage EURORDIS in new projects of integration of rare disease European research infrastructures and study design
- RD-Connect, an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research. EURORDIS ensures the involvement of patient organisations in capacity building on registries, biobanking and omics and directly contributes to developing registry, biobanking and omics infrastructures and their integration. EURORDIS ensures a strong interaction and coordination of the RD-CONNECT network with other initiatives within and beyond Europe, as well as the dissemination of the RD-CONNECT project outcomes at the international level (WP1, WP7).
- BBMRI Consortium, ensuring patient representation in its governance
- EuroBioBank, promoting specific agreements between patient organisations and EuroBioBank by providing information, template agreements and advice

### SHAPING AND PROMOTING RESEARCH POLICY SUPPORTING CLINICAL RESEARCH

**| Promote research on rare diseases as a policy and budget priority at international, EU and national levels with an integrated approach through:**

- Dissemination of EURORDIS policy fact sheets and reference papers on research
- Participation in and support of the development of the International Rare Disease Research Consortium (IRDiRC), of which EURORDIS is a member of the Executive Committee, a member and Chair of the Therapies Scientific Committee and Working Groups on Ethics & Legal Affairs, Registries, Regulatory Affairs, Biomarkers. Take part in all meetings and activities
- Increased participation in the ERA-Net project E-Rare involving Member States
- Participation in research policy activities related to national plans for rare diseases (EUROPLAN)

**| Promote patient rare disease registries and data collection**

- Engage in the development of the EU platform on Rare Disease Registries at the Commission Joint Research Centre through its Advisory Board, based on the EPIRARE Book and EURORDIS 10 key principles, EUCERD Recommendation on registries and patient's preferred policy scenario on Registries, while making the link with the servicing of European

### SUPPORTING CLINICAL RESEARCH

**| Promote medicines adaptive pathways to patients, so to increase clinical research for unmet needs, speed development, reduce costs and improve outcomes:**

- Participate as full Partner in the IMI Consortium led by EMA 'Accelerated Development of Appropriate Patient Therapies: a Sustainable Multi-stakeholder Approach from Research to Treatment', a support action involving EURORDIS, European Patient Forum,



EUnetHTA, NICE, HAS, ZIN, CASMI, TI Pharma / Esher and NEWDIGS

- Take part in the EMA-EFPIA Working Group facilitated by Tapestry on 'The Journey to Improve Outcomes'. Get involved in all relevant activities and concrete processes to support its implementation such as listed in the section of this Action Plan in 'Promoting Drug Development & Access to Treatments'
- Bring these innovative policy approaches and activities to the IRDiRC

#### **Promote patient engagement in clinical research and medicine development on rare diseases all along the life cycle:**

- Create and lead a consortium to apply to IMI 'Knowledge Repository to Enable Patient Focused Medicine Development' through a project entitled 'Enabling Patient Engagement & Improving Patient Outcomes: a Sustainable Structured Multi-Stakeholder Approach from Research to Treatment', bringing together EURORDIS, EPF as co-lead, EATG, EGAN, IPPOSI, EMA, NICE, AIFA, TI Pharma, Centre de Sociologie des Organisation of Paris, University of Manchester, University College London and University of Copenhagen
- Promote adoption of the EURORDIS Charter for Collaboration between Patient Organisations and Sponsors of Rare Disease Clinical Trials; facilitate the implementation of the Charter with the support of a Mentor

#### **Support specific actions in Rare Cancers:**

- Participation in the Rare Cancer Europe network co-founded by EURORDIS
- Collaboration with the European Society of Medical Oncology (ESMO)
- Support coordination between the Commission Expert Group on Rare Diseases and the Commission Expert Group on Cancer Control to maximise synergies of these two EU policy areas
- Collaborate with the European Cancer Patients Coalition (ECPC) on Personalised Medicines and Rare Cancer Registries position paper
- Support a EURORDIS Task Force on Rare Cancers (future Community Advisory Board Rare Cancers) composed of 8 elected representatives of member patient groups, to inform, advise and lead EURORDIS policy in rare cancer area, including the representative in Commission Expert Group
- Support rare cancer patients community and capacity building through training activities, RareConnect, programmes with European Federations and communication
- Increase relevant visibility of EURORDIS' involvement in rare cancer activities on the EURORDIS website
- Increase membership of rare cancer patient groups

## **GAINING KNOWLEDGE ON RARE DISEASE TREATMENTS**

#### **Develop working relations with experts in outcomes research to better integrate patients' views on R&D of medicines, their benefit/risk evaluation and their value for society**

- Maintain partnership with the International Society of Pharmacoeconomics and Outcomes research and co-chair its Patient-Centred Special Interest Group

#### **Promote rare disease patient spontaneous reporting on suspected adverse drug reactions of orphan and non-orphan drugs by:**

- Maintaining webpages on EURORDIS website containing explanation on: why it is important to report adverse drug reactions (ADRs); explanation of the work done by regulatory authorities when receiving reports from patients; explanation of the pharmacovigilance system as a whole; and links to reporting sites in Europe, by country/ language
- Deployment of activities in the WebRadr & SCOPE Joint Action as Advisor
- Providing information to EURORDIS online patient communities and other social media
- Posting announcements in EURORDIS eNews to encourage members to become pharmaco-vigilant and informing them of where to gain knowledge regarding compassionate use
- Organising and participating in sessions on pharmacovigilance at rare disease conferences
- Training of patients advocates through webinars and/or eLearning and/or session at EURORDIS Summer School
- Participating in the Joint Action on Pharmacovigilance Advisory Committee (SCOPE)

#### **Develop activities within the Drug Information, Transparency and Access Task Force**

- Launch new survey 'Tell us how you take your treatment', including off-label use
- Prepare possible extension of the survey on off-label use in rare diseases to all other EU Member States
- Continue the activities in parallel to the workplan of the PCWP
- Liaise with EMA and heads of medical agencies for more transparency of compassionate use programmes. Support the creation of a compassionate use facilitation group. Propose guidelines to companies and Member States for the organisation of compassionate use programmes. Publish a Q&A on the subject
- Maintain pages on EURORDIS website containing information on compassionate use programmes with links to the competent department in national agencies

- Explore the development of a ranking system for the healthcare industry, in order to promote best practices
- Maintain a sub-group of volunteers on specific needs for blind and vision-impaired patients
- Create online video tutorials on how to search information on main websites (EU portal on CT, database on suspected adverse drug reactions, EudraPHARM)

**Explore feasibility and resources to conduct research on the use of NATC products (Natural, Alternative, Traditional, Complementary products and also vitamins, food supplements, etc) with rare disease patients in line with our proposal for a research priority in Horizon 2020**

## PROMOTING DRUG DEVELOPMENT & ACCESS TO TREATMENTS

**Expand activities on medicines development, information and access & prepare a long term representation and leadership of EURORDIS in the EMA Committees:**

- Participate in the EMA Committee for Orphan Medicinal Products (COMP) with two representative members and one permanent observer
- Contribute to the EMA Pharmacovigilance and Risk Assessment Committee (PRAC) as external experts for rare disease therapies
- Participate in the EMA Paediatric Committee (PDCO) with one representative member and one alternate and one observer
- Participate in the EMA Committee for Advanced Therapies (CAT) with one representative member, one alternate and one observer
- Participate in EMA Human Scientific Committees' Working Party with Patients' and Consumers' Organisations (more commonly known as the Patients' and Consumers' Working Party or PCWP), with one representative member, one alternate and one observer
- Mentoring patients on the CHMP procedure for oral explanations used for risk/ benefit assessment at the time of marketing authorisation
- Participate in EMA user testing group of the European Clinical Trials Register (EUCTR), European Database for Suspected Adverse Drug Reactions and European Database of Authorised Medicines
- Consultation on new EMA intranet for delegates
- Identify and support patient experts for protocol assistance/scientific advice, scientific advisory groups of CHMP
- Support EURORDIS patient representatives in EMA Scientific Committees and Working Parties with the EURORDIS Therapeutic Action Group (TAG) via monthly conference calls and sharing information, agendas, reports, providing mutual support and by discussing main issues

- Participation of patients in EMA multiHTA parallel scientific advice and SEED (Shaping European Early Decision)
- Review all orphan drug designation applications and protocol assistance dossiers. Review of designation criteria at the time of marketing authorisation and reports on significant benefit, paediatric investigation plans for rare diseases, including waivers and deferrals and advanced therapy (gene, cell and tissue engineering) applications
- Review and validate all public Information on rare disease therapies disseminated by EMA at the time of designation (PSOs) and marketing authorisation (EPARs, Package Leaflets, Significant Benefit Public Reports)
- Take action to enhance collaboration between EMA and FDA on rare therapy development beyond orphan drug designation

**Investigate and propose solutions to European and national medicines supply shortages:**

- Lead the group drafting a common position of patients' and healthcare professionals' organisations on medicines supply shortages. Create a European/ international coalition of interested parties
- Advocate for an improved European coordination of medicines supply shortages, due to both technical and economic factors
- Pursue action to include medicine supply shortages on the agenda of Member States

**Explore difficulties patients may have in accessing medicines or other types of medical care:**

- Maintain and further develop the Access Campaign to collect feedback from national alliances, European federations, DITA task force members and patients at large on difficulties in accessing treatments in the context of the economic and financial crisis
- Develop a report and recommendations based on the results of the Access Campaign survey to be available on EURORDIS website and widely disseminated to relevant stakeholders
- Plan and take appropriate action for the future organisation of the 6th EURORDIS Survey on Access to Orphan Drugs in the European Union

**Engage in Health Technology Assessment (HTA) activities & prepare long-term representation and leadership of EURORDIS in HTA:**

- Participate in the newly established permanent European HTA Network as member and co-chair of their Stakeholders' Forum (implementation of the EU Directive on Patients' Rights in Cross-Border Care).
- Promote the engagement of patient representatives in its strategic vision, policies and guidelines, methodological approaches and assessment procedures, particularly to address rare diseases challenges
- Support and advise the dialogue of national alliances with HTA Agencies

### Capacity building of our members and volunteers on clinical trials, drug development and EU regulatory processes:

- Organise EURORDIS ExPRESS Summer School 2015 in Barcelona in June with a new format to train 25–30 new patient advocates representing a diversity of diseases and geographical locations
- Collaborate with COST to incorporate 10–15 researchers in the new format of the EURORDIS Summer School
- Collaborate with ECRIN to incorporate 10 non-rare disease patient representatives from the therapeutic indication areas of nutrition and medical devices, in addition to rare diseases
- Continue to improve and contribute to the Training Resources section of the EURORDIS website that currently provides access to all presentations from the Summer School in downloadable pdf form, as well as video recordings of the speakers. Online training tools include a preparatory section for the Summer School and the e-learning platform for continued evaluation of learning
- Take part in the DIA EuroMeeting 2015 Paris in April with speakers, session chairs, contribution to programme committee, a DIA Patient Fellowship Programme for approximately 40 fellows and a patient fellows booth
- Empower our volunteers via regular information and monthly therapeutic activity Reports, call

for volunteers, support of Drug Information Transparency & Access (DITA) Task Force with more volunteers and organisation of one or two workshops of the DITA Task Force annually

- Participate in the 'European Patients' Academy on Therapeutic Innovation' (EUPATI) from 2012 to 2016, an IMI-funded project gathering a consortium led by European Patients' Forum (EPF) with other European umbrella patient organisations
- Creation of material for training of audiences in the EUPATI. Training material will be freely available on the EUPATI website
- EURORDIS is also responsible for delivery of the EUPATI face-to-face events when required

### Promote dialogue with pharmaceutical & biotech companies involved in rare disease therapy development:

- EURORDIS Round Table of Companies: consolidate membership and organise two workshops in 2015: one in Brussels in February on 'Rare Diseases: Going Global!' and the second in Barcelona in September on 'Patient Relevant Outcomes and Patient Reported Outcomes'
- Expand direct dialogue with pharmaceutical and biotech companies
- Strengthen the dialogue with EFPIA–EuropaBio Task Force on Orphan Drugs

## Cross-Cutting Priorities

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### Implement EURORDIS Strategy 2010–2015:

- Improve planning and anticipation of major EURORDIS activities such as ECRDs, Membership Meeting, Rare Disease Day, EURORDIS Round Table of Companies Workshops, RareConnect, EURORDIS Patient Voices, major advocacy campaign and new projects
- Develop the Strategy 2015–2020 in 2015 and adopt at Board Meeting July 2015
- Continue collection of EURORDIS Indicators and analysis

### Develop EURORDIS strategic partnerships:

- Maintain strategic partnership with NORD (USA), renewed for 2014–2019
- Maintain a partnership with CORD (Canada), the Japanese Patients Association – JPA (Japan) and establish partnerships with Rare Voices Australia, the Russian Patients Union and Chinese Organisation for Rare Diseases, in addition to being open to new partnerships with other international patient organisations
- Develop strategic partnership with ORPHANET
- Maintain partnership with DIA–Europe
- Implement strategic partnership with the European Society for Human Genetics, EuroGentest,

International Society of Social Workers, European Institute Women's Health, International Society For Pharmacoeconomics and Outcomes Research ISPOR, Health Technology Assessment International (HTAi), European Society of Medical Oncology (ESMO), European Hospital and Healthcare Federation (HOPE), etc

- Consolidate partnership with the members of the EURORDIS Round Table of Companies

### Maintain and support the EURORDIS Panel of Experts composed of expert individuals from a range of diverse backgrounds sharing the mission and values of EURORDIS, in order to provide, when needed, a comprehensive, multidisciplinary approach to complex issues

### Create EURORDIS International Circle of Ambassadors aiming to bring together community leaders from Europe and North America to promote rare diseases as an international cause, raise the profile of EURORDIS in North America and Europe; assist in building a donor base of individual people, foundations and corporations beyond the health sector



### **| Increase EC support to EURORDIS activities through:**

- Application 2015 for Operating Grant Specific Grant Agreement 2016
- Application for 2nd Joint Action of the Commission Expert Group on Rare Diseases
- Prepare for the 1st Joint Action of the Commission Expert Group on Rare Cancers
- Application for Public Health Project or Tender in DG Sanco Work Programme 2014–2015 as project leader or partner
- Application for a Patient-Driven Health Research Project in DG Research FP 7 Work Programme 2014 or 2015, as project leader or partner
- Study feasibility and apply for grant application to other programmes in other DGs in support of activities foreseen in EURORDIS Strategy 2015
- Application for patient-driven health research project at IMI 2

### **| Develop and diversify private funding:**

- Maintain overall support level from industry donors spread between 45 companies and a variety of activities within the confines of the EURORDIS Policy of Relationship with Commercial Companies, EMA Policy on Prevention of Conflict of Interest and EAHC rules
- Engage corporate and foundation donors beyond the pharmaceutical industry to support EURORDIS projects & actions. Priority: EURORDIS Membership Meeting; the website, eNews, eLearning, EURORDIS TV and InfoHub, all in seven languages; the RareConnect project and social media; support to European federations & networks; EURORDIS Patient Voices pilot; and Rare Diseases International
- Organise the EURORDIS Black Pearl Gala Dinner for Rare Disease Day on 24 February 2015 in Brussels and plan event for 2016.
- Launch the EURORDIS International Circle of Ambassadors, bringing together 6 – 8 community leaders from Europe and North America
- Conduct 1 – 2 electronic giving appeals to raise funds for specific projects 2015.
- Seek in-kind contributions from individuals and companies outside the health sector to help meet objectives for EURORDIS projects and actions, (ex: communications consulting, tools).

### **| Implement the Staff Strategy & Organisation & Evolution 2011–2013**

- Maintain organisation in 3 Units: Governance & Public Affairs, Operations & Knowledge Management, Finance & Support Services
- Create the position of Online Community Manager (WebRadr)
- Create the position of Patient Engagement Manager
- Create the position of Patient Voices Manager
- Create the position of Public Affairs Manager (for ADAPT-SMART project)
- Update the coordination processes: operating grant steering committee meetings (4 per year), all staff monday meetings (2 times a month), finance meeting (monthly), resource development meeting (quarterly & mini monthly), advocacy & strategy meetings (monthly), editorial meetings (monthly) and management meetings (monthly)

### **| Seek alternative human resources:**

- Seek opportunities to secure seconded staff
- Create EURORDIS internship opportunities, unpaid and paid, for up to five interns per year for periods of 1 to 6 months

### **| Maintain procedures in finance, human resources and office support services**

- Maintain decentralised structure with offices in Paris (main office), Brussels (European public affairs), Barcelona (web communications & RareConnect) and a presence in London (EMA) and Geneva (Rare Diseases International), with integrated operations through work processes, IT standards/ intranet, voice, data and internet communication
- Explore the pros and cons of a presence in Moscow
- Explore the pros and cons of expanding the RareConnect team through partnership with national alliances in Europe
- Explore the pros and cons of a presence in New York
- Reinforce the Barcelona presence with a contracted office space
- IT support: equipment, services, virtual office open to volunteers
- EURORDIS contact database management fully operational
- EURORDIS funding database

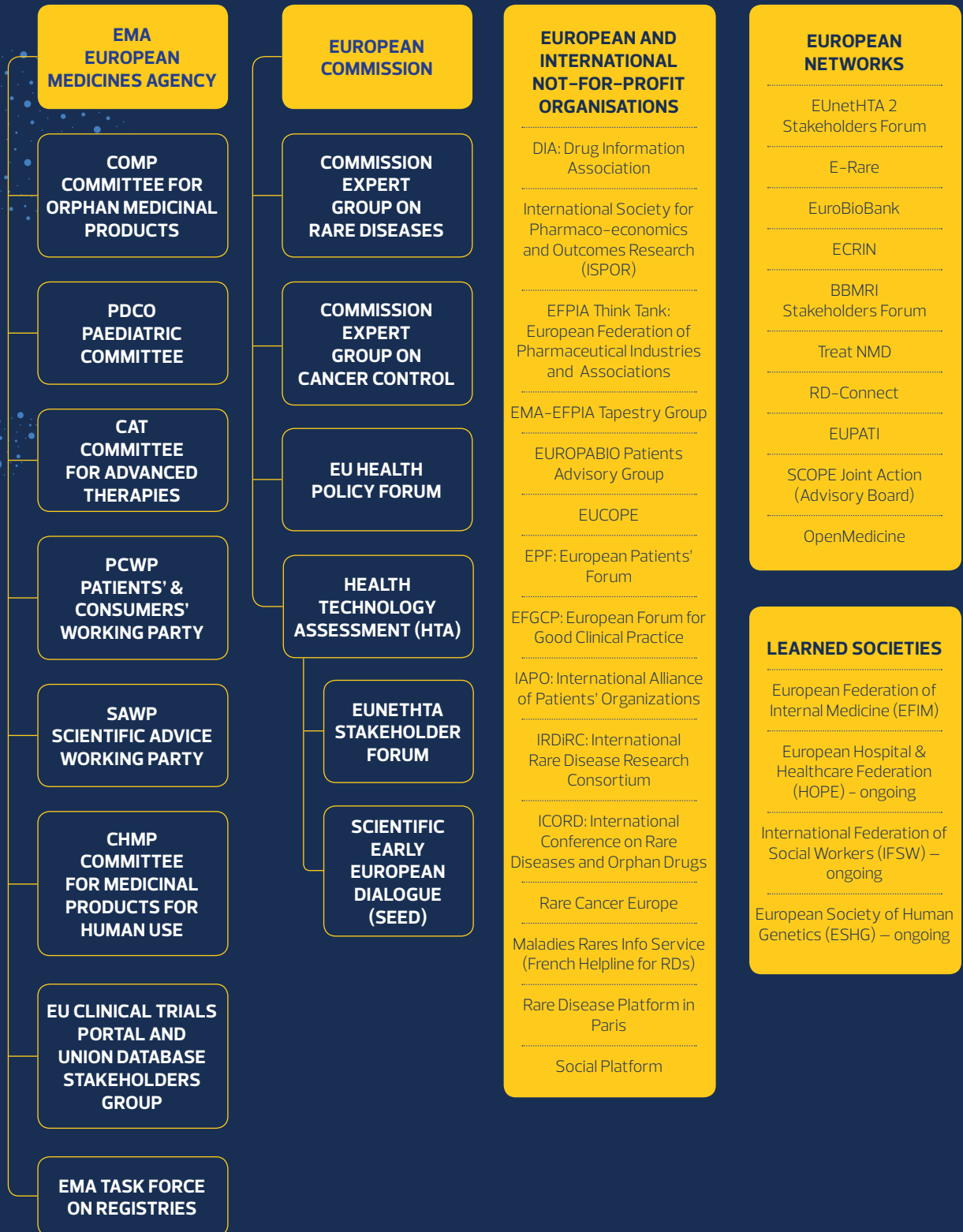
# GOVERNANCE CHART 2015



**EURORDIS**  
Rare Diseases Europe

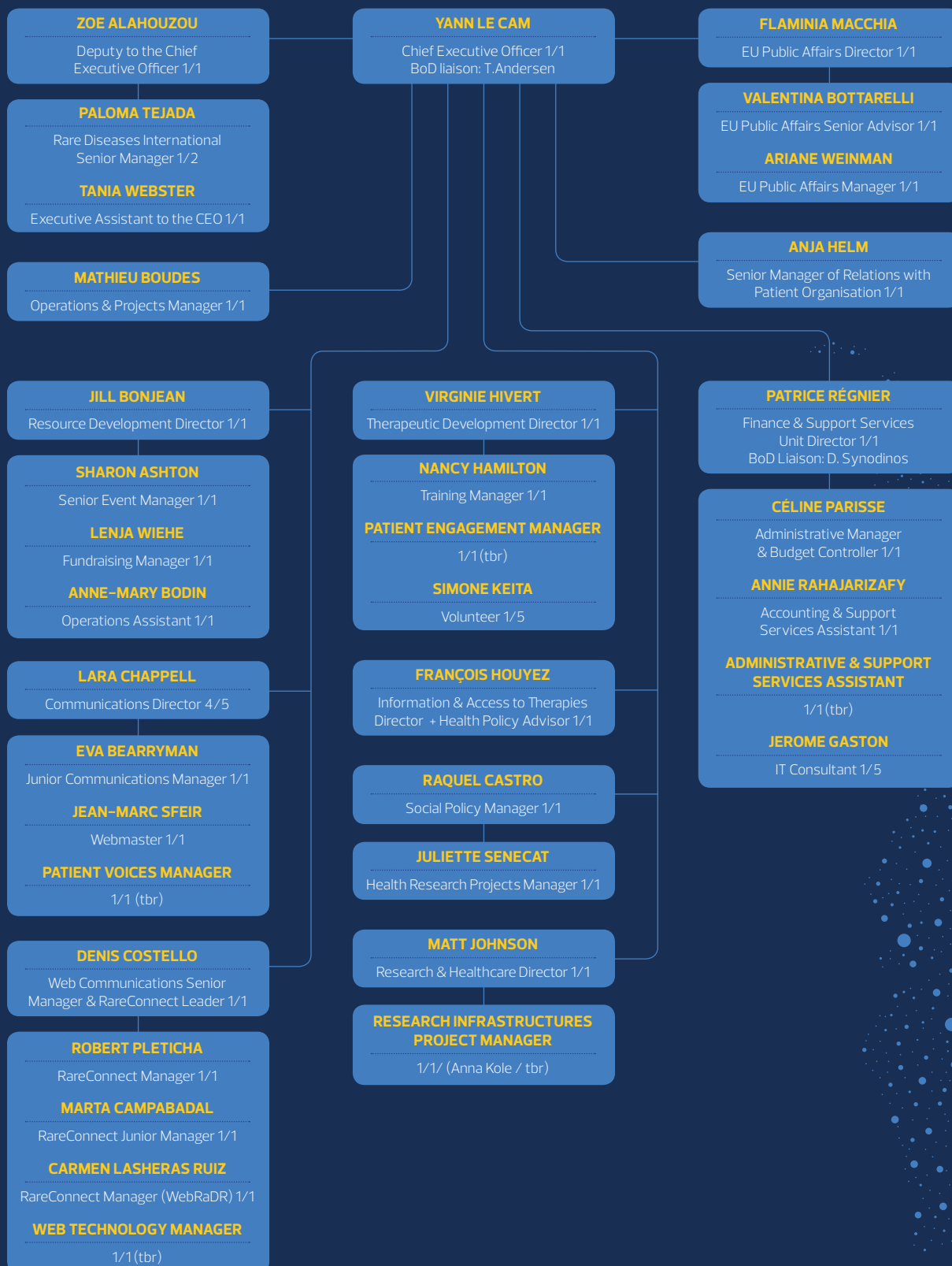


# EXTERNAL REPRESENTATION CHART 2015

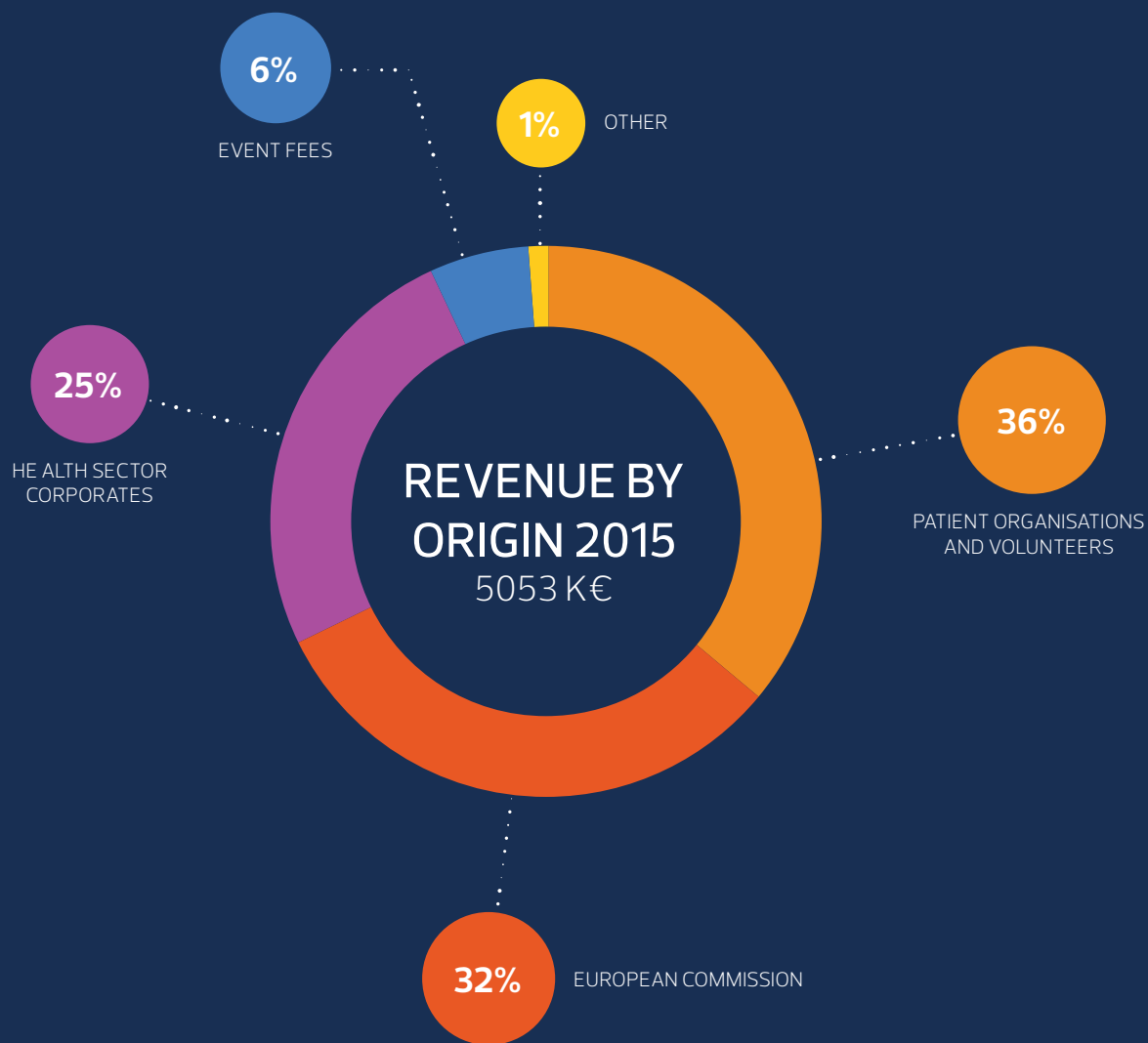


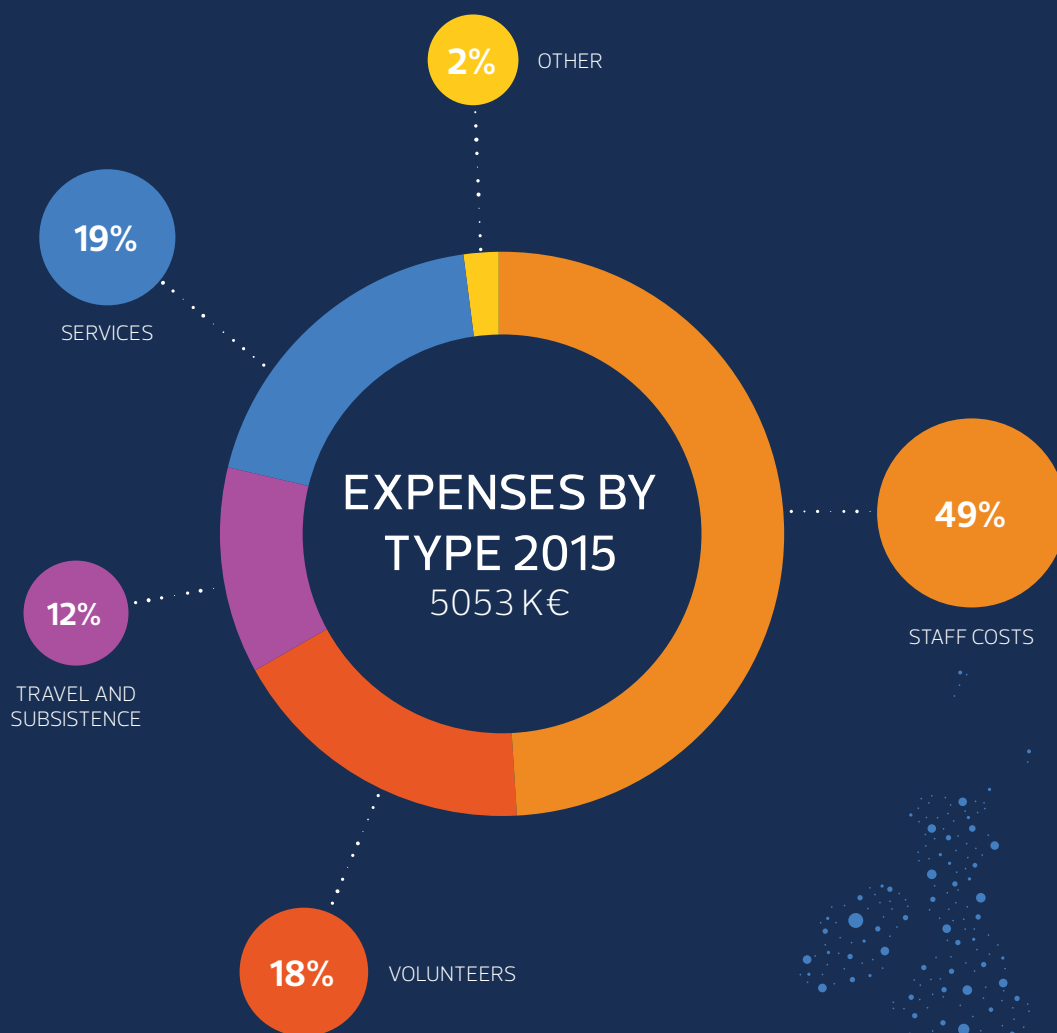


# TEAM CHART 2015



# REVENUE & EXPENSES 2015





# ACRONYMS & DEFINITIONS

## EURORDIS INTERNAL COMMITTEES & TASK FORCES

<b>BoD</b>	Board of Directors (of EURORDIS)
<b>BoO</b>	Board of Officers (of EURORDIS)
<b>CEF</b>	Council of European Federations of Rare Diseases
<b>CNA</b>	Council of National Alliances (of Rare Diseases' patient associations)
<b>DITA</b>	Drug, Information, Transparency & Access (Task Force of EURORDIS)
<b>EPAC</b>	European Public Affairs Committee (includes current and some former Board members, TAG members and Eurordis managers)
<b>ERTC</b>	EURORDIS Round Table of Companies (with pharma & biotech developing Orphan Drugs)
<b>PAG</b>	Policy Action Group (of EURORDIS) – Brings together Eurordis' representatives (mainly volunteers) of the Commission Experts Group on Rare Diseases (former EUCERD)
<b>PAG-RC</b>	Policy Action Group – Rare Cancers (of EURORDIS) – supports the volunteers on the Commission Expert Group on Rare Cancers
<b>PAC-RBP</b>	Rare-BestPractices Patient Advisory Council
<b>PAC-RD Connect</b>	RD Connect Patient Advisory Council
<b>SPAG</b>	Social Policy Advisory Group
<b>TAG</b>	Therapeutic Action Group (of EURORDIS) – Brings together Eurordis' representatives (mainly volunteers) in EMA scientific committees

## PROJECTS OF EURORDIS OR IN WHICH EURORDIS IS INVOLVED

<b>BBMRI Stakeholders' Forum</b>	Biobanking and Biomolecular Resources Research Infrastructure
<b>ECRIN</b>	European Clinical Research Infrastructures Network
<b>E-Rare</b>	Network of ten partners – public bodies, ministries and research management organisations – from eight countries, responsible for the development and management of national/regional research programs on rare diseases
<b>EunethTA Forum</b>	Support effective HTA collaboration in Europe that brings added value at the European, national and regional levels
<b>EUROBIOBANK</b>	European Network of DNA, cell and tissue banks for rare diseases
<b>EUROPLAN</b>	Fostering National Plans in Europe
<b>EURORDIS Summer School (ESS)</b>	4 day training on clinical trials for beginners. Since 2008, takes place each year in Barcelona, Spain.
<b>EUPATI</b>	Innovative Medicines Initiatives Joint Undertaking "Fostering Patient Awareness on Pharmaceutical Innovation"
<b>EJA</b>	Joint Action on Rare Diseases of the EU Committee of Experts on Rare Diseases; Funded by EC and by Member States, divided in work packages corresponding to specific activities, e.g. continuity of Europlan (Work Package 4); developing guidelines for social services dedicated to RDs (Work Package 6)
<b>EYRD</b>	European Year for Rare Diseases
<b>IRDiRC</b>	International Rare Disease Research Consortium
<b>Rare! Together</b>	Project to promote European disease-specific federations
<b>RDD</b>	Rare Disease Day
<b>RDI</b>	Rare Diseases International
<b>TREAT-NMD</b>	Translational Research in Europe – Assessment and Treatment of Neuromuscular diseases

## EURORDIS & EUROPEAN MEDICINES AGENCY (EMA)

<b>CAT</b>	Committee for Advanced Therapies – Michele Lipucci di Paola represents Eurordis
<b>CHMP</b>	Committee for Human Medicinal Products
<b>COMP</b>	Committee of Orphan Medicinal Products – Lesley Greene is Vice-Chair and Birthe Byskov Holm represents Eurordis as well – Maria Mavris is Observer
<b>PCWP</b>	Patients and Consumers Working Party – Richard Webst and François Houjé represent EURORDIS
<b>PDCO</b>	Paediatric Drugs Committee – Tsveta Schyns represents Eurordis
<b>PRAC</b>	Pharmacovigilance and Risk Assessment Committee
<b>SAWP</b>	Scientific Advice Working Party



## EUROPEAN COMMISSION

<b>CHAFEA</b>	Consumers, Health and Food Executive Agency
<b>DG Enterprise and Industry</b>	Directorate General Enterprise and Industry
<b>DG Sanco / DG Sante</b>	Directorate General Health and Consumers = DG Sanco / now Directorate General Health and Food Safety = DG Sante
<b>DG Research</b>	Directorate General Research

## EURORDIS & EUROPEAN COMMISSION

<b>Commission Experts Group on Rare Diseases (2013–2016) – CERD</b>	8 patients' representatives included 2 representatives of EURORDIS and 2 Observers
<b>EUCERD (2010–2013)</b>	EU Committee of Experts on Rare Diseases (8 Eurordis' representatives and 2 observers)
<b>EU HPF</b>	EU Health Policy Forum

## EURORDIS & NON GOVERNMENTAL PARTNERS

<b>DIA</b>	Drug Information Association
<b>CORD</b>	Canadian Organization for Rare Disorders / Chinese Organization for Rare Disorders
<b>EFGCP</b>	European Forum for Good Clinical Practices
<b>EFPIA</b>	European Federation of Pharmaceutical Industries and Associations
<b>EPF</b>	European Patients' Forum
<b>EPPOSI</b>	European Platform for Patients' Organisations, Science and Industry
<b>EUROPABIO</b>	The European Association for Bioindustries
<b>IAPO</b>	International Alliance of Patients' Organizations
<b>ICORD</b>	International Conference on Rare Diseases and Orphan Drugs
<b>INSERM</b>	French National Institute for Health and Medical Research
<b>JPA</b>	Japan Patients Association
<b>LEEM</b>	Les Entreprises du Médicament (French Pharmaceutical Companies Association)
<b>MRIS</b>	Maladies Rares Info Services (French helpline for rare diseases)
<b>NORD</b>	National Organization for Rare Disorders (USA) – Eurordis' counterpart in the US
<b>RVA</b>	Rare Voices Australia
<b>RPU</b>	Russian Patients Union

## MISCELLANEOUS

<b>CAVOMP</b>	Clinical Added Value of Orphan Medicinal Products
<b>CoE</b>	Centre of Expertise
<b>ECRD</b>	European Conference on Rare Diseases and Orphan Products
<b>ERN</b>	European Reference Network
<b>EUNRDHL</b>	EU Network for Rare Diseases Helplines
<b>HTA</b>	Health Technology Assessment
<b>MAPPS</b>	Medicine Adaptive Pathways to Patients
<b>MEP</b>	Member of the European Parliament
<b>MOCA</b>	Mechanism of Coordinated Access to orphan medicinal products
<b>EU MS</b>	Member State (of the European Union)
<b>PLWRD</b>	People Living with a Rare Disease
<b>TRP</b>	Therapeutic Recreation Programme



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