



EUROPEAN ORGANISATION FOR RARE DISEASES

ACTIVITY REPORT 2012 & WORKPLAN 2013

**THE VOICE OF
RARE DISEASE
PATIENTS IN EUROPE**

eurordis.org



EURORDIS
Rare Diseases Europe



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ACTIVITY REPORT

2012 & WORKPLAN 2013

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THE VOICE OF
RARE DISEASE
PATIENTS IN EUROPE

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"BROTHERHOOD AND SISTERHOOD, WAR AND
PEACE" EMILIA, TADAS & MARIA, MPS VI, LITHUANIA.
IMAGE OF RARE DISEASE DAY 2012.
PHOTO BY RAMUNE SLIUOZAITE.

FOREWORD

BY THE PRESIDENT
AND THE CHIEF EXECUTIVE OFFICER

In 2012, EURORDIS reached over 500 members, bringing our membership to 544 members in 49 countries (24 EU countries) and including 32 National Alliances and 36 European Federations, which overall represent more than 1500 patient organisations throughout the whole of Europe and cover over 4000 different rare diseases. EURORDIS' membership base has been growing steadily with 56 new members in 2012 only. EURORDIS has reached its initial vision of creating a Europe-wide rare disease community of patient groups and individuals able to take action on behalf of the 30 million people living with rare diseases in Europe.

New national rare disease alliances have been created in Slovakia, Czech Republic and Finland. EURORDIS is strengthening its support to Central & Eastern Europe, South-Caucasia and Russia with a combination of actions including patient fellowships, country visits, co-organisation and participation to local conferences, dissemination of information, exchange of experience and advice on their strategy to promote rare diseases as a public health priority in their countries. ECRD 2012 Brussels was interpreted in Russian and the Russian version of the EURORDIS eNews and Website were launched in 2012, reaching out to the as many as 250 million Russian speakers worldwide.

EURORDIS and NORD are scaling up RareConnect.org, our multilingual platform of moderated social networks of patients and families, that is provided in 5 languages and, as of April 2013, is made up of 40 international online communities which involve over 300 patient groups, 90 volunteer moderators and 4300 individual members.

EURORDIS activities to empower patient advocates included the continued improvement of its website with new sections & features, the launch of the weekly eNews in April 2012 replacing the monthly Newsletter to further engage with our community, the constant expansion of use of social media tools, the launch of EURORDIS TV in February 2013 and the consolidation of the EURORDIS Summer School as well as Online Learning Tools in the Website Training section.

EURORDIS Strategy 2010-2015 is well on track and on time. After three years of implementation, EURORDIS has reviewed its progress by the end of 2012 in order to adjust its objectives for the next three years. EURORDIS focuses on European activities having an impact on people living with rare diseases in all European countries as much as on national activities influencing the European level, while promoting international activities having an impact on patients in Europe.

Rare Disease Day further expanded internationally in 2012 with 63 countries participating and in 2013 with over 70 countries. The event continues to grow with increased local events and media outreach. Rare Disease Day has now become an essential leverage to raise public awareness and promote national policies. The theme of "Solidarity" in the field of Rare Diseases with the slogan "Rare Disorders without Borders" have reinforced the necessity of meaningful policy on rare diseases.

The 6th European Conference on Rare Diseases and Orphan Products organised in May 2012 in Brussels (ECRD 2012 Brussels) was attended by 650 participants from 45 countries

and covered topics on rare diseases and orphan products and policies and challenges at the national and European levels with an international perspective. The conference included tutorials, poster sessions and networking opportunities and was organised for the first time, with a difference format based on new partnerships involving all key stakeholders such as EUCERD, the EMA, Industry, Learned Societies, Rare Disease National Alliances in Europe and NORD, and was co-organised with the DIA. The next ECRD 2014 Berlin is already being planned for 8-10 May 2014. EURORDIS is now also partnering with NORD and DIA to co-organise an annual US Conference on Rare Diseases and Orphan Products, providing a transatlantic policy forum.

These European Conferences are coordinated with EURORDIS' intense involvement in promoting national strategies on rare diseases (eg in EUCERD activities & EUCERD Joint Action / EuroPlan) as well as in stimulating rare disease therapy development and access (eg in EMA & its Scientific Committees, EUCERD, IRDiRC).

EURORDIS and its National Alliances are organising 25 national conferences in EU Member States and across Europe in 2012 & 2013 so to promote good national measures and a dialogue between all stakeholders. ECRD 2012 Brussels and ECRD 2014 Berlin are part of a broader strategy to push toward concrete implementation of the new EU rare disease policy framework both at European and national levels across the European continent. ECRDs and these national conferences are focused on the promotion of national plans, Centers of Expertise and European Reference Networks, patient registries and biobanks, translational research from bench to the bedside, information and medical education, services to patients and their families and carers and patient empowerment.

In 2010, EURORDIS made a long-term commitment to the development of national strategies across Europe. This includes a combination of actions blossoming in 2012 & 2013 as we get closer to the 2013 deadline for all EU Member States to adopt a national plan: more intense work with the EURORDIS Council of National Alliances, use of Rare Disease Day to raise political and public awareness, new sections on our website, new EURORDIS Policy Fact Sheets. The EURORDIS Membership Meetings 2010 Athens, 2011 Amsterdam, 2012 Brussels, 2013 Dubrovnik are dedicated to building capacities of patient advocates in national plans and strategies and learning from each other.

The EU Committee of Experts on Rare Diseases (EUCERD) is central in our action. The fact that EURORDIS has 4 patient representatives and 3 alternates, one being elected Vice Chair, and is a full NGO Partner in the EUCERD Joint Action, enable EURORDIS to take full responsibilities in promoting and implementing rare disease policies as well as to engage the patient community into these processes. The EUCERD is playing an essential role in implementing the Commission Communication of 2008 and the Council Recommendation on Rare Diseases adopted in 2009. "The State of the Art of Rare Disease in Europe" is a now reference. The EUCERD provides guidance in the development of national strategies including an EUCERD Recommendation on Indicators for National Plans to be adopted in 2013. The EUCERD Recommendations on "Quality Criteria for Centres of Expertise on Rare Diseases in Member States" and "European Reference Net



YANN LE CAM
Chief Executive Officer

works on Rare Diseases" will be complemented by an "EUCERD Recommendation on Registries & Data Collection", the cornerstones of the future EU healthcare and research framework EURORDIS is promoting.

Lack of equitable access to orphan medicines across Europe continues to be a high concern aggravated by the impact of the financial and economic crisis. EURORDIS dedicates its Round Table of Companies activities and a significant part of its advocacy to this top issue in 2012 & 2013, focusing on the promotion of the Clinical Added Value of Orphan Medicinal Products (CAVOMP), Mechanism of Coordinated Access to Orphan Products and European Transparent Value Framework, Compassionate Use and Early Access Programmes, Off-label Use, Shortage of Medicines, Progressive Patients Access to Medicines (Adaptive Licencing), Patients Reported Outcomes, utility of rare disease therapies, regulatory and financial environments to stimulate rare disease therapy developments, and partnership between all stakeholders to reach these goals.

EURORDIS' participation in the European Medicines Agency's Scientific Committees (COMP, PDCO, CAT) is very intense with 386 dossiers reviewed in 2012. A EURORDIS representative is Chair of the Patients & Consumers Working Party and another one has been elected Vice Chair of the COMP. EURORDIS continues to prepare for an active role in the assessment of risk & benefits of medicines both for marketing authorisation and for pharmacovigilance. EURORDIS is training volunteers and staff to take an active part in the Health Technology Assessment of orphans and rare disease therapeutic interventions. EURORDIS is a full member of the EUnetHTA Stakeholder Forum and is commencing a collaboration with the HTAi and ISPOR.

The International Rare Disease Research Consortium (IRDiRC) has now been officially launched and is operational, and the 1st IRDiRC Conference took place in Dublin in April 2013. EURORDIS is involved in its Executive Committee, Scientific Committees and several Working Groups. EURORDIS' two important Reference Papers "Why invest in Rare Disease Research" and "Patients' Priorities and Needs for Rare Disease Research" are providing a support to shape our positions on behalf of patients. EURORDIS has also initiated a Joint Declaration with NORD and CORD on 10 Key Principles on Rare Disease Registries and a Joint Statement on IRDiRC Policy Paper.

Rare diseases are an international public health challenge and international policy convergence and collaboration can enhance European activities in research, healthcare organisation, information and therapy development. EURORDIS is actively involved in the International Conferences on Rare Diseases and Orphan Drugs (ICORD), held in Tokyo in January 2012 and Saint Petersburg in October 2013. EURORDIS will initiate in 2013 Rare Diseases International with its partners around the world as the network to speak with one global voice on behalf of people living with rare diseases.

EURORDIS was able to grow in 2012 in spite of a challenging economic environment thanks to actions to diversify its public funding and new initiatives in fund raising. The EURORDIS Black Pearl Gala Dinners 2012 and 2013 in Brussels were a success. Funding remains a critical issue to sustain our development. EURORDIS will continue to diversify and consolidate its resources in 2013.

Our community is governed by common values and modes of action. We believe in solidarity and equitability, social justice and fairness. We work through patients' empowerment and capacity building of patient advocates, exchange and mutual support, gathering of experience and networking, partnership with all stakeholders. We are audacious and innovative. We proactively collect facts and data to support the promotion of policy. We trust that facts and words shape policy and actions which result in better outcomes. We know what we want for patients and believe that a patient driven agenda has the capacity to catalyse all interested parties and support public policy.

TERKEL ANDERSEN
President



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.

EURORDIS' MISSION



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

544 member patient organisations

49 countries
(24 European Union Member States)

32 National Alliances

KEY FIGURES 2012

Over **1,000** patient groups represented

132 volunteers

Over **4,000** rare diseases represented





STRATEGIC APPROACH 2010–2015

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

1

- 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

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

2

- A Public Health priority in European countries (Member States and beyond)
- National Plans in each Member State with patient-centred 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

3 | 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

■ 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

4 | 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
- RARECONNECT™
- COMMITTEES, TASK FORCES, GROUPS



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
- **INDUSTRY:** Through EURORDIS Round Table of Companies and other initiatives
- **ACADEMIA:** For education and capacity building and for social & policy research





EURORDIS
Rare Diseases Europe

ACTIVITY REPORT

2012



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HIGHLIGHTS 2012

- **EURORDIS** celebrated reaching 500 members in 2012. With 56 new members in total, membership reached 544 members in 49 countries
- Rare Disease Day 2012 was organised in collaboration with partners in 63 countries worldwide. Over 1000 events took place and a Rare Disease Day European Symposium was organised in Brussels and attended by 120 people including representatives from the Commission, Industry, patient organisations and academia
- The 1st **EURORDIS** Awards for Outstanding Accomplishments in the field of Rare Diseases were presented during the 1st **EURORDIS** Black Pearl Gala Dinner which celebrated Rare Disease Day 2012
- The 14th Workshop of the Council of National Alliances (29 National Alliances for Rare Diseases) was held for the first time in conjunction with the 5th Workshop of the Council of European Federations (36 rare disease-specific networks). **EURORDIS** also continued the "Programme of Support to European Federations"
- The European Network of Rare Disease Help Lines officially applied for the reservation of a unique European 116 call number for rare disease help lines
- RareConnect.org continued to expand with 19 new online patient communities launched in 2012 reaching 35 global communities in collaboration with 270 patient groups and 3126 members
- The new weekly eNews replaced the monthly newsletter. **EURORDIS** eNews is circulated to almost 8000 subscribers
- The **EURORDIS** website and eNews were officially launched in Russian reaching out to as many as 250 million Russian speakers
- The 6th European Conference on Rare Diseases & Orphan Products – ECRD 2012 Brussels took place with a record 650 participants from 45 countries. The Conference was organised with a new format, partnering with the DIA & NORD to consolidate the event as the major rare disease policy forum
- **EURORDIS** continued to support the promotion of national plans on rare diseases with National Alliances through the EUCERD Joint Action Work Package 4 (EUROPLAN) facilitating the ongoing organisation of 25 EUROPLAN National Conferences including guidance for national plans, indicators as well as patient advocate capacity building, exchange and networking
- The 2nd EUCERD Recommendation on "Improving Informed Decisions based on the Clinical Added Value of Orphan Medicinal Products (CAVOMP) Information Flow" was adopted
- The EUCERD Joint Action Work Package 6 on "Specialised Social Services and integration of Rare Diseases into Social Policies and Services" was launched providing information and guidance on Respite Care Services, Therapeutic Recreation Programmes, Resource Centres and Adapted Housing Services
- Two new EUCERD Recommendations: "Rare Disease European Reference Networks" and "Common Principles and Consensus on Patient Registries and Data Collection for Rare Diseases" were prepared and are to be adopted in 2013
- The 5th **EURORDIS** Summer School for patient advocates was held in Barcelona in collaboration for the first time with the two training projects, EUPATI and ECRIN-IA. 154 patient advocates have been trained so far
- The **EURORDIS** Round Table of Companies (ERTC) counted 38 member companies and organised its 15th and 16th workshops on the benefits of partnering with patients and the value of orphan drugs respectively
- 386 dossiers on orphan drugs, advanced therapies and paediatric investigation plans were reviewed as part of participation in the European Medicines Agency's Scientific Committees
- **EURORDIS** representatives were officially appointed as members of the Governing Body and the Therapeutics Scientific Committee of International Rare Disease Research Consortium (IRDiRC)



ADVOCACY

Sustaining Rare Diseases as an EU Public Health Priority

EU Committee of Experts on Rare Diseases (EUCERD)

The seven¹ patient representatives to the **European Union Committee of Experts on Rare Diseases (EUCERD)** proposed by EURORDIS and appointed by the European Commission in 2010 continued their advocacy and advisory role to EU officials on behalf of the rare disease community in 2012, and participated in the following EUCERD meetings:

Plenary meetings:

All appointed patient representatives participated in the EUCERD plenary meetings on 26-27 January, 20-21 June and 14-15 November.

Working groups/workshops meetings:

- 19-20 November, Workshop on Genetic Testing in Ispra, Italy. Participants are preparing a report on genetic testing on offer in Europe
- 20-21 June, Informal Meeting on European Reference Networks in Luxembourg

The most important achievements of the EUCERD in 2012 was the adoption of the second EUCERD Recommendation on “Improving Informed Decisions based on the Clinical Added Value of Orphan Medicinal Products (CAVOMP) Information Flow” (September 2012). Throughout 2012, EURORDIS also contributed to the preparation of the EUCERD Recommendations on “European Reference Networks for Rare Diseases” and the EUCERD Recommendations on “Common Principles and Consensus on Patient Registries and Data Collection for Rare Diseases”, scheduled to be adopted during the first semester 2013.

The patient representatives at the EUCERD were also particularly active in their advisory role to the European Commission regarding:

- National Plans for Rare Diseases
- Rare Disease Day and the European Conference on Rare Diseases and Orphan Products
- Integration of rare diseases into several policies and services

All reports and recommendations produced by the EUCERD in 2012 are available on www.eucerd.eu

¹ Eight patient representatives were appointed to the EUCERD in 2010, amongst them Torben Grønnebæk (Rare Disorders Denmark) who sadly passed away in 2012

Rare Disease Day European Symposium, Brussels

A Rare Disease Day European event highlighting the 2012 Rare Disease Day theme of *Solidarity* was held on 29 February at the International Press Centre Residence Palace in Brussels. The one-day symposium was attended by 120 people, including representatives from the Commission, Industry, patient organisations, and academia. Keynote speakers included the Honourable First Lady of Georgia, Sandra E. Roelofs; Director General for Health and Consumers at the European Commission, Paola Testori Coggi; and Director of Health Research, DG Research, Ruxandra Draghia-Akli. Nessa Childers, MEP, delivered the opening speech.

The Symposium reflected the 2012 Rare Disease Day slogan “Rare but Strong Together” by focusing on EU solidarity in the field of rare diseases. Four European Reference Networks that successfully collaborate and pool expertise were presented: Treat NMD/Care NMD; TAG (Together Against Genodermatoses); Euro Wabb (EU registry for rare diabetes); and ENERCA (European reference network for rare anaemias).

At 12:00 (GMT) the Symposium attendees raised and joined their hands in a symbolic act demonstrating solidarity with rare disease patients around the world. The event was live streamed and broadcast via the EURORDIS website and the Rare Disease Day Facebook page.



Support to National Plans for Rare Diseases in Europe

In 2012, EURORDIS activities in support of National Plans and Strategies in Europe were focused on the implementation of the work programme of the EUCERD Joint Action for Rare Diseases (EJA) Work Package 4 “Support for the implementation of plans or strategies at MS level” (EUROPLAN) in which EURORDIS facilitates the organisation of 20 EUROPLAN National Conferences in the European Union as well as 5 other national conferences in European countries outside the EU. Following the same methodology used in the first EUROPLAN project, the conferences are organised by National Alliances and involve all relevant stakeholders. A team of 10 Advisors, all from National Alliances, support the staff in this endeavour and are in charge of advising the preparation of 2 to 3 conferences.

Organisation of National Conferences

In 2012 EURORDIS launched the Call for Expression of Interest for the organisation of National Conferences.

Nineteen National Alliances (NAs) and patient groups were selected to organise EUROPLAN National Conferences on RDs in the EU27, out of which 6 National Alliances are organising a EUROPLAN National Conference for the first time. The other National Alliances were involved in the EU DG SANCO co-funded project “EUROPLAN” (2008-2011) and plan to organise a second EUROPLAN conference in order to further consolidate relations and actions with policy decision makers towards the implementation of National Plans in their country.

Contacts were held with member patient groups and National Alliances in the EU in order to identify the partner for the organisation of the 20th EUROPLAN National Conference. In January 2013, it was agreed with the Lithuanian representative at the EUCERD that a EUROPLAN Conference on National Plans on Rare Diseases will be held during the EU Presidency of Lithuania together with Latvia and Estonia, open to more Member States in the region. Furthermore, 5 additional National Alliances have been selected from

non EU countries to organise a EUROPLAN National Conference in their own countries: Georgia, Russia, Serbia, Switzerland and Ukraine.

Two first EUROPLAN conferences were held in 2012, in Sweden and in Greece.

Additionally, a set of guiding tools and documents organised along the thematic structure of the EU main documents, were developed or updated. All documents have been collected in an “Info Pack” for Conference organisers and available on the EURORDIS website in the section: “National Rare Disease Policy”: www.eurordis.org.

Guidelines for Conference Organisers and Content Guidelines

Throughout the year 2012, EURORDIS developed the following essential documents of the “Info Pack” and disseminated them to conference organisers: a Common Format of National Conferences; 6 Content Guidelines for the 6 thematic workshops of each national conference; 6 template PowerPoint presentations for the 6 thematic workshops. The above were finalised and disseminated to conference organisers in 2012.

The first draft of the following documents was also developed:

- Guidelines for Chairs and Rapporteurs
- Template of the Final Report

The “Content Guideline” of this Info Pack is based on the essential EU policy documents, including EUCERD Recommendations and EUROPLAN documents. It will be reflected in the layout of the 6 workshops of each National Conference. The process of analysing the above material and compiling the guidance documents for National Conferences required

several months of work, before the conferences commenced. It involved mainly EURORDIS staff, with extensive consultations with volunteers, EUROPLAN-EURORDIS Advisors and Conference organisers, as well as EJA partners.

Selection of EUROPLAN Indicators for EUROPLAN National Conferences

Although not originally planned, EURORDIS and its 10 Advisors started in 2012 a process of selection of core common indicators for RD National Plans from the list of EUROPLAN Indicators (developed in the EUROPLAN project in 2010), with the aim to include them in the conference material. The selected core Indicators are meant to be utilised by Member States for data collection when monitoring the implementation of National Plans or Strategies on Rare Diseases. On the basis of common criteria, each Advisor reviewed the full list of EUROPLAN Indicators with the support of their own National Alliance and at least one contact person with experience in the area of health planning/rare diseases at national level. This approach will lead to the selection of core common indicators for RD national plans and strategies and will cross over with the Delphi method applied by the WP4 leader, the Italian Istituto Superiore di Sanità (ISS), to select core indicators in an effort to support EU Member State administrators.

The EUCERD Joint Action for Rare Diseases (EJA) brings together Member States (public institutions) and EURORDIS as the full partner NGO and is funded by the European Commission (60%), Member States and EURORDIS. Work Package 6 is led by Domenica Taruscio of the ISS.

Rare Diseases: An International Public Health Priority

EURORDIS continued to work on the “International Paper” in 2012 with the objectives to: 1. directly promote rare diseases as an international public health priority towards relevant institutions at international level; 2. provide an advocacy tool for patient groups to advocate towards their national authorities thereby serving as a basis for patient empowerment locally; and 3. enhance international cooperation in the field of rare diseases.

After a meeting in July 2012 with the Chair of IAPO (the International Alliance of Patients’ Organisations), Dr Durhane Wong-Rieger, and IAPO’s public affairs staff, it was decided to hold the adoption and publication

of the Joint Declaration and launch a survey within Europe, through EURORDIS membership, the National Alliances and the European Federations, as well as in other regions of the world. This survey will sharpen the main messages of the Joint Declaration and will increase ownership of the declaration by patient groups around the world.

The Strategic Partnerships between EURORDIS and the US National Organization for Rare Disorders (NORD) continued to play a key role in promoting rare diseases as a public health priority with notable areas of collaboration in 2012 being: the joint EURORDIS-NORD online patient community portal,

RareConnect™, which in 2012 launched 19 new patient communities online; the organisation of the second US Conference on Rare Diseases and Orphan Products in Washington (October 2012); partnering for the European Conference on Rare Diseases and Orphan Products in Brussels in May 2012 and May 2014 in Berlin; and the first EURORDIS “Black Pearl” Gala Dinner which was a resounding success.

In October 2012, an official partnership was sealed between EURORDIS and the Canadian Organization for Rare Disorders (CORD) to further enhance our collaboration on international advocacy activities especially in the following areas: Rare Disease Day, with CORD taking the lead in Canada; Support to national plans for rare diseases; Advocacy coordination and possible co-signing of Joint Position Papers together with NORD; and Collaboration on Conferences, such as the European Conference on Rare Diseases and Orphan Medicinal Products.

These official partnerships have worked to increase coordination and common action by the three organisations on advocacy actions and joint projects at an international level. In 2012, this included, at the initiative of EURORDIS, the adoption of a EURORDIS-NORD-CORD Joint Declaration on 10 Key Principles of Rare Disease Patient Registries, and a Joint Contribution to the Policy Paper of the International Rare Diseases Research Consortium.



Improving Access to Orphan Drugs

Clinical Added Value of Orphan Medicinal Products (CAVOMP)

Advocacy activities continued in 2012 in view of the creation of a mechanism of exchange of information and common assessment on the Clinical Added Value of Orphan Medicinal Products (CAVOMP, previously known as CAVOD) at EU level. EURORDIS' work in 2012 was as intense as in 2011, including liaising with high officials in the European Commission, requesting meetings on a regular basis in Brussels, liaising with the European Medicines Agency (EMA), National Authorities, National Health Technology Assessment Bodies, as well as pharmaceutical companies; EURORDIS also offered a platform for discussion at the EURORDIS Round Table of Companies on Improving Access to OMPs. This intense advocacy work has progressively led to the

creation of a consensus on the CAVOMP and to the unanimous adoption (26 out of 27 Member States and 1 abstention) of the EUCERD recommendation on the CAVOMP in September 2012.

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. After contributing in 2011 to the definition of key concepts, glossary, prevalence of therapeutic indication and innovative tools for discussions on pricing, EURORDIS has been



involved as the stakeholder representing patients (on behalf of the European Patients' Forum - EPF) in the Working Group on the MoCA throughout 2012, participating in all meetings and conference calls, drafting the Work Package 3 on Individual access, contributing to the other two Work Packages, as well as to the recommendations and Final Report, and liaising with the European Commission and other stakeholders, both within and outside the Working Group.

Differential Pricing

EURORDIS participated in a series of Discussions on the subject of "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. EURORDIS actively participated, bringing in the patients' perspective, in the discussions around the "non-paper" on "Is differential pricing a way forward for better access to pharmaceuticals in Europe?", while liaising with different stakeholders on this approach to tackle inequalities of access to medicinal products, within and beyond Europe. EURORDIS has been instrumental in the reflection process - still going on

at EU level – on the differential pricing approach as a way to decrease inequalities in access for patients to medicinal products. The system to be established at EU level is still being discussed and defined but could lead to the creation of some clusters of countries, by level of GDP and population, to which the same pharmaceutical product would be sold at different price levels, according to the needs and ability to pay of the national health budgets. This approach would respond to the principle of distributive justice while opening to the industry the poorest markets, which would otherwise remain closed. The differential pricing system could not co-exist with the External Reference Pricing system, by which national decisions are often based on the lowest price decided by a previous country. EURORDIS' proposal consists in proposing that the "reference price" from which to differentiate could usefully be the price as defined through the MoCA process, namely through the value-pricing system based on the following three main elements: the European Transparent Value Framework, the volume (prevalence of the therapeutic indication) and the agreed post-marketing research activities.

Adaptive Licensing

In 2012, EURORDIS has been, and will continue to be in the coming years, instrumental in the elaboration of the concept and piloting of “adaptive licensing” 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) on this subject and has been given a platform for presenting the concept at the EURORDIS Round Table of Companies. EURORDIS has now engaged a

dialogue with several companies in orphan medicinal products as well as with the European Federation of Pharmaceutical Industries and Associations (EFPIA) to further explore the feasibility of this initiative within the current legislation or, potentially, at the time of the next revision of the EU Pharmaceutical Legislation.

Improving Access to Quality Care

Promoting Cross-Border Healthcare and Mobility of Patients

The First phase of the long-lasting advocacy activities on cross-border healthcare and patient mobility has been achieved with the adoption of the EU Directive on Patients' Rights in Cross-Border Healthcare in April 2011. EURORDIS has been instrumental in placing the focus of the Cross-Border Healthcare Directive on rare disease patients and the specificities of rare diseases. 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.

Throughout 2012, EURORDIS has kept up the pressure on the Second phase of its Advocacy activities: the implementation of the Directive for the maximum benefit of rare disease patients. A whole Theme of the European Conference on Rare Diseases in May 2012 was dedicated to the implementation of the Cross-border Healthcare Directive. To help EURORDIS members and National Alliances best advocate for patients' rights for the transposition of the Directive on Cross-border Healthcare, a Questions and Answers document was prepared and translated in 7 languages (Bulgarian, English, French, German, Greek, Italian and Spanish). This document was prepared in consultation with lawyers and experts in the field, to provide advocates with an effective tool when meeting with their authorities.

The transposition of the Directive into national legislation will end in 2013 and EURORDIS will remain vigilant in monitoring the actual functioning of rare disease patients' mobility.



European Reference Networks (ERN)

Within the framework of both the EU Directive on Patients' Rights in Cross-Border Healthcare and the reflection process that commenced in 2005 on Centres of Expertise and European Reference Networks (ERN), EURORDIS has contributed substantially, from the very beginning, to the development of the process leading to the finalisation of the EUCERD Recommendation on ERN at the end of December 2012 and its adoption in early 2013.

EURORDIS has put forward throughout 2012, and in different fora, its patient-centred vision of the overall system to be achieved at EU level, aimed at establishing a limited number of ERN, covering all rare diseases, gathered by therapeutic areas, starting from the pilot networks and widening in a step-wise approach to the most mature centres wishing to establish a reference network at European level. The overarching goal of the whole system is, in EURORDIS' opinion, to reach an overall system where no rare disease patient will be left outside and each patient will “find a home”, including undiagnosed patients.

Other Issues

Revision of the EU Clinical Trials Directive

Following on the work carried out by EURORDIS in 2011, the advocacy work on the revision of this Directive has continued and intensified in 2012 through liaising with individual MEPs, participating in conferences and meetings, presenting the rare disease community needs and expectations in the field of clinical trials.

The focus of EURORDIS advocacy activities is mainly: the need for gathering the scarce existing expertise on rare diseases at EU level and ensuring that this limited knowledge reaches the Reporting Member States, including the aspects that are left to the national level, in order to deliver a well-informed opinion on the authorisation or not of the clinical trial in question.

This intense advocacy by EURORDIS has led a group of MEPs to co-sign a specific amendment on clinical trials in rare diseases to be presented to the Environment, Public Health and Food Safety Committee and if adopted at Committee level, to be voted in plenary.

Revision of the Data Protection Directive

The Revision of the Data Protection Directive has been identified as a key issue with high rare disease specificity. Therefore, EURORDIS has been carrying out an internal reflection process on this revision, in particular in relation to patient registries and mobility of data while protecting patients' privacy.

This reflection, undertaken together with other relevant groups such as the EPIRARE project (the European Platform for Rare Disease Registries), has led to the definition of a number of amendments that were sent to the relevant Members of the European Parliament. After the publication of the Report by MEP Jan Philipp Albrecht, the need for an urgent Statement in favour of striking the right balance between data protection and protection of research activities in Europe was discerned.

Transparency Directive

The Transparency Directive was first adopted in 1989. This important legislation governs the time limits for Member States/Health service coverage authorities and the holder of a marketing authorisation to agree on a price and/or the reimbursement/coverage

of medicinal products. It also delineates how the process should be transparent.

However, as revealed by the EURORDIS surveys on Price and Availability of Orphan Medicines, the time limits are rarely respected, and many patients struggle for access for long periods of time.

As the European Court of Justice had to judge several cases, and as public health is suffering from such delays, the Commission took the initiative to revise the rules and adopted, in March 2012, a proposal for a Revision of the Transparency Directive. In June 2012, EURORDIS sent a letter to MEP Ms Antonia Parvanova, the Rapporteur from the European Parliament in charge of this file, thereby proposing a number of amendments aimed at the following objectives:

- Preventing the “post-code lottery”: the regionalisation of healthcare can prevent patients in some regions within the same Member State from accessing a medicine reimbursed in another region of their own country
- Demanding from marketing authorisation holders a greater transparency on the cost structure of the medicine
- Clarifying the terms “marketing of a medicine” and “placing on the market of a medicine”
- Preventing treatment interruption for an effective treatment obtained via a compassionate programme prior to a marketing authorisation due to the post-marketing authorisation processes for pricing and reimbursement
- Making the list of medicines included in the health insurance system transparent and accessible by the public, as well as their prices

In November 2012, EURORDIS came back to the Rapporteur with some further elements of reflection in order to ensure overall coherence between the different mechanisms and instruments being discussed at EU level but in different fora, all aimed at improving access to medicinal products for patients. As EURORDIS' representatives are often the ones participating in all the on-going initiatives, it is also an important responsibility to keep in mind and respect the “full picture”.

Visit http://ec.europa.eu/enterprise/sectors/healthcare/competitiveness/pricing-reimbursement/transparency/index_en.htm to learn more.

Conflict of Interest at the European Medicines Agency (EMA)

During 2012, the issue of the roles and mandates of patient representatives within the Scientific Committees of the European Medicines Agency, as well as the clarification of their nomination and conflict of interest requirements, became a matter of priority for the efficiency of their activities in these committees. Together with other organisations, EURORDIS participated in the shaping of the overall position of patient and consumer groups working as experts within the EMA Committees, and also developed a specific contribution to the debate from the rare disease patient perspective. After meeting with Guido Rasi, the Executive Director of the EMA, in London in July 2012, EURORDIS elaborated a letter explaining the challenges faced by patient representatives and the necessary adjustments that should be made to the current EMA policy on Conflict of Interest in order to ensure substantial contribution of patient experts at the EMA. This process will continue in 2013.



The EU Public Health Programme 2014-2020: “Health for Growth”

In May 2012, EURORDIS proposed specific Rare Disease amendments in order to demand for appropriate funding to support actions contributing to the objectives of the Commission Communication on Rare Diseases: Europe’s Challenges and the Council Recommendation on an action in the field of Rare Diseases, including (but not only) for the creation of European Reference Networks, information and registries. These amendments were accepted and tabled by MEP Christian Busoi.

>> The legislative process has been stalled due to major disagreements amongst Member States concerning the overall budget of the European Union and therefore also on the envelopes for specific programmes.

The EU Research Programme 2014-2020: “Horizon 2020”

In June 2012, EURORDIS proposed several amendments to the Carvalho Report on the Implementation of the Regulation for the establishment of Horizon 2020, the new EU Research Framework Programme for 2014-2020. The most important amendments were accepted and tabled by MEPs Françoise Grossetête and Frédérique Ries. These amendments have been adopted and incorporated into one long amendment resulting from the discussions and the agreement amongst the main political groups. With the adoption of these amendments, the original Commission proposal on Horizon 2020 is more aligned with EURORDIS’ priorities on rare disease research, as well as with the IRDiRC objectives.

>> The legislative process has been stalled due to major disagreements amongst Member States concerning the overall budget of the European Union and therefore also on the envelopes for the specific programmes.

Personalised Medicines

EURORDIS has been following the creation and development of the European Alliance on Personalised Medicines (EAPM) with interest, given that personalised medicines represent an emerging area in therapeutics. EURORDIS currently participates in the discussions and meetings organised by the EAPM with the status of observer. This topic will certainly become an increasing priority in the coming months and years.





INFORMATION & NETWORKING

Membership Base – 544 Members

56 new members joined in 2012 (34 full members and 22 associate members). New countries represented: Belarus, Macedonia, South Africa.

At the end of 2012, EURORDIS had 544 members in 49 countries, 36 of which are European countries, 24 being members of the European Union.

EURORDIS Membership Meeting 2012, Brussels

In 2012 the EURORDIS Membership Meeting took place in Brussels on 23 May. This one-day meeting took place back-to-back with the European Conference on Rare Diseases & Orphan Products (ECRD). The meeting included the General Assembly of EURORDIS, which approved the Activity report and Financial Report 2011, as well as the Action Plan & Budget 2012. Four new Board members were elected.

Following the General Assembly, 6 Learning from Each Other Forums and 6 Capacity building Workshops took place, allowing patient representatives to exchange experiences and learn about topics such as the Directive on Cross Border Health Care, Centres of Expertise, Registries, and Compassionate Use Programmes for Orphan Drugs.

Council of National Alliances

EURORDIS supports a network of 32 National Alliances for Rare Diseases, of which 29 constitute the Council of National Alliances (CNA). Three new National Alliances joined the Network in 2012 from the Czech Republic, Finland and Slovakia.

The CNA's main activities in 2012 included the work on National Plans for Rare Diseases, notably in the framework of the EUCERD Joint Action on Rare Diseases, as well as the preparation and coordination of Rare Disease Day 2013.

In 2012, one CNA Workshop took place, which for the first time was held partly in common with the Council of European Federations (CEF), to allow cross-cutting discussions on common issues.

The first day of the workshop gathered 28 participants and focused on the following topics:

- National Plans for Rare Diseases
- Overview of National Plans

- Guidance to organise EUROPLAN 2012-2015 national conferences
- National initiatives to support the common 116 rare disease helpline number
- Rare Disease Day 2013: preparation

The second day, held together with the CEF, gathered 40 participants and focused on:

- The creation of an Interest Group for translational research
- Access to treatment in the context of the financial crisis
- Specialised Social Services
- Results of the EURORDIS Off Label survey
- Cross-Border Healthcare: State of implementation at national level



Council of European Federations & Networks

For the first time, part of the meeting took place in conjunction with the CNA (Council of National Alliances) to discuss cross-cutting issues and share experience. **This part of the meeting focused on:**

- The creation of an Interest Group for translational research
- Access to treatment on the context of the crisis
- Specialised Social Services
- Results of the EURORDIS Off Label survey
- Cross Border Healthcare – State of implementation at national level

The second part of the meeting was dedicated to:

- European Reference Networks
- Support to European RD Federations
- RareConnect™ for European RD Federations

- The next day, the representatives participated in a one-day capacity building session entitled "Research coordination & promotion"

For the third year, EURORDIS continued the programme "Support to European Rare Disease Federations", which aims at supporting the smallest and/or youngest organisations facing difficulties in financing their network meetings (Board meetings, Network meetings, Conferences etc). EURORDIS granted a total of 27400 € in 2012 to 15 European RD Federations to help them organise their different meetings.

Find more information at:
 raretogether.eurordis.org

Rare!Together

Rare!Together aims at helping in the creation, operation and management of European Rare Disease Federations, in particular through the website <http://raretogether.eurordis.org>. This website is continuously updated in order to remain a good practice reference handbook and toolkit for existing European Federations and for patient organisations planning to set up their European Federation.

The mentoring aspect of Rare!Together continued with three young Federations, through regular advice and support via telephone, e-mails and participation in workshops and meetings.



European Network of Rare Disease Help Lines

In July 2012, EURORDIS, on behalf of the Network of Rare Diseases Help Lines, wrote to the European Commission DG Connect to reserve a "116" unique call number for rare disease help lines that operate in Europe. Five services of social interest already exist, such as a "116" number for missing children, and to increase the visibility of help lines the network proposed to submit this request. The decision should be made in 2013 by the COCOM, the Communication Committee.

A fifth Caller Profile Analysis was conducted in November 2012: 12 help lines participated, for a total of 1676 calls or emails received, which addressed 474 distinct rare diseases. In average the time needed to respond was 23 minutes (median 15) with one-fourth of the enquiries requiring more than 25 minutes to respond.

Rare Disease Online Communities



RareConnect™, the online patient community portal rareconnect.org continues to grow. Launched in 2010 by EURORDIS in partnership with the US National Organization for Rare Disorders (NORD), the project has helped patients and families connect through an online social network and support each other and share vital experiences on aspects of living with a rare disease. RareConnect™ is a multilingual platform in 5 languages (English, French, German, Italian, and Spanish). Organised into disease-specific communities, this platform also provides links to quality information and involves patient associations in the 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.

By the end of 2012, RareConnect™ successfully created 35 disease-specific online communities. These communities saw international partnerships emerge from 277 different patient organisations who formally partook in the setup and governance of each community.

During 2012 the RareConnect™ website received over 140 000 visits from 189 countries which led to 3500 patients or caregivers signing-up to participate in the service.

Of the 35 live communities, 19 were launched during 2012 for the following diseases:

- Dravet syndrome
- Ehlers-Danlos syndrome
- Moebius syndrome
- Paraneoplastic Neurological syndromes

- Pulmonary hypertension
- Narcolepsy
- Multiple System Atrophy
- Trimethylaminuria
- Rett Syndrome
- DysNet
- Neuroacanthocytosis
- Alstrom Syndrome
- Evans Syndrome
- Mastocytosis and Mast Cell Activation Disorders
- Fibromuscular Dysplasia
- Coats Disease
- Lipoprotein Lipase Deficiency
- WHIM Syndrome
- Cryoglobulinemia

The RareConnect™ network is also composed of 100 moderators nominated by the various participating patient groups. 6 capacity-building webinars took place during the year in order to train and empower moderators in the governance and management of a successful online patient community. Topics discussed during these webinars included moderation best-practices, updates to the platform, updates to the RareConnect™ Charter for online communities, and training in the use of other Social Media tools to promote RareConnect™. In May, in Brussels, a workshop was dedicated to the RareConnect™ project and social media use and was included as a satellite workshop in the EURORDIS Membership Meeting. In September, EURORDIS hired a full-time RareConnect™ Junior Manager to join the RareConnect™ team in order to support its growth.



Find more information at:

rareconnect.org



EURORDIS Website

The EURORDIS website "eurordis.org" continued to sustain increased visitor activity throughout 2012. The total number of website visitors increased by 10% in 2012 compared to 2011. There were 249,721 visits from over 190 countries throughout the year. The contents of the website became fully available in Russian, bringing the total number of languages of the EURORDIS website to seven: English, French, German, Italian, Portuguese, Russian and Spanish. The number of visitors increased in all seven language sections.

A new section was added to the EURORDIS website in 2012. The Specialised Social Services section provides a list of identified services in Europe, and will be updated regularly to provide information and developments relating to these services for rare diseases.

Find more information at:

eurordis.org

The website was also tweaked to improve access to information. Certain key content was reorganised to improve visibility and a scrolling tab was added to the homepage to allow easier access to the recent eNews lead stories.



EURORDIS eNews

In April 2012, the EURORDIS newsletter evolved from a monthly to weekly format, allowing us to communicate breaking news of interest to the rare disease community more rapidly and frequently. Each weekly eNews features an in-depth, lead article devoted to relevant topic areas, including policy developments relating to rare diseases and/or orphan medicinal products, advances in European national rare disease plans, updates in orphan medicine regulation, Rare Disease Day, EURORDIS' involvement in national and European-level projects, and other EURORDIS initiatives, activities, and new services. Other regular sections of the eNews allow us to report frequently on developments, activities and events in specific areas including: the RareConnect™ online rare disease communities; National Plans; the Rare Disease blog; What's New at EURORDIS; News from EURORDIS Members; the European Conference on Rare Diseases and Orphan Drugs; and more. The weekly eNews is disseminated in seven languages: English, French, German, Italian, Portuguese, Russian and Spanish, and has 7,500 subscribers. In 2012, content from the eNews began to be regularly communicated via social media tools, including Facebook and Twitter.



EURORDIS Website & eNews in Russian



In 2012 EURORDIS proudly launched its website and eNews in Russian. Over a year in the making, the new website is designed to strengthen ties with rare disease patients throughout the Russian Federation and to increase the capacity of Russian-language patient advocates to improve conditions for people living with rare diseases. There are as many as 250 million Russian speakers, located primarily in Russia, Belarus, Kazakhstan, Uzbekistan, Ukraine, Kyrgyzstan, Moldova and Azerbaijan. EURORDIS is working closely with patient groups in several of these countries. The new website provides Russian patients, healthcare practitioners and other stakeholders with accurate, comprehensive, up-to-date policy and information of interest. The website section in Russian will continue to expand and become progressively available in 2013.

The addition of Russian to the existing EURORDIS languages of English, French, German, Italian, Portuguese and Spanish would not have been possible without the generous support of the Association of International Pharmaceutical Manufacturers (AIPM) or all the volunteers who worked to make the Russian-language EURORDIS website and eNews a reality.

EURORDIS Social Media

EURORDIS continued to leverage existing social media tools in order to communicate and interact with the rare disease community, and social media activity was robust in 2012. EURORDIS has its own Facebook page (facebook.com/rarediseaseday), Twitter account (twitter.com/rarediseaseday), Flickr account (flickr.com/photos/eurordis) (for photographs), YouTube channel (youtube.com/eurordis) (for videos), and SlideShare account (slideshare.net/eurordis) (for slideshow presentations).

In 2012, EURORDIS Facebook posts and updates received 246 000 views and generated 10 000 interactions with Facebook subscribers. The EURORDIS Facebook page received 3 500 “likes”. EURORDIS Twitter was also active, with 526 tweets sent to over 2 200 followers. EURORDIS produced 32 videos and made them available via the EURORDIS YouTube channel. Altogether, these videos were viewed 13 177 times and in 136 different countries. EURORDIS also shared over 70 slideshows via SlideShare.



Finally, throughout 2012, the EURORDIS/NORD sponsored blogging platform, rarediseaseblogs.net, received an increased number of contributions from opinion leaders and patient advocates, featuring commentary on rare disease and orphan drug policies and issues. There was a lengthy blog series on access to the medicines for Fabry and Pompe diseases in Holland led by patient advocate Cees Smit as well as a number of updates on the evolution of Rare Disease National Plans in various European Union Member States.



EURORDIS Policy Fact Sheets

The EURORDIS Policy Fact Sheets provide comprehensive, validated information on specific topics relevant to rare diseases. In 2012, we added a new Policy Fact Sheet on Newborn Screening. Several existing Fact Sheets were revised and updated in 2012 in order to provide the latest information available. Updated Fact Sheets include: Centres of Expertise; European Network of Rare Disease Help Lines; National Help Lines; Orphanet; Registries; Respite Care; and Therapeutic Recreation. The Fact Sheets are freely available on the EURORDIS website.





Rare Disease Day 2012



Rare Disease Day

29 February 2012 marked the fifth annual Rare Disease Day. Rare Disease Day was launched in 2008 and is celebrated each year on the last day of February – a rare day for rare people. Rare Disease Day 2012 continued the momentum that has been steadily building since 2008, with active participation in a record-breaking 63 countries, including all 27 European Union Member States. There was participation for the first time in several countries including Chile, Costa Rica, Czech Republic, Guatemala, Pakistan, United Arab Emirates, and Venezuela. Around the world over 1000 unique events were staged on or around Rare Disease Day by patient organisations and other partners in order to raise awareness for rare diseases. EURORDIS organised a one-day symposium in Brussels entitled Rare Diseases: an EU Model of Solidarity (for further information refer to the “Rare Disease Day European Symposium, Brussels” in the Advocacy section of this report).

The Rare Disease Day website (rarediseaseday.org) also received a record number of visits in 2012. The number of visits to the site doubled from 2011 during the months leading up to Rare Disease Day. There were 89,000 visits to the site in January and February 2012 alone. The Rare Disease Day 2012 website featured a calendar of events organised by country, allowing visitors to find activities taking place near them. The website also provided information on the purpose and goals of the Rare Disease Day campaign and offered free electronic downloads for patient organisations and other stakeholders to adapt and use, including logos, banners, badges, and posters. The Rare Disease Day poster was printed and distributed to all EURORDIS member associations and other partners.

In 2012, Rare Disease Day introduced the campaign of Raise and Join Your Hands in celebration of Rare Disease Day. Participants were urged to raise and join hands in solidarity at 12:00 (GMT) on 29 February or on the occasion of their Rare Disease Day event. Almost 100 different photographs were sent to the Rare Disease Day website documenting participants around the world taking part in this campaign. The Raise and Join Your Hands campaign was renewed in 2013.

An especially successful feature of the Rare Disease Day 2012 campaign was the creation of the official Rare Disease Day video, designed to raise awareness for the Rare Disease Day 2012 theme Rare but Strong Together. Translated into 12 languages, this video, produced by Animo Communications with in-kind support of Burson-Marsteller (Brussels), received over 100,000 views and was “liked” on Facebook over 9000 times. The video was ranked first on YouTube in the NGO category.

The Friends of Rare Disease Day dedicated website section received 376 responses, including the French Ministry of Health, Harvard Medical School, and the European Medicines Agency. Those who signed up as Friends of Rare Disease Day committed to displaying the Rare Disease Day logo on their website and providing a link to the Rare Disease Day website.

The Rare Disease Day Social Media activity was particularly strong in 2012. There were 13 400 Facebook fans who “liked” Rare Disease Day 2012. There were 3 127 people following the Rare Disease Day 2012 campaign on Twitter. There were 423 photos added to the Rare Disease Day 2012 Flickr account, and 88 videos posted to the Rare Disease Day 2012 YouTube Channel.



Find more information at:
rarediseaseday.org



The first **EURORDIS Black Pearl Gala Dinner**, celebrating Rare Disease Day: Solidarity & Hope for Rare Disease Patients throughout Europe was held in Brussels, Belgium on 29 February 2012.

Over two hundred people attended the event – Brussels community members, national and international officials, researchers and other professionals striving

EURORDIS *Black Pearl* Gala Dinner

to make treatments available to rare disease patients, individuals living with a rare disease, family and friends. They joined together to lift up the rare disease cause and recognise recipients of the 2012 EURORDIS Awards in addition to raising funds for EURORDIS' actions to empower rare disease patients throughout Europe.

2012 marked the first EURORDIS Awards for outstanding accomplishments in the field of rare diseases, which were presented in Brussels on Rare Disease Day.

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 100 nominations received from EURORDIS members, volunteers and staff, with the aim of promoting leadership and the highest achievements in favour of people living with rare diseases.

The 2012 EURORDIS honorees were as follows:

• EUROPEAN RARE DISEASE ACHIEVEMENT AWARD

Kerstin Westermark, MD, PhD

In recognition of her expertise, dedication and more than a decade of support to those with rare diseases, as the Swedish delegate and Chairperson of the Committee for Orphan Medicinal Products at the European Medicines Agency in London.

• POLICY MAKER AWARD

Androulla Vassiliou, JD

In recognition of her invaluable contribution to the rare disease community as Commissioner for Health and Consumer Policy from February 2008 to end of 2009, when she supported the adoption of the Commission Communication: "Rare Diseases: Europe's Challenge", and the Council Recommendation on Rare Diseases which has laid the groundwork to improve conditions for rare disease patients in all Member States of the European Union.

• SCIENTIFIC AWARD

Professor Alain Fischer and Professor Maria Grazia Roncarolo

On behalf of the San Raffaele Telethon Institute for Gene Therapy, Italy and the Hôpital Necker - Enfants Malades, France and in recognition of their scientific

EURORDIS Awards

research and European collaboration that has resulted in the successful development of the first gene therapy for rare diseases related to Severe Combined Immuno Deficiencies, which has set the stage for the extension of gene therapy to other genetic diseases with a high unmet medical need and for which there is currently no cure.

• PATIENT ORGANISATION AWARD

French Muscular Dystrophy Association (AFM-Telethon)

In recognition of its unmatched support for the cause of rare diseases, rare disease patients' organisations throughout France and EURORDIS, as well as its outstanding success in increasing awareness, raising funds and widely supporting research.

• VOLUNTEER AWARD

Michele Lipucci Di Paola, PhD

In recognition of his unshakable commitment to improving conditions for people affected by rare diseases in Italy and at the European level.

• COMPANY AWARD

CSL Behring

In recognition of the company's long-standing commitment to treatment of rare disease patients, and most recently for its advances to benefit people living with primary immunodeficiency diseases and secondary immune-deficiencies.

Sigma-Tau Pharmaceuticals

In recognition of being an early leader in developing medicines for rare diseases and for remaining an exemplary partner in the rare disease community.

Shire

In recognition of Shire's pioneering initiatives, particularly for patients with Hunter Syndrome and Gaucher Disease, and for the company's ongoing commitment to people living with rare diseases throughout Europe.

• MEDIA AWARD

BBC

In recognition of more than three decades of support, raising awareness on rare diseases and covering rare disease issues.



Brussels 23-25 May 2012

European Conference on Rare Diseases & Orphan Products

HEALTH POLICY & HEALTH CARE

6th European Conference on Rare Diseases & Orphan Products – ECRD 2012 Brussels

The 6th European Conference on Rare Diseases & Orphan Products – ECRD 2012 Brussels “A better future for patients: Shaping together the Agenda 2020” was held in Brussels from 23 to 25 May 2012.

Highlights of this successful conference include:

- Highest participation since the first ECRD was held in 2001
- Participant representation from a record 45 countries including 25 countries from the European Union
- 17 of the 45 countries represented were Eastern European states
- A comprehensive conference programme was proposed: 131 speakers/chairs, 36 sessions

A total of 649 participants attended the event (as compared to 545 in 2010, 420 in 2007 and 320 in 2005). 131 speakers and session chairs participated with representation from all of the official conference partners including the EUCERD, EMA/COMP, EBE and EuropaBio, Orphanet, DIA and the ESHG.

Participants attended from 45 countries (as compared to 41 in 2010, 35 in 2007 and 24 in 2005). **In terms of the participants’ categories, the composition of ECRD 2012 was as follows:**

- 43% patient representatives
- 32% healthcare professionals
- 19% pharmaceutical and health industry representatives
- 6% policy makers/other

The ECRD 2012 offered 40 full patient advocate fellowships including 3 carers/enablers for wheelchair users. An additional 2 persons from Georgia were granted special registration fee waivers. Fellowship recipients represented a total of 14 countries from Eastern & Central Europe (Bosnia, Slovakia, Ukraine, Georgia, Macedonia, Bulgaria, Russia, Czech Republic, Estonia, Hungary, Romania, Belarus, Serbia and Lithuania).

Seven conference themes were proposed:

- Theme 1:** National Plans for Rare Diseases (transversal theme)
- Theme 2:** Centres of Expertise & European Reference Networks
- Theme 3:** Information & Public Health
- Theme 4:** Research from Bench to Bedside
- Theme 5:** Orphan Products & Rare Disease Therapies: Access

Theme 6: Orphan Products & Rare Disease

Therapies: Regulatory

Theme 7: Patients' Empowerment

181 poster submissions were received based on a pre-defined set of quality criteria. 155 posters were accepted by the ECRD 2012 Programme Committee and presented at the Conference.

A special ECRD 2012 supplement of the online Orphanet Journal of Rare Diseases was published with a selection of 42 abstracts from speakers' presentations and posters. A 50-page Executive Summary of the ECRD 2012 was published online and a printed version made available.

The EURORDIS Annual General Assembly took place on the pre-conference day of the ECRD 2012.

Country Visits to Belarus, Russia, Serbia and Iceland

The Byelorussian MPS Organisation organised a conference entitled "Medical care for patients with MPS and other genetic diseases" that took place in the Mother & Child Hospital of Minsk, 20 and 21 September.

It was the first time that a conference in Belarus gathered patients and doctors, with the support of the Ministry of Health. About 100 participants, mostly young doctors and patients and their families, met and listened to presentations from MDs, Ministry of Health, Researchers and the EURORDIS Representative, Anja Helm. EURORDIS participated with a presentation on the importance of patient participation in advocacy, research and awareness.

In June 2012, EURORDIS Board Member, Vlasta Zmazek represented EURORDIS at the the First Eurasian Conference on Rare Diseases & Orphan Products and Third All Russia Conference on Rare

Diseases and Rarely Used Medical Technologies which was held in Moscow, Russia. In the same month she also represented EURORDIS at a conference on "Rare neurological diseases in children - early recognition, diagnosis and treatment options" in Novi Sad, Serbia. It was organised by "Life"- Association against child rare diseases, and constituted the very first conference for some rare diseases in Serbia.

In May 2012, EURORDIS President Terkel Andersen represented EURORDIS at the Nordic conference on Rare Diseases in Reykjavik, Iceland. The conference on Nordic collaboration on Rare Diseases was organised as a follow up of the RareLink@ project and of a mapping study of RD resources in the Nordic countries. Terkel Andersen was invited to make the key note speech at the opening of the conference.



Specialised Social Services

In 2012 EURORDIS continued its focus on Specialised Social Services (Respite Care Services, Therapeutic Recreation Programmes, Resource Centres and Adapted Housing Services), mainly through its involvement in Work Package 6 of the EUCERD Joint Action Working for Rare Diseases (EJA). Within the context of the EJA, EURORDIS elaborated a concept paper entitled 'Rare Diseases: Addressing the Need for Specialised Social Services and Social Policies'. The definition of these services was refined and spread via the concept paper and several presentations done at EURORDIS events (ECRD, CNA, CEF, ENRDHL). In parallel, EURORDIS updated the policy Fact Sheets on Respite Care Services and Therapeutic Recreation Programmes and continued to distribute them throughout the year.

In July, a survey was developed and conducted with National Alliances and European Federations, in order

to start the mapping of Specialised Social Services integrating people living with rare diseases in Europe. The map of these services, in addition to definitions, factsheets and testimonies of beneficiaries and volunteers can be found on the EURORDIS website in the designated section entitled “Services to Patients: Specialised Social Services”, which was launched in December 2012.

Country visits to Agrenska Respite Care Services (Sweden), and Frambu Resource Centre (Norway) were also carried out in 2012 in order to collect extensive information on each of the services, which will be used to develop case study documents. A script guide of 55 questions has been developed in order to facilitate the collection of information during such country visits.

Finally, in December, a Workshop on ‘Guiding Principles for Specialised Social Services’ was organised at NoRo Centre, Romania, with 28 participants from 16 countries to reach consensus on baseline guiding principles, to be compiled in a document which will be distributed to EJA and EURORDIS partners in 2013.

The EUCERD Joint Action: Working for Rare Diseases brings together Member States (public institutions) and EURORDIS as the full partner NGO. It is funded by the European Commission (60%), Member States and EURORDIS. Work Package 6 “Provision of Specialised Social Services and Integration of Rare Diseases into Social Policies and Services” is dedicated to i) mapping and follow-up on Respite Care Services (RCS), Therapeutic Recreation Programmes (TRP), Resource Centres (RC) and other rehabilitation services, such as Adapted Housing (AH); ii) collecting information on training of Social Services Providers and iii) developing social guidelines in the field of rare diseases. The Leader of Work Package 6 is EURORDIS Board Member Dorica Dan.



RESEARCH, DRUGS & THERAPIES

EURORDIS intensified its direct role in advancing research in the field of rare diseases by expanding its efforts in strengthening ties with the research community driven 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 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.

Shaping and Supporting Research Policy

EURORDIS representatives were officially appointed members of the governing bodies of the **International Rare Diseases Research Consortium (IRDiRC)** and participated in their first meetings. Béatrice de Montleau, Board Member of EURORDIS, represented EURORDIS at the 1st meeting of the Executive Committee of the Consortium, held in Brussels in January 2012; and at the 2nd meeting that took place in September 2012, in Evry, France, kindly hosted by the IRDiRC member AFM (Association Française des Myopathies).

In 2012, the Executive Committee worked on the completion of the by-laws of the Consortium, outlined in its "Governance Paper", and of the development and finalisation of the Policy Paper, laying out objectives, policies and guidelines of IRDiRC funded research projects. The leading Committee also undertook to organise the first high level scientific conference of the Consortium, scheduled for April 2013 in Dublin.

Maria Mavris, EURORDIS' Therapeutic Development Director, was appointed as member of the Therapeutics Scientific Committee of the IRDiRC. The Committee held its first meeting in May 2012, Milan.

The Therapeutics Committee, as well as the other two IRDiRC Scientific Committees, worked to shape the content of the IRDiRC Policy Paper in view of its adoption by the Executive Committee.

EURORDIS provided inputs and support to its representatives in IRDiRC, in particular by helping them prepare the meetings and providing regular contribution to the key policy documents. In particular, EURORDIS actively contributed to shaping the policy paper by submitting policy proposals and amendments that aim to ensure the presence of rare disease patients in the decision-making process of the IRDiRC.

EURORDIS is also a member of the External Advisory Board of E-Rare, which is 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.

Promoting the Development of Effective Rare Disease Patient Registries

Given the importance of registries and within the perspective of creating a European Platform for Rare Disease Registries, EURORDIS conducted an online survey to gather patients' thoughts and expectations on this topic. The EPIRARE Patient Survey was composed of 14 questions (plus an open commentary) and it focused on issues closely regarding patients' interests and views on registries. The online survey was available in 11 languages: English (EN), French (FR), Italian (IT), German (DE), Spanish (ES), Portuguese (PT), Greek (EL), Romanian (RO), Czech (CS), Danish

(DA) and Hungarian (HU). This survey was conducted as part of the European **Platform for Rare Disease Registries project (EPIRARE)**, a three-year project co-founded by the European Commission's DG Health and Consumers within the EU Programme of Community Action in the field of Public Health. The Project and corresponding survey were presented at the 2012 European Conference on Rare Diseases in Brussels. Preliminary results of over 4 000 responses to the survey were presented at the "Rare Disease and Orphan Drug Registries" International Workshop on 8-9

October, 2012 and the EU Committee of Experts on Rare Diseases (EUCERD) Workshop on Rare Disease Registration 13 November 2012 in Luxembourg.

Following this thorough consultation, EURORDIS, the National Organization for Rare Disorders (NORD) and the Canadian Organization for Rare Disorders (CORD) released a **Joint Declaration of 10 Key Principles for Rare Disease Patient Registries** on behalf of an estimated 60 million people living with rare diseases in Europe and North America. It is expected that these common reflections and principles will serve as a reference to all other stakeholders when shaping policies and taking actions in the field of Rare Disease Patient Registries particularly in the EUCERD, national plans and IRDiRC.

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 TREAT-NMD Global Database Oversight Committee (TGDOC). The TREAT-NMD has moved out of the EC FP6 funding period and transitioned into the TREAT NMD Alliance remaining a non-legal entity. From 2012 the Alliance has expanded its formal collaboration to engage in additional strategic partnerships with universities, patient organisations and pharmaceutical companies across the world. Composed of representatives of the TREAT-NMD network plus representatives of patient organisations and each national registry, this Committee is responsible for reviewing all requests for data from the global database.

Promoting the Development of Sustainable and Integrated European Biobanks

As the former coordinator of **EuroBiobank**, EURORDIS continues its strong commitment in promoting and actively participating in the first network of biobanks dedicated to rare diseases and participating in its governance. 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.

EURORDIS continued to advocate on behalf of this patient perspective both by promoting policy recommendations in the field of biobanking and liaising with the **Biobanking and Biomolecular Resources Research Infrastructure (BBMRI) Consortium**. Over the past 3 years BBMRI has grown into a 54-member consortium with more than 225 associated organisations (largely biobanks)

from over 30 countries, making it one of the largest research infrastructure projects in Europe. In 2012 BBMRI was implemented under the ERIC (European Research Infrastructure Consortium) legal entity with headquarters in Graz, Austria, responsible for coordination of the activities of National Nodes established in participating countries. By December 2012, 14 countries (Austria, Bulgaria, Czech Republic, Estonia, Finland, France, Greece, Italy, Latvia, Malta, the Netherlands, Norway, Spain, and Sweden) had signed the Memorandum of Understanding (MoU) where they express their aim to establish BBMRI as an ERIC and become Members of BBMRI-ERIC. EURORDIS continued to raise the importance of supporting biobanks for rare disease research to European and national level policy makers.



Promoting the Sustainability, Integration and Harmonisation of Data Infrastructures

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.

Funded by the EU Seventh Framework Programme under the International Rare Diseases Research Consortium (IRDiRC), 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 neurogenerative and neuromuscular disorders. The RD-Connect project supports the overarching IRDiRC goals of developing the means to diagnose all rare diseases and producing 200 new therapies by the year 2020.

EURORDIS encouraged opportunities for patient involvement throughout each of the RD-Connect project work packages including the preparation and delivery of educational materials and to 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. Because of its direct involvement in ongoing projects supporting the development of biobanks and registries, EURORDIS contributes to RD-Connect work packages focusing on these types of infrastructures. 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.

Health Technology Assessment

Health Technology Assessment (HTA) plays an increasing role throughout Europe for the evaluation of and access to therapeutic interventions for rare disease patients. It will play an even bigger role within the mechanism of the Clinical Added Value of Orphan Medicinal Products Information Flow (CAVOMP) (see Advocacy section).

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

EURORDIS is one of the 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.



The first Joint Action ended in 2012. Some 53 HTA agencies participated in one of the EUnetHTA activities. One of its outcomes is a core HTA report, namely a document that describes the technology in question, including its current use, safety, effectiveness / accuracy, costs and economic evaluation, ethics, and organisational, social and legal aspects.

Joint Action 2 will start in 2013 and will apply the tools created by the first Joint Action in various Member States. EURORDIS will participate in the EUnetHTA stakeholders' forum.

Visit www.eunetha.eu to learn more.

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 Party:

- Committee for Orphan Medical Products (COMP)
- Paediatric Committee (PDCO)
- Committee for Advanced Therapies (CAT)
- Patients' and Consumers' Working Party (PCWP)

The dedicated patient representative volunteers contributed this year to the examination of a total of 386 dossiers as part of the scientific committees they belong to. These include: 191 dossiers for orphan drug designations, 17 classification or certifications by the CAT and 178 Paediatric Investigation Plans by the PDCO.

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

In addition to these permanent activities at 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 2012, 17 patient representatives attended meetings of the SAWP for Protocol Assistance. Protocol Assistance is scientific advice for orphan medicinal products and is a way for the company developing the medicine to obtain scientific and regulatory advice on the manufacture, 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 the clinical trials. The process of Scientific Advice/Protocol Assistance is recommended in order to avoid major objections regarding the design 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 where EURORDIS is a member and which is a unique forum where all scientific committees of the agency meet with patients and consumers, held four meetings in 2012. This working party organises the involvement of patients and consumers in all of the EMA activities (in 2011 a total of 423 patients/consumers were involved

in one of the EMA activities). It proposes exploratory ways of involving patients with scientific committees where patients are not yet represented (such as the Committee for Human Medicinal Products), discusses the policy on conflict of interest, negotiates important aspects in involving patients such as special needs for patients who attend meetings at the agency. It constitutes the overarching body that supervises the involvement of patients in all committees and meetings of the EMA.

In 2012, the main topic was the implementation of the new European legislation for pharmacovigilance, probably the main change in the legal framework of pharmaceuticals since 1995. It entered into play on 12 July 2012. Of particular importance, measures to increase transparency on risks and how the regulators assess risks and communicate on them, and also the new roles patients and their organisations can play to contribute to a safer use of medicines. The DITA task force (see below) was key in providing the opinion of EURORDIS in all the guidelines that were developed in 2012.



For example, the EMA now has the right to organise public hearings to discuss a safety related issue in the context of the therapeutic benefit of a medicine. To best proceed with this new mode of communication/action, the EMA management board has engaged in an indepth reflection on how to organise such hearings. On 21 March the management board invited EURORDIS Director of Health Policy to present his own experience and views on public hearings.

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. One example is the consultation on a risk management plan for a new medicine to treat a rare cancer that could cause

harm to the foetus during pregnancy, and therefore measures on how to best minimise this risks were discussed and EURORDIS contributed.

Another important activity in 2012 was an invitation to EURORDIS to express its views on the public release of individual patients' data from clinical trials. Following a decision of the European Ombudsman, the agency will release that information in 2013, and it is first necessary to decide how to release this information without impairing data privacy protection, among other risks. EURORDIS will contribute to shaping this EMA policy.

The Drug Information, Transparency and Access Task Force (DITA)

In 2012, the Orphan Drugs and Paediatric Drugs task forces were dismissed. The EURORDIS Drug Information, Transparency and Access (DITA) task force continues, and involves a maximum of 18 patient representatives and volunteers.

The composition of the DITA task force was renewed following a call to EURORDIS members, with 8 volunteers who were members since 2007 who will continue, and 6 newcomers (who all attended EURORDIS Summer School on regulatory affairs).

After the EURORDIS Summer School in 2013 a new call will enlarge the composition of the task force up to 18 members.

The DITA task force met twice in 2012. Its main activities in 2012 were:

- to conduct a pilot survey on the experience of patients with off-label use in rare diseases

- to contribute to all EMA guidelines for the implementation of the pharmacovigilance legislation (9 guidelines, 57 comments, and series of workshops at the agency)
- to contribute to DG Enterprise consultation on pharmacovigilance fees and opinion on a "reasonable compensation" for the work of volunteers
- to give an opinion on braille requirements for distributors of medicine (with the creation of a working group focusing on information needs for people with impaired vision)

Regarding the **pilot survey on off-label use** in rare diseases, the outcomes were presented at the joint meeting between healthcare professionals, patients and consumers at the EMA, and at the joint workshop of the Council of National Alliances and the Council of European Federations in October 2012. The survey should be expanded in 2013, with the creation of a database to collect the valuable scientific information provided by those who responded to the survey.

The DITA task force continued to advise the EMA on the new functions for the public part of its European register on clinical trials (EUDRACT register) and its European pharmacovigilance database Eudravigilance.

DITA reviewed 9 European Public Assessment Reports and 15 package leaflets.

Interestingly, many years after the European marketing authorisation for thalidomide to treat multiple myeloma, Japanese patients' organisations contacted EURORDIS to seek guidance for their own negotiation on a risk management plan for thalidomide.

EURORDIS Capacity Building & Training for Patient Advocates

EURORDIS Summer School

In 2012, EURORDIS continued its capacity building & training activities for patient advocates through holding the highly successful EURORDIS Summer School for the 5th year in a row; expanding its online e-learning tool; and continuing involvement in the DIA Patient Fellowship Programme. In addition, 2012 was the first year of collaboration with two training projects: the European Patients' Academy on Therapeutic Innovation (EUPATI) and the European Clinical Research Infrastructure Network – Integrating Activity (ECRIN-IA).

In 2012, a Training Manager, Nancy Hamilton, was employed, to work on all training projects including EUPATI, ECRIN and the Summer School.

The 5th EURORDIS Summer School Session was held in Barcelona in June 2012 and once again aimed at training patient representatives in clinical trials, drug development and EU regulatory processes. As in previous years, this four-day training gathered together a dynamic group. 2012 gathered 37 participants representing 15 countries and 39 different diseases.

EURORDIS is involved in training patient advocates in the areas of clinical trials, drug development and EU regulatory processes via the EURORDIS Summer School.

As in previous years, a combination of small group sessions and formal presentations was used to introduce the concepts and terminology of clinical trials and to explain the roles of patient representatives at the European Medicines Agency both as committee members and as invited external experts involved in protocol assistance or communication activities for medicinal products.

This year, for the first time, non-rare disease patients were included as part of our collaboration with the European Clinical Research Infrastructure Network – Integrating Activity (**ECRIN-IA**) which develops e-services, education material to train professionals and patients associations, and communication 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 device (WP5), and nutrition (WP6). The inclusion of these non-rare disease patients was seamless and added an extra dimension for all participants involved.

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 and are also available for viewing in the Training section of the website.

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. This project was developed throughout 2011 and started in 2012.

The Consortium comprises a unique combination of pan-European patient organisations, 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 DIA Patient Fellowship Programme enabled 35 patient representatives from 16 different European countries to participate in the DIA 23rd EuroMeeting in Copenhagen on 26-28 March 2012. In addition, 16 representatives of patient organisations were speakers in the EuroMeeting.

This annual high-level congress 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, innovative therapies and meet with many different stakeholders. Since 2008 patient fellows have had a dedicated booth supported by EURORDIS in the exhibition hall, which has helped increase their visibility.



EURORDIS Round Table of Companies (ERTC) 2012 workshops

"The Value of Partnering in Rare Disease Therapies"

01 MARCH 2012, BRUSSELS: 72 ATTENDEES
FROM 11 COUNTRIES

Held for the first time in the Belgian capital, this workshop explored the benefits of working with patients along the treatment lifecycle in the areas of clinical trials, regulatory affairs and health technology assessment. The need for patients to be empowered and therefore trained was emphasised and the EURORDIS Summer School and EUPATI project were described.

"The Value of Orphan Drugs: Efficient for the Patients, Useful to Society?"

28 SEPTEMBER 2012, BARCELONA: 74 ATTENDEES
FROM 11 COUNTRIES

In the current economic climate, the actual value of orphan drugs is misunderstood by society. The concept of value has different meanings for different stakeholders including clinical efficacy, economic value and improvements to the quality of life for patients. One strong recurrent message was the need for early dialogue between all parties involved, in order to improve access to treatment for patients, adaptive licensing being a potentially powerful way forward.

In 2012, the ERTC comprises:

- 38 members
- 3 new members: Biogen Idec, Inc.; Lysogene; Raptor Pharmaceuticals
- 1 upgrade: Aegerion Pharmaceuticals, Inc from the Sapphire level up to the Emerald level of membership



Charter for Clinical Trials on Rare Diseases

The aim of the EURORDIS Charter for Clinical Trials on Rare Diseases is to regulate the relationship between a clinical trial sponsor and the patient organisations representing the disease concerned by the study. Pharmaceutical companies developing medicinal products for rare diseases are formally invited to adhere to the Charter. To date, seven pharmaceutical companies have signed the Charter and two Agreements of Understanding have been drawn up. All are available on the EURORDIS website.

In 2012, patient representatives from Tuberous Sclerosis Complex (TSC) met with the sponsor of three clinical trials and one compassionate use to discuss the development programme and propose necessary

changes. Contacts were made with other patient representatives from other diseases to establish working groups with the sponsors of clinical trials for their diseases. eTSC became a legal entity with 16 countries headquartered in Germany. They had their first General Assembly in Naples in September 2012 and elected a Board from 7 different countries. They also regularly interact with the US TSC group and did so twice in 2012.

The Charter and its outcomes were presented in a session of the 6th European Conference on Rare Diseases and Orphan Products in May in Brussels.

Compassionate Use

After the survey regarding pharmaceutical companies' experience of compassionate use and the presentation of its results at the EURORDIS Round Table of Companies in November 2011, the main actions in this domain in 2012 were a request to the Heads of Medicines Agency (28 June in Copenhagen) to increase the transparency of compassionate use programmes authorised by their respective agencies. The proposal was made to inform the public of such programmes for example on the website of the heads of medicines agencies.

After two sessions on this matter at the 6th European Conference on Rare Diseases and Orphan Products in Brussels, EURORDIS collaborated with the EMA to prepare a list of national contacts for compassionate use programmes. EURORDIS also presented its data and views on compassionate use to the Gianni Benzi 5th Foresight Training Course in Warsaw, 29-30 September.



GOVERNANCE, ORGANISATION & FUNDING

Board of Directors

EURORDIS Board member, Torben Grønnebæk, sadly passed away in early 2012. Birthe Byskov Holm replaced Torben Grønnebæk, as the representative of Rare Diseases Denmark.

Three new members joined the EURORDIS Board of Directors in 2012, having been elected at the General Assembly: Simona Bellagambi, representing the Italian National Alliance (UNIAMO); Vlasta Zmazek, representing the Croatian Alliance for Rare Diseases, and Geske Wehr (elected for one year) representing

Selbsthilfe Ichthyose e.V from Germany. In addition, Béatrice de Montleau and Avril Daly were re-elected as Directors at the General Assembly 2012. The Board is composed of 12 members from 10 European countries.

The Board of Officers is composed of Terkel Anderson (President), Avril Daly (Vice-President), Christel Nourissier (General Secretary), John Dart (Deputy General Secretary) Dimitrios Synodinos (Treasurer), Dorica Dan (Officer).



Staff Organisation

The team comprised 29 people, 24 full-time equivalent (FTE) as of December 2012. The team is composed of paid staff, one office volunteer, one consultant, and one freelance staff. 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 is based in Barcelona. The Chief Executive Officer shares his time between the Paris and Brussels offices.

The following are the main changes in human resources in 2012:

- Annie Rahajarizafy, Administrative & Accounting Assistant, has left EURORDIS
- Eliza Latxague, Accounting and Administrative Manager, has left EURORDIS
- Justine Evans, Web Content Manager, has left EURORDIS

- Nina Miandabu, Accountant, has joined EURORDIS
- Raquel Castro, Social Policy and Specialised Social Services Manager, has joined EURORDIS
- Zoe Alahouzou, Deputy to the CEO, has joined EURORDIS
- Nancy Hamilton, Training Manager, has joined EURORDIS
- Robert Pleticha, RareConnect™ Project Manager, has started working from Barcelona
- Marta Campabadal, RareConnect™ Junior Manager, has joined EURORDIS
- Louise Taylor Communications & Development Writer, has joined EURORDIS
- Jean-Marc Sfeir, Webmaster, has joined EURORDIS
- Minna Granger, Director of Outreach & Partnership and Communications & Development Unit Director, has joined EURORDIS

Volunteers

Volunteers of EURORDIS are active patient advocates, 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.

EURORDIS is proud to rely on 37 dedicated volunteer patient advocates, one permanent office volunteer and 94 volunteer moderators of Online Communities of Rare Disease Patients, within "RareConnect™".

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, the DITA Task Force, and speak at international conferences.

Fourteen volunteer patient advocates represent the patients in EU high-level Committees of the European Medicines Agency and the European Commission.

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), and Mrs Lesley Greene (EURORDIS, UK) who is the Vice-Chair of the COMP.

Dr Maria Mavris, Drug Development Programme Manager, is observer.

■ EMA Paediatric Committee (PDCO):

Dr Tsveta Schyns (European Network for Research on Alternating Hemiplegia, ENRAH, Belgium), full member, and her alternate, Dr. Gérard Nguyen, Rett Syndrome Europe.

■ EMA Committee for Advanced Therapies (CAT):

Dr. Michele Lipucci di Paola (Associazione Veneta Lotta Talassemia, Italy).

■ EMA Patients' and Consumers Working Party (PCWP):

Lise Murphy, the PCWP Co-Chair, (Marfan Syndrome, Sweden) and Richard West (UK Behçets Syndrome Society).

François Houyez, Information & Access to Therapies Director, is a member of PCWP and supports the volunteers.

Our Representatives at the EU Committee of Experts on Rare Diseases (EUCERD) of the European Commission

The eight seats (four full members and four alternates) for patients are held by EURORDIS representatives. Yann Le Cam is the only staff member; other representatives are volunteers, all being patients or parents of patients:

- Dorica Dan (Romanian Prader Willi Association and Rare Diseases Romania)
- Jan Geissler (Leukämie-Online e.V, Germany)
- Alastair Kent (Genetic Alliance UK and Rare Disease UK)
- Christel Nourissier (EURORDIS and Prader Willi France)
- Bianca Pizzera (International Patient Organisation for Primary Immunodeficiencies, IPOPI, Italy)
- Dr Gabor Pogany (William Syndrome and Rare Diseases Hungary)

Flaminia Macchia, European Public Affairs Director, and Ariane Weinman, European Public Affairs Manager, are both observers on the EUCERD and support the group of volunteers.

Torben Grønnebaek (Wilson Disease and Rare Disorders Denmark) was nominated to the EUCERD in 2010. He passed away in early 2012, leaving behind an invaluable heritage of courage. EURORDIS wishes to pay tribute to his outstanding contribution and dedication.



Finance and Support Services

- 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 two months to review progress on implementation, deliverables and budget
- Quarterly collection of activity indicators
- Management of human resources-related activities, such as recruitments and regulations
- Management of office support such as IT infrastructure, contact database, office supplies
- Management of legal and fiscal matters related to contractual matters

Contract Grants

New

- Operating Grant for year 2012 ("EURORDIS_FY2012"), single beneficiary, DG Health and Consumers, 12 months
- 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
- ECRD2012 European Conference on Rare Diseases 2012, DG Sanco, 12 months, 2012

Renewed

- RareConnect™ Online Patients Communities, The French Pharmaceutical Companies Association Foundation, under the auspices of the Fondation de France (Leem Fondation), 12 months

Ongoing

- Advocacy and core activities, AFM-Téléthon, 48 months 2010-2013
- Rare!Together, Medtronic Foundation, 24 months 2011-2012
- EPIRARE - European Platform for Rare Diseases Registries, associated beneficiary, DG Health and Consumers, 30 months, 2011-2013

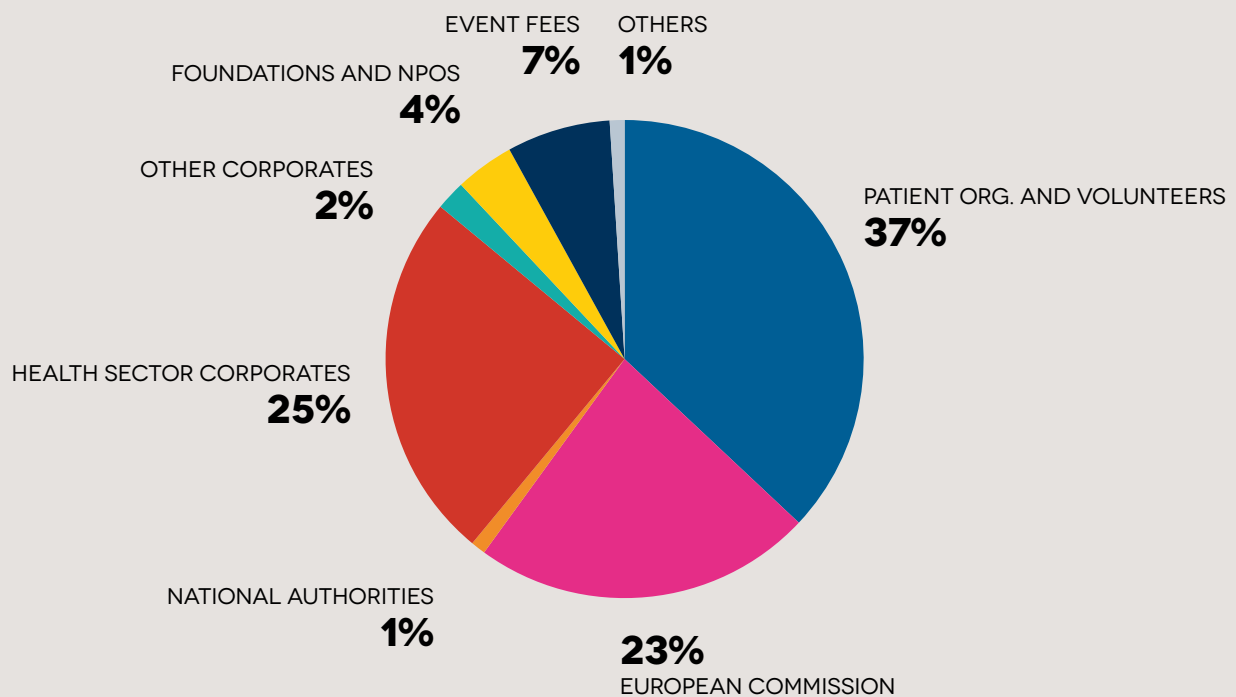


REVENUE AND EXPENSES

2012 (IN EUROS, PROVISIONS EXCLUDED)

REVENUE BY ORIGIN 2012 = 4,038 K€

- Patient Organisations and Volunteers
- European Commission
- National Authorities
- Health Sector Corporates
- Other Corporates
- Foundations and Not-for-Profit Organisations
- Event Fees
- Others

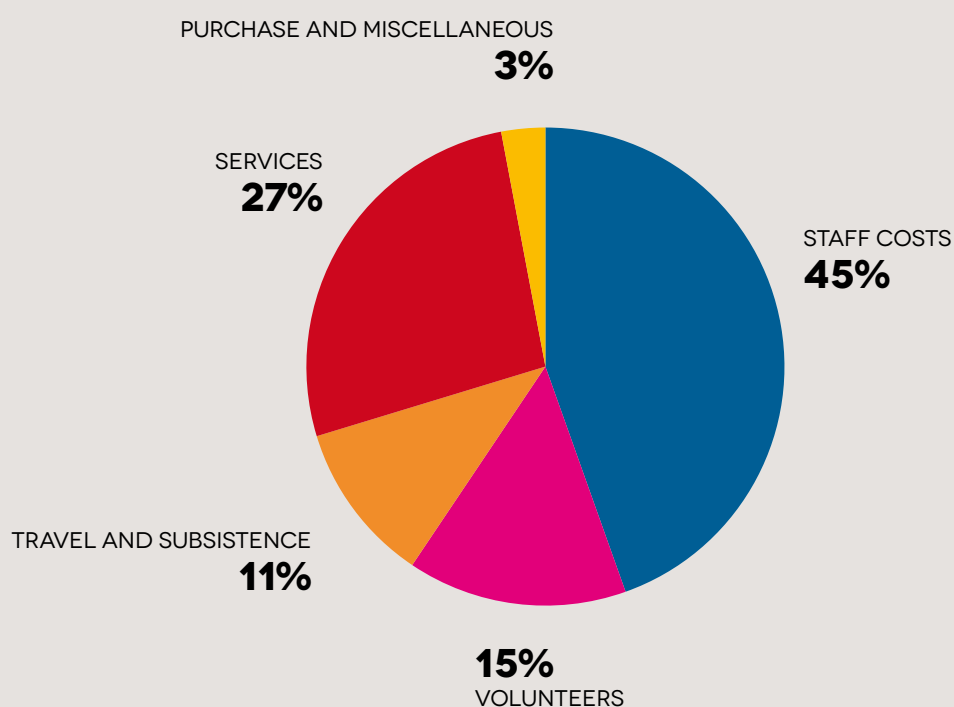




EURORDIS
Rare Diseases Europe

- Staff Costs
- Volunteers
- Travel and Subsistence
- Services
- Purchase and Miscellaneous

EXPENSES BY TYPE 2012 = 3,843 K€



BOARD OF DIRECTORS

MAY 2012 – MAY 2013



PRESIDENT

MR TERKEL ANDERSEN *
• PRESIDENT

Danish Haemophilia Society

DENMARK

DIRECTORS

MS SIMONA BELLAGAMBI	UNIAMO – Federazione Italiana Malattie Rare	ITALY
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MS DORICA DAN * • OFFICER	Romanian Prader Willi Association	ROMANIA
MR JOHN DART * • DEPUTY GENERAL SECRETARY	DEBRA International	UNITED KINGDOM
MS BIRTHE BYSKOV HOLM	Rare Disorders Denmark	DENMARK
MS BÉATRICE DE MONTLEAU	AFM - Associations Française contre les Myopathies	FRANCE
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*Member of the Board of Officers

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	UNIONE ITALIANA ITTIOSI
	VISUS - ASSOCIAZIONE TRA AFFETTI DA RETINITE PIGMENTOSA
LATVIA	ASSOCIATION OF PEOPLE WITH SPECIAL NEEDS "MOTUS VITA"
	RARE DISEASE ASSOCIATION "CALADRIUS"
LUXEMBOURG	ASSOCIATION LUXEMBOURGEOISE D'AIDE POUR LES PERSONNES ATTEINTES DE MALADIES NEURO-MUSCULAIRES ET DE MALADIES RARES
	EEN HÄERZ FIR KRIIBSKRANK KANNER ASBL
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MALAYSIA	MALAYSIA METABOLIC SOCIETY
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MEXICO	PROYECTO PIDE UN DESEO MEXICO IAP
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	NETHERLANDS ASSOCIATION OF PATIENTS WITH STERNO COSTO CLAVICULAR HYPEROSTOSIS
	NEUROFIBROMATOSE VERENIGING NEDERLAND
	OSCAR NEDERLAND



CONFERENCES 2012

Participation of EURORDIS Representatives in Conferences and Workshops

▼ 6ème Rencontre de la Plateforme Maladies Rares : « La Directive européenne relative aux droits des patients sur les soins de santé transfrontaliers : quelles conséquences pour les malades et les centres experts en France? », Paris, France, 17 December

Yann Le Cam: “Les attentes des patients vivant avec une maladie rare” (Expectations from the patients living with rare diseases).

▼ Symposium on Rare Diseases of Metabolism: “Assessing the involvement of all stakeholders to improve healthcare” (organised by the Faculdade de Ciências Médicas of Lisbon, the Chronic Diseases Research Center and the Portuguese CDG association), Lisbon, Portugal, 14 December

Marta Campabadal: Presentation of RareConnect™

▼ Workshop on Specialised Social Services for Rare Diseases, (Work Package 6 of the EUCERD Joint Action), Zalau, Romania, 6-7 December

Dorica Dan: leader of the Work Package

Raquel Castro: project manager of the Work Package on Specialised Social Services

▼ World Orphan Drug Congress Europe, Geneva, Switzerland, 29-30 November

Yann Le Cam: “Providing a new paradigm of rare diseases, treatment development and access in Europe?”

Maria Mavris: “Patients involvement in EMA regulatory procedures for rare disease therapies”

▼ EMA Workshop on Clinical Trial Data and Transparency, London, UK, 22 November

François Houÿez represented EURORDIS

▼ 16th International Conference on Pharmaceutical Medicine – ICPM 2012 – and Xth Congress of AMIFE (Asociación de Medicina de la Industria Farmacéutica en España), Barcelona, Spain, 14-16 November

Denis Costello: “EURORDIS - Promoting Rare Disease Research”

▼ EFGCP Workshop on “Options and Challenges for Ethical Assessment in the Clinical Trial Regulation Proposal”, Brussels, Belgium, 8 November

Flaminia Macchia: “Patients’ Needs and Expectations from the Review of the Clinical Trials Directive?”

▼ 1st European Days of Albinism, Paris, France, 27-28 October

Anja Helm: “Networking between rare disease patients organisations in Europe”

▼ D’genes: V Congreso Nacional de Enfermedades Raras, Totana, Spain, 26-27-28 October

Terkel Andersen, keynote speaker: “Retos y oportunidades para las personas con enfermedades raras en Europa”

▼ 3rd Pan European Conference on Haemoglobinopathies and Rare Anaemias, Limassol, Cyprus, 24-26 October

Jonh Dart: “Activities and vision of the European Organisation for Rare Diseases”

▼ NORD DIA US Conference on Rare Diseases, Washington DC, USA, 22-24 October

Flaminia Macchia: Opening Session: Rare Diseases as an International Public Health Priority

Maria Mavris: Closing Ceremony

2nd Annual Orphan Drug Congress 2012, Barcelona, Spain, 17-18 October

Denis Costello: "RareConnect™- Online Patient Communities and Social Media"

12th France-USA AGIPHARM Conference: "Politics, Health, Attractiveness", Paris, France, 11 October

Flaminia Macchia represented EURORDIS

RIME: Réunion d'Information, Alliance Maladies Rares, Paris, France, 10 October

Béatrice de Montleau: Presentation of IRDiRC – International Rare Disease Research Consortium

Christel Nourissier and Ariane Weinman: Presentation of EUROPLAN II

Conference on ePatients organised by Telethon Italia and Orphanet Italy, Rome, Italy, 10 October

Denis Costello: "RareConnect™ - Online Patient Communities and Social Media"

6th Annual DIA Clinical Forum: "The Empowered Patient", The Hague, Netherlands, 8-10 October

Maria Mavris: "Training patient advocates as partners in research"

Denis Costello: "Patients' Self-reported Outcomes: The EURORDIS RareConnect™ Project"

EPIRARE: Rare Disease and Orphan Drug Registries International Workshop, Italian National Institute of Health, Rome, Italy, 8-9 October

Monica Ensini: "Governance, anyone?"

"Preliminary Results of the EPIRARE Patient Survey on Rare Disease Registries conducted by EURORDIS"

Flaminia Macchia also represented EURORDIS

"La Centralità della Toscana Nella Ricerca Mondiale su una Malattia Rara: l'Alcaptonuria", Florence, Italy, 3 October

Monica Ensini: "Registri di pazienti e biobanche: strumenti chiave per la ricerca sulle malattie rare"

Annual TOPRA Regulatory Affairs Symposium 2012, University College Dublin, Ireland, 1-3 October

Denis Costello: Presentation of the RareConnect™ project

EDRIC Membership Meeting – an organisation for people affected by Dysmelia, Malmö, Sweden, 28- 29 September

Rob Pleticha: Presentation of RareConnect™ - International DysNet Community

Fondation Maladies Rares, Paris, France, 27 September

Christel Nourissier: "Advocacy for research and empowerment of our 528 patient organisation"

2nd edition of Media Trip for Healthcare journalists: "How to Communicate Good Health?" – session organised for Romanian healthcare journalists by Tarus Media, European Parliament, Brussels, Belgium, 26 September

Jill Bonjean: Presentation of EURORDIS with an emphasis on Communication tools

Public Hearing on maintaining or not the reimbursement of Pompe and Fabry treatments in the Netherlands, Amsterdam, 21 September

Yann Le Cam: European patients' views

"Vital Problems of Rendering Medical Assistance to Patients with MPS and Other Rare Genetic Diseases", Minsk, Republic of Belarus, 20-21 September

Anja Helm: Presentation of EURORDIS with an emphasis on "the Power of Patient Organisations"

Irish Institute of Human Genetics, Dublin, 3 September

Avril Daly: Presentation of EURORDIS with an emphasis on European policies for rare diseases

Croatian Haemophilia Society Summer School, Šibenik, Croatia, 1 September

Vlasta Zmazek: Presentation of EURORDIS

Press Conference: "Malattie Rare: oltre la Spending Review. Il Valore sociale delle associazioni di settore", Italian Senate, Rome, Italy, 31 July

Terkel Andersen, spokesperson

First meeting of the Therapeutics Committee of the International Rare Disease Research Consortium (IRDiRC), Milan, Italy, 19 July

Maria Mavris represented EURORDIS

2012 World Congress of the World Federation of Hemophilia, Paris, France, 8-12 July

Paloma Tejada: "The European Organisation for Rare Diseases - Working together: Learning from each other"

First Eurasian Conference on Rare Diseases & Orphan Products and Third All Russia Conference on Rare Diseases and Rarely Used Medical Technologies, Moscow, Russia, 21-23 June

Vlasta Zmazek represented EURORDIS

Meeting of the Heads of Medicines Agencies (HMA 2), Copenhagen, Denmark, 19-20 June

François Houÿez represented EURORDIS

Council of Europe: Symposium on Biobanks and Research on Biological Materials of Human Origin, Strasbourg, France, 19-20 June

Monica Ensini represented EURORDIS

Meeting of the «CISS - Collectif Interassociatif Sur la Santé » (French healthcare organisations group), Paris, France, 10 June

François Houÿez: Presentation of EURORDIS

1st European Conference on Aniridia, Oslo, Norway, 8-10 June

Rosa Sanchez de Vega: "Current EURORDIS' actions to promote rare disease research policies"

▼ EurActiv Lunch Debate: “Translating research to health innovation: How can forthcoming legislation pave the way for better care in rare diseases?” Brussels, Belgium, 7 June

Flaminia Macchia, [Panelist](#)

▼ Annual General Meeting 2012 of GIRP (umbrella organisation of pharmaceutical full-line wholesalers in Europe), Lisboa, Portugal, 4-5 June

Flaminia Macchia [represented EURORDIS](#)

▼ Conference on Paediatric Neurological Diseases, Novi Sad, Serbia, 1-2 June

Vlasta Zmazek: [Presentation of EURORDIS](#)

▼ Nordic conference on Rare Diseases in Reykjavik, Iceland, 31 May – 1 June

Terkel Andersen: [“Rare Diseases in Europe – EURORDIS”](#)

▼ Informal meeting of the Committee for Orphan Medicinal Products (COMP), Copenhagen, Denmark, 23-24 May

Birthe Holm: [attended as Vice-Chair of the COMP and EURORDIS’ representative](#)

▼ “The Future of Health Research and Innovation in Europe: The Need for Strategic Action”, co-organised by the European Commission and the Alliance for Biomedical Research in Europe, Brussels, Belgium, 23 May

Béatrice de Montleau: [“Rare Diseases: European Coordination at a Glance”](#)

▼ 2012 HAE Global Conference (Hereditary Angioedema), Copenhagen, Denmark, 17 - 20 May

Birthe Holm: [“Presentation of EURORDIS Advocacy Actions”](#)-[“Reimbursement of medication and handling the regulatory authorities”](#)

▼ US National Organization for Rare Disorders (NORD) Corporate Council, Washington D.C.; USA, 15 May

Yann Le Cam: [“NORD & EURORDIS: An International Collaboration”](#)

▼ Conference at the University of Augsburg: “Patient organizations, health movements and medical research: varieties, effects and future of civil society engagement in science, technology development and research policies”, Augsburg, Germany, 10-12 May

Christel Nourissier: [“Advocacy for research and empowerment of our 510 patient organisation membership: the role of EURORDIS”](#)

▼ EU High level Conference: “EU Health Programmes: results and future perspectives”, organised by the European Commission, Brussels, Belgium, 3 May

Yann Le Cam: [“The European Integrated Rare Diseases Strategy: Success and Perspectives”](#)

▼ Training programme for patients’ organisations by EmPatient: Capacity Building in information technologies, Paris, France, 25 April

David Oziel: [“RareConnect – Online Rare Disease Patients’ Communities”](#)

▼ X Jornada de Actualización en Genética Humana de la AEGH – Genética: Especialidad y Calidad asistencial: ¿Qué significa ser genetista en los tiempos que corren?, Madrid, Spain, 20 April

Rosa Sanchez de Vega [represented EURORDIS](#)

▼ World Orphan Drug Congress, Washington D.C. , 11-13 April 2012

Yann Le Cam: [“The European Integrated Rare Diseases Treatment Paradigm in an International Perspective”](#)

▼ International Symposium on Hepatic Glycogen Storage Diseases, Lyon, France, 3-6 April

Christel Nourissier: [“Presentation on EURORDIS with an emphasis on research”](#)

▼ Eduthera 2012: “Les nouvelles technologies de l’information au service de l’éducation des patients” (new information technologies for patients’ trainings), 3 April

David Oziel: [“Les cybercommunautés de maladies rares”](#) (Rare Disease online patient communities)

▼ 24th Annual DIA EuroMeeting, Copenhagen, Denmark, 26-28 March

Denis Costello: [“Information to Patients: Strategic Recommendations For Future Debate”](#)

Gérard Nguyen: [“Patient Outcomes Research: E-patient”](#)

Yann Le Cam: [“What Would the Integrated Rare Diseases Treatment Paradigm Look Like?”](#); [“Adaptive licensing: a useful concept for introducing drugs?”](#)

Michele Lipucci di Paola, Maria Mavris and Ariane Weinman [attended as EURORDIS’ representatives](#)

▼ Balkan Patient Meeting: “Together To Success”, Sofia, Bulgaria 23-25 March

Christel Nourissier: [“How does Europe support progress today for people living with rare diseases?”](#)

Vlasta Zmazek [also represented EURORDIS](#)

▼ Annual Workshop ENPR–EMA: European Network of Paediatric Research at the EMA, London, UK, 22 March

Richard West [represented EURORDIS](#)

▼ 3rd European Narcolepsy Day: Hot topics in European Narcolepsy Research & European Narcolepsy Association Meeting, Bologna, Italy, 17-18 March

Rob Pleticha: [Social Media and Patient Networking for Narcolepsy Patient Groups](#)

▼ IAPO (International Alliance of Patients’ Organizations) 5th Global Patients Congress - London, UK - 17-19 March

Jill Bonjean [represented EURORDIS](#)

▼ RE-ACT Congress: International Congress on Research and Orphan Diseases, Basel, Switzerland, 29 February - 2 March

Maria Mavris: [“Patients and Scientists’ involvement in the Orphan Drug development process”](#)

Launch of the “Fondation Maladies Rares” (French Foundation for Rare Diseases) under the auspices of the French Ministry of Education and Research, and Ministry of Work, Employment and Health, Paris, France, 29 February

Christel Nourissier represented EURORDIS

First International Workshop on Clinical Practice Guidelines on Rare Diseases, Rome, Italy 23-24 February

Monica Ensini: “Clinical practice guidelines on rare diseases and quality of care: Patients’ expectations”

EFC Research Forum Stakeholders’ Conference: “Research and social innovation: the potential for European foundations to pave the way”, Barcelona, Spain, 9–10 February

Jill Bonjean and Denis Costello attended on behalf of EURORDIS

ESMO – Rare Cancers Europe: Consensus Meeting and Conference: “Improving the Methodology of Clinical Research on Rare Cancers”, Brussels, Belgium, 9 February

Yann Le Cam: “Significant Benefit and Clinical Added Value in the Context of Access to Orphan Drugs”

Welcome and Introduction together with Dr. Casali, European Society for Medical Oncology (ESMO)

ICORD: International Conference on Rare Diseases and Orphan Drugs, Tokyo, Japan, 4-5 February

Yann Le Cam: “The Value of and Promotion of Basic Research in RD – The future of International Collaboration”

“Patient Groups: Their Connection and Needs”

Swiss Parliament: “Table ronde – Communauté d’intérêts maladies rares: Une stratégie nationale pour la Suisse?” Berne, Switzerland, Monday 30 January

Christel Nourissier: “Les Plans nationaux pour les maladies rares en Europe” – National Plans for Rare Diseases in Europe

ECPC and EURORDIS Joint Workshop on Rare Disease/ Rare Cancers Patient Registries, European Parliament, Brussels, Belgium, 25 January

Yann Le Cam, Flaminia Macchia and Monica Ensini represented EURORDIS

Launch of the Finnish Alliance for Rare Diseases, Helsinki, Finland, 21 January

Terkel Andersen: “Rare Diseases 1981-2011: Achievements of three decades of collaboration and cross sector involvement”

EMA Workshop – “Significant benefit of orphan drugs: concepts and future developments”, London, UK, 12 January

Yann Le Cam: “Significant Benefit in the Context of Access to Orphan Drugs”

EMA Workshop - “A model for a fruitful interaction between CAT (Committee of Advanced Therapies) and its Stakeholders”, London, UK, 12 January

Michele Lipucci di Paola and Monica Ensini represented EURORDIS



ACKNOWLEDGEMENTS

EURORDIS would like to thank the following organisations and companies for their financial support in 2012

NOT-FOR-PROFIT ORGANISATIONS AND PUBLIC ENTITIES

AFM – TÉLÉTHON

The “Association Française contre les Myopathies” (AFM – Téléthon), for the annual core activities grant and the office space they make available to the organisation free of charge



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 » and Integration of Rare Diseases into Social Policies”



EUROPEAN COMMISSION

European Commission (DG Health and Consumers) for its essential contribution to the following projects:

- The Operating Grant for year 2012 (EURORDIS_FY2012)
- The European Conference on Rare Diseases and Orphan Medicinal Products 2012, Brussels (ECRD2012)
- The European Platform for Rare Diseases Registries (EPIRARE)
- The EUCERD Joint Action Working for Rare Diseases (EJA)



Co-funded by the Health Programme of the European Union

European Commission (DG Research) for its essential contribution to the following projects:

- 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 Understanding (IMI-JU) project: European Patients' Academy on Therapeutic Innovation (EUPATI)



AIPM, ASSOCIATION OF INTERNATIONAL PHARMACEUTICAL MANUFACTURERS



For co-funding the Russian version of the EURORDIS Website and eNews

EFPIA, EUROPEAN FEDERATION OF PHARMACEUTICAL INDUSTRIES AND ASSOCIATIONS



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

FONDATION DU LEEM – LES ENTREPRISES DU MEDICAMENT



For co-funding the RareConnect™ Online Patients Communities

MEDTRONIC FOUNDATION



For co-funding of the Rare!Together Project

FUNDACIÓ DOCTOR ROBERT UNIVERSITAT AUTÒNOMA DE BARCELONA CASA CONVALESCÈNCIA



For the office space they make available to the RareConnect™ staff free of charge and for their support to the EURORDIS Summer School

SCIENCE MUSEUM COSMO CAIXA



For providing the venue of the EURORDIS Summer School free of charge

ACKNOWLEDGEMENTS

PHARMACEUTICAL AND BIOTECH SECTOR COMPANIES

EURORDIS has diversified its corporate sponsorship from 37 to 39 different companies.

EURORDIS believes that diversification of funding is a key success factor to minimise potential conflict of interest with donors.

Companies have supported EURORDIS through the EURORDIS Round Table of Companies¹, the EURORDIS Membership Meeting 2012 Brussels, the RareConnect™ Online Patients Communities², the EPIRARE project and the EURORDIS Gala Dinner³ which funds actions to reduce isolation of people living

with rare diseases, 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

Alexion Pharma



Bayer Healthcare



Celgene International



GlaxoSmithKline



Pfizer



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

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

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

OTHER PHARMACEUTICAL COMPANIES

> Listed in alphabetical order.

	 Dompé farmaceutici	
		
		
		
		
		
		
		
		
		
		

ACKNOWLEDGEMENTS

OTHER HEALTH SECTOR CORPORATES

IDIS

For EURORDIS Black Pearl Gala Dinner 2012 donation



MARK KRUEGER & ASSOCIATES, INC

For EURORDIS Black Pearl Gala Dinner 2012 donation and support to the ERTC Workshop Welcome Dinners



OTHER CONTRIBUTORS

BURSON-MARSTELLER

For the in-kind contribution of the development and production of the Rare Disease Day 2012 video



ERNST AND YOUNG

For EURORDIS Black Pearl Gala Dinner 2012 donation



MULTIBURO

For the discounted pricing of the office space made available to the Public Affairs team based in Brussels





EURORDIS
Rare Diseases Europe

WORKPLAN 2013

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EURORDIS
Rare Diseases Europe

ACTION PLAN

2013



Advocacy

Advocacy Issues

- **Turning rare diseases into a sustainable public health priority in the European Union**
- **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 drugs and treatments for rare diseases**
- **Improving quality and access to rare disease diagnosis and care**
- **Accessing cross-border healthcare and making possible patient mobility**
- **Promoting research and bridging patients and researchers**
- **Addressing the issues of genetic testing, genetic counselling & newborn screening**

Advocacy Actions

- **Support implementation of the EU policy framework adopted in 2008 “Commission Communication on Rare Diseases: Europe’s Challenges” and in 2009 “Council Recommendation on an Action in the Field of Rare Diseases” through:**

- Participation of four EURORDIS’ patient representatives and their three alternates in the EU Committee of Experts on Rare Diseases (EUCERD)
- Support the implementation of the EUCERD Road Map
- Support implementation of the EUCERD Joint Action as a Full Partner across all Work Packages (EURORDIS is the NGO representing patients in this Joint Action involving EC and Member States) and disseminate its outcomes
- Participation of the seven patient representatives and additional EURORDIS patient representatives in EUCERD’s key activities such as National Strategies / National Plans & Indicators, Centres of Expertise & European Reference Networks, Cross-Border Healthcare, Rare Disease Registries & Data Collection, Access to Rare Disease Therapies, Newborn Screening, Emergency Cards, Specialised Social Services and Integration of Rare Diseases into Social Policies and Services
- Support the patient representatives in EUCERD with the EURORDIS Policy Action Group (PAG), public affairs staff, volunteers, information sharing and reports
- Contribute to EUCERD with concrete policy proposals in order to address patient needs when implementing the strategy foreseen in the EU policy framework

- Lead & support EUCERD collaboration in specific areas, eg: Rare Disease Day, ECRD, Access to Medicines, Social Policy, and International dimension
- Contribute actively to the “State of the Art of Rare Disease Activities in Europe” through direct EURORDIS contributions and involvement of its National Alliances
- Disseminate EU policy outcomes, monitor progress, promote new strategies and innovative solutions, to integrate all main stakeholders at EU and national levels, and share common objectives through ECRD 2012 Brussels Executive Summary, upcoming ECRD 2014 Berlin, internal dissemination, EURORDIS eNews and EURORDIS Website

- **Maintain rare diseases as a sustainable policy and budget priority in the EU policy programmes for the period 2014- 2020:**

- As a top public health priority in the 3rd EU Public Health Programme, called “Health for Growth” & Annual Work Programmes
- As a top research priority in the 8th EU Research & Technology Framework Programme called “Horizon 2020” & Annual Calls
- Through advocacy actions to European Commission, European Parliament, Council and EU Presidency

- **Advocate on the implementation of the EU Directive on Cross-Border Healthcare (Patients’ Mobility) adopted in 2011 providing a legal basis for patients’ rights for cross-border care and for the establishment of European Reference Networks aimed at addressing specific needs of rare disease patients and families:**

- Empower patient advocates for the national transposition (October 2013) through Q&A document
- Gather and exchange experience of National Alliances working on the national transposition through emails and webinars

- **Advocate to improve access to orphan drugs in the context of the financial and economic crisis to prevent or limit impact of cut back measures:**

- Support and Monitor implementation of the Clinical Added Value of Orphan Medicinal Products (CAVOMP) based on the EUCERD Recommendation, in conjunction with EMA, EUneHTA, MEDEV, the EuropaBio-EBE Taskforce on Rare Diseases and Orphan Drugs
- Develop adapted measures: Adaptive Licensing for Orphan Medicinal Products, Compassionate Use and Early Access Management Plans, Relative Effectiveness of Orphan Medicinal Products, and Off-label Use of Medicines in Rare Indications
- Contribute to the new approaches to Benefit/Risk evaluation, taking into consideration the patient values & patients’ preferred treatment options, in order to

improve the patient information and to help medical decisions

- Contribute to a Mechanism of Coordinated Access (MOCA) to Orphan Drugs, developed within the EU Stakeholders Forum on Corporate Responsibility in Pharmaceuticals, promoting collaboration between Member States on Orphan Drugs Pricing and Reimbursement based on a common European Transparent Value Framework
- Contribute to new approaches on methodology and criteria for the evaluation of the value of Orphan Drugs, for potential use at European and National levels
- Promote Conditional Pricing in Member States & National Plans on Rare Diseases
- Adopt a Declaration on Improving access to Orphan Medicinal Products in a context of European economic crisis and austerity measures in EU Member States
- Advocate for the adoption of the revision of the Transparency Directive to accelerate the placing on the market of new medicines
- In emergency situations, support actions to defend patients' access to rare disease therapies

■ **Advocate on the specificity of rare disease therapy availability through a European Policy Discussion Event for Rare Disease Day 2013 on the future EU Regulation on Transparency**

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

- The international spreading of Rare Disease Day
- NGO status at the Council of Europe
- Elaboration of a strategy toward WHO
- The adoption of a Joint Declaration "Rare Diseases: an International Public Health Challenge" by major rare disease umbrella patient organisations
- The development of "Rare Diseases International" as an informal network of rare disease patient organisations
- The inclusion of this theme in the IRDiRC Conference 16-17 April 2013 Dublin
- The support of ICORD as a tool to expand the rare disease movement at the international level (ICORD 2013 St Petersburg)

■ **Advocate in support of rare disease research: A high priority in the Strategy 2010-2015:**

- Promote a more favourable research policy framework for rare diseases: dissemination at EU and national level of the two reference papers "Why Research on Rare Diseases?" and "Patients' Priorities and Needs for Rare Disease Research"
- Support the development of the International Rare Disease Research Consortium (IRDiRC) of which EURORDIS is a member of the Executive Committee and a member of the Therapies Scientific Committee.

Take part in all meetings and activities. Promote IRDiRC objectives and contribute to IRDiRC policy papers

- Promote a joint paper of EUCERD and COMP to DG Research on strategy & priorities for the EU Programme "Horizon 2020"
- Empower patient advocates through Policy Factsheets and capacity building workshops at the EMM 2013 Dubrovnik
- Explore feasibility and resources to conduct research on the use of NATC products within rare disease patients (Natural, Alternative, Traditional, Complementary products and also vitamins, food supplements, etc) in line with our proposal for a research priority in Horizon 2020

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

- Elaboration of EURORDIS positions on genetic testing and screening
- Development of a knowledge base and shaping future EU legislation on anti-genetic discrimination
- Contribution to the ongoing reflection processes on personalised medicines

Information & Networking

Community Building

■ **Maintain the EURORDIS Membership over 500 members and ensure regular interaction**

■ **Launch new process of regular membership reassessment of eligibility criteria**

■ **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 2013 Dubrovnik on 30 May-1 June with "Capacity Building" Workshops and a "Learning From Each Other" Forum**

■ **Build capacities of the European network of 29 National Alliances through:**

- 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 one day Workshop of the Council of National Alliances
- Organising telephone or web based conference calls on specific topics
- Increasing 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

- Encourage contacts between patient organisations in same countries and the creation of new national rare disease alliances where there are none yet, in particular in Slovenia, Ukraine and Poland

■ **Build capacities of the European network of 35 disease specific European / International Federations & Networks through:**

- Sharing information, experience, guidance 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
- Organising a two day workshop of the Council of European Federations, including a 1 day training
- Sharing experience and good practices through the Rare!Together website
- Providing regular advice to European Federations & Networks
- Expanding the EURORDIS Programme to Support European Federations & Networks with seed 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 Caucasias and support their actions to raise public awareness, promote policy on rare diseases and create national alliances:**

- Provide EURORDIS Patient Advocate Fellowships for the EURORDIS Membership Meeting 2013 Dubrovnik to patient advocates from: Bosnia-Herzegovina, Croatia, Hungary, Macedonia, Montenegro, Serbia, and Slovenia.
- Maintain dissemination of Decide Topics available in 22 languages
- 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 as well as the EURORDIS-EURORPLAN conferences on national RD strategies.
- Expand EURORDIS Website, eNews and main documents available in Russian
- Organise country visits

■ **Support EURORDIS Volunteers involvement through:**

- Strengthening the identification, recruitment and support to volunteers to be increasingly involved in EURORDIS activities, participating in NGO partnerships and representing patients in European Commission and EMA working groups and committees
- Briefing on key topics, access to shared reference documents and public presentations

■ **Expand RareConnect™, the EURORDIS-NORD Social Networks of Online Patients Communities in five to seven languages:**

- Maintain & support the 32-35 existing Online Patient Communities in conjunction with the over 230 patient groups involved and volunteer moderators
- Launch 25 new Online Patient Communities in conjunction with NORD and the existing patient groups for these rare diseases, adapting the tool to each specific community needs in conjunction with relevant patient groups
- Continue to provide operational support to moderators through the “RareConnect™ Moderator Manual”, regular webinars, mailing list and direct support from staff. Focus includes: Identifying quality medical information online, referring patients to resources and specialists (where available), promotion and regular maintenance of their community
- Promote the service at large and each online community
- Specifically pilot communities on very rare diseases and on rare cancers
- Develop new features to make the service more vibrant including the addition of two new languages, access from individual mobile devices, access to full text research articles via partnership with PatientInform, integration of InfoHub
- Maintain the human translation in the current five languages (EN, FR, DE, ES, IT) with possible addition of two languages in 2013 (PT, RU)
- Elaborate and adopt a strategic plan for RareConnect™ for the period 2014-2015
- Involve National Alliances and European Federations
- Promote upgraded version of the EURORDIS Guidelines on Good Practices on Social Media

Informing & Raising Awareness

■ **Organise Rare Disease Day 2013: on the theme/ slogan “Rare Disorders without Borders” keeping the overarching theme of “Solidarity” for the next 4 years:**

- Info Pack, Poster & Slogan, dedicated Website, patient testimonies, social media

- Expand to 60+ countries in Europe and at the international level
- Continue to expand the raising hands campaign and tie it in to social media campaign
- Update and upgrade of rarediseaseday.org and make country events more visible
- Storify: A digest of social media and Country pages information disseminated in the weeks leading up to Feb 28

Possibly:

- A promotional video (only if pro bono/ volunteer support is secured)
- RDD logo projection on public buildings or hospitals (if agreements with ENDESA electricity company are secured)
- Paid media coverage in international daily newspaper (i.e. Metro). Try to negotiate a package deal across several countries or free supplement (ie. Media Planet) paid for with industry advertising

■ **Organise a Policy Discussion Event in Brussels on the occasion of Rare Disease Day 2013**

■ **Plan Future Rare Disease Days:**

- RDD2014: Info Pack, Poster & Slogan, Website, patient testimonies, social media
- RDD2014: Plan Rare Disease Day Event in Brussels
- Take actions toward a European Year for Rare Diseases (2019)
- Take actions to promote Rare Disease Day as an official WHO Day

■ **Improve content quality and information architecture of EURORDIS Website:**

- Centered on target audiences: patient organisations & advocates, patient and families. Other stakeholders (secondary)
- Improve navigation and user friendliness of website with (i) revised content, (ii) changes on Home Page, side bar, main and secondary menu, (iii) new features, i.e. interactive maps, video presentations, online training tools, tags
- Maintain quality, updated information in six languages (EN, FR, DE, ES, IT, PT) in all sections, expand the section in Russian
- Further develop content and facilitate access via three focal points on homepage: Core – EURORDIS.org including specialised services, Satellites – (eNews, Rare Disease Day, RareConnect™, Help Lines), Social media – (Facebook, Twitter, Flickr, YouTube, Rare Disease Blogs)
- Within core website consolidate issues of strategic importance e.g. EU policy, National policies, Centres of Expertise & European Reference Networks, Registries

& Biobanks, Gene Testing, New Born Screening, Orphan Drugs and other Rare Disease Therapies (including new pages on pharmacovigilance, on off-label use of medicines, on compassionate use of medicines), Access to Medicines & CAVOMP & HTA, Specialised Social Services, Patient training, Living with a Rare Disease, Learning From Each Other, Membership, Volunteers

- Promote the International Rare Disease Blog supported by EURORDIS and involve NORD, CORD, National Alliances in Europe, European Federations, volunteers and key actors
- Maintain specifications for technical backend maintenance of EURORDIS.org and RareDiseaseDay.org
- Better organise the publications section so that important documents such as Q&A, Fact Sheets are easily accessible
- Develop and implement a Strategy and Internal organisation for more vibrant Social Media such as the Blog, Facebook, Twitter, Flickr, YouTube

■ **Improve access to patients' generated knowledge through Social Media:**

- Explore integration of RareConnect™ with Researcher/Patient exchange platform in partnership with academic research team
- Continue Social Media and Patient Empowerment Webinar Series to moderators and patient groups

■ **Develop and launch EURORDIS TV:**

- To provide video footage of: News & Current Affairs, Living with Rare Diseases, Patient Groups, Research, Health Policy, Orphan Drug & Therapies, EURORDIS speeches & events, conference presentations, training or capacity building videos, Rare Disease Day events through the EURORDIS website
- Record videos – in house and outsourced - to present key policy issues / papers / EURORDIS Summer School and EMM testimonies to also be provided on EURORDIS TV

■ **Publish EURORDIS eNews in 7 languages (EN, FR, DE, SP, IT, PT, RU) on a weekly basis all year round (except August), 45-48 issues**

■ **Organise Video & Photo Contest 2013**

■ **Develop photo exhibition projects for exhibition in European Institutions, European events, national events, and on the web**

■ **Identify key advancements benefiting People Living with Rare Diseases in the last 15 years and develop into communication tools for 2014**

Information Services to Patients

■ Improve access to and quality of information through Rare Disease Help Lines:

- Disseminate Policy Fact Sheets and guideline on the creation & development of national help lines and the European Network of Help Lines
- Develop the European Network of Rare Diseases 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
 - ▶ Maintain shared tools, e.g., common software for the collection of data on enquiries, develop new functions and update disease classification, and organise the annual caller profile analysis
 - ▶ Organise local trainings on shared tools and searching quality information on the web
 - ▶ 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)
 - ▶ Advocate and implement the EU wide unique 116 number if accepted by Commission
 - ▶ Link to National Plans
 - ▶ Link to ERNs

■ Improve access to quality information sources on the web for patients, families and relatives as well as structured access and use of information for patient advocates:

- Provide a search service on EURORDIS Website to provide user friendly access
- Brand and promote the service
- Promote the EURORDIS quality criteria for rare disease web services
- Provide online tutorials on searching validated information on the web
- Update and deepen the Review of 40 web services providing essential information on rare diseases and related research, treatments, drugs, services
- Create online video tutorials on how to search information on main websites
- Work with EURORDIS Contact Database to provide disease targeted access to EURORDIS members website
- Launch this new service in early 2013

Health Policy & Healthcare Services

Promoting Rare Disease Health Policy Development

■ Organise the 7th European Conference on Rare Diseases and Orphan Products– ECRD 2014 Berlin on May 8-10th 2014:

- The ECRD is the unique platform/forum across all rare diseases, across all European countries, bringing together all stakeholders - academics, health care professionals, industry, policy makers, patient representatives. It provides 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 and national levels
- Prepare the Programme and the Programme Committee:
 1. Coordinate the multiple partnership with DIA, NORD, EUCERD, EMA, Orphanet, ESHG, EBE/EFPIA-EuropaBio. 2. Create Programme Committee (Co-chairs: Lesley Greene, Wills Hughes-Wilson and Ségolène Ayme) and Develop Programme. 3. Promote ECRD & registrations in six languages. Plan Call for Posters, renew EURORDIS Patient Advocates Fellowship Programme, organise logistics of sessions selected to be simultaneously translated, develop communication around the event
- Explore feasibility of Investors' Forum to be included in ECRD 2014
- Plan satellite workshops for partners eg. EUCERD, Orphanet, EPIRARE

■ Support the National Alliances for their action in advancing national rare disease policy through the following activities:

- EUCERD Joint Action / Work Package 4 – EUROPLAN II:
 - ▶ 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.
 - ▶ Support 4 National Alliances (or Patient Group) in non EU countries to facilitate the organisation of a EUROPLAN National Conference
 - ▶ The EUROPLAN National Conferences use the same format and methodological tools derived from EUROPLAN I; however the organisers can adapt the layout of the Conferences to the most pressing needs of the RD 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 10 EURORDIS Advisors team up with the EURORDIS public affairs staff

- ▶ The 10 EURORDIS Advisors participate in the EUROPLAN capacity building workshops together with representatives of national authorities
- ▶ Collaboration between the 10 EURORDIS Advisors and EURORDIS staff/ volunteers to select European indicators for monitoring national plans and synthesise recommendations identified in EUROPLAN I; work on common policy scenarios for small EU countries (less than 5 million)
- ▶ Actively contribute to the identification of main indicators for national plans and to the drafting of the EUCERD Recommendation on Indicators
- EURORDIS Membership Meeting 2013 in May in Dubrovnik
 - ▶ Information & experience sharing across members of EURORDIS on national planning
 - ▶ Capacity building workshop for rare disease patient advocates: promoting national plans, proposing adequate measures, playing an active role in national plan steering committees, being trained in new policy areas
- EURORDIS Policy Fact Sheets for Patients' 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; New Born Screening; and Genetic Testing & Counselling
- Targeted communication
 - ▶ Promote exchange of information and sharing of experiences through the website by expanding the more user-friendly web sections on EU and national policies, the eNews and the participation in national conferences or other meetings
 - ▶ Develop new ways to exchange information, experience, good measures, and concerns between National Alliances, the 10 EURORDIS Advisors and the Staff – such as “national plan mailing list”, regular conference calls and country visits

Putting Rare Disease Patients at the Heart of the Healthcare System

■ Promote policy on European Reference Networks & Centres of Expertise & Expert Networks & Healthcare Pathways on rare diseases:

- Promote the long term EURORDIS strategic vision on European Reference Networks on Rare Diseases addressing patient needs and contribute to the EUCERD Recommendation on European Reference Networks – principles, scope of activities, quality criteria, selection, priorities, evaluation
- Disseminate the Declaration of Common Principles on Centres of Expertise & European Reference Networks
- Disseminate the Patients' Preferred Policy Scenario on Centres of Expertise
- Disseminate “Good Practices of Collaboration between Patient Organisations in Centres of Expertise and European Reference Networks”, based on POLKA
- Develop future possible transversal actions to support the collaboration between European Federations, patients organisations or communities and European Reference Network (ERNs), based on the new EU policy of ERNs in rare diseases, in key areas such as governance of ERN, disease registries, biological repositories, clinical trials, treatment protocol trials, standards of diagnosis & care, standards of social & supportive care, information to patients, outreach to patients
- Organise a workshop at the Membership Meeting 2013 Dubrovnik
- Organise a theme in ECRD 2014 Berlin on European Reference Networks
- Maintain a dedicated Website section on Centres of Expertise and European Reference Networks

■ Promote policy on newborn screening, gene testing, pre-implantation diagnostics:

- Promote awareness and citizens' debates on gene testing and newborn screening through the promotion of four Decide Topics, and disseminate the identified Patient Preferred Policy Scenario
- Contribute to the EUCERD Recommendation on Newborn Screening and promote the importance of follow up and support of families after diagnosis
- Create a dedicated website section and disseminate information, including better promotion of information available from EuroGeneTest, European Society of Human Genetics and International Society of Neonatal Screening

Integrating Rare Diseases into Social Policy and Specialised Services to Patients

■ Promote integration of the challenges faced by people living with rare diseases 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": mapping existing Specialised Social Services and collecting guiding principles for these services; developing case studies on experienced services based on information collected during country visits; addressing issues related to the training of Specialised Social Services providers
- Also within the EJA, EURORDIS will coordinate with the work package on National Plans, ensuring the integration of the different issues concerning social challenges into the content outline - minimum requirements and recommended content for the workshops of EUROPLAN National Conferences
- 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 rare diseases and their relatives and on the need to integrate rare diseases into existing social policies and services
- Follow up of the EU project BURQOL-RD and dissemination of relevant findings on the social economic burden and health related quality of life in patients with rare diseases in Europe
- Perform preliminary research and action plan for a EURORDISCare 4 Survey on the Social Burden and Financial Burden of Rare Diseases for Patients and Families – concept, research plan, organisation, academic partners, funding – for a possible start in 2014 and implementation & dissemination over three years

■ Stimulate the development and the improvement of access to Respite Care Services, Therapeutic Recreation Programmes, Adapted Housing & Related Services and Resource Centres for Rare Diseases:

- Development of EURORDIS website sections for Respite Care Services, Therapeutic Recreation Programmes, Adapted Housing Services and Resource Centers under a new section dedicated to Specialised Social Services containing all relevant definitions, information and contacts collected
- Mapping of existing services and update on services' contacts and 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 Centers derived from country visits performed within the EJA; the organisation of a workshop on training of Specialised Social Services providers

- Advocate for the creation of new Respite Care Services, Therapeutic Recreation Programmes, Adapted Housing Services and Resource Centres through the Policy Fact Sheet, the capacity building workshop at the EURORDIS Membership Meeting 2013 and the monitoring of measures adopted in national plans
- Encourage EURORDIS staff, volunteers and members to volunteer at Summer Camps and members of EURORDIS Round Table of Companies to financially support the Summer Camps or participate as volunteers
- Organisation of a workshop on training of Specialised Social Services providers

Research, Drugs & Therapies

Involvement with Research Publications

■ Link with Orphanet Journal of Rare Diseases

■ Member of the Editorial Board for OrphaNews Europe

■ Involvement on Editorial Board of the Journal Expert Opinion on Orphan Drugs (due to be launched in January 2013)

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 the EURORDIS Reference Papers on "Why Research on Rare Diseases?" and "Patients' Priorities and Needs for Rare Disease Research"
- Dissemination of EURORDIS Policy Fact Sheets on Research
- Organise workshop on research policy in the Membership Meeting 2013 Dubrovnik
- Organise a theme in ECRD 2014 Berlin on Research Policy
- Participation in the International Consortium for Rare Disease Research (IRDIRC) with involvement in Executive Committee and Therapies Scientific Committee

- Participation in the ERA-Net project E-Rare involving Member States
- Support and participate in the preparation of the 2014 International Congress on Research of Rare and Orphan Diseases “RE(ACT)”, organised in Switzerland by the BlackSwan Foundation
- Participation in research policy activities related to national plans for rare diseases (EUROPLAN)

■ **Partnering with EuroBioBank (European Network of DNA, Cell and Tissue banks for Rare Diseases) which has been transferred from EURORDIS to the Italian Telethon Foundation. Participate in its governance. Promote policy recommendations and liaise with BBMRI Consortium**

■ **Promote the development of rare disease patient data collection and registries, their good governance across Europe, and optimal use aimed at patients’ interest, in particular through the completion of the project EPIRARE:**

- Analysis of the outcomes of the EURORDIS patients’ survey on Rare Disease Registries (i.e. assessing patients’ existing knowledge and expectations of Registries)
- Contribute to the definition of a minimum common data set for rare disease registries
- Contribute to the analysis of the European legal framework and solutions regarding patient registries
- Organisation of EURORDIS-EPIRARE Workshop on Policy Scenarios for an EU Rare Disease Registration Repository Platform in April 2013
- Contribute to the public consultation on the Revision of the EU Directive on Data Protection and follow up the legislative process with regards to rare disease needs
- Participation in EPIRARE Workshop in May 2013 on Data Quality
- Participation to the 2nd International Workshop on Rare Diseases and Orphan Drugs Registries in October 2013
- Collaboration with the ISS Summer School on Rare Disease Registries
- Disseminate the EURORDIS-NORD-CORD Joint Declaration on 10 Key Principles on Rare Diseases Registries
- Contribute to the elaboration of the EUCERD Recommendation on Data Collection and Registries for Rare Diseases

■ **Participate in the development of new long term Infrastructure projects on Registries, BioBanks and –omics-. EURORDIS’ contribution focuses on patient involvement in these projects (e.g. RD-Connect: An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research)**

Supporting Clinical Research

■ **Promote good practices for clinical research on rare diseases:**

- 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
- Collaborate with ECRIN Project
- Contribute to the revision of the EU Directive on Clinical Trials

■ **Maintain EURORDIS’ involvement in EPPOSI Board and in different Thematic Programmes & Workshops such as Rare Diseases, Chronic Diseases, HTA, Health Innovation**

■ **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 EUCERD and the European Partnership for Action Against Cancer (EPAAC) to maximise synergies of these two EU policy areas
- Collaborate with the European Cancer Patients Coalition (ECPC) on position paper on Personalised Medicines and Rare Cancer Registries
- Support rare cancer patients community and capacity building through training activities, RareConnect™, programmes with European Federations, communication
- Increase relevant visibility of EURORDIS’ involvement in rare cancer activities on the EURORDIS website

■ **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: Platform for sharing best practices for management of rare diseases. The Evaluation of Centres of Expertise will be carried out within this project by the Health Research Projects Manager, a position to be created within 2013
- How to better use scientific progress in biomedical research to translate results into tangible benefits for RD patients and to offer high quality information to patients and health-care professionals
- How to promote and support a consistent level of healthcare services for RD patients in the EU while implementing the EU Directive on Cross-border Healthcare

■ **Initiate collaboration with the European Network on Clinical Ethics to inform and build capacities of our members as well as to develop joint activities**

Gaining Knowledge on Rare Disease Treatments

■ **Promote rare disease patient spontaneous reporting on suspected adverse drug reactions of orphan and non-orphan drugs (EURORDIS DIOD project):**

- Creation of web pages on EURORDIS website containing explanation 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, links to reporting sites in Europe, by country/ language
- Information to EURORDIS online patient communities and other social media
- Articles in EURORDIS eNews to encourage members to become pharmaco-vigilant
- Presentation of the pharmacovigilance system and spontaneous reporting tools to rare disease specific federations
- Organisation of a workshop at the EURORDIS Membership Meeting 2013 Dubrovnik
- Organisation and participation in sessions on pharmacovigilance in rare disease conferences such as DIA EuroMeeting, Orphan Drug Congress, Gianni Benzi Foresight Training
- Training of patients advocates through webinars and/ or eLearning and/or session at EURORDIS Summer School

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

- Disseminate the results of the off-label use survey 2012 and prepare the extension, publish articles and discuss with regulators
- Prepare possible extension of the survey on off-label use in rare diseases conducted in 2012 to all other EU MS and possible creation of a database to document this. Engage dialogue with the Patients' and Consumers' Working Party (PCWP), the Committee for Orphan Medicinal Products (COMP), the Pharmacovigilance Risk Assessment Committee (PRAC) and the Committee for Medicinal Products for Human Use (CHMP) on off-label use
- Continue the activities in parallel to the work plan of the PCWP
- Organise the task force to respond to patients' requests for help in accessing treatments
- 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

- Propose a ranking system for the healthcare industry
- Creation of a sub-group of volunteers on specific needs for blind and vision-impaired patients
- Create on-line video tutorials on how to search information on main websites (EUDRACT, database on suspected adverse drug reactions, EUDRAPARM) as part of the EURORDIS InfoHub service

Promoting Drug Development & Access to Treatments

■ **Expand activities on drug development, information and access:**

- Participate in EMA Committee for Orphan Medicinal Products (COMP) with two representative members and one permanent observer
- Participate in EMA Pharmacovigilance and Risk Assessment Committee (PRAC) as external experts for RD Therapies
- Participate in EMA Paediatric Committee (PDCO) with one representative member and one alternate
- Participate in the EMA Committee for Advanced Therapies (CAT) with one representative member and one alternate
- 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 and one alternate and (PCWP)
- Participate in EMA Working Group on Clinical Trials in Third Countries, in the user testing group of the European Register for Clinical Trials, the European Database for Suspected Adverse Drug Reactions, the European Database of Authorised Medicines, the EMA web site
- 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) sharing information, agendas, reports, providing mutual support, by discussing main issues
- Review all orphan drug designation applications (ODD), orphan protocol assistance (PA), review of designation criteria at the time of marketing authorisation and reports on significant benefit, all paediatric investigation plans (PIP) for rare diseases, PIP waivers and deferrals, all gene, cell therapy & 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

■ **Capacity building of our members and volunteers on clinical trials, drug development, EU regulatory affairs:**

- Organise EURORDIS Summer School 2013 Session in Barcelona in June to train 30 to 40 new patients' advocates representing a diversity of diseases and geographical locations
- Collaborate with ECRIN to incorporate 10 non-rare disease patients' representatives from the therapeutic indication areas of nutrition and medical devices
- Provide the E-Learning platform in English with sections on Methodology, Statistics and Ethics, using learning approaches such as lessons, glossary, case studies, relevant documents and a self-evaluation questionnaire
- Take part in the DIA EuroMeeting 2013 Amsterdam in March with speakers, session chairs, 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; Organise one or two workshops of the DITA Task Force annually
- Participate in the "European Patients' Academy on Therapeutic Innovation" (called EUPATI or Patient's Academy, from 2012 to 2016), an IMI-funded project gathering a Consortium led by European Patients' Forum (EPF) with other European umbrella patient organisations. EUPATI deliverables that involve EURORDIS in 2013 include: the EUPATI Curriculum and Syllabus for the Tool Box-due in February 2013, the development of material for different audience types, and the planning and organisation of face-to-face meetings
- Create partnership for Rare Disease Patient Advocates' Capacity Building by consolidating and trying to create new collaborations to enable the participation of patient advocates in capacity building opportunities such as: the DIA EuroMeeting 2013 and other DIA major meetings in clinical trials, regulatory affairs, registries; the European Society of Human Genetics Conferences; HTA trainings and conferences; World Orphan Drug Congress - Washington and Geneva 2013; ISS Summer School on Rare Disease Registries

■ **Create the EURORDIS Scientific Advisory Committee composed of academic leaders and patient advocates in rare disease research, from different backgrounds and competences, different**

therapeutic areas, different countries, sharing the mission and values of EURORDIS and based on EURORDIS Position Papers in Research published in 2010 and 2012

■ **Collaborate with HTAi on adapted HTA for Rare Diseases**

■ **Support and advise the dialogue of National Alliances with HTA Agencies**

■ **Plan and take appropriate action for the future organisation of the 6th EURORDIS Survey on Access to Orphan Drugs in the European Union**

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

- EURORDIS Round Table of Companies: consolidate membership and organise two workshops in 2013: one in Brussels in February and the second in Barcelona in September
- Expand the direct dialogue with pharmaceutical and biotech companies
- Strengthen the dialogue with EBE-EuropaBio, EFPIA and national pharmaceutical associations

Cross-Cutting Priorities

■ **Implement EURORDIS Strategy 2010-2015:**

- Midway 2010-2013 Progress Report of Implementation Strategy 2010-2015
- Adjustment of EURORDIS Strategy 2010-2015 based on progress report & BoD Staff-Seminar July 2012 Monticchiello
- Develop Operational Units Action Plan 2013-2015 (three years) for each Unit
- Develop financial orientations 2014-2015
- Improve planning anticipation of major EURORDIS activities such as ECRDs, Membership Meeting, Rare Disease Day, EURORDIS Round Table of Companies Workshops, RareConnect™, EURORDISCare, major advocacy campaign, new projects

■ **Develop EURORDIS Strategic Partnerships:**

- Prepare Strategic Partnership with AFM-Téléthon for 2014-2017 based on Collaborative Agreement 2010-2013 and respective strategies of AFM & EURORDIS
- Assess strategic partnership with NORD (USA) and consolidate common actions based on Strategic Partnership Memorandum signed in 2009, for 2010-2015
- Implement a Partnership with CORD (Canada)
- Consolidate Partnership with the members of Round Table of Companies

- Establish partnership with relevant Russian organisation(s)
- Establish partnership with the Japanese Patients Association, JPA (Japan)
- Maintain global partnership with DIA
- Develop Strategic Partnership with ORPHANET
- Develop Strategic Partnership with the European Society for Human Genetics

■ Increase EC support to EURORDIS activities through:

- Proposal for optimal usage of the Operating Grant as a financial instrument in 3rd Public Health Programme, "Health for Growth"
- Application 2013 for Operating Grant 2014
- Application 2013 for Conference Grant ECRD 2014 Berlin
- Application for a Patient Driven Public Health Project in DG Sanco Work Programme 2013-2014 as project leader or partner
- Application for a Patient Driven Health Research Project in DG Research FP 7 Call 2013 or 2014 as project leader
- Study feasibility and apply for grant application to other Programmes in other DGs in support of activities foreseen in EURORDIS Strategy 2015

■ Develop and diversify private funding:

- Maintain overall support level from industry donors spread between approximately 40 companies and a variety of activities, within 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 in supporting EURORDIS' projects & actions. Priority to co-funding of the EUCERD Joint Action Work Package 4 (EURORPLAN) for the 20 national conferences and for Work Package 6 on Specialised Social Services; RD-Connect Project; EURORDIS Membership Meeting; the Website & eNews & eLearning & EURORDIS TV & EURORDIS InfoHub in seven languages; the RareConnect™ project & Social Media; Support to European Federations & Networks; EURORDISCare 4
- Organise the EURORDIS Black Pearl Gala Dinner for Rare Disease Day on 26 February 2013 in Brussels "Hope and Solidarity for Rare Disease Patients throughout Europe" and plan Gala Dinner 2014: Gala Committee, Patrons, Master of Ceremony & Script, Performances, Policy makers & Opinion Leaders, Corporate Partnership, Table & Seat sales, Photo Exhibition, Website & Newsletter & Programme & other communication tools, logistical organisation for 250 participants
- Develop a plan for individual donors to be implemented from 2014

- Promote in-kind support from private partners (ex: travels, communication tools)

■ Implement the Staff Strategy & Organisation & Evolution 2011-2013:

- Complete the new organisation in 4 Units: Governance & Advocacy & Membership, Communications & Development, Operations, Finance & Support Services
- Consolidate the capacities & roles of the Unit Directors
- Create the position of Scientific Director
- Create the position of Resource Development Director
- Create the position of Health Research Projects Manager
- Revise and improve the coordination processes: Operating Grant Steering Committee Meetings, All Staff Meetings, Finance & Funding Meetings, Monday Meetings, Advocacy & Strategy Meetings

■ Seek alternative human resources:

- 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
 - ▶ Revise and adopt additional procedures in the field of Finance, Human Resources and Office Support Services
 - ▶ Secure VAT exemption for ECRD 2014 Berlin
 - ▶ Maintain the decentralised structure from Paris (Main Office), Brussels (European Public Affairs), London (EMA), Barcelona (Web Communications & RareConnect™) with integrated operations through work processes, IT standards & intranet, voice & data internet communication
 - ▶ IT support: equipment, services, virtual office, open to volunteers
 - ▶ EURORDIS Contact Database Management fully operational
 - ▶ Continue 5th year of collection of EURORDIS indicators on activity and results

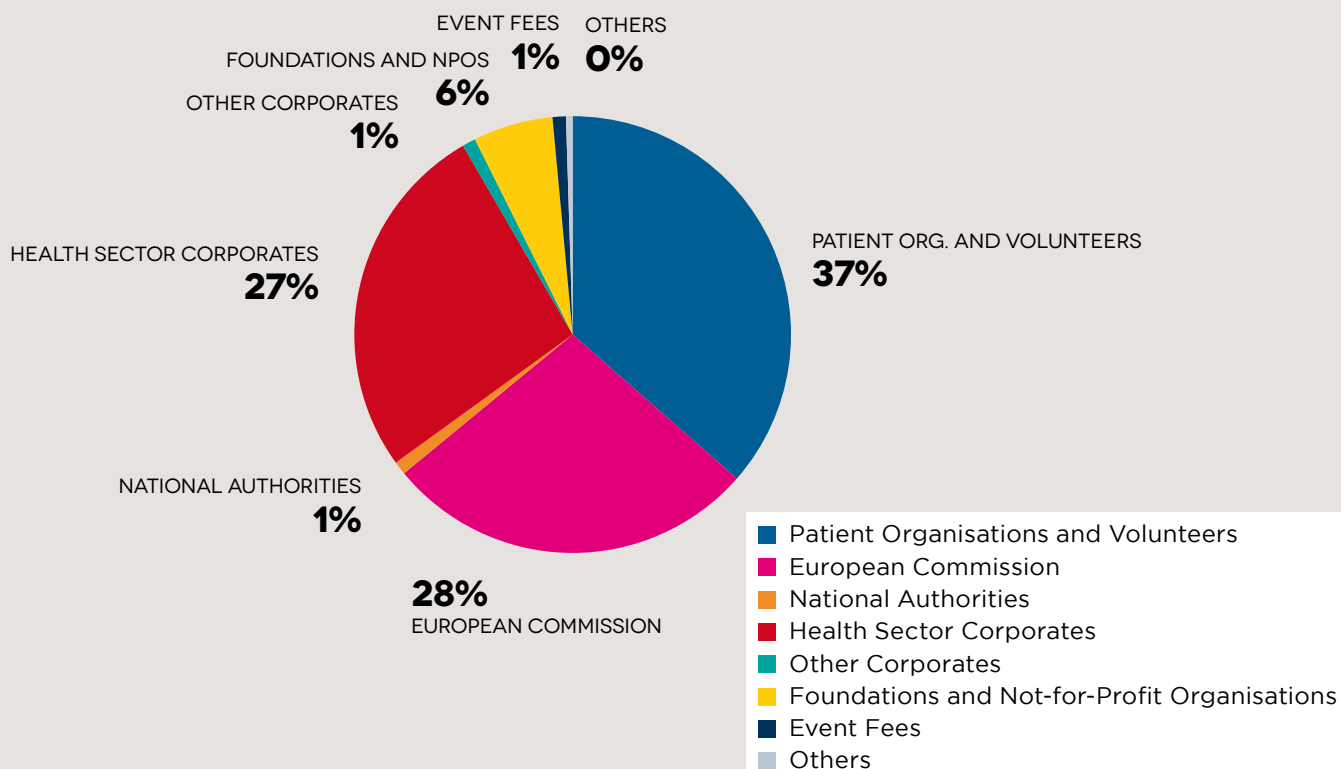
BUDGET 2013

(IN EUROS, PROVISIONS EXCLUDED)

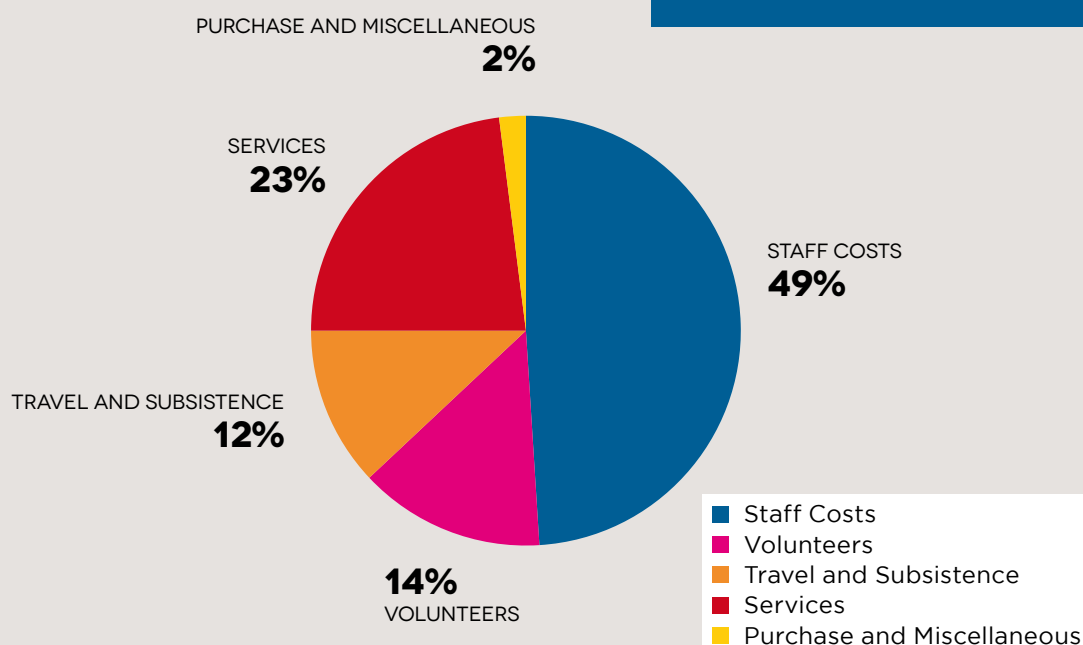


EURORDIS
Rare Diseases Europe

REVENUE BY ORIGIN 2013 = 3,992 K€



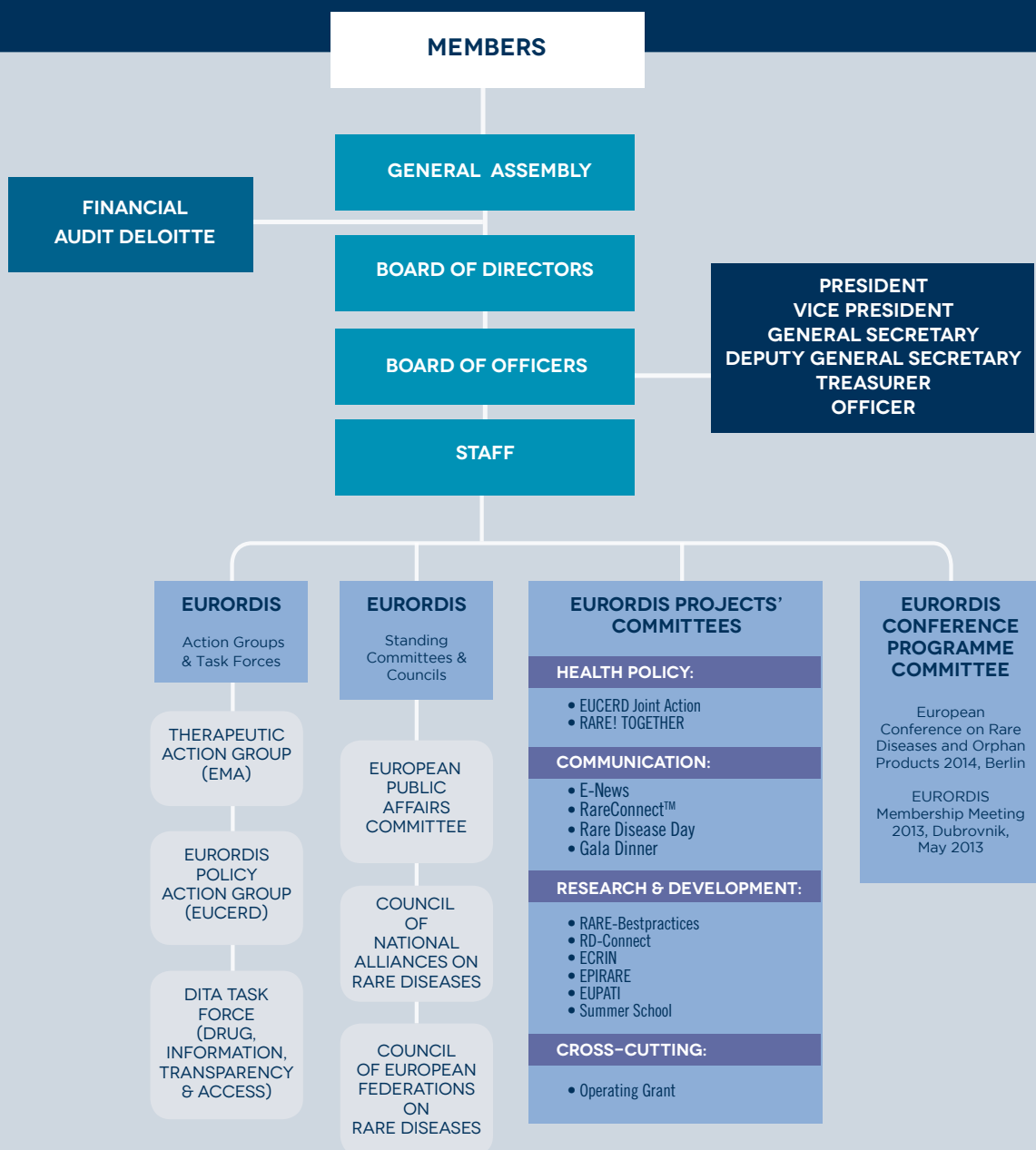
EXPENSES BY TYPE 2013 = 4,098 K€



2013 GOVERNANCE CHART



EURORDIS
Rare Diseases Europe

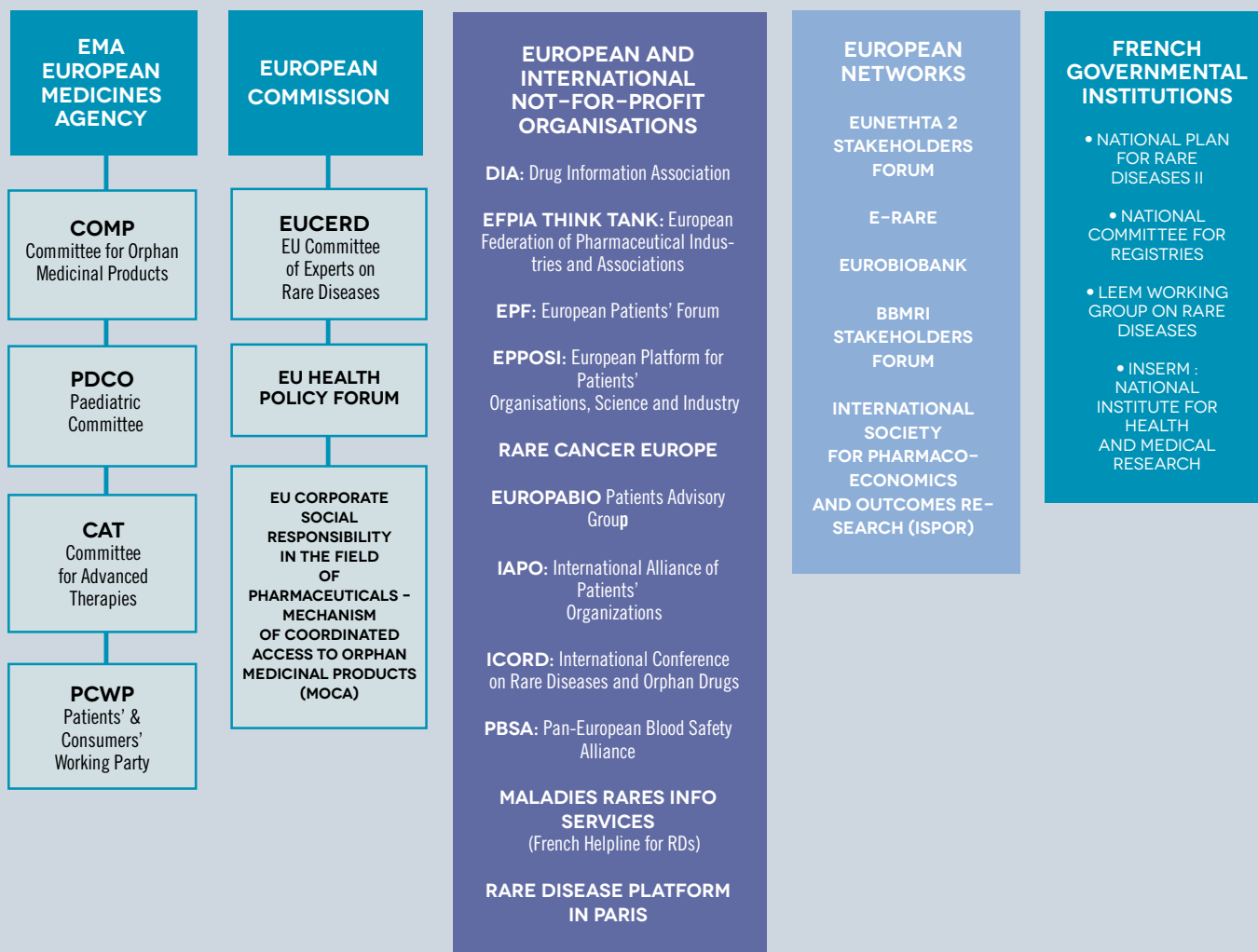


EXTERNAL REPRESENTATION CHART 2013

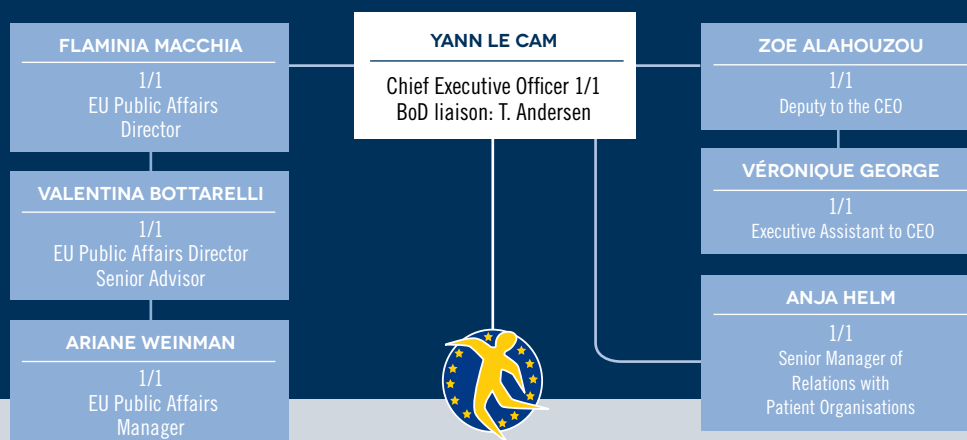
GOVERNMENTAL INSTITUTIONS

NON-GOVERNMENTAL ORGANISATIONS

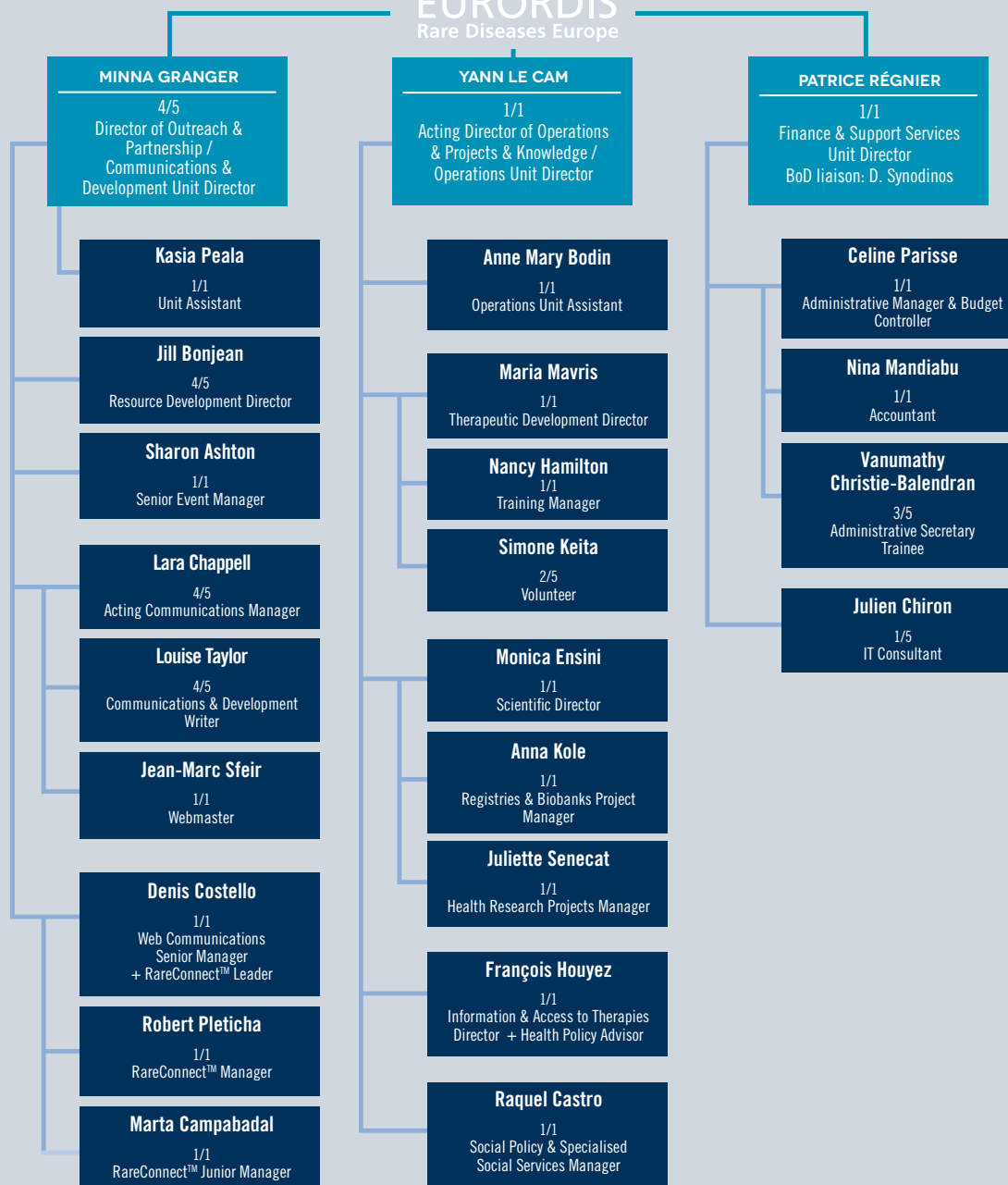
EXTERNAL NETWORKS



2013 TEAM CHART



EURORDIS Rare Diseases Europe



NOTES



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ACTIVITY REPORT 2012

& WORKPLAN 2013

THE VOICE OF RARE DISEASE PATIENTS IN EUROPE

EUROPEAN ORGANISATION FOR RARE DISEASES



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