



**EURODIS**  
Rare Diseases Europe

& WORKPLAN 2012

EUROPEAN ORGANISATION FOR RARE DISEASES

# ACTIVITY **REPORT** 2011

**The Voice of Rare  
Disease Patients  
in Europe**



“Geschwisterliebe” Felix, Tuberous Sclerosis, Germany. Image of Rare Disease Day 2011.  
Photo by Jeannette Bobos.





**EURORDIS**  
Rare Diseases Europe

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# Foreword

by the President  
and by the Chief Executive Officer

**EURORDIS**, its members, volunteers, Board, staff and partners celebrate 15 years of achievements.

As we reached over 500 members in 48 countries, covering more than 4,000 different rare diseases, **EURORDIS** has achieved its initial vision of creating a European 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 today.

**EURORDIS'** member base has grown steadily with 50 new members in 2011 alone. Our membership includes 28 National Alliances and 33 European Federations, overall representing more than 1500 patient organisations. New national rare disease alliances have been created in Austria and Russia in 2011. **EURORDIS** is strengthening its support to Central and Eastern Europe, South-Caucasia and particularly Russia with a combination of actions including patient fellowships, country visits, co-organisation and participation in local conferences, dissemination of information and exchange of experience, content and advice for their strategy to promote rare diseases as a public health priority in their countries. The **EURORDIS** Newsletter is now also available in Russian and our website in Russian is currently under construction. **EURORDIS** and the US National Organization for Rare Disorders (NORD) are scaling up their web portal of disease-specific social networks for patients and families through continuous enhancement of rareconnect.org with 12 new communities launched in 2011 and 20 more planned for 2012.

After ensuring the development and coordination of the Eurobiobank network for 11 years, **EURORDIS** decided to step down and transfer this responsibility to the Italian Telethon Foundation at the end of 2011. **EURORDIS** will maintain its involvement in this and other rare disease research networks supported by the European Commission. In addition, **EURORDIS'** activities to empower patient advocates in research include the Summer School, online learning tools under the website's Training Resources section, as well as involvement, from 2012 onwards, in two new projects, the European Patients Academy on Therapeutic Innovation (EUPATI) and the European Clinical Research Infrastructure Network (ECRIN), to share good practices and develop educational material in the field of clinical trials.

In 2011 and into 2012, **EURORDIS** is focusing on European as well as national activities that influence the lives of rare disease patients on a European level while also promoting those international actions that impact people living with rare diseases in Europe.

The EU Directive on Cross-Border Healthcare was adopted at the beginning of 2011. **EURORDIS'** front line advocacy consolidated rare diseases in this important EU legislation. It provides a robust base for a future European Reference Network on Rare Diseases that would organise expertise on a European level and bring it to patients locally. We are now advocating on its transposition into national policies in order



Yann Le Cam

to preserve and possibly increase support for rare disease patients' mobility in accessing diagnosis and care.

**EURORDIS'** advocacy actions on research policy have significant impact. **EURORDIS** disseminated two important Position Papers: "Why invest in Rare Disease Research" and "Patients' Priorities and Needs for Rare Disease Research". The rare disease research budget has reached its highest ►



Terkel Andersen

► level ever in the 7th EU Research Framework and they are a priority in "Horizon 2020", the EU research framework programme 2014-2020. The International Rare Disease Research Consortium (IRDiRC) is now officially launched and **EURORDIS** is involved in its Executive and Scientific Committees.

**EURORDIS** will amplify its advocacy activities so to further promote rare diseases as a priority of high community added value in the 3rd EU Public Health Programme which has an increased budget for the period 2014-2020. This includes among others: new position papers, coordinated advocacy actions, communication to media, organisation of a Rare Disease Day Symposium in Brussels on this topic.

The EU Committee of Experts on Rare Diseases (EUCERD) in which **EURORDIS** has four patient representatives and three alternates, one being elected Vice Chair, is now central in our action. The EUCERD is playing an essential role in the implementation of the Commission Communication of 2008 and the Council Recommendation on Rare Diseases adopted in 2009. Its first outputs have been visible through its report "the State of the Art of Rare Disease Activities in Europe" and the adoption of its first EUCERD Recommendation on "Quality Criteria for Centres of Expertise for Rare Diseases in Member States".

In 2010, **EURORDIS** made a long term commitment to the development of national strategies across Europe. This includes a combination of actions: closer work with the **EURORDIS'** Council of National Alliances, use of Rare Disease Day to raise political and public awareness, new sections on our website, **EURORDIS** Policy Fact Sheets and the **EURORDIS** Membership Meeting 2011 in Amsterdam and 2012 in Brussels, entirely dedicated to building capacities of patient advocates in national plans.

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** dedicated its Round Table of Companies activities and a significant part of its advocacy to this top issue in 2011 and continues to do so in 2012, focusing on the promotion of the Clinical Added Value of Orphan Drugs (CAVOD), compassionate use and early access programme, the utility of rare disease therapies, regulatory and financial environments to stimulate rare disease therapy developments, and the partnership between all stakeholders to reach these goals.

**EURORDIS'** participation to the European Medicines Agency's (EMA) Scientific Committees - the Committee for Orphan Medicinal Products (COMP), the Paediatric Committee (PDCO), the Committee for Advanced Therapies (CAT) - is very intensive with more than 400 dossiers reviewed. A **EURORDIS** representative is Chair of the Patients & Consumers Working Party and another is Vice Chair of the COMP. **EURORDIS** continues to prepare for an active role in the assessment of risk and 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 orphan drugs and rare disease therapeutic interventions. **EURORDIS** is also a full member of the European network of Health Technology Assessment (EUnetHTA) Stakeholder Forum.

Despite the challenging economic environment, **EURORDIS** was still able to grow from 2010 to 2011 thanks to the continued support of the AFM - Téléthon, the European Commission and other valued donors. Increased funding is a critical issue to sustain our development. **EURORDIS** will continue to diversify its public funding and will take new initiatives in fundraising. The first **EURORDIS** Gala Dinner in 2012 is an important step in this direction.

(April 2012)

Terkel Andersen  
President

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Yann Le Cam  
Chief Executive Officer



## 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.



**EURORDIS**  
Rare Diseases Europe

- **Today it is supported** by its members and by the Association Française contre les Myopathies, AFM - Téléthon, the European Commission, not-for-profit organisations 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. ”

### → Key figures 2011

**492** member patient organisations

**46** countries (24 EU countries)

**28** national alliances

**3,094,000 €** in Revenue

Over **1,000** patient groups represented

**59** volunteers

Over **4,000** rare diseases represented

**23** staff members



Find more information at:  
[www.eurordis.org](http://www.eurordis.org)



# Strategic Approach 2010-2015

## EURORDIS' Position in 2015

- **EURORDIS** 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 and families living with a rare disease in Europe
- **EURORDIS** is facilitating the effective implementation of European regulations and strategies at national levels in more policy areas for the benefit of patients and families
- **EURORDIS** has developed enriched and more supportive capacity building relationships with its members and empowerment of volunteers
- **EURORDIS** is more sustainable in terms of human, financial, organisational resources and governance
- **EURORDIS** has consolidated its activities in drug development, centres of expertise, patients' advocates empowerment and public awareness
- **EURORDIS** has implemented a strategy and activities along the main lines of the Commission Communication 2008 and the Council Recommendations 2009 on Rare Diseases for an optimum impact at EU and national levels
- **EURORDIS** has aligned its Strategy with its Unit work plans, its National Alliances, its European Federations, and its Strategic partnerships for an optimum synergy based on same goals and indicators



## Strategic orientations

- **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**
- **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**

- 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 those of Eastern Europe at large and Mediterranean region at large
- Raising rare disease awareness amongst general public (incl. international Rare Disease Day)
- Rare diseases become an international movement and gain visibility and influence in international instances (World Health Organisation, United Nations, Organisation for Economic Co-operation and Development, World Bank, etc.)
- Production, sharing and accessibility of patient-generated knowledge

- 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, quality of life recognised as a major goal for indicators
- Development / Consolidation of European Networks integrative of European and National levels:
  - European Reference Networks of Centres of Expertise
  - European Networks of Research
  - 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, Standard of Screening Diagnostic and Care, Quality of Life)

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

- Maintain a high level of legitimacy and credibility by maintaining a high level of consent amongst **EURORDIS'** members
- An integrative (in main areas of activities) and supportive volunteer programme well recognised inside and outside
- Intensify capacity-building and networking with and between the National Rare Diseases Alliances and European Rare Diseases Federations for improved efficacy and decentralisation
- 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

## A special focus on research

### The role of EURORDIS shall aim at:

- Higher public awareness in support of rare disease research
- More favourable research policy framework for rare diseases
- Development of **EURORDIS'** interactions with the research community and learned societies
- Promotion of the development of European Research Networks and European Research Infrastructure for rare diseases
- Promotion of the participation of patients in research and therapeutic developments – which enhances capacity building of patient representatives
- 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

### EURORDIS aims 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, scope and quality of life in a holistic approach
- Educational information on the management of specific symptoms which are common 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, etc.)
- 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 local respite centres and their services
  - Production and availability of validated and updated information on therapeutic recreation programmes
  - Promotion of research on quality of life in EU framework programmes in research and public health

► **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)
- Integrative IT infrastructure, database and tools
- In-kind external resources





## Community Structure Overview

- National Alliances on Rare Diseases
- European Rare Diseases Federations or Networks
- Patient Organisations' Members other than above
- Patients' Organisations and Patients' Outreach in All Europe (48 countries)
- Committees, Task Forces, Groups



## Strategic Partnerships

- AFM - Téléthon: Association Française Contre les Myopathies
- NORD: US National Organization for Rare Disorders
- ECPC: European Coalition of Patients with Cancer (on hold)
- ORPHANET: The web server of medical experts generated and validated information
- INDUSTRY through EURORDIS Round Table of Companies and other initiatives
- ACADEMIA for education and capacity building and for social & policy research

# Highlights 2011



## Advocacy

- EUCERD adoption of first Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases in Member States, with active participation from **EURORDIS**
- Adoption of the EU Cross-Border Healthcare Directive with a special focus on rare diseases and European Reference Networks
- Preparation of two new projects as part of the EUCERD Joint Action on Rare Diseases: Specialised social services and social policies & National conferences to discuss National Plans for Rare Diseases in 20 countries
- Contribution to the feasibility study to support the creation of a mechanism aimed at assessing the clinical added value of Orphan Drugs at EU level
- Contribution to the public consultation on the revision of the EU Clinical Trials Directive

## Information & Networking

- Organisation of Rare Disease Day 2011 in collaboration with partners in 56 countries worldwide
- 50 new members in 2011, bringing up total membership to 492 members in 46 countries
- Organisation of the **EURORDIS** Membership Meeting in Amsterdam
- 12th and 13th Workshop of the Council of National Alliances (25 National Alliances for Rare Diseases in Europe)
- 4th Workshop of the Council of European Federations (35 rare disease-specific networks)
- Continuation of "**EURORDIS** Programme of Support to European Federations"
- Counselling and support to three young rare disease Federations, as part of the Rare!Together project
- Launching of 12 new Online Patient Communities and continuous enhancement of rareconnect.org
- **EURORDIS** website recorded more than 18,000 visitors a month from 190 countries
- **EURORDIS** e-Newsletter added Russian as a seventh language
- **EURORDIS** Photo and Video Contest attracted entries from more than 25 countries



## Health Policy & Health Care Services

- Completion of the Patients' Consensus on Preferred Policy Scenarii for Rare Disease Projects (POLKA) project
- 206 deliberative policy debates Decide sessions organised in 2011 and a total of 320 Decide sessions, involving 1,704 participants in 22 countries, organised since the beginning of the project
- First field evaluation of Centres of Expertise involving rare disease patients and health professionals simultaneously in three Member States (UK, France and Denmark)



## Research, Drugs & Therapies

- Review of 377 dossiers on orphan drugs, advanced therapies and paediatric investigation plans, as part of participation in the European Medicines Agency's Scientific Committees
- **EURORDIS** Position paper "Patients' Priorities and Needs for Rare Disease Research in 2014-2020"
- Official creation of International Rare Disease Research Consortium (IRDiRC) and active participation of **EURORDIS** in preparatory meetings
- Administrative coordination hand-over of EuroBioBank to the Italian Telethon Foundation
- **EURORDIS** involvement in two new research projects to share good practice and develop educational material in the field of clinical trials
- Fourth **EURORDIS** Summer School for patient advocates held in Barcelona
- Development of an e-learning tool covering clinical trials design, statistics and ethics
- 14th and 15th workshops of the **EURORDIS** Round Table of Companies (ERTC)
- Compassionate use survey of 64 pharmaceutical companies



# Advocacy



## Sustaining Rare Diseases as an EU Public Health Priority

### EU Committee of Experts on Rare Diseases (EUCERD)

The eight 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 2011, and participated in the following EUCERD meetings:

#### Plenary meetings:

All appointed patient representatives participated in the EUCERD plenary meetings on March 22-23 and October 24-25.

#### Working groups/workshops meetings:

- 21-22 March: EUCERD Workshop on Centres of Expertise and European Reference Networks, in Luxembourg.
- 8 September: EUCERD Workshop on Centres of Expertise and European Reference Networks, in Luxembourg.
- 4 October: EUCERD Workshop on Rare Diseases Registries, in London. Patient representatives were involved, including Lesley Greene (COMP co-chair and speaker), Bianca Pizzera and Birthe Holm (COMP), as well as Fabrizia Bignami, Monica Ensini and Maria Mavris (COMP, CAT), and Ulrike Pypops (CF Association Belgium).
- EUCERD Workshop on Healthcare data management. Two patient representatives participated, Christel Nourissier and Gabor Pogany.

The most important achievement of the EUCERD in 2011 was the unanimous adoption of the Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases in Member States. This is the first set of recommendations adopted by this Committee. In order to share knowledge and expertise more efficiently, the EUCERD recommendations seek to introduce harmonious standards of quality practices by elaborating criteria for the Member States to incorporate into their process to designate Centres of Expertise.

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
- "Evaluation of population newborn screening practices for rare disorders in Member States of the EU"

All reports and recommendations produced by the EUCERD in 2011 are available on [eucerd.eu](http://eucerd.eu)

### EUCERD Joint Action on Rare Diseases

**EURORDIS** was involved in the preparation and elaboration of the "EUCERD Joint Action: working for Rare Diseases" (EJA) aimed at supporting the implementation of the Commission Communication "Rare Diseases: Europe's Challenges" and the Council Recommendation on Rare Diseases. The project, that will run from 2012-2015, brings together Member states and **EURORDIS** as the full partner NGO. It is funded by the European Commission (50%), Member states and **EURORDIS**.

**EURORDIS** Board Member, Dorica Dan, is the Leader of the Work Package: "Specialised Social Services and Integration of Rare Diseases into Social Policies and Services". This

part of the Joint Action is aimed at mapping specialised social services for rare disease patients and planning for integration of rare diseases into mainstream social policies and services.

**EURORDIS** is also involved in the Work Package on National Plans for Rare Diseases (EUROPLAN II), aimed at supporting the establishment of National Plans and strategies at Member State level by 2013, through national conferences, educational tools, methodological support, indicators and spreading of good practices.

## Rare Disease Day European Symposium, Brussels

A Rare Disease Day European event entitled, "European Symposium - Rare but Equal- Addressing Health Inequalities for Rare Disease Patients in Europe," was held on February 28th in Brussels at the International Press Centre Residence Palace, next to the European Commission and the European Parliament.

86 people attended, including representatives of the European Commission, Industry, Belgian patient organisations and authorities, European Federations and Member State Permanent Representations. There were delegates from Italy, Germany, the Netherlands, Switzerland, Norway, the United Kingdom and all the way from Romania and Bulgaria.

The attendance of MEP Antonyia Parvanova, (ALDE – Bulgaria) shadow rapporteur of the Cross-Border Healthcare Directive, was very timely given that the Directive was adopted that same day by the Council of Ministers.

The Symposium included presentations of three EU initiatives to address health inequalities: The EU Solidarity in Health Initiative, the EU Disability Strategy and the new EU Directive on Patients' Rights in Cross-Border Healthcare. This is the first time **EURORDIS** engaged with the Directorate for Employment & Social Affairs and the Directorate for Justice, Fundamental Rights & Citizenship. The rest of the programme showed prevalent inequalities through case studies, surveys and patient testimonies.

At the event, the European Commission announced the key results of the Eurobarometer survey on public opinion about rare diseases. The event was streamed live on the **EURORDIS** website and on the Rare Disease Day Facebook Group throughout the day.



## Making Rare Diseases a Public Health Priority in all Member States

### Advocacy support to National Plans for Rare Diseases

**EURORDIS** facilitated information exchange and networking about National Plans for Rare Diseases at the Council of National Alliances (May and November 2011). In January 2011, the last of the 15 EUROPLAN-EURORDIS National Conferences was held in Ireland. These Conferences were organised in partnership with the National Alliance in each country and in collaboration with the national competent authorities and main stakeholders. The Irish Conference concluded a one-year process that gathered 2,200 participants from all over Europe to discuss national plans on rare diseases in their respective countries.

**EURORDIS** produced the Final Report summarising the key outcomes of the 15 EUROPLAN National Conferences that was presented at the final conference of the EUROPLAN project in Rome in February 2011.

During the latter half of the year, the preparation of a second series of National Conferences, "EUROPLAN II", which is to be organised within the framework of the EUCERD Joint Action: working for Rare Diseases (see above), commenced. A call for applications addressed to **EURORDIS** National Alliances interested in organising a conference in 2012-2013 was launched and resulted in the selection of 20 of them. Also, the process for recruiting 9 EUROPLAN-EURORDIS Advisors began at the end of 2011.

A brand new section on national rare disease policies was developed on the **EURORDIS** website and will be launched in early 2012.

### Policy Fact Sheets for Rare Disease Patient Advocates

A new fact sheet on Newborn Screening was developed as part of the series of Policy Fact Sheets to empower patient advocates at EU, national and local level. These fact sheets are concise summaries of many of the policy

issues that correspond to the Council Recommendation on Rare Diseases and that are useful in advocating for National Plans.

## Advocacy

### Rare Diseases: An International Public Health Priority

- **EURORDIS** has initiated and developed a Joint Declaration, together with other umbrella patient organisations from North America, South America, Asia, Australia and New Zealand, called "Rare Diseases: An International Public Health Challenge."

The on-going Strategic Partnership between **EURORDIS** and the US National Organization for Rare Disorders (NORD) plays a key role in this respect with increasing coordination between the two organisations on advocacy actions and joint

projects. These include the promotion of the cause of rare diseases at the international level; a stronger collaboration on Rare Disease Day internationally; the launch of 12 new international rare disease online patient communities; the organisation of the first US Conference on Rare Diseases and Orphan Products in Washington in October 2011 and partnering for the European Conference on Rare Diseases and Orphan Products in Brussels in May 2012.



### Improving Access to Orphan Drugs

#### Clinical Added Value of Orphan Drugs (CAVOD)

Advocacy activities continued, in view of the creation of a mechanism of exchange of information and common assessment on the Clinical Added Value of Orphan Drugs (CAVOD) at EU level. **EURORDIS'** work in 2011 involved liaising with the European Commission, the European Medicines Agency (EMA), National Authorities, National Health Technology Assessment Agencies, pharmaceutical companies; as well as offering a platform for discussion at the **EURORDIS** Round Table of Companies.

A Call for Proposals was published by the European Commission which selected the consulting firm Ernst & Young to carry out a Feasibility Study on the Mechanism to Implement the CAVOD. **EURORDIS** participated in this

process and all the relevant meetings throughout 2011. The feasibility study has been presented on several occasions, including at the Committee of Orphan Medicinal Products and at the **EURORDIS** Round Table of Companies, and made publicly available by the European Commission in early December 2011. **EURORDIS** organised bilateral and multilateral meetings with different stakeholders, on a regular basis, and is involved in the drafting group of the future EUCERD recommendation on CAVOD.

#### Mechanism of Coordinated Access to Orphan Medicinal Products

**EURORDIS** has participated in the Mechanism of Coordinated Access to Orphan Drugs of the EU Transparency Platform since its creation in 2010. Throughout 2011 **EURORDIS** contributed to the definition of key concepts, glossary, prevalence of therapeutic indication and innovative tools for discussions on pricing.







## Improving Access to Quality Care

### Promoting Cross-Border Healthcare and Mobility of Patients

The long-lasting advocacy activities on cross-border healthcare and patient mobility have 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 work that **EURORDIS** has been carrying out on European Reference Networks for Rare Diseases, as well as on the cooperation between Member

States on Health Technology Assessment, is closely linked to the patient mobility and cross-border healthcare dimension. **EURORDIS** has been organising meetings with Commission officials and national authorities, both directly and through National Alliances, in order to understand how to optimise the national transposition of the Directive for the maximum benefit of rare disease patients. The advocacy activities in this area will continue in 2012 as Member States have 30 months to adapt their national legislation accordingly.

### Other issues



### Revision of the EU Clinical Trials Directive

In May 2011, **EURORDIS** sent a contribution to the European Commission's Public Consultation on the EU Clinical Trial Directive. Work on the revision of this Directive will continue and intensify in 2012.

**EURORDIS** has also been promoting the position and main points of concerns of rare disease patients regarding the scope of the coordinated assessment procedure for the authorisation of clinical trials and the composition of the ethics committees, at several proceedings, including the EFPIA Think-Tank Workshop at the European Parliament (1st December 2011).

### 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** carries out an internal reflection process on this revision, in particular in relation to patient registries.

### Rare Cancers

Rare Cancers are incorporated in all **EURORDIS** activities, from activities on orphan drugs, paediatric use of medicines, advanced therapies, to inclusion into national strategies and plans on rare diseases as well as strategies on Centres of Expertise or Registries. **EURORDIS** is calling for future coordination between the EU policy framework "European Partnership Against Cancer" and the EU Strategy on Rare Diseases, in particular through the EUCERD.



**EURORDIS** is consolidating its advocacy activity on rare cancers, mainly towards industry and patient groups and is allocating significant resources and efforts supporting and advising rare cancer patient groups to gain access to orphan drugs.

The dialogue with European Cancer Leagues, the European Society for Medical Oncology and the European Cancer Patient Coalition has been enhanced by the creation of Rare Cancers Europe.

# Information & Networking

## Membership base - 492 members

50 new members joined in 2011 (27 full members and 23 associate members). New countries represented: Brazil, Hong Kong, Mexico.

At the end of 2011, **EURORDIS** had 492 members in 46 countries, 34 of which are European countries, 24 being members of the European Union.

## EURORDIS Membership Meeting - Amsterdam 2011

In 2011 the **EURORDIS** Membership Meeting took place in Amsterdam, 13 and 14 May.

This two-day meeting, gathered 244 participants from 31 countries, around the theme "The Patient's Voice in National Plans for Rare Diseases".

The meeting included the **EURORDIS** General Assembly, which elected four new directors. The membership meeting consisted of one public conference and seven workshops:

- Four capacity building sessions on: Newborn Screening Centres of Expertise & Networks of Experts, Registries, Research

- Three Brainstorming & Policy Development sessions: Social aspects; Training, Information & Education of Medical Doctors; Prevention

Pre-conference sessions on May 13 included the Workshop of the Council of National Alliances, an Online Patient Communities Workshop, Decide sessions and a meeting of Dutch Patient Organisations organised by the Dutch Alliance VSOP and the Dutch Steering Committee for Orphan Drugs.



## Council of National Alliances

**EURORDIS** supports a network of 28 National Alliances for Rare Diseases, of which 25 constitute the Council of National Alliances (CNA). Two new National Alliances joined the Network in 2011: Austria and Russia.

In 2011, the Council of National Alliances met twice. The 12th Workshop was held in May in Amsterdam and the 13th Workshop was in November in Paris. Both workshops covered the following topics:

- The EUROPLAN Project and National Plans for Rare Diseases
- Rare Disease Day 2011 wrap up and preparation of Rare Disease Day 2012

- European Conference on Rare Diseases & Orphan Products 2012: the new format of ECRD and how National Conferences can be coordinated with European Conferences.

- Improving access to medicines; CAVOD and other specific measures that need to be integrated into National Plans

- International Rare Disease Research Consortium: advocacy to Member States' Ministers of Research to join the International Consortium as Funding Members

- EU Directive on Cross Border Health Care: Working together on the transposition into national legislation and policy

- National Alliances' involvement with regulatory affairs and contact with national medicines agencies



## Council of European Federations

The 4th Workshop of the Council of European Rare Disease Federations (CEF) met in June-July 2011 for a two day meeting gathering 18 representatives of European Federations and Networks.

The first day focused on Rare Disease Day - How can European Federations get involved; Impact of Cross-Border Healthcare Directive on rare disease patients; Centers of Expertise & European Reference Networks; the new European Clinical Trials Registry, Online Patient Communities. The second day was a capacity building on Clinical Trials.

In 2011, **EURORDIS** continued the pilot project "EURORDIS Programme of Support to European Federations" which intends to provide seed money to the smallest and/or youngest organisations which often have great difficulties in financing their networking activities.

Through this support programme, **EURORDIS** granted a total of 13,200 € to six European Rare Disease Federations.

## Rare!Together

Rare!Together aims at helping in the creation, operation and management of European Rare Disease Federations, in particular through the website [raretogether.eurordis.org](http://raretogether.eurordis.org). This website 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 the telephone, e-mails and participation in workshops and meetings.

Find more information at:  
[raretogether.eurordis.org](http://raretogether.eurordis.org)





## Information & Networking



## Rare Disease Online Communities - rareconnect.org

**EURORDIS**, in partnership with the US National Organization for Rare Disorders (NORD), has created the RareConnect project to help patients and families connect through an online social network, support each other and share vital experiences on aspects of living with a rare disease. The online patient community portal [rareconnect.org](http://rareconnect.org) is a multilingual platform in five languages (English, French, German, Spanish and Italian). 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.

There are 15 communities in total. These communities saw international partnerships emerge from 110 different patient organisations who formally partook in the setup and governance of each community.

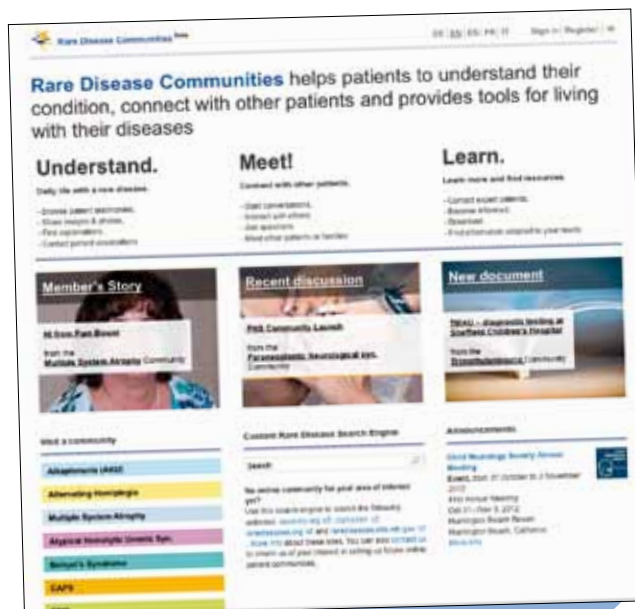
During 2011 the RareConnect website received over 70 000 visits from 166 countries which led to 900 patients or caregivers signing-up to participate in the service.

Of the 15 communities, 12 were launched during 2011 for the following diseases:

- Alkaptonuria
- Alternating Hemiplegia
- Atypical Hemolytic Uremic Syndrome
- Behçet's Syndrome
- Congenital Disorders of Glycosylation
- Cystinosis
- Epidermolysis Bullosa
- Familial Mediterranean Fever
- Glut 1 Deficiency
- Hereditary Spastic Paraplegia
- Multiple Myeloma
- Waldenstrom Macroglobulinemia

Three training workshops took place in order to further involve patient organisations in the governance and management of future online communities. In May, in Amsterdam, a workshop was dedicated to the RareConnect project and social media use and was included as a satellite workshop in the **EURORDIS** Membership Meeting. At the end of October, **EURORDIS** welcomed 23 patient representatives to Paris for a day, outlining how new patient groups can take part in the project. Finally, at the end of November, 12 moderators of current communities met in Barcelona to discuss their community's progress while learning about finding quality medical information and clinical trials online.

Find more information at:  
[rareconnect.org](http://rareconnect.org)



## Mailing Lists

In addition, **EURORDIS** continued its maintenance of 30 Mailing lists (listservs) for patient advocacy and patient support through a partnership with Médicalistes. In 2011, there were seven advocacy and governance mailing lists and 23

disease-specific mailing lists. The most active mailing lists are for Behçet's, European Public Affairs Committee and National Plans.



Find more information at:  
[eurordis.org](http://eurordis.org)

## EURORDIS Website - eurordis.org

The **EURORDIS** website, [eurordis.org](http://eurordis.org), recorded on average over 18,800 visitors a month from 190 countries over the course of the year. Russian was added as a new language allowing visitors to access the website, download information about **EURORDIS**, sign up for our monthly newsletter and access a recent archive of the e-Newsletter in the new language. The **EURORDIS** website is now available in seven languages (English, French, Spanish, Italian, German, Portuguese and Russian).

A new online learning module was developed to provide an accessible tool that can fulfil immediate training requirements for all patient organisation representatives. Several new video recordings from the **EURORDIS** Summer School were made available to help train patient representatives on various topics related to the orphan drug regulatory process. Lastly, a new section was added to explain and show the activities relating to the Decide policy debate sessions as part of the POLKA project.

## Rare Disease Blog - rarediseaseblogs.net



The EURORDIS/NORD sponsored blogging platform, [rarediseaseblogs.net](http://rarediseaseblogs.net), received more contributions from opinion leaders and patient advocates, featuring commentary on rare disease policy and orphan drugs. There were also more LIVE blogs directly written on-site from various congresses throughout the world. This sparked interesting debates and discussions on topics such as fundraising and communication.

Find more information at:  
[rarediseaseblogs.net](http://rarediseaseblogs.net)

## Information & Networking

### EURORDIS Social Media

**EURORDIS** continued to regularly communicate its activities and share information via Facebook ([facebook.com/eurordis](https://facebook.com/eurordis)) and Twitter ([twitter.com/eurordis](https://twitter.com/eurordis)). Our 2011 Social Media activity can be summarised in the following statistics:

- **EURORDIS'** updates on Facebook received 150,000 views generating 680 interactions with Facebook subscribers
- By the end of the year, **EURORDIS** had 2,000 people who had "liked" our page
- **EURORDIS** posted 156 tweets on Twitter to over 1,000 followers
- **EURORDIS** produced 30 videos which were made available on the YouTube channel [youtube.com/eurordis](https://youtube.com/eurordis) which generated 11,535 views from 121 countries
- **EURORDIS** also shared over 1,000 photos from its many activities via the social media photo sharing site Flickr [flickr.com/photos/eurordis](https://flickr.com/photos/eurordis) which generated close to 3,000 views
- **EURORDIS** shared over 70 slideshow presentations via [slideshare.net/eurordis](https://slideshare.net/eurordis)



### EURORDIS E-Newsletter

In 2011, **EURORDIS** produced and disseminated 10 monthly issues of the e-Newsletter in six languages (English, French, Spanish, Italian, German and Portuguese) to 8,200 subscribers. Starting with the November 2011 issue, we added one additional language (Russian).

Three special e-mailings in Russian were sent out to announce the new version of the Newsletter and to encourage Russian-speaking rare disease patients and other stakeholders to subscribe. The e-Newsletter was also promoted in key rare disease conferences in Russia. A landing page in Russian has been added to [eurordis.org](https://eurordis.org) with a sign-up box, the presentation brochure and a Newsletter archive in Russian.

### Presentation Brochure

A new presentation brochure describing **EURORDIS'** mission and action was adapted and translated into the seven languages of the **EURORDIS** website, including Russian.



### Photo & Video Contest

The **EURORDIS** Photo & Video Contest was held in March-April 2011. The contest was promoted through the **EURORDIS** website, the **EURORDIS** and Rare Disease Day Social Media as well as the **EURORDIS** e-Newsletter. The contest attracted 94 photo entries and 43 video entries. Photo submissions were received from 20 different countries, and showcased over 25 different rare diseases. Video submissions were received

from more than 25 different countries, and showcased more than 21 different rare diseases. The winning prizes went to people living with Achondroplasia and Moebius Syndrome respectively. All entries can be viewed on **EURORDIS** Flickr gallery and YouTube Channel.





## Rare Disease Day

Rare Disease Day 2011 took place on 28 February 2011. **EURORDIS** and its National Alliances, together with another 31 patient groups acting as country organisers, mobilised hundreds of patient organisations throughout five continents.

Patient organisations from 56 countries joined the campaign in 2011 (including 27 in Europe). Newcomers included Mexico, Thailand, Panama, Armenia, Uruguay, Nepal, Dubai, Morocco, Peru and Iran. In line with the 2011 theme "Inequalities" and the slogan: "Rare but Equal", **EURORDIS**



organised a one-day symposium in Brussels in partnership with the European Commission. (for further information refer to "Rare Disease Day European Symposium, Brussels" in the Advocacy section of this report).

The website [rarediseaseday.org](http://rarediseaseday.org) received 50,000 visits from 150 countries in two months. The website contained a calendar of events organised by country. In addition, it

included a downloadable toolkit with an information pack, logo, poster and other materials accessible to organisers.

292 individuals, patient organisations, industry, health professionals, research and public authorities signed up as 'Friends of Rare Disease Day'.

Other interesting features in 2011 included a special section on the website on "Rare Diseases and Health Inequalities" featuring more than 50 testimonies from patients about their experience of inequality in access to treatments, care and social services.

986 posters were printed and sent out to member patient organisations, National Alliances, European Federations, researchers, policy-makers, industry and other relevant stakeholders.

On-line news articles were collected through a media monitoring service and a newsfeed displayed them on the home page of the website.

## Rare Disease Day Social Media

Rare Disease Day activities and general information was regularly shared via:

Facebook ([facebook.com/rarediseaseday](https://facebook.com/rarediseaseday)), Twitter ([twitter.com/rarediseaseday](https://twitter.com/rarediseaseday)), Youtube ([youtube.com/rarediseaseday](https://youtube.com/rarediseaseday)), Flickr ([flickr.com/photos/rarediseaseday](https://flickr.com/photos/rarediseaseday)). Our Rare Disease Day social media activity can be summarised in the following statistics:

- Total 14,000 fans on Rare Disease Day Facebook 2010 and 2011
- 15,000 followers on Twitter
- Total 432 photos and 100 videos shared through the "Tell Your Story" section of the website



Help Lines  
for Rare  
Diseases

## European Network of Rare Disease Help Lines

Signatures of letters of Agreement between help lines and the European Network of Help Lines for Rare Diseases was coordinated by **EURORDIS**. These agreements define the membership status of each help line as a full or associate member or as an observer.

A fourth Caller Profile Analysis was conducted in November 2011: 11 help lines participated, for a total of 1,739 calls or emails received, which addressed 394 distinct rare

diseases. It was the first time that it was possible to analyse for which disease questions came in, since 7 out of 10 help lines are now using the Orpha Codes for rare diseases.

Other activities of the European Network of Rare Disease Help Lines have been suspended since August 2010 due to a lack of operating funds. **EURORDIS** is looking to secure adequate funding from public or private sources from 2012 - 2013 onwards.

# Health Policy & Health Care Services

## Patients' Consensus on Preferred Policy Scenarios for Rare Diseases project— POLKA

The POLKA project, which started in 2008 was completed on the 31 August, 2011. The objectives of the POLKA project were to democratise, enlarge, and promote the opinions of rare disease patients' key health policies.

The project was based on three pillars, two of which were conducted until August 2011 and are described in detail below. A third, the 5th European Conference on Rare Diseases which took place in 2010, was included in last year's report.

### Deliberative patients' debates – Decide

Six new Decide kits were created on the following topics chosen for their importance to rare disease patients: newborn screening, cross border health care, the cost of care (the case of orphan drugs), stem cell research, diagnosis information and counselling, pre-implantation genetic diagnosis. The topics have been translated into most European languages (21 total, which is a first for **EURORDIS**). We also contributed to the overhaul of the dedicated website: [playdecide.eu/getinvolved/projects/](http://playdecide.eu/getinvolved/projects/)

In total, 320 sessions have taken place, involving 1,704 participants (most of whom are patients or patient representatives) from 22 countries. Hungary, Norway and Spain organised almost half of the sessions.

### European Reference Networks and Centres of Expertise for rare diseases

The objective was to foster the involvement of patient representatives in shaping the policy of European Reference Networks for Rare Diseases and continue advising the European Commission on its policy regarding these networks.

To analyse the implication of patient organisations in existing European Reference Networks (ERN), **EURORDIS** used its involvement as a partner or as a member of the Advisory Board of some ERNs funded by the European Commission, and conducted interviews with ERN academic leaders and patient organisation representatives. On the basis of these interviews, guidelines on good practices for collaborating and a template Work Package for future collaboration between ERNs and patient groups have been developed and adopted by the **EURORDIS** Council of European Federations.

A total of 207 Danish, British and French patients, patient representatives and health care professionals, were involved in this qualitative process (based on Delphi methods). The recommendations delivered by this project are the direct expression of their discussion and opinions. To our knowledge, this marks the first "field" evaluation of Centres of Expertise, simultaneously in three EU Member States involving both patients and health care professionals. The results were presented to the EUCERD and contributed to the definition of quality criteria for Centres of Expertise.



## Collaboration with European Reference Networks

- European Network on Rare Paediatric Neurological Diseases (nEUroped) to develop a network of communication and information sharing across the field of rare nervous system disorders in children, characterised by paroxysmal attacks.

- European Registry and Network for Intoxication-type Metabolic Diseases (E-IMD) to promote health for individuals affected with rare organic acidurias (OADs) or urea cycle defects (UCDs).

## Country visits to Austria, Germany, Georgia, Latvia and Russia

In March 2011, **EURORDIS** met with 19 patient representatives, policy makers and health care professionals in Riga. The purpose was to learn more on the reality of rare diseases in Latvia and to envisage future working relations.

**EURORDIS** partnered with the Georgian Foundation for Genetic and Rare Diseases in organising the Second South Caucasia Conference on Rare Diseases and Orphan Drugs on 27-28 October 2011, meeting with patient representatives, policy makers and healthcare professionals in Tbilisi.



## Therapeutic Recreation Programs

In the Spring, employees of members of the **EURORDIS** Round Table of Companies were encouraged to volunteer and share their skills at therapeutic recreation summer camp programmes as part of the European Year of Volunteering 2011.

In October, **EURORDIS** presented future partnership developments at the European Network of Hole in the Wall Camps' meeting at Bator Tabor camp in Hatvan, Hungary. Members of the Therapeutic Recreation Camp European

Network include camps in France, Ireland, Hungary, Italy, and the United Kingdom.

At the **EURORDIS** Round Table of Companies meeting in November 2011, two representatives from the Hole in the Wall Camps' network discussed volunteer opportunities and the needs of therapeutic recreation programs while highlighting the vital service these camps provide for young people with rare diseases.





# Research, Drugs & Therapies

## EURORDIS Position Paper “Patients’ Priorities and Needs for Rare Disease Research (2014-2020)”

In 2011, **EURORDIS** developed a second Position Paper on rare disease research, which coupled with the 2010 Position Paper “Why Invest in Rare Disease Research”. Relevant arguments were collected in favour of targeted and increased investment in rare disease research at national and EU levels.

The new Position Paper is the result of extensive consultations within the **EURORDIS** Public Affairs Committee and with members (presentation and discussion at the

**EURORDIS** Membership Meeting in May), as well as externally with the EU Public Affairs Committee, selected scientists, experts and stakeholders.

The Position Paper adopted in October 2011, is publicly available in the Library of the **EURORDIS** website.

## International Consortium for Rare Disease Research (IRDiRC)

**EURORDIS** representatives participated in the high-level preparatory meetings in view of the creation of an International Consortium for Rare Disease Research (IRDiRC), led by the European Commission and the US National Institutes of Health. **EURORDIS** was present at the two meetings organised in 2011, in Washington DC in April for the official launch, and in Montreal, Québec, Canada in October 2011, for further consolidation of policy and governance aspects.

**EURORDIS** appointed representatives in working groups of the IRDiRC and actively contributed to shaping the newly established Consortium 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’ involvement in biobanking activities: the EuroBioBank Network ([eurobiobank.org](http://eurobiobank.org))

After ensuring the administrative coordination of the network for 11 years, **EURORDIS** has decided to step down and transfer this responsibility to the Italian Telethon Foundation, at the beginning of 2012.

By the end of 2011, over 400,000 rare disease samples (DNA, Cell and Tissue) were available across the EuroBioBank Network. An indicator of the network’s importance is the sample activity: 25,600 samples were collected and 7,600 samples distributed. Research conducted with samples from the network resulted in the publication of 43 scientific articles acknowledging EuroBioBank.

Further work on quality control was performed in 2011, enabling the EuroBioBank members to explore the possibility of accreditation of their biobanks, as well as to determine the long-term degree of satisfaction and scientific value of the samples sent out a few years ago. In 2011, with the end of its participation in the TREAT-NMD Network of Excellence, the EuroBioBank network prepared for the future by applying to the FP7 Health’s Call for Proposal.

## Collaboration with rare disease research networks

**EURORDIS** collaborated in the following rare disease research networks supported by the European Commission:

### ECRIN-IA

The European Clinical Research Infrastructures Network is a network of excellence gathering the main public clinical research centres. The goal is to share good practices and methodologies facilitating international clinical trials in all fields. ECRIN and **EURORDIS** have been collaborating since the onset of this network more than five years ago. As a patient representative, **EURORDIS** has been invited to participate in the ECRIN-IA (Integrated Activity) project as a partner for the next five years.

### E-RARE

E-Rare 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. **EURORDIS** is a member of the External Advisory Board.

### TREAT-NMD

TREAT-NMD (Translational Research in Europe – Assessment and Treatment of Neuromuscular Diseases) was established as an EU funded 'Network of Excellence' with the remit of 'reshaping the research environment' in the neuromuscular field. From January 2007 to December 2011, **EURORDIS** was

involved as a partner, representing EuroBioBank to support the biobanking, as well as some of the patient registries activities of the project.

The European TREAT-NMD network has developed into a global organisation bringing together leading specialists, patient groups and industry representatives to ensure preparedness for trials and therapies of the future, while promoting best practices today. With the end of the five-year EC funding, TREAT-NMD pursues its missions as a self-sustained organisation called the "TREAT-NMD Alliance". More information: [treat-nmd.eu/](http://treat-nmd.eu/)

### EUPATI

The European Patients' Academy on Therapeutic Innovation (EUPATI) is 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 will start in 2012.

The Consortium comprises a unique combination of pan-European patient organisations, academic and not-for profit organisations and 16 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 and 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: [patientsacademy.eu](http://patientsacademy.eu).

## 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 Medicinal 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 377 dossiers as part of the scientific committees they belong to. These include: 166 dossiers for orphan drug designations, 24 classification or certifications by the CAT and 187 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 2011, 16 patient representatives attended meetings of the SAWP for Protocol Assistance. Protocol Assistance is scientific advice for orphan drugs which helps the company make sure that it performs the appropriate tests and studies, so that no major objections regarding the design of the tests are likely to be raised during evaluation of the marketing authorisation application. ➤

## Research, Drugs & Therapies

► In addition, two patients participated at the COMP on disease-specific issues and one consultation was carried out Europe-wide with disease-specific patient groups in order to assist the Committee to arrive at the best decision concerning the product in question.

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

Three **EURORDIS** Task Forces involving a total of 34 patient representatives and volunteers in the areas of Drug Information, Transparency and Access (DITA); Orphan Drugs and Paediatric Drugs were continued, as well as the organisation of two meetings for the DITA task force.

The Drug Information, Transparency and Access (DITA) Task Force met three times in 2011. It exchanged information on the following subjects: Fabrazyme and Replagal shortages, involvement of patients' organisations in the activities of their national regulatory agencies, self-reporting of adverse drug reactions, off-label use of medicines for rare diseases, compassionate use programmes for orphan drugs, the situation regarding some treatments for Friedreich Ataxia in France, the EMA workshop on clinical development and scientific advice in ophthalmology, the EMA SAG meeting

about a new medicine for Addison's diseases and **EURORDIS** contribution to the European Commission DG Enterprise consultation on a possible revision of the Transparency Directive 89/105 EEC, and the new Pharmacovigilance legislation. An important discussion with the Agency was about the new policy on Conflicts of Interest, which will continue in 2012.

The DITA Task Force also advised the EMA on its new functions for the public part of its European register on clinical trials and its European pharmacovigilance database Eudravigilance.

DITA reviewed eight European Public Assessment Reports and 12 package leaflets. The Committee of Human Medicinal Products (CHMP) requested an opinion for a benefit/risk evaluation on four occasions.

Richard West from Behçet's Syndrome Society UK, was appointed as **EURORDIS** representative in the Working Party with Patients' and Consumers' Organisations (PCWP) by the Board of Directors.



## EURORDIS Summer School for advanced patient advocates

The 4th **EURORDIS** Summer School Session was held in Barcelona in June 2011 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 of 43 participants representing 19 countries and 24 different rare diseases.

A combination of small group sessions and formal presentations are 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.

## 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. In addition, all presentations have been recorded and are available for viewing in the Training Resources section of the **EURORDIS** website.

All of the above experiences will be further developed to include additional topics and to target different disease groups through the EUPATI project (see Collaboration with rare disease research networks in this section).





## 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, six pharmaceutical companies have signed the Charter and two Agreements of Understanding have been drawn up. All are available on the **EURORDIS** website.

In 2011, patient representatives from Tuberous Sclerosis Complex 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.

## Health Technology Assessment

Health Technology Assessment (HTA) plays an increasing role throughout Europe for evaluation of and access to therapeutic interventions for rare disease patients. It will play an even increasing role within the mechanism of the Clinical Added Value of Orphan Drugs (CAVOD) (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.

Three patient representatives from the **EURORDIS** staff and Summer School attendees were identified and selected to participate in a four-day training at the University of Hall in Tyrol, Austria.

**EURORDIS** published an article "Health Technology Assessment and patients' rights: "Assess versus access?" in *Pharmaceuticals Policy and Law*, 13 (2011) 169-176, IOS Press.

## The EUnetHTA Joint Action

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. **EURORDIS** is one of the four patient representative organisations at the EUnetHTA Stakeholders Forum.



## Research, Drugs & Therapies



### Drug Information Association (DIA) Patient Fellowship Programme

The DIA Patient Fellowship Programme enabled 28 patient representatives, including nine speakers, from 14 countries to participate in the DIA 23rd EuroMeeting in Geneva on 28-30 March.

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.

## Compassionate Use

**EURORDIS** conducted a survey regarding pharmaceutical companies' experience of compassionate use during the last three years. A questionnaire was sent to 64 companies which either obtained a market authorisation for a designated orphan drug over the last three years and/or were a member of EBE/EuropaBio or of the **EURORDIS** Round Table of Companies. Responses were obtained from 17 companies

for 19 products, of which information on compassionate use was available for nine. The compassionate use programmes for the nine products mentioned above, were available in 42 countries, representing a total of 72 different programmes. The full results were presented at the **EURORDIS** Round Table of Companies Workshop on Compassionate Use on 21 November 2011.

### EURORDIS Round Table of Companies (ERTC) - 2011 workshops



#### → “Compassionate Access to Rare Disease Therapies”

**21 November 2011, Paris: 82 attendees from 16 countries**

The main objective of the meeting was to take stock of the progress made and discuss the difficulties encountered, six years after the inclusion of Compassionate Use in the European legislation, and the 2004 ERTC Workshop on this topic.

#### → “Mechanisms for the Implementation of the Clinical Added-Value (Relative Efficacy or Relative Effectiveness) of Orphan Drugs, so called CAVOD”

**27 May 2011, Barcelona: 84 attendees from 20 countries**

The main objective of the meeting was to discuss the preliminary results of the Feasibility Study on CAVOD performed by Ernst & Young with all stakeholders (see Advocacy section)

#### → In 2011, the ERTC comprises:

- 37 members
- Five new members: Actelion, Aegerion, ARIAD, GSK Rare Diseases and Médunik Canada
- Two upgrades: Orphan Europe and Swedish Orphan–Biovitrum from 5,000 € to 10,000 €



# Governance, Organisation & Funding

## Board of Directors

A new member, Dimitrios Synodinos representing the Greek National Alliance (PESPA), was elected at the General Assembly 2011. Dorica Dan, Rosa Sanchez de Vega and Torben Gronnenbaek were re-elected as Directors at the General Assembly 2011. The Board is composed of 12 members from nine European countries.

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

Flavio Minelli stepped down after six years on the Board of Directors of **EURORDIS**.

## Staff Team Organisation

The team comprised 23 people 20 full-time equivalent (FTE) as of December 2011. The team is composed of paid staff, one office volunteer, one consultant and one free-lance staff. Most staff is 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 Chief Executive Officer shares his time between the two offices.

### The following are the main changes in human resources in 2011:

- Nathacha Appanah, Communications Assistant, has left **EURORDIS**
- Monica Ensini, Registry and BioBanks Manager, has joined the team
- François Faurisson, Clinical Research Advisor, returned to INSERM

- Fabrizia Bignami, Therapeutic Development Director, has left **EURORDIS**
- Leif Jiskoot, Health Policy POLKA Assistant, has left **EURORDIS** after the end of the POLKA project
- Charles-Edouard Brossard, Administrative Secretary Trainee, has left the team to be replaced by Vanumathy Christie-Balendran for a two-year period
- Lara Chappell has joined the team as Communications Assistant, part-time (80%)





## Governance, Organisation & Funding

### Volunteers

**EURORDIS** counted 59 volunteers in 2011, including one permanent office volunteer, six volunteer translators and 52 volunteer patient advocates.

The **EURORDIS** volunteer patient advocates play an active role in our advocacy activities aimed at shaping European healthcare policies for rare diseases. They participate in committees, working groups, task forces and they speak

at international conferences. Most of these volunteers are either patients or parents of patients.

We are proud and very grateful to count 14 volunteers representing **EURORDIS** and the interests of 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):

Mrs Lesley Greene (**EURORDIS**, UK) and Ms Birthe Byskov Holm (Danish Osteogenesis Imperfecta Society and Rare Disorders, Denmark), who is the Vice-Chair of the COMP. Dr Maria Mavris, Drug Development Programme Manager, is an 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):

Patient representatives Dr Fabrizia Bignami (Therapeutic Development Director), full member and her alternate Dr Michele Lipucci di Paola (Associazione Veneta Lotta Talassemia, Italy).

#### → EMA Patients' and Consumers Working Party (PCWP):

Ms Lise Murphy, the PCWP Co-Chair, (Marfan Syndrome, Sweden), Mr Richard West (UK Behçets Syndrome Society), appointed in September 2011, and Mr François Houyez, Health Policy Director.

### 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)
- Torben Grønnebaek (Wilson Disease and Rare Disorders Denmark)
- 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 (Williams Syndrome and Rare Diseases Hungary).

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





## 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 2011 ("EURORDIS\_FY2011"), single beneficiary, DG Health and Consumers, 12 months
- Epirare - European Platform for Rare Diseases Registries, associated beneficiary, DG Health and Consumers, 30 months, 2011-2013

### → Renewed

- Web2.0, The French Pharmaceutical Companies Association (LeeM), 12 months
- Summer School Session 2011, Drug Information Association Philanthropy Programme

### → Ended

- POLKA, main partner, DG Health and Consumers, 36 months 2008-2011
- EuroPlan, associated partner, DG Health and Consumers, 36 months 2008-2011
- nEUroped, associated partner, DG Health and Consumers, 36 months 2008-2011
- Treat-NMD (EuroBioBank), associated partner, DG Research, 60 months 2007-2011

### → Ongoing

- Advocacy and core activities, AFM-Téléthon, 48 months 2010-2013
- Rare!Together, Medtronic Foundation, 24 months 2011-2012

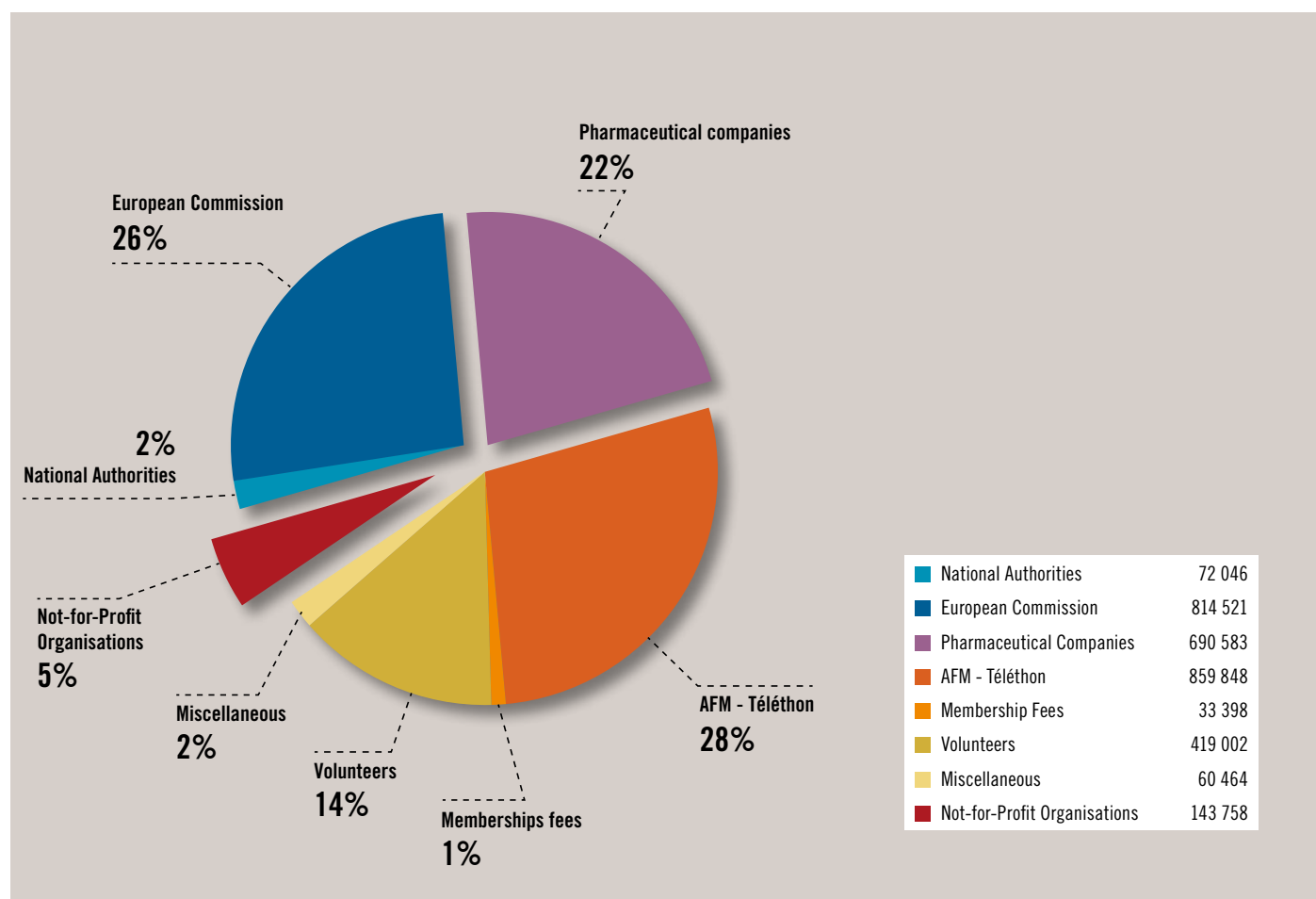


# REVENUE and expenses 2011

(in euros, provisions excluded)



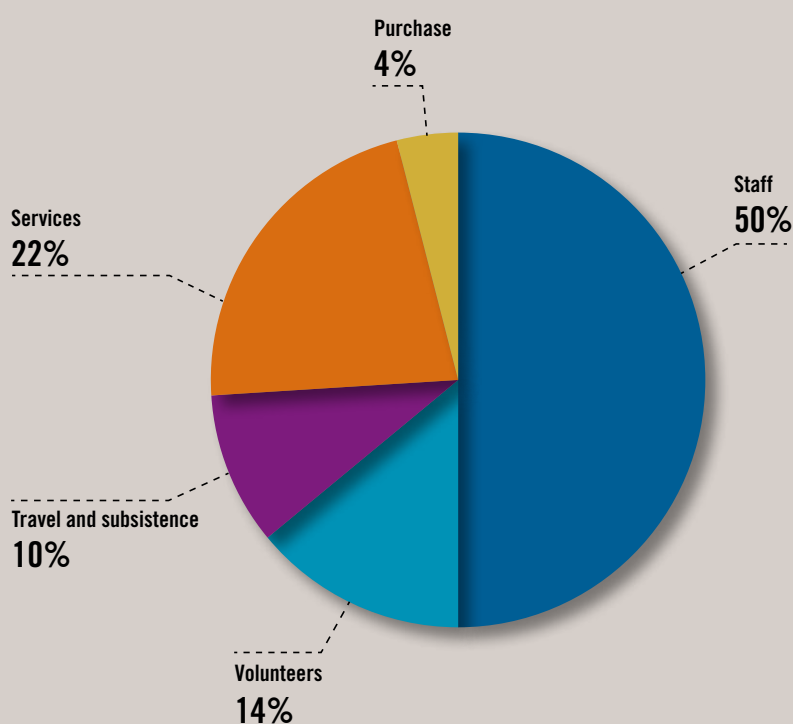
**Revenue by origin 2011 = 3,094 k€**





→  
**Expenses by Type 2011 = 3,148 k€**

■ Staff	1 578 839
■ Volunteers	419 002
■ Travel and subsistence	320 892
■ Services	694 136
■ Purchase	135 144



# BOARD OF DIRECTORS

May 2011 – May 2012



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# Conferences 2011

→ “Access to Medicines in Europe”: Meeting of the EC Project group on a Mechanism of Coordinated Access to Orphan Medicinal Products, Warsaw, Poland, 14 December

**Yann Le Cam represented EURORDIS.**

→ European Haemophilia Consortium Roundtable of Stakeholders: “Supporting R&D in rare diseases: barriers and incentives”, Brussels, Belgium, 7 December

**Yann Le Cam: “EU Regulation on Orphan Drugs: 11 years on what challenges remain for the patients”.**

→ “Clinical Trials- New Therapies”, Greek Alliance of Rare Diseases, Athens, Greece, 17 December

**Dimitris Synodinos represented EURORDIS**

→ NORD (National Organization for Rare Disorders) New York Regional Membership Meeting, New York, December 1-2, 2011

**Rob Pleticha: “RareConnect – Rare Disease Online Communities”**

→ EUROPABIO Workshop on “The Benefits of a Simplified and Coherent Clinical Trials Framework in Europe, European Parliament, Brussels, Belgium, 1 December

**Flaminia Macchia: “Patient’s perspective: a viewpoint from RDs patients on the Revision of the Clinical Trials Directive”**

→ EngageHealth: “The future of healthcare in Europe, closing the stakeholder gap”, Brussels, Belgium, 28 November

**Denis Costello: “Working across borders for patients with rare diseases”**

→ “Autoimmune Diseases, Today and Tomorrow”, Meeting of the Greek Alliance for Rare Diseases, Athens, Greece, 25-26 November

**Dimitris Synodinos represented EURORDIS**

→ XIV Meeting of the Italian Human Genetics Society, Milan, Italy, 13-16 November

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

→ The International ITP (Idiopathic Thrombocytopenic Purpura) Assembly 2011, Brussels, Belgium, 11-12 November

**Rob Pleticha: “Social media and patient networking for rare disease advocacy groups”**

→ TREAT-NMD (Neuromuscular Diseases) Global Conference, Geneva, Switzerland, 8-11 November

**Fabrizia Bignami and Anne-Mary Bodin attended as EURORDIS representatives**

→ Second FEDER-CREER School (Escuela de Formación FEDER-CREER), Burgos, Spain, 6 November

**Rosa Sanchez de Vega: “Situación de las Enfermedades Raras en Europa” (state of the art on rare diseases in Europe)**

→ All-Russia Civil Conference of rare diseases patients “Together for the benefit patients with rare diseases”, Samara, Russia, 4-5 November

**Anja Helm and François Houÿez: “Presentation of EURORDIS”, “Patient Advocacy”**

→ RARE 2011 – The Eurobiomed meeting on rare diseases and EUCERD/Eurobiomed event: “Shared data to improve healthcare management for rare diseases”, Montpellier, France, 2-4 November

**Christel Nourissier: « Du coût au prix des médicaments : les étapes et les processus de décision » (from cost to price of drugs: steps and decision making processes)**

**Christel Nourissier: Panelist in the Round Table: “Improving Access to Expertise and Quality Care”, 4 November**

**Gabor Pogany: Panelist in the Round Table: “Ensuring Visibility of Rare Diseases in Health Information Systems”, 4 November**

→ 2nd South Caucasian Conference on Rare Diseases and Orphan Drugs, Tbilisi, Georgia, 27-28 October

**Yann Le Cam: “The European Organisation for Rare Diseases: Sharing Experience on Integrating Patients & Parents Voice in the Policy Making Process”**

→ Workshop “Advanced Therapy Medicinal Products: from Promise to Reality” co-organised by the European Medicines Agency (EMA) and the European Society of Gene & Cell Therapy (ESGCT), Brighton, UK, 27 October

**Michele Lipucci di Paola represented EURORDIS in his capacity of member of the EMA Committee for Advanced Therapies (CAT)** ➤



→ “European Actions to improve the life of patients living with rare diseases”, Executive Agency for Health and Consumers, Luxembourg, 25 – 26 October 2011

Testimonials by patients or family members on concrete impact of EU funding/ policies in everyday life – representatives of **EURORDIS**:

**Avril Daly**, Ireland, patient living with Retinitis Pigmentosa, member, Irish National Alliance

**Lesley Greene**, UK, mother of a child with Cystinosis, member of the EMA Committee for Orphan Medicinal Products

**Yann Le Cam**, France, father of a child living with Cystic Fibrosis, Chief Executive, Rare Disease Europe (**EURORDIS**)

Video stories of **Béatrice de Montleau**, **Tsveta Schyns** and **Karen Aiach**

**François Houÿez**: **EURORDIS**: “A European organisation of patients in action”

→ “Together Against Genodermatoses” – under the patronage of French Health Minister, Mr. Xavier Bertrand, Hôpital Necker, Paris, France, 13-14-15 October 2011

**Anja Helm**: “Presentation of **EURORDIS**”

**Rob Pleticha**: “RareConnect Online Patient Communities”

**John Dart** and **Flaminia Macchia** participated in the Round Table

→ **NORD Rare Diseases/Orphan Products Summit**, Washington D.C., USA, 11-13 October

**Yann Le Cam**: “A patient’s perspective: International development and access to medicines”

**Sharon Ashton** also represented **EURORDIS**

→ **International Rare Disease Research Consortium (IRDiRC)**: European Union – USA Bilateral Workshop on Rare Diseases and Orphan Products, Montreal, Canada, 8 - 9 October

**Béatrice de Montleau**, **Valentina Bottarelli**, **Marlene Haffner** and **Prof. Josep Torrent Farnell** represented **EURORDIS**

→ **European Haemophilia Consortium Conference**, Budapest, Hungary, 7 -9 October

**Gabor Pogany** was in charge of the **EURORDIS** booth

→ **14th European Health Forum**: “European health in 2020 and beyond”, Gastein, Austria, 5-8 October

“**European Rare Disease Policy**” Session at Gastein”, 6 October

**Christel Nourissier**: “Actions and positions of patient organisations in helping shape rare diseases policies at European and Member States level”

→ **EUCERD/EMA Workshop**: “Towards a public-private partnership for registries in the field of rare diseases”, EMA London, 4 October

**Lesley Greene**, co-chair of the session: “Overview of the current situation and identified issues for discussion between stakeholders”

**Fabrizia Bignami**, **Monica Ensini**, **Maria Mavris**, **Birthe Holm**, **Bianca Pizzera** and **Ulrike Pypops** represented **EURORDIS** as experts

→ **5e Journée de la Myopathies et des Maladies Rares (5th Day for Rare Diseases and Neuromuscular Diseases)**, Luxembourg, 1 October

**Paloma Tejada**: Presentation of the “**International Rare Disease Day**”

→ **The European Multidisciplinary Cancer Congress**, Stockholm, Sweden, 23 – 27 September

**Advocacy and Ethics Session**: “How Healthcare Professionals and Patient Groups are Using Social Media”

**Denis Costello**: “Rare Cancer Communities Participating in the **RareConnect.org** project”

→ **The Post-Approval Summit – Europe**, Zürich, Switzerland, 21-22 September

**Fabrizia Bignami**, **Monica Ensini** and **Maria Mavris** represented **EURORDIS**.

→ **EFPIA Patient Think Tank meeting**, Brussels, Belgium, 22 September

**Lise Murphy**: “Recent developments and impression of patient involvement in the EMA”

→ **Health Activism in Europe Today**, EPOKS - European Patient Organisations in Knowledge Society - Participative Conference, 14-16 September, Lancaster, UK

**David Oziel** and **François Faurisson** represented **EURORDIS**

→ **9th EUROBIOBANK Network Annual Meeting & Treat-NMD WP04.1 Meeting**, Paris, France, 8-9 September

Co-organised by **Fabrizia Bignami** and **Anne-Mary Bodin**

→ **EUCERD Workshop** on a **EUCERD Recommendation on Quality Criteria for National Centres of Expertise for Rare Diseases**, European Commission, Luxembourg, 8 September

**Dorica Dan**, **Yann Le Cam**, **Christel Nourissier** and **Gabor Pogany** represented **EURORDIS**

→ 4th FORESIGHT TRAINING COURSE – Gianni Benzi Foundation - Evidences for rational therapies: from new born to elderly population, Rome, Italy, 1-3 September

**Round Table 3: Access to therapies for patients and Health professionals**

**Simona Bellagambi: “Access to therapy in rare conditions”**

→ EPIRARE Kick off Meeting, Rome, Italy, 11-12 July

**Fabrizia Bignami and Monica Ensini represented EURORDIS**

→ XIIe Forum des Associations (Orphanet –Alliance Maladies Rares: “Partager les données de santé pour une meilleure prise en charge”, Paris, France, 30 June

**Monica Ensini represented EURORDIS**

→ Opening of “The pilot Centre for Rare Diseases and virtual Rare Disease Training E-University” (NoRo project), Zalau, Romania, 28-29 June

**The Centre is managed by Dorica Dan**

**Christel Nourissier presented at the opening: “State of the art of Rare Diseases in Europe”**

→ 5th Europaediatrics Congress, Vienna, Austria, 23-26 June

**Dorica Dan: “The role of family and patient associations: the case of rare diseases”**

→ Meeting of the Greek Alliance of Rare Diseases (PESPA), Athens, Greece, 22 June

**Dimitris Synodinos represented EURORDIS**

→ Consensus Workshop of the EU Network of Experts on Neonatal Screening, Luxembourg, 20-21 June

**Fabrizia Bignami represented EURORDIS**

→ 6ème Conférence Nationale de Santé: “Le patient au centre des préoccupations”, droits des patients, rôle des associations de patients, importance de l’alliance des patients”, Luxembourg, 15 June

**Yann Le Cam: “Vers un réseau d’associations de patients”**

→ The 16th Congress of European Hematology Association, London, UK, 9-11 June

**Denis Costello: “Social media and patient networking”**

→ Joint Informal Meeting of EMA CHMP-COMP-PDCO-CAT, Budapest, Hungary 31 May & 1 June

**Lesley Greene and Birthe Holm represented EURORDIS**

→ Joint **EURORDIS/Ernst&Young** - 14th ERTC Workshop: “Mechanisms for the Implementation of the Clinical Added-Value (Relative Effectiveness) of Orphan Drugs, CAVOD”, Barcelona, Spain, 27 May

**Fabrizia Bignami: “How can patients contribute to the CAVOD mechanism?”**

→ Ecsite Annual Conference 2011, Warsaw, Poland, 26-28 May

**François Houyez represented EURORDIS**

→ 37th Panhellenic Medical Conference (organized by Tuberous Sclerosis Association of Greece, The Greek Alliance for Rare Diseases and The Hellenic Paediatric Neurology Association), Athens, Greece, 20 May

**Dimitris Synodinos represented EURORDIS**

→ NORD Corporate Council Meeting, Washington D.C., 17 May

**Yann Le Cam: NORD & EURORDIS: “A new international collaboration”**

**Fabrizia Bignami, Jill Bonjean and Sharon Ashton also represented EURORDIS**

→ E-Health Week, Budapest, Hungary, 10-12 May

**Gabor Pogany: “The Perfect” Handling of Patient Data – the Patients’ Perspective”**

→ EUneHTA Stakeholder Forum Meeting, Brussels, Belgium, 3 May

**Fabrizia Bignami and Maria Mavris represented EURORDIS**

→ Heads of Medicines Agencies Meeting, Visegrad, Hungary, 28-29 April

**Lise Murphy: “EMA model of working with patients and consumers”**

- **Second All-Russian Conference for Rare Diseases and Rarely Used Medical Technologies, Saint-Petersburg, Russia, 21-22 April**  
**Yann Le Cam: "EU Policy on Rare Diseases and the Involvement of Russia"**  
**Yann Le Cam: "The European Organisation for Rare Diseases: Sharing Experience in Promoting Interest of Patients with Rare Diseases"**
- **2nd EU-US Meeting for the International Consortium on Rare Diseases Research, Washington D.C. USA, 5-8 April**  
**Yann Le Cam and Valentina Bottarelli represented EURORDIS**
- **Stakeholder Workshop on the Revision of the Clinical Trials Directive, European Commission, Brussels, Belgium, 31 March**  
**Fabrizia Bignami and François Faurisson represented EURORDIS**
- **High-Level Lunch on "The Development of Paediatric Medicines: the Trends and the Challenges", Brussels, Belgium, 29 March**  
**Flaminia Macchia represented EURORDIS**
- **DIA EuroMeeting 2011, Geneva, Switzerland, 28-30 March**  
**Maria Mavris: "Patients: an increasingly influential voice in drug development. Patients: their voice"**  
**Yann Le Cam: "Contribution of Patient's Organisations in the Decision Making of Benefit/Risk Assessment"**  
**Yann Le Cam, Chair of the session: "Can we reach some harmonization across Europe on HTA Review, assessment of value product, added value product and reimbursement decision process"**
- **EUCERD workshop on initiatives and incentives in the field of centres of expertise for rare diseases and European collaboration between centres of expertise, Luxembourg, 21-22 March**  
**Gabor Pogany: Co-chair of Session 1 on Centres of Expertise**  
**Yann Le Cam, Co-chair of Session 2 on European collaboration between expert centres, healthcare pathways and European Reference Network**  
**Christel Nourissier and Bianca Pizzerra participated as EURORDIS' representative**
- **1st Cyprus National Scientific Conference on Rare Disorders, Cyprus, 18-19 March**  
**Mirando Mrcic: "EURORDIS Overview"**
- **XVI Telethon Italia Convention, Riva del Garda, Italy, 7-8 March**  
**Terkel Andersen represented EURORDIS**
- **Rare Disease Day: "All-Russia conference on rare diseases", Moscow, Russia, 1 March**  
**Yann Le Cam: "The European Organisation for Rare Diseases: Sharing Experience in Promoting Interest of Patients with Rare Diseases"**
- **International Rare Disease Day 2011, Brussels, Belgium, 28 February**  
**Yann Le Cam: "Rare but Equal: Addressing Health Inequalities for Rare Disease Patients in Europe"**
- **International Rare Disease Day 2011, Budapest, Hungary, 26 February**  
**Yann Le Cam: "Rare but Equal: Addressing Health Inequalities for Rare Disease Patients in Europe"**
- **EUROPLAN project, Final Conference, Rome, Italy, 25 February**  
**Yann Le Cam: "The National EUROPLAN Conferences: main results and key questions for discussion"**  
**With the participation of Valentina Bottarelli and EURORDIS Advisors to EUROPLAN:**  
**Simona Bellagambi, Britta Berglund, Avril Daly, Dorica Dan, Mirjam Mann and Christel Nourissier**
- **International Rare Disease Day 2011, Meeting in Switzerland, organised by ProRaris, Swiss Parliament, Berne, Switzerland, 19 February**  
**Christel Nourissier: "Challenges and perspectives in the fight against rare diseases: State of the play in Europe and in Switzerland"**
- **TRANSFoRm Stakeholders Meeting, Brussels, Belgium, 9 February**  
**Tsveta Schyns represented EURORDIS**
- **"Forum des Métiers de l'Événementiel" (Meeting and Event Career Forum), RECIPROS: Salon Bedouk, Paris, France, 3 February**  
**Sharon Ashton presented her experience as Event Manager at EURORDIS**
- **Kick-off meeting of E-IMD (European Registry and Network of Expertise for Intoxication Type Metabolic Diseases), Luxembourg, 1-2 February**  
**Lut de Baere: "Presentation of the European Registry and Network of Expertise for Intoxication Type Metabolic Diseases"**
- **Rare Diseases Platform Workshop on Research, Paris, France, 20 January**  
**Christel Nourissier and Bianca Pizzerra represented EURORDIS**





# ACKNOWLEDGEMENTS

EURORDIS would like to thank the following organisations and companies for their highly valued support in 2011

## Not-for-Profit Organisations and Public Entities

### AFM - Téléthon

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



### INSERM

INSERM for the secondment of the Clinical Research Advisor of **EURORDIS**



### EUROPEAN COMMISSION

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

- The Operating Grant "European Organisation for Rare Diseases" (**EURORDIS** FY2011)
- Patients' Consensus on Preferred Policy Scenarios for Rare Diseases (POLKA)
- The European Project for Rare Diseases National Plans Development (EuroPlan)
- The European Network on Rare Paediatric Neurological Diseases (nEUroped)
- The European Platform for Rare Diseases Registries (Epirare)



**European Commission (DG Research)** for its essential contribution to the neuromuscular network TREAT-NMD supported under the Sixth Framework Program



## Acknowledgements

### Medtronic Foundation



Co-funding of Rare!Together project

### AIPM , Association of International Pharmaceutical Manufacturers



Co-funding of Russian version of the E-Newsletter

### LEEM - Les entreprises du médicament



Co-funding of RareConnect (Online Patient Communities)

### DIA, Drug Information Association



Co-funding of Summer School 2011

### Everylife Foundation for Rare Diseases



Co-funding of the EURORDIS Gala Dinner

### Fundació Doctor Robert Universitat Autònoma de Barcelona Casa Convalescència



Volunteering in the context of the Polka project and the Summer School 2011

### Science Museum Cosmo Caixa



Venue of Summer School free of charge

## Pharmaceutical and Biotech Companies

**EURORDIS** has diversified its corporate sponsorship from 32 to 37 different companies.

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

Corporate companies have supported **EURORDIS** through the **EURORDIS** Round Table of Companies<sup>1</sup>, RareConnect™ The Online Patients Communities<sup>2</sup>,

Polka and Epirare projects, the **EURORDIS** Membership Meeting 2011 Amsterdam and the **EURORDIS** Gala Dinner<sup>3</sup>, which funds actions to reduce isolation of people living with rare diseases and their families, to increase awareness of rare diseases and to build capacities.

The breakdown of company's donations by project is detailed on our website on the "Corporate" tab of the "Financial Information" section<sup>4</sup>.

### The Five Top Donors

CSL Behring

Biotherapies for Life™ **CSL Behring**

GlaxoSmithKline



Novartis



Shire



Sigma-Tau Pharmaceuticals



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

2 - <http://www.rarediseasecommunities.org/en/article/project-funding>

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

4 - <http://www.eurordis.org/financial-information-and-funding>

## Acknowledgements

### The other pharmaceutical companies

The 32 other donors are listed in alphabetical order.



## Other contributors

Air France

For flight tickets free of charge



Mark Krueger & Associates, Inc

For recognition of support of the ERTC Workshop  
Welcome Dinners

Mark  
Krueger & Associates, Inc.



Emilia, Achondroplasia, Germany  
Winner of the EURORDIS 2011 Photo Contest. Photo by: Anna Spindelndreier



**EURORDIS**  
Rare Diseases Europe

## Workplan 2012

WORKPLAN 2012

## The Voice of Rare Disease Patients in Europe

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# Our Action Plan for 2012



## 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
- Improving quality and access to rare disease care
- Accessing cross-border healthcare and making possible patient mobility
- Promoting research and bridging patients and researchers
- Starting to address 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” and in 2009 “Council Recommendation on 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 these seven patient representatives and additional **EURORDIS**’ patient representatives in EUCERD’s key activities such as National Strategies / Plans & Indicators, Centres of Expertise & European Reference Networks, Rare Disease Registries & Databases & Standardisation, Access to Rare Disease Therapies & CAVOD, New Born Screening, Social Specialised Services and Integration of Rare Diseases into Social Policies and Services
- Support these patient representatives with the **EURORDIS** Policy Action Group (PAG), staff, volunteers, information sharing and reports
- Contribute to EUCERD with concrete policy proposals so to address patients needs when implementing the strategy foreseen in this policy framework
- Lead & support EUCERD collaboration in some areas eg: Rare Disease Day, ECRD & Transatlantic Forum, Access to Medicines, Social Policy, 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
- Use ECRD 2012 Brussels to disseminate EU policy outcomes, to monitor progress, to promote new strategies and innovative

solutions, to integrate all main stakeholders at EU and national levels, and share common objectives

- Communication through the **EURORDIS** eNews, **EURORDIS** Website

→ Promote the development of National Strategies & Action Plans on Rare Diseases in all Member States by 2013 through:

- Active participation in the EUCERD Joint Action / EuroPlan Work Package through the working group of 10 **EURORDIS** Volunteer Advisors and the support of the **EURORDIS** European Policy Staff : methodology recommendation for the development of plans, identification & promotion of best measures for national policies, development of policy indicators, disseminate information on national plans
- Organisation of 20 new national conferences in 2012 & 2013 in Belgium, Croatia, Cyprus, Denmark, France, Georgia, Greece, Hungary, Ireland, Italy, Netherlands, Portugal, Romania, Russia, Serbia, Spain, Sweden, Switzerland, Ukraine, United Kingdom, organised by National Alliances with **EURORDIS** in the framework of EuroPlan to promote national rare diseases plans. Dissemination of their synthesis with key recommendations, together with the outcomes of the 15 national conferences organised in 2010 & 2011
- Take additional initiatives to promote national plans by 2013 within EUCERD as well as direct actions between **EURORDIS** and National Alliances
- Active **EURORDIS** support in several Member States & EU Presidency (2012 Denmark & Cyprus, 2013 Ireland & Lithuania)
- Update and develop the section dedicated to national policies & plans in **EURORDIS** Website
- Update the existing **EURORDIS** Policy Fact Sheets and create new ones
- Communication through the eNews, the Website, the ECRD 2012 Brussels



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

- As a higher public health priority in the 3rd EU Public Health Programme
- As a higher research priority in the 8th EU Research & Technology Framework Programme called "Horizon 2020"
- Through position papers and advocacy actions to European Commission, European Parliament, Council and EU Presidency
- Through the Rare Disease Day Symposium on 29 February 2012 in Brussels
- Through the ECRD organised in 2012 in Brussels for this purpose

→ **Advocate on the implementation of the EU Directive on Cross-Border Healthcare (Patients' Mobility) adopted in 2011 providing a sustainable legal and financial instrument for European Reference Networks and patients' mobility across EU so to promote policy measures addressing the specific needs of rare disease patients and families. Empower patient advocates for the national transposition before end 2013 with a Q&A document and a Capacity Building Workshop**

→ **Advocate on access to Rare Disease treatments, in particular orphan drugs, in the context of the financial and economic crisis to prevent or limit impact of cut back measures**

→ **Advocate on key points of the European Commission's proposal to revise the Transparency Directive to speed up access to medicines**

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

- The international spreading of Rare Disease Day
- The application for NGO status at the Council of Europe
- The elaboration of a strategy toward WHO

- The coordination of a Joint Declaration "Rare Diseases: an International Public Health Challenge" to be adopted by major rare disease umbrella patient organisations

- The development of "Rare Diseases International" as an informal network of rare disease patient organisations

- The planning of the Transatlantic Forum on Rare Diseases and Orphan Products 2013 in Croatia to bring together all key players from Europe, USA and Canada

- The support of ICORD as a tool to expand the rare disease movement at the international level such as ICORD 2012 Tokyo and ICORD 2013 Shanghai

→ **Advocate in support of rare disease research. A high priority in the Strategy 2010-2015. Based on 2010 survey on the involvement of patient organisations in research, Rare Disease Day 2010, Symposium in Brussels in March 2010, elaboration of reference paper, advocacy actions, EURORDIS is taking activities in research policy to a new level in 2011 & 2012 with several actions: (see also section on Shaping Research Policy)**

- Promote a more favourable research policy framework for rare diseases: dissemination 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 Scientific Committee Therapies. Contribute to the IRDiRC Policy Paper. Take part in all meetings and activities. Promote IRDiRC.

- Promote a joint paper of EUCERD and COMP to DG Research on strategy & priorities for the EU Programme "Horizon 2020"

→ **Build capacities of our members to advocate on research policy Explore the feasibility for first petition from EU citizens on rare diseases**

## Information & Networking

### Community Building

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

→ **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 2012 Brussels on 23 May with six "Capacity Building" Workshops and six "Learning From Each Other" Forums**

→ **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 one 1.5 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

- Creating a **EURORDIS** "Learning From Each Other" Short Term

# Our Action Plan for 2012

► Fellowships 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 Czech Republic, Slovenia, Ukraine

## → 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 On Line Patient Communities
- Organising a two day workshop of the Council of European Federations
- Sharing experience and good practices through the Rare!Together wiki guide
- Mentoring of one or two new 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
- Creating a **EURORDIS** "Learning From Each Other" Short Term Fellowships 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, Russia and Caucasia and support their actions to raise public awareness, promote policy on rare diseases and create national alliances:

- Give priority in **EURORDIS** Patient Advocate Fellowships Programmes for the **EURORDIS** Membership Meeting & ECRD 2012 Brussels, as well as in fellowships for EPPOSI Workshops and DIA EuroMeeting 2012 Copenhagen
- Maintain dissemination of Play Decide Games available in 22 languages
- Official **EURORDIS** endorsement, promote and take part in national conferences across Central & Eastern Europe on request of our members
- Support Russia with the 2nd All Russia Conference on Rare Diseases & Orphan Drugs in Moscow organised by the Russian alliance of "Patients with Rare Diseases", the 3<sup>rd</sup> "LifeLine" Conference on Rare Diseases & Orphan Drugs in Saint Petersburg or Moscow organised by Genetics and the 1<sup>st</sup> Euro-Asia Conference on Rare Diseases & Orphan Drugs organised in Moscow
- Support the 3<sup>rd</sup> South Caucasia Conference on Rare Diseases & Orphan Drugs

- Expand **EURORDIS** Website, eNews and main documents available in Russian

- Planning of possible development of **EURORDIS** Website and eNews in Polish

- Organise country visits

## → Strengthen 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

## → Support EURORDIS' Volunteers involvement with short briefing material on key topics, access to shared resources of reference documents and public presentations

## → Expand "RareConnect", the EURORDIS-NORD Social Networks of Online Patients Communities and Advocates Communities in five languages (EN, FR, DE, ES, IT):

- Maintain & support the 15 existing Online Patient Communities in conjunction with the over 122 patient groups involved and volunteer moderators
- Launch 20 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
- Consolidate methodology for operational support to create a community, recruit train and support moderators, promote a community
- Promote the service at large and each online community
- Specifically pilot communities on very rare diseases, on rare cancers, on managing clinical features common to several rare diseases or on common topics across several diseases (linked to topics in **EURORDIS** work programme)
- Develop new features to make the service more vibrant
- Maintain the human translation in 5 languages service to enhance networking
- Planning of possible addition of two languages in 2013 (PT, RU)
- Elaborate and adopt a strategic plan for "RareConnect" jointly developed by **EURORDIS** and NORD
- Update, revise with moderators and adopt an upgraded version of the **EURORDIS** Guidelines on Good Practices on Social Media



## Informing & Raising Awareness

### → Organise Rare Disease Day 2012: on the theme of “Solidarity & Rare Diseases” focusing on “Rare but strong together”

- Info Pack, Poster & Slogan, dedicated Website, patient testimonies, social media
- Create a Rare Disease Day Video Clip in several languages (in kind support)
- Expand to 60+ countries in Europe and at the international level

### → Organise a one day Rare Disease Day Symposium in Brussels on Wednesday 29 February on “Rare but Strong Together” aimed at consolidating rare diseases as a priority in the 3rd EU Public Health and in the “Horizon 2020” EU Research Programmes, both for 2014-2020, and at promoting EU budget for rare diseases based on the high community added value

#### → Plan Future Rare Disease Days:

- RDD2013: Info Pack, Poster & Slogan, Website, patient testimonies, social media
- RDD2013: Create a Rare Disease Day Video Clip in several languages
- RDD2013: Plan Rare Disease Day Symposium in Brussels
- Take actions to declare a European Year for Rare Diseases (2016? 2018?)
- Take actions to promote Rare Disease Day as an official WHO Day

#### → Improve content quality and navigation of EURORDIS Website:

- Centred on target audiences: patients organisations & advocates, patient and families, stakeholders
- Improve navigation and friendly use of website with revised or new features
- 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), Social media – (Facebook, Twitter, Flickr, Rare Disease Blogs)

- Within core website consolidate issues of strategic importance e.g. EU policy, National plans, Centres of Expertise & European Reference Networks, Orphan Drugs, Access to Medicines & CAVOD & HTA, Specialised Social Services, Patients’ trainings, Living with a Rare Disease, Learning From Each Other, Membership, Volunteers
- Stimulate the International Rare Disease Blog supported by EURORDIS-NORD
- Maintain specifications for technical backend maintenance of EURORDIS.org and RareDiseaseDay.org

### → Publish four issues of the e-Newsletter until April 2012, and replace by eNews

### → Create EURORDIS eNews, a weekly communication to our members and subscribers (patient groups, patient advocates, healthcare professionals or researchers leaders, policy makers, regulators, companies, partners, media) to increase our outreach, maximise the efficacy and effectiveness of our communication, streamline our work, reduce our costs. Create the lay out and customised tool. Produce a minimum of 30 eNews in 7 languages

### → Organise Video & Photo Contest. Presentation at ECRD 2012 Brussels

### → Continue the photo project of 30 families affected by 30 different rare diseases in the 28 Member States of the European Union and two other European countries, illustrating 30 million people affected by rare diseases, for exhibition in European Institutions, European events, national events, and on the web. Complete with text and video testimonies

### → Identify key advancements benefiting People Living with Rare Diseases in the last 15 years

## 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
- Re-launch progressively the European Network of Rare Diseases Help Lines:
  - Governance & business meeting, develop membership base
  - Maintain shared tools eg. common software for the

collection of data on enquiries 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 (orphan or not and off label)
- Apply for the EU wide unique 116 number

## Our Action Plan for 2012

### → 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 on-line tutorials on searching validated information on the web
- Provide a search service on **EURORDIS** Website to provide user-friendly access
- Elaborate the **EURORDIS** quality criteria for rare disease web services
- Update and deepen the Review of 40 web services providing essential information on rare diseases and related research, treatments, drugs, services
- Create on-line video tutorials on how to search information on main websites

- Work with **EURORDIS** Contact Database to provide disease targeted access to **EURORDIS** members website
- Brand this new service, launch at ECRD 2012 and scale up a communication through 2012.

### → Improve access to patients' generated knowledge through Social Media:

- Promote the good use of social media to access patient generated knowledge
- Organise Social Media and Patient Empowerment Webinar Series with five modules: Developing a social media strategy and guidelines for your patient group, Structuring your social media communication, Allocating patient group's resources to effectively use social media, Using social media to build a disease community, Harnessing social media to spread quality information.

## Health Policy & Healthcare Services

### Promoting Rare Disease Health Policy Development

#### → Organise the 6th European Conference on Rare Diseases and Orphan Products in Belgium – ECRD 2012 Brussels on 23-25 May 2012:

- 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
- Finalise the Programme with the Programme Committee. Coordinate the multiple partnership with DIA, NORD, EUCERD, EMA, Orphanet, ESHG, EBE/EFPIA-EuropaBio. Promote ECRD & registrations in six languages. Organise selection of Posters submitted. Organise the **EURORDIS** Patients Fellowship Programme. Organise the logistic of one plenary, 36 Sessions & four Tutorials & four networking activities, 125 speakers & moderators, 700 participants, simultaneous translations up to six languages, communication around the event
- Organise the "Investors' Forum" on 23 May
- Organise satellite workshops for partners eg EUCERD, Orphanet, Epirare

#### → Plan the 1st Transatlantic Forum on Rare Diseases and Orphan Products in May 2013 in Dubrovnik, Croatia, organised by EURORDIS with NORD and CORD and co-organised with the DIA in partnership with EUCERD, EMA, FDA, NIH, Orphanet, EFPIA-EuropaBio, PhrMa for 250-300 participants as a working forum focused on selected priorities

#### → Empower the national alliances for their action in support of national rare disease plan:

- Involve them in all aspects of the EUCERD Joint Action / Work Package EuroPlan (methodology, recommendations, good practices, indicators, national conferences) through the working group of 10 **EURORDIS** Volunteer Advisors, all from different national alliances, and the support of the **EURORDIS** European Policy Staff
- Dedicate the next two **EURORDIS** Membership Meeting 2012 & 2013 to capacity building of national plans and make it a central theme in ECRD 2012 Brussels
- Organise the **EURORDIS** Membership Meeting 2012 Brussels on information & experience sharing across Europe as well as capacity building of rare disease patient advocates to promote national plans, to propose adequate measures and to play an active role in national plan steering committees
- Disseminate the **EURORDIS** Policy Fact Sheets on important policy and services expected by patients and families, derived from the Commission Communication, Council Recommendations and national plans at key events eg ECRD 2012 Brussels, Membership Meeting 2012, every national conference
- Maintain new ways to exchange information, experience, good measures, concerns between rare disease national alliances, the 10 patient advocates involved in promotion or implementation of national plans and European Public Affairs Staff – such as "national plan mailing list", regular conference calls and visits
- 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





# Putting Rare Disease Patients at the Heart of the Healthcare System

→ **Continue the dissemination of the deliverables of the “Patients’ Preferred Policy Scenarios” project (POLKA) in particular (a) the Play Decide Games so to stimulate civil society debates and citizens informed opinion on stem cell research, neo-natal screening, gene testing & counselling, centres of expertise for rare diseases, access to cross border healthcare and patients’, upper limit on orphan drug pricing, and (b) the Delphi-like method on the evaluation of centres of expertise by patients and clinical experts**

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

- Develop a long term strategic vision on European Reference Networks on Rare Diseases addressing patients’ 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
- Progressively withdraw from direct participation in disease specific European Reference Networks and rather collaborate with European/International Federations & Networks to take over or play an active role.

- Develop future possible long term 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, standard of diagnosis & care, standard of social & supportive care, information to patients, outreach to patients

- Organise a theme in ECRD 2012 Brussels on European Reference Networks

- Organise a workshop at the Membership Meeting 2012 Brussels

- Maintain a dedicated Website section on Centres of Expertise and European Reference Networks

→ **Promote policy on newborn screening, gene testing, pre-implantation diagnostic:**

- Promote awareness and citizens’ debates on gene testing and newborn screening through the promotion of four Play Decide Games, and disseminate the identified Patient Preferred Policy Scenario
- Contribute to the draft EUCERD Recommendation on Newborn Screening
- Promote the importance of post-screening follow up and support of families after neo-natal screening & diagnosis
- Disseminate the most relevant points for patient advocates of the Report on Newborn Screening led by ISS involving **EURORDIS**
- 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 Neo-Natal 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 (project 2012-2014), **EURORDIS** leads the Work Package on “Specialised social services and integration of rare diseases into Social Policies and Services” such as social guidelines, information & training of social service providers to address complex needs
- Promote EU and national policies for social research and quality of life studies
- **EURORDIS** Position Paper on the social challenges faced by people living with rare diseases and their relatives and why integrate rare diseases into existing social policies
- Collaborate with the EU project BURQUOL to develop indicators for quality of life
- Perform preliminary research and action plan for a possible **EURORDISCare 4** Survey on the Social Burden and Financial Burden of Rare Diseases for Patients and Families – concept, research

plan, organisation, funding – for a possible start in 2013 and implementation & dissemination over three years

→ **Stimulate the development of and improve access to Respite Care Services:**

- Share experiences through the dissemination of current guidelines and the elaboration of detailed common guidelines through a wiki tool
- Provide updated online information resources
- Promote the creation of new services through the Policy Fact Sheet and monitoring of measures adopted in national plans

→ **Stimulate the development of and improve access to Therapeutic Recreation Programmes:**

- Share experiences through the dissemination of current guidelines and their improvement through a wiki tool
- Provide updated online information resources

## Our Action Plan for 2012

- Promote the creation of new services through the Policy Fact Sheet and monitoring of measures adopted in national plans
- ➔ **Stimulate the development of and improve access to Adapted Housing & Related Services:**
  - Mapping of existing policy and services in EU Member States
  - Analyse good practices experiences and elaborate guidelines
- Promote the creation of Adapted Housing & Related Services through a new Policy Fact Sheet and monitoring of measures adopted in national plans
- Encourage **EURORDIS** Staff, Volunteers and Members to volunteer in Summer Camps and members of **EURORDIS** Round Table of Companies to financially support the Summer Camps or participate as Volunteers

## Research Drugs & Therapies

### Shaping and Supporting Research Policy

- ➔ **Promote Rare Disease Research as a Policy and Budget priority at International, EU and national levels within 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 a theme in ECRD 2012 Brussels on Research Policy
  - Organise workshop on research in the Membership Meeting 2012 Brussels
  - Participation in the International Consortium for Rare Disease Research (IRDiRC)
  - Participation in the ERA-Net project E-Rare involving Member States
  - Support and participate in the European conference "Re-ACT" dedicated to research advancements and results in rare diseases, organised in Switzerland
  - Participation in research policy activities and activities related to national plans
- ➔ **Transfer EuroBioBank, the Rare Disease Biological Resource Banks and European Networking, from EURORDIS to the Italian Telethon Foundation to take it to a new level. Participate in its governance. Promote policy recommendations and liaise with BBMRI Consortium.**
- ➔ **Promote the development of rare disease registries, their good governance across Europe, and optimal use geared at patients' interest, in particular through the project Epirare:**
  - Contribute to the mapping and identification of best practices for international and European registries
  - Contribute to the definition of a common data set for rare disease registries
  - Contribute to identification of best ICT options and their interoperability
  - Contribute to the analysis of the European legal framework and solutions
  - Start elaboration of preferred policy scenarios
- ➔ **Participate to the development of new long term Infrastructure projects on Registries and BioBanks focusing EURORDIS contribution on patient involvement**
- ➔ **Contribute to the Revision of the EU Directive on Data Protection**

### Supporting Clinical Research

- ➔ **Promote good practices for clinical research on rare diseases:**
  - Promote adoption of the **EURORDIS** Good Practices 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 Patient Partner EU Project
  - Collaborate with ECRIN Project
  - Contribute to the revision of the EU Directive on Clinical Research
- ➔ **Promote rare disease patient spontaneous reporting on adverse events of orphan and non-orphan drugs as well as off-label uses of medicines in rare indications. Explore methodologies to collect patients' reported outcomes**
- ➔ **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
- Organisation of a Workshop on Registries for Rare Diseases & Rare Cancer at the European Parliament in partnership with the European Cancer Patient Coalition and Rare Cancer Europe
- Participation in a Workshop on Clinical Trials in Rare Cancers organised by Rare Cancer Europe and contribution to the Consensus Statement on Clinical Trials in Rare Cancers
- Support to coordination between EUCERD and the European Partnership for Action Against Cancer to maximise synergies of this two EU policy areas

→ **Participate in the development of new long term projects on the methodologies to create and review best clinical practices, focusing EURORDIS contribution on patient involvement**

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

→ **Contribute to the Revision of the EU Directive on Clinical Trials**

## Promoting Drug Development & Access to Treatments

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

- Participate in EMA Committee for Orphan Drugs (COMP) with two representatives and one permanent observer
- Participate in EMA Paediatric Drugs 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 Patients & Consumers Working Party with one representative and one alternate and support (PCWP) Co-Chair
- Participate in EMA Working Group on Clinical Trials in 3rd Countries
- Link-in patient expertise for Protocol Assistance/Scientific Advice, Scientific Advisory Groups of CHMP, Risk Management, Risk Communication, Pharmacovigilance
- 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 plan (PIP) for rare diseases, paediatric research 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 and Health Technology Assessment:**

- Organise **EURORDIS** Summer School 2012 Session in Barcelona in June to train 30 to 50 new patient advocates in priority from national alliances, European federations, Central & Eastern

Europe and patient groups from targeted disease areas

- Provide the E-Learning platform in English with three sections on Methodologies, Statistics and Ethics and 20 modules with concepts, basic knowledge, glossary, case studies, self evaluation questionnaire
- Take part in the DIA EuroMeeting 2012 Copenhagen in March with speakers, a DIA Patient Fellowship Programme for about 40 fellows, a Patient Fellows Booth Stand
- Empowerment of our volunteers: Information and Monthly Reports, Call for Volunteers, Support the three Task Forces on Orphan Drugs, Paediatric Drugs and Drug Information Transparency & Access; Organise one or two workshops of the Task Force for Drug Information Transparency & Access
- Participate in the "European Patients' Academy on Therapeutic Innovation" (called EUPATI, from 2012 to 2017), an IMI funded project gathering a Consortium led by European patient Forum with other European umbrella patient organisations
- Train 10 ECRIN Fellows in **EURORDIS** Summer School

→ **Improve access to orphan drugs:**

- Promote the Common Scientific Assessment of the Clinical Added Value of Orphan Drugs (CAVOD) through collaboration between Member States, at EUCERD and European Commission, in conjunction with EMA, EUnetHTA and the pharmaceutical umbrellas as well as HTAi and INAHTA;
- Promote European Evidence Generation Plans on Orphan Drugs and relative effectiveness evaluation
- Contribute to the new approaches to Risk & Benefits Assessments, in order to take into consideration the patient values & patients' preferred treatment options
- Promote Conditional Pricing in Member States
- Contribute to a Mechanism of Coordinated Access (MOCA) to Orphan Drugs between Member States with possible innovative approaches on Pricing, within the EU Stakeholders Forum on Corporate Responsibility in Pharmaceuticals
- Promote Early Access Management Plans on Orphan Drugs
- Interact with the EUnetHTA & Participate as a member of its Stakeholders Forum

# Our Action Plan for 2012

- Facilitate citizens debates and identification of Patient Preferred Scenario on orphan drug pricing based on a Play Decide Game

## → Organise the 6th EURORDIS Survey on Access to Orphan Drugs in the European Union

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

- **EURORDIS** Round Table of Companies: consolidate membership and organise two workshops in Brussels in March and in Barcelona in September / October 2012

- Deepen the direct dialogue with international big pharmaceutical companies

- Strengthen the dialogue with EBE-EuropaBio, EFPIA and national pharmaceutical associations

## Cross-Cutting Priorities

### → Implement EURORDIS Strategy 2010-2015:

- Develop Operational Units Action Plan 2013-2015 (three years) for each Unit
- Improve planning anticipation of major **EURORDIS** activities such as ECRDs, Transatlantic Forums, Membership Meetings, Rare Disease Day, ERTC Workshops, RareConnect, **EURORDISCare**, new projects

### → Develop EURORDIS Strategic Partnerships:

- Consolidate Strategic Partnership with AFM-Téléthon (based on Collaborative Agreement 2010-2013)
- Expand common actions with NORD (USA) (based on Strategic Partnership Memorandum signed in 2009)
- Establish a Partnership with CORD (Canada)
- Consolidate Partnership with the members of Round Table of Companies
- Establish partnership with Russian Patient Union and Genetic
- Establish global partnership with DIA
- Develop Strategic Partnership with ORPHANET
- Develop Strategic Partnership with the European Human Genetic Society, European Academy of Paediatricians, International Society of Neo-Natal Screening, the European Society of Medical Oncology, the Standing Committee of Physicians in Europe or other essential learned societies

### → Increase EC support to EURORDIS activities through:

- Advocacy actions on Operating Grant in 3rd Public Health Programme
- Application 2012 for Operating Grant 2013
- Application 2012 for Conference Grant 2013
- Plan application for a Patient Driven Public Health Project in DG SanCo Work Programme 2013 as project leader or partner
- Plan application for a Patient Driven Health Research Project in DG Research FP 7 Call 2013 as project leader or partner
- Study feasibility of 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 among 38 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 for the 20 national conferences and for the Work Package on Social Challenges, the 1st Transatlantic Forum, the Website & eNews & Rare InfoSearch service in seven languages, the RareConnect & Social Media, **EURORDISCare** 4

- Organise the **EURORDIS** Black Pearl Gala Dinner for Rare Disease Day on 29th February 2012 in Brussels "Hope and Solidarity with Rare Disease Patients in Europe" and plan Gala Dinner 2013: 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, logistic organisation for 250 participants

- Complete the planning in 2012 to launch individual giving program in 2013

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

### → Implement the Staff Strategy & Organisation & Evolution 2011-2013, including:

- A new organisation in four Units: Governance & Advocacy, Operations, Communications & Development, Finance & Support Services
- Full review of job descriptions, reshuffling of tasks & priorities, job evolutions
- Recruitments of a Deputy to the CEO, a Director for Operations, a Social Policy & Specialised Social Services Manager, a Training Manager

### → Seek opportunities to secure seconded staff

### → Create EURORDIS Internship Opportunities, unpaid and paid, for up to five interns per year for periods of 1 to 6 months

### → Revise and adopt additional procedures in the field of Finance, Human Resources and Office Support Services

### → Secure VAT exemption for Transatlantic Forum 2013 with Croatia and start negotiation with Germany for ECRD 2014



→ Upgrade the Brussels Office with more office space & better IT

→ Maintain the decentralised structure from Paris (Main Office), Brussels (European Public Affairs), London (EMA), Barcelona (Web Communications & Rare Connect) and Moscow (Russian language & Liaison with Russia and the Eastern Partnership States) with integrated operations through work processes, IT standards & intranet, voice & data internet communication

→ IT support: equipment, services, intranet sharing, virtual office, open to volunteers

→ Priority: EURORDIS Contact Database Management fully operational

→ Continue 4th year of collection of EURORDIS indicators on activity and results

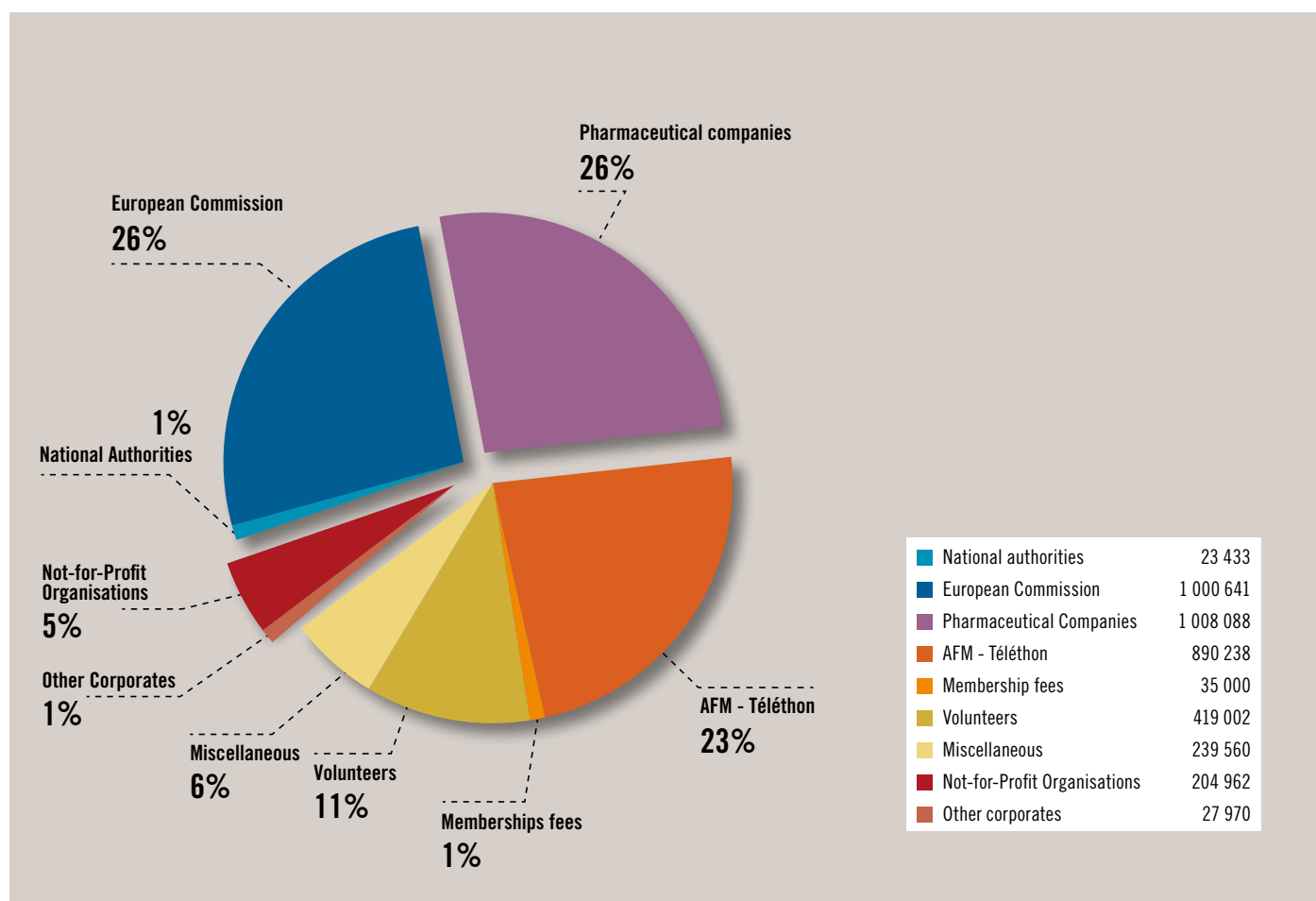


# BUDGET 2012

(in euros, provisions excluded)

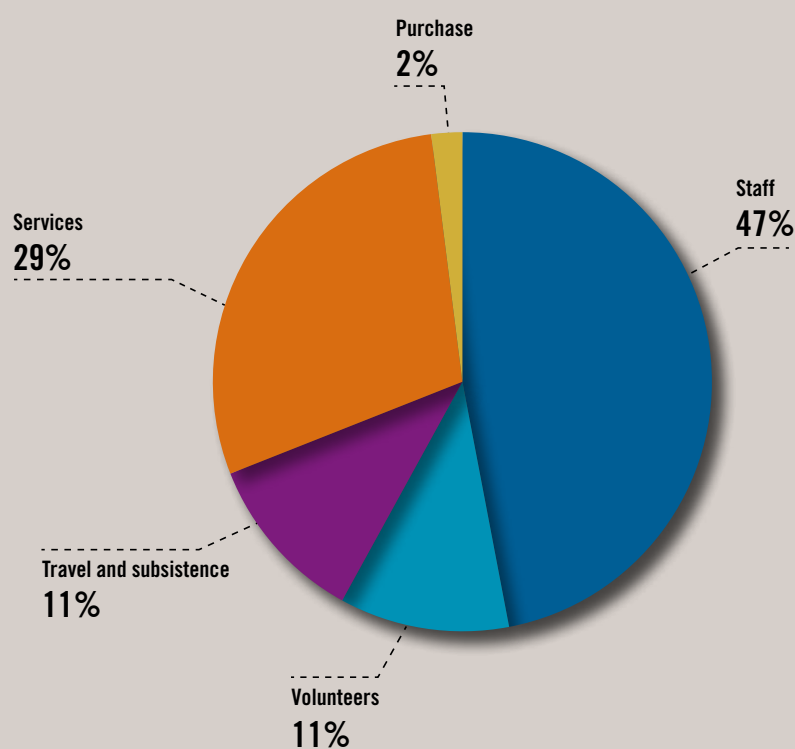


**Revenue by origin 2012 = 3,849 k€**

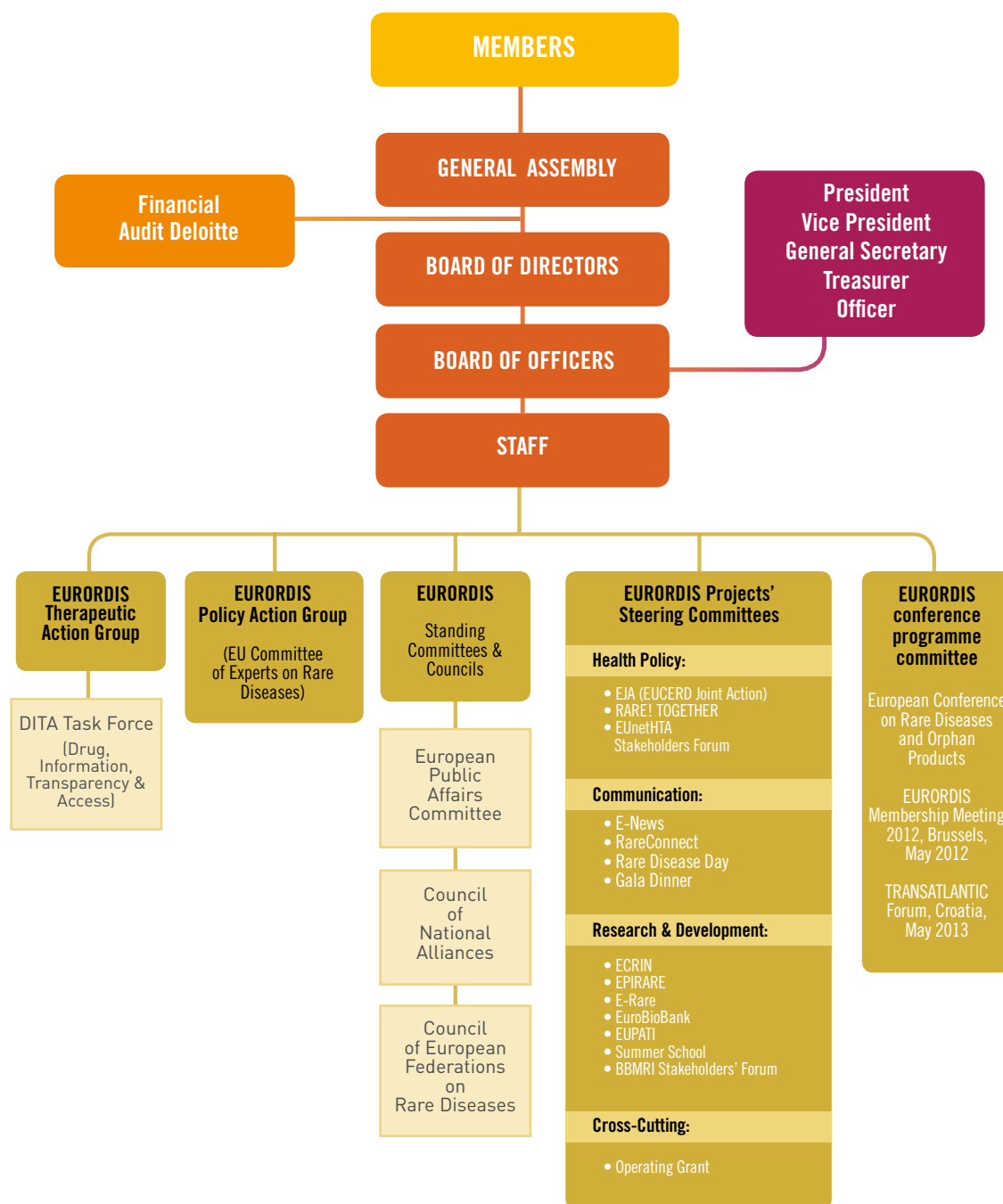


→  
**Expenses by Type 2012 = 3,847 k€**

■ Staff	1 804 861
■ Volunteers	419 002
■ Travel and subsistence	443 154
■ Services	1 105 748
■ Purchase	74 390





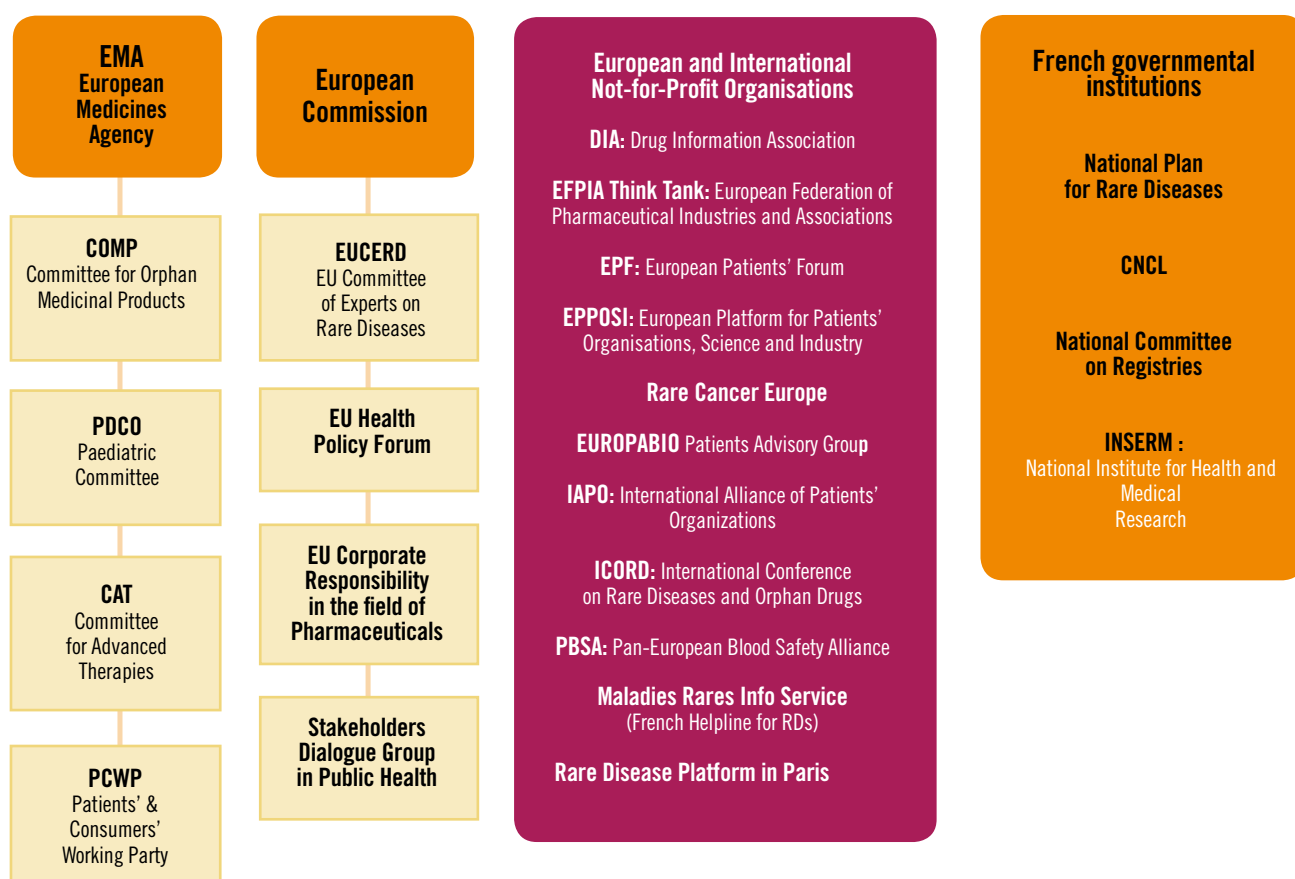
# GOVERNANCE CHART 2012



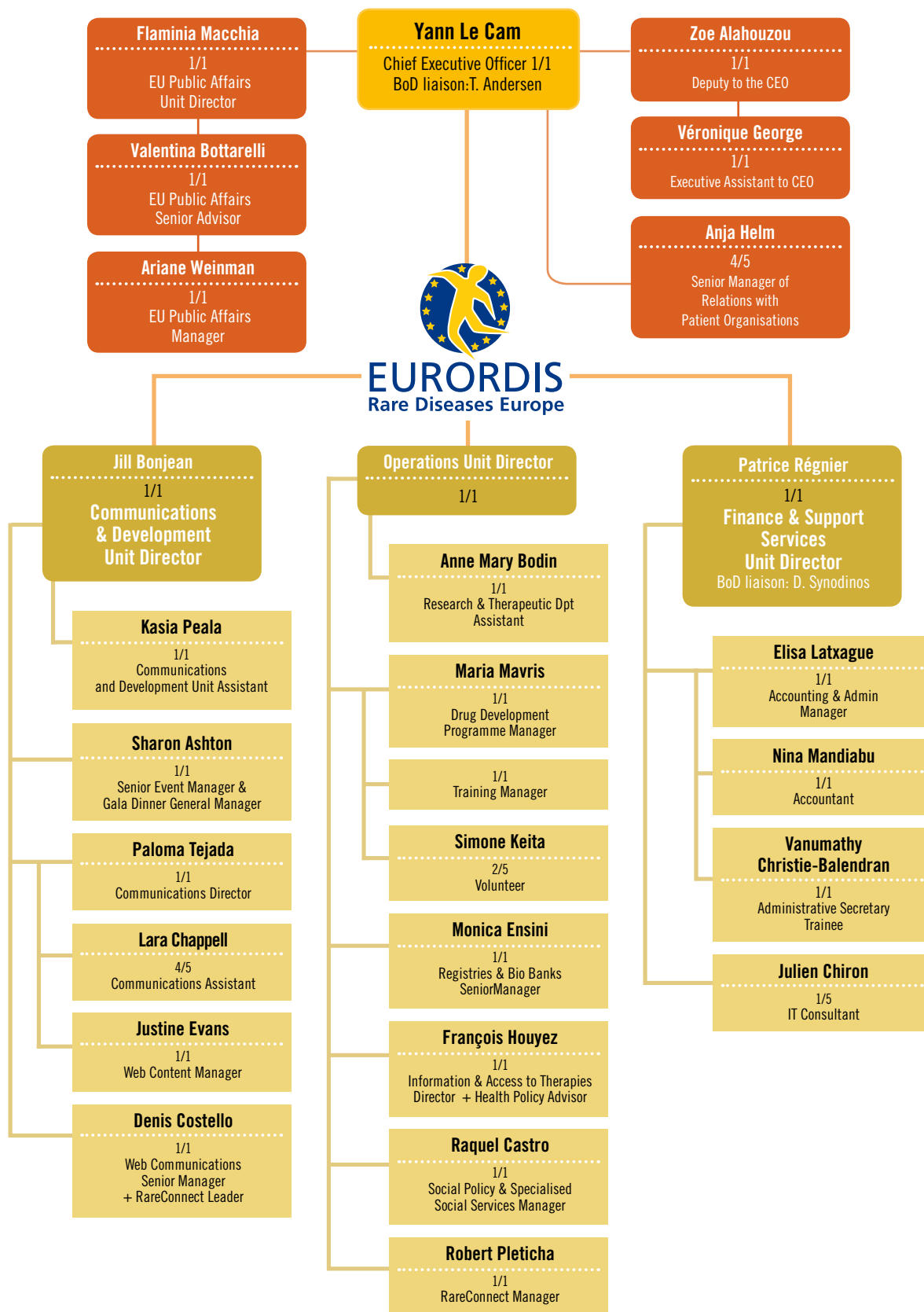


# EXTERNAL REPRESENTATION CHART 2012

→ **Governmental Institutions**   
→ **Non-Governmental Organisations** 



# TEAM CHART 2012





**EURORDIS**  
Rare Diseases Europe

Comité éditorial  
Eurordis

Conception et réalisation

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