



**EURODIS**  
Rare Diseases Europe

EUROPEAN ORGANISATION FOR RARE DISEASES

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2010

ACTIVITY REPORT

WORK PROGRAMME  
2011

[www.eurordis.org](http://www.eurordis.org)

# THE VOICE OF RARE DISEASE PATIENTS IN EUROPE



Photo credits:

**Cover:** Lou, Morsier Syndrome, Belgium. Image of Rare Disease Day 2010. Photo by Luc Boland.

**This page:** Ilona and Claudia, Spinal Muscular Atrophy, Type II, Germany

"Together we have more power and more fun" Winner of the EURORDIS Photo Contest 2010 Photo by Ilona Brand.

# 2010

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# FOREWORD

## BY THE PRESIDENT AND BY THE CHIEF EXECUTIVE OFFICER

In 2010, [EURORDIS](#) focused both on activities having an impact for people living with rare diseases in all European countries as much as on national activities influencing the European level, while promoting international activities having an impact on rare disease patients in Europe.

[EURORDIS'](#) front line advocacy on the EU Directive on Cross-Border Health Care resulted in its adoption. This Directive consolidates rare diseases in EU legislations. It provides a robust base for future European Reference Networks on Rare Diseases in order to organise expertise at European level and bring it to patients locally. New patients' mobility rights for access to diagnosis are granted.

[EURORDIS'](#) awareness and advocacy actions on research policy, undertaken in 2010, had significant impact. The rare disease research budget will reach its highest level ever in the Work Program 2012 of the 7th EU Research Framework. Research funding bodies of the European Union, USA and other countries are launching the new International Rare Disease Research Consortium, whose main objective is to deliver by 2020 diagnostic tests for all rare diseases and 200 new therapies for patients affected by rare diseases.

[EURORDIS](#) will amplify its advocacy activities in order to further promote rare diseases as a higher priority with increased budget for the period 2014-2020 in the 8th EU Research Framework and 3rd EU Public Health Programs. This will include new position papers, coordinated advocacy actions with national alliances, European federations and our members, outreach to the Council of the European Union and Members of the European Parliament. Rare Disease Day will grow internationally in 2011 and 2012. From 2012, [EURORDIS](#) will organise the European Conference on Rare Diseases and Orphan Products annually, with a consolidated partnership of key stakeholders.

The creation of the EU Committee of Experts on Rare Diseases (EUCERD) and the appointment by the European Commission of 8 patient representatives – all nominees of [EURORDIS](#) – was a milestone of 2010. A [EURORDIS'](#) representative was elected Vice Chair of this policy and scientific Committee. The EUCERD had its first meeting in December 2010. It is playing an essential role to implement the Commission Communication of 2008 and the Council Recommendation on Rare Diseases adopted in 2009. Its first outputs should be visible in 2011 and its role will grow in 2012-2014.

The European Conference on Rare Diseases (ECRD 2010 Krakow) organised by [EURORDIS](#) and its partners gathered 545 participants from 41 countries and attracted the highest ever participation from Central and Eastern Europe. Entitled "From Policies to Effective Services for Patients," this conference was part of a broader strategy to push towards the concrete implementation of the new EU rare diseases policy framework, both at European and national levels, across the European continent.

ECRD 2010 Krakow was connected to [EURORDIS'](#) intense involvement in the EUROPLAN project. Activities in 2010 included the production of over 80 concrete recommendations and practical methodological advice to guide the elaboration of national plans in the 27 Member States by 2013, as well as the adoption of indicators to monitor their implementation. In this context, [EURORDIS](#) and 15 of its national alliances have organised 15 national conferences based on a common agenda and documentation. The conferences gathered a total of 2 200 participants -all stakeholders- to discuss national strategies. Both ECRD 2010 Krakow and the 15 national conferences focused on the promotion of national plans, centers of expertise and European Reference Networks, patient registries and biobanks, translational research from the bench to the bedside, information and medical education, services to patients, families and carers.

In 2010, [EURORDIS](#) made a long-term commitment to the development of national plans 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 in our website, [EURORDIS](#) Policy Fact Sheets. In 2011, the [EURORDIS](#) Membership Meeting is dedicated to building capacities of patients advocate in national plans, and the new Joint Action on Rare Diseases, co-funded by the European Commission and Members States will provide concrete support actions for national plans.





EURORDIS' membership base is steadily growing with 447 members in 44 countries. Some new EU Members States are getting stronger on rare disease policy like Bulgaria, Croatia (accessing country), Hungary, Czech Republic and Poland. New national rare disease alliances have been created in Cyprus and in Switzerland. One is under development in Estonia. EURORDIS is strengthening its support to Central and Eastern Europe, the South Caucasus and particularly Russia with a combination of actions including patient fellowships, country visits, co-organisation and participation to local conferences, dissemination of information, exchange of experience, and advice on their strategy to promote rare diseases as a public health priority in their countries.

EURORDIS activities to empower patient advocates included the re-development of its website launched in 2010 to be further improved through 2011, the consolidation of the EURORDIS Summer School with the addition of regular webinars as well as a new EURORDIS e-Learning School.

Lack of equitable access to orphan medicines across Europe, aggravated by the impact of the financial and economic crisis, continues to be a high concern. EURORDIS dedicated its Round Table of Companies activities and a significant part of its advocacy work to this priority issue in 2010 and 2011, focusing on the promotion of the Clinical Added Value of Orphan Drugs (CAVOD). In 2011, new actions on compassionate use and early-access programs and planning of new EU mechanisms for coordinated access to orphan drugs will be initiated. EURORDIS' participation in the European Medicines Agency's Scientific Committees (COMP, PDCO, CAT) is very heavy with over 500 dossiers reviewed annually. A EURORDIS representative has been elected Chair of the Patients' & Consumers' Working Party. EURORDIS is preparing for a future 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 be active in the Health Technology Assessment of orphans and rare disease therapeutic interventions. EURORDIS is now a full member of the EUnetHTA Stakeholder Forum.


EURORDIS' members adopted its new Strategy 2010-2015, which is well advanced in its implementation. EURORDIS Strategic Partnership with NORD is increasingly important. In 2011 and 2012 EURORDIS will maintain its enthusiasm and innovative spirit launching new activities on patient registries, on specialised social services and integration of rare diseases into social policies, as well as re-launching the European Network of Rare Disease Help Lines and Information Centers. EURORDIS and NORD will scale up their web portal of social networks of patients and families launching regularly new rare disease communities and will plan for new web services for patients' online self-registration and patients' self-reported outcomes.

EURORDIS was able to grow in 2010 in spite of a challenging economic environment. Funding will be a critical issue to sustain our development. EURORDIS will continue to diversify its public funding, apply to private foundations and take new fundraising initiatives such as the EURORDIS Gala Dinner on February 29, 2012 in Brussels.



# EURORDIS IN BRIEF 2010

- 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.
- It was founded in 1997 by four patient groups from different therapeutic fields: the Association Française contre les Myopathies (AFM), Vaincre la Mucoviscidose, Ligue nationale contre le Cancer (LNCC), and AIDES Fédération.
- Today, it is supported by its members and by the Association Française contre les Myopathies (AFM), the European Commission, corporate foundations and the health industry.
- EURORDIS is the voice of 30 million patients affected by rare diseases throughout Europe.



## → Key figures 2010 :

- 447 member patient organisations
- 44 countries (25 EU countries)
- 23 national alliances
- More than 800 patient groups represented
- More than 4000 rare diseases represented
- 28 staff members (24,5 FTE)
- 62 volunteers
- 3 553 000 € in Revenue

## → Our mission :



### EURORDIS' MISSION IS:

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.

### TO THIS END, EURORDIS UNDERTAKES ACTIVITIES ON BEHALF OF ITS MEMBERS, NOTABLY IN FAVOUR OF:

- Empowering rare disease patient groups;
- Advocating rare diseases as a public health issue;
- Raising public rare disease awareness, and also that of national and international institutions;
- Improving access to information, treatment, care, and support for people living with rare diseases;
- Encouraging good practices in relation to these;
- Promoting scientific and clinical rare disease research;
- Developing rare disease treatments and orphan drugs;
- Improving quality of life through patient support, social, welfare, and educational services.

# STRATEGIC APPROACH 2010-2015

**EURORDIS IN 2015 HAS CONSOLIDATED ITS POSITION AS THE REFERENCE ORGANISATION FOR RARE DISEASES IN EUROPE AND IS RECOGNISED AS AN ACTOR IN WORLDWIDE PROCESSES HAVING AN IMPACT ON PATIENTS LIVING WITH RARE DISEASES IN EUROPE**

- Rare Diseases as a Public Health priority in Europe (8<sup>th</sup> EU Framework Programme for Research & Technology, 3<sup>rd</sup> 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 Eastern Europe and the Mediterranean region
- Raising rare disease awareness amongst the general public (including international Rare Disease Day)
- Rare diseases become an international movement and gain visibility and influence in international instances (WHO, UN, OECD, WB...)
- Production, sharing and accessibility of patient-generated knowledge

## → EURORDIS' Position in 2015

<b>EURORDIS</b> has consolidated its position as the reference organisation for rare diseases in Europe and is recognised as an actor in worldwide processes having an impact on patients and families living with a rare disease in Europe	<b>EURORDIS</b> is facilitating the effective implementation of European regulations and strategies at national level in more policy areas for the benefit of patients and families	<b>EURORDIS</b> has developed more supportive capacity building relationships with its members and fostered the empowerment of volunteers	<b>EURORDIS</b> is more sustainable in terms of human, financial, organisational resources and governance	<b>EURORDIS</b> has consolidated its activities in drug development, centres of expertise, empowerment of patient advocates and public awareness	<b>EURORDIS</b> 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	<b>EURORDIS</b> 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
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## **EURORDIS IN 2015 IS FACILITATING THE EFFECTIVE IMPLEMENTATION OF EUROPEAN REGULATIONS AND STRATEGIES AT NATIONAL LEVEL IN MORE POLICY AREAS FOR THE BENEFIT OF PATIENTS AND FAMILIES**

- A Public Health priority in European countries (Member States and beyond)
- National Plans in each Member State with patient-centred approaches including centres of expertise, research, medicines, registries, information, quality of life recognised as a major indicator
- Development and consolidation of European Networks integrated at 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 member patient organisations on the effective implementation of rare disease regulations and policies (evaluation process) and remaining unmet needs (research budget, Centres of Expertise, Standard of Screening, Diagnostics and Care, Quality of Life)

## A SPECIAL FOCUS ON RESEARCH THE ROLE OF EURORDIS SHALL AIM AT:

2010

- A greater public awareness in support of rare disease research
- A 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 enhance capacity building of patient representatives
- Promotion of the participation of patient representatives in ethics committees in clinical research and human genetics

## A SPECIAL FOCUS ON INFORMATION AND QUALITY OF LIFE THE ROLE OF EURORDIS SHALL AIM AT:

2010

### BEING A DIRECT OPERATOR IN THE FOLLOWING FIELDS:

- Rare disease specific help lines in national languages linked in a European Network
- Patient-based knowledge on care, coping and quality of life, generated and shared with 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 research and public health framework programmes

## EURORDIS IN 2015 HAS DEVELOPED MORE SUPPORTIVE CAPACITY BUILDING RELATIONSHIPS WITH ITS MEMBERS AND FOSTERED THE 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 of the organisation
- Intensify capacity-building and networking with and amongst National Rare Disease Alliances and European Rare Disease Federations for improved efficacy and decentralisation
- Member patient organisations as key links to families and patients to generate and access relevant customised information
- Capacity building, networking, sharing experience and common tools, giving an easy access to good practices to empower patient advocates



## EURORDIS IN 2015 IS MORE SUSTAINABLE IN TERMS OF HUMAN, FINANCIAL AND ORGANISATIONAL RESOURCES AND GOVERNANCE

- Well established shared values and governance processes
- Leadership sustainability of the Board of Directors
- Decreased and better-balanced workload inside the staff, more high level volunteers, efficient balance of workload between staff and volunteers
- Web communications central in strategy, organisation and 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



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## COMMUNITY STRUCTURE OVERVIEW

- National Rare Disease Alliances
- European Rare Disease Federations or Networks
- Patient Organisations' Members other than above
- Patient Organisations and patient outreach in all Europe (48 countries)
- Committees, Task Forces, Groups

## STRATEGIC PARTNERSHIPS

- AFM : Association Française Contre les Myopathies
- NORD : US National Organization for Rare Disorders
- ECPC: European Cancer Patient Coalition (on hold)
- ORPHANET: the reference website for validated information on rare diseases and orphan drugs
- INDUSTRY through EURORDIS Round Table of Companies and other initiatives
- ACADEMIA for education and capacity building and for social policy research

# HIGHLIGHTS

## 2010

### ADVOCACY

- Promoting better quality and access to healthcare services: Continued and successful discussions to promote the adoption of the EU Directive on Cross-Border Healthcare
- Improving access to orphan drugs: Actions to support the implementation of [EURORDIS'](#) proposal for European collaboration on common scientific assessment of the Clinical Added Value of Orphan Drugs (CAVOD)
- Promoting national plans and strategies across Europe including 15 National Conferences on Rare

Diseases organised in 15 countries by national rare disease alliances in partnership with national competent authorities and stakeholders, in the framework of the EC-funded project EUROPLAN, which gathered over 2 200 participants

- Contributing to the long-term governance of rare disease strategy and policies: 8 patient representatives have been appointed to the EU Committee of Experts on Rare Diseases (EUCERD) which held its 1<sup>st</sup> meeting in December 2010



### INFORMATION & NETWORKING

- Organising Rare Disease Day 2010 in collaboration with partners in 46 countries worldwide – compared to 30 countries in 2009
- 447 members in 44 countries including 44 new members and 2 new countries
- Consolidation of the Network of 23 National Rare Diseases Alliances with two workshops of the Council of National Alliances and two new Alliances in Cyprus and Switzerland
- Consolidation of the European Network of 25 European Rare Diseases Federations and Networks with the 3<sup>rd</sup> Workshop of the Council of European Federations and a new pilot program of grants to support their activities

- Expanding the 30 patient mailing lists and launching the new “Rare Disease Communities” portal in partnership with NORD – first two pilot online patient communities: CAPS and VHL

- [EURORDIS](#) website consolidated in 6 languages and with new sections recorded more than 17,000 visits a month

- [EURORDIS](#) e-Newsletter, featuring new design and layout, serviced 7 800 subscribers monthly

- Photo and Video Contest attracted 272 photo entries and 36 video entries

## HEALTH POLICY & HEALTH CARE SERVICES

- The 5<sup>th</sup> European Conference on Rare Diseases (ECRD 2010 Krakow) in May “From Policies to Effective Services for Patients” gathered 545 participants from 41 countries with 71 speakers, 24 sessions, 5 languages and a poster session
- Creating six new Play Decide sessions, translated in 21 languages, 114 sessions played across Europe
- Analysing the involvement of patient organisations in the activities of Centres of Expertise in France, Denmark and UK using an online survey
- Disseminating updated best practice guidelines modified to reflect the range of different types of respite care services

## RESEARCH, DRUGS & THERAPIES

- Raising awareness: “Bridging patients and researchers to build the future agenda for rare disease research in Europe” through Rare Disease Day 2011 and a series of events and publications
- Conducting a EURORDIS survey on “European Rare Disease Patient Groups in Research: Current Role and Priorities for the Future” and disseminating results
- Organising two successful Workshops of the EURORDIS Round Table of Companies, with over 60 participants each
- Conducting the 5<sup>th</sup> EURORDIS Survey on Patients’ Access to Orphan Drugs in Europe, in collaboration with 10 national rare disease alliances
- 3<sup>rd</sup> EURORDIS Summer School held in Barcelona in September 2010 with 34 fellows
- Finalising and adopting a Reference Paper arguing in favour of investing in rare disease research
- Over 440 000 rare disease samples available across the EuroBioBank Network

## GOVERNANCE, ORGANISATION & FUNDING

- Formal unanimous adoption of the EURORDIS Strategy 2010-2015 by members
- New and enlarged Paris Offices in the Rare Disease Platform
- Significant evolution of staff: 4 new permanent positions and 3 replacements
- Consolidation of volunteers’ involvement with 62 volunteer positions
- Renewal of EURORDIS-AFM Téléthon Collaborative Agreement to guarantee independence in all governance and advocacy actions, as well as some core funding
- Renewal of European Commission Operating Grant

# ADVOCACY

## 2010

REPRESENTING PATIENTS WITHIN EUROPEAN  
GOVERNMENT INSTITUTIONS AND ADVOCATING  
FOR POLICIES WHICH ADDRESS THE NEEDS OF  
PATIENTS AND THEIR FAMILIES



## SUSTAINING RARE DISEASES AS AN EU PUBLIC HEALTH PRIORITY EU COMMITTEE OF EXPERTS ON RARE DISEASES (EUCERD)

The European Commission appointed eight patient representatives to the European Union Committee of Experts on Rare Diseases (EUCERD) on 27 July 2010. All candidates were proposed by EURORDIS in order to form a consistent group representing people living with rare diseases. They are patients or parents of patients affected by different diseases, in different countries and recognised for their long experience in patient advocacy. Yann Le Cam was elected Vice Chairperson of the EUCERD.

The work consisted in participation to the meetings of the newly created EUCERD; advocacy activities and advice to EU officials for the appropriate functioning of the Committee; creation of Working Groups on priority topics, such as Centres of Expertise, for the rare disease community.

All eight patient representatives attended the first meeting of the EUCERD on 9-10 December. Two meetings are planned in 2011 as well as several workshops in priority areas.



Yann Le Cam



Christel Nourissier



Torben Grønnebak



Dorica-Dan



Jan Geissler



Alastair Kent



Bianca  
Pizzera-Plantanida



Gábor Pogány



## EUROPEAN JOINT ACTION ON RARE DISEASES

EURORDIS representatives participated in the preparation of the Joint Action on Rare Diseases which will support the work of the EUCERD for the period 2012-2014. EURORDIS played a leading role in the Work Packages on Specialised Social Services and Integration of Rare Diseases into

Social Policies and Services and on National Plans or Strategies. These activities will be intensified in 2011.

## RARE DISEASE DAY EUROPEAN EVENT

Preparatory work to organise a European event, focusing on rare diseases and health inequalities for Rare Disease day. EURORDIS met with policy officers responsible for the Communication from the Commission "Solidarity in health: Reducing health inequalities in the EU" at the DG Employment and Social Affairs Unit on 'Active Ageing, Pensions, Healthcare, Social Services'. Further discussions with officials in charge of the

Cross-Border Health Care file at the DG Health and Consumers and with officials responsible for the European Disability Strategy 2010-2020 at the DG Justice, Fundamental Rights and Citizenship. EURORDIS liaised with MEP Antonia Parvanova's office in order to organise and secure her participation at the Rare Disease Day 2011 Symposium in Brussels.

## MAKING RARE DISEASES A PUBLIC HEALTH PRIORITY IN ALL MEMBER STATES

### ADVOCACY SUPPORT TO NATIONAL PLANS FOR RARE DISEASES

EURORDIS exchanged information and networked about national plans for rare diseases at the Council of National Alliances (May and December 2010). In May 2010, in Krakow, we organised the EUROPLAN Workshop with the EUROPLAN project leaders of the Italian Istituto Superiore di Sanità (ISS), to present the EUROPLAN Recommendations to National Health Authorities responsible for developing and implementing national plans or strategies on rare diseases.

We helped organise 15 EUROPLAN-EURORDIS National Conferences in partnership with the national alliance in each country and in collaboration with the national competent

authorities and main stakeholders, gathering a total of 2 200 participants.

EURORDIS participated directly in the development of the second French Plan on Rare Diseases and its negotiation with French authorities and stakeholders.

We gave particular support to National Plans on Rare Diseases in Ireland, Romania, Spain and Croatia.

A new section on national policies was developed on the EURORDIS website (<http://www.eurordis.org/content/europlan-guidance-national-plans-and-conferences>).

## POLICY FACT SHEETS FOR RARE DISEASE PATIENT ADVOCATES

We continued the production of a 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 for Rare Diseases. In 2010, the following ones have been developed:

- Respite Care Services (May 2010)
- Rare Disease Research (June 2010)
- Therapeutic Recreation Programmes (November 2010)
- Orphanet (November 2010)
- Registries (December 2010)



## 15 CONFERENCES TO PROMOTE NATIONAL PLANS ON RARE DISEASES IN THE FRAMEWORK OF THE EUROPLAN PROJECT

2010

In 2010, 15 National Conferences on Rare Diseases have been organised by EURORDIS and its National Alliances in their respective countries. The conferences were set in the context of the EC-funded EUROPLAN project 2008-2011 and led by Istituto Superiore di Sanita in Italy. They gathered the main stakeholders in the field of rare diseases, amounting to more than 2200 participants in the 15 European countries involved.

EURORDIS, as one of EUROPLAN's main partners and leader of Work Package 8, coordinated and supervised the organisation of the conferences with the support of six advisors from their respective National Alliances: Simona Bellagambi, UNIAMO, Italy; Britta Berglund, Rare Diseases Sweden, Sweden; Avril Daly, GRDO, Ireland; Dorica Dan, RONARD, Romania; Mirjam Mann, ACHSE, Germany; Christel Nourissier, Alliance Maladies Rares, France.

The National Conferences, one of EUROPLAN's main deliverables, helped achieve the project's main objective: supporting national authorities to develop National Plans or Strategies on Rare Diseases. In particular, EUROPLAN National Conferences were designed and organised to promote and assess the transferability at national level of the European policy guideline documents.

The Conferences were organised from May to December 2010 in Bulgaria, Denmark, Romania, France, Spain, Sweden, Italian, the Netherlands, Greece, Germany, Croatia, Hungary, Poland and UK. The Irish Conference was postponed to January 2011.

EURORDIS provided the organisers with the final texts of the EUROPLAN Recommendations and the EUROPLAN Indicators to assess the implementation of National Plans which EURORDIS contributed actively to develop and finalise in 2010. In particular, the EUROPLAN Recommendations were presented and analysed by national health authorities on 13 May 2010, during a satellite workshop organised at the European Conference on Rare Diseases in Krakow.

In each country, a multi-stakeholder Steering Committee prepared the Conference. Each National Conference was configured in plenary session and constituted of five workshops on the following themes: Governance of a National Plan; definition, codification and inventorying of rare diseases; research; standards of care, Centres of Expertise and European Reference Networks; patient empowerment.

15 Final Reports in English were produced by Conference organisers, based on a common template which mirrored the common outline of the Conferences. The Final Synthesis of the 15 National Reports was started in 2010 and finalised in April 2011.

Based on the EUROPLAN approach, other events have been organised in Switzerland, Austria and Cyprus.

## RARE DISEASES: AN INTERNATIONAL PUBLIC HEALTH PRIORITY

EURORDIS is progressively expanding its efforts to make rare diseases an international public health priority.

The ongoing Strategic Partnership between EURORDIS and the US National Organization for Rare Disorders (NORD) plays a key role in that respect with increasing coordination between the

two organisations on advocacy actions in 2010. These include: the promotion of rare diseases at the international level; a stronger joint support to promote Rare Disease Day internationally; the launch of international rare disease online communities; and the planning of a Conference on Rare Diseases and Orphan Drugs in Washington DC in October 2011.

## IMPROVING ACCESS TO ORPHAN DRUGS

### CLINICAL ADDED VALUE OF ORPHAN DRUGS

Continued advocacy activities to support EURORDIS proposal for the establishment of a Working Party for European collaboration towards a common scientific assessment of the Clinical Added Value of Orphan Drugs (CAVOD). This work will be done at the time of marketing authorisation, based on new data generated by an agreed post-licensing European research plan on risks, benefits and effectiveness. It will help know the value of the product in real-life use, identify its right place on the therapeutic strategy, promote best possible medical practice and make the best use of limited healthcare resources. Our work involves liaising with pharmaceutical companies,

the European Commission, the European Medicines Agency, national authorities, national Health Technology Assessment agencies.

A Call for Proposal was published by the European Commission which selected the consulting firm Ernst & Young to carry on a Feasibility Study on the Mechanism to Implement the CAVOD. EURORDIS followed the process through and participated in meetings organised by Ernst & Young aimed at identifying the most appropriate tool to achieve better cooperation at EU level for facilitating patients' access to orphan drugs.

### SURVEY ON PATIENTS' ACCESS TO ORPHAN DRUGS IN EUROPE

In September 2010, EURORDIS and 10 rare disease National Alliances joined forces to obtain information about the real-life situation of European patients with respect to access to 60 orphan drugs. The idea behind this collaborative work was to find out whether orphan drugs that had received EU market authorisation since the implementation of the EU Regulation on Orphan Medicinal Products in 2000 were actually

available for rare disease patients within their own countries. The results of this survey were presented at the 13th EURORDIS Round Table of Companies workshop held in Paris on 13 December 2010. They have also been widely used by the European Commission, at national conferences on rare diseases and have prompted additional studies by research academic teams.

### CHARTER FOR CLINICAL TRIALS IN RARE DISEASES

EURORDIS promotes, amongst patient groups and whenever appropriate in different arenas, the Charter for Clinical Trials in Rare Diseases

to regulate the relationship between a clinical trial promoter and the patient organisations representing the disease concerned by the study.

## IMPROVING ACCESS TO QUALITY CARE

### PROMOTING CROSS-BORDER HEALTHCARE AND MOBILITY OF PATIENTS

The long-lasting advocacy activity on the Cross-Border Healthcare Directive, intensified throughout 2010, has been successfully achieved in 2011 with the final adoption, by the Parliament first and the Council later, of the Directive on Patients' Rights in Cross-Border Healthcare. This, de facto, creates a new right for European patients to choose where to get the best possible healthcare service they need. The work performed in 2010 involved continuous exchange with Members of the European Parliament (the rapporteur and other key MEPs), approaching national ministries, exchanges and meetings with the European Commission's officials in charge of the file.

For months, the main focus of the debate on the Cross-Border Healthcare Directive has been placed on rare disease patients and the specificities of rare diseases: it has been widely recognised that the Directive would benefit primarily patients

living with rare diseases for which the highest level of expertise and healthcare cannot be found in each EU Member States for every disease. Therefore, the added value of patient's mobility for rare diseases patients is particularly high. Another important element of this Directive is the provision concerning the European Reference Networks of Centres of Expertise (ERN): Article 12 provides a legal basis for the ERN and an invitation from the Commission to Member States to support their development, in particular in the area of rare diseases. The Directive also creates a legal basis for cooperation between Member States on Health Technology Assessment.

The advocacy activities in this area will continue in 2011 as Member States have 30 months to modify their national legislation accordingly. EURORDIS will remain vigilant to help ensure the best possible transposition into national laws and policies.

## OTHER ISSUES

### INFORMATION TO PATIENTS

EURORDIS has clarified its position on Information to Patients through a document elaborated in March 2010.

EURORDIS has proposed two amendments to the Fjellner Report in April 2010, both through the European Patients Forum and directly to selected Members of the European Parliament.

### 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 and the European Society for Medical Oncology is being strengthened.

### ORGAN DONATION AND TRANSPLANTATION

EURORDIS has updated the Expert Group on Organ Donation and Transplantation and followed closely the legislative progress until final adoption

of the EU Directive on Organ Donation and Transplantation in May 2010.



# INFORMATION & NETWORKING

**BRINGING THE RARE DISEASE COMMUNITY TOGETHER  
ENABLING PATIENTS TO SHARE AND LEARN FROM  
EACH OTHER  
INFORMING, EDUCATING AND RAISING AWARENESS  
ABOUT RARE DISEASES**

# 2010



## MEMBERSHIP BASE - 447 MEMBERS

44 new members joined in 2010 (28 full members and 16 associate members). New countries represented: Algeria and Georgia.

At the end of 2010, EURORDIS has 447 members in 44 countries, out of which 34 European countries and 25 from the European Union.

## COUNCIL OF NATIONAL ALLIANCES

Consolidation of the Network of 23 National Rare Disease Alliances.

Two Workshops of the Council of National Alliances focusing on the organisation of Rare Disease Day; the exchange of experience on national plans (including the Recommendations and Indicators for National Plans for Rare Diseases

and organisation of 15 national conferences) in the framework of the EUROPLAN project; and the survey on availability of orphan drugs.

Two new National Alliances joined the CNA in 2010: Cyprus, created in September 2010 by 5 national groups, and Switzerland, launched in June 2010 by 42 national patient organisations.



## COUNCIL OF EUROPEAN FEDERATIONS

The 3rd European Workshop of the Council of European Rare Disease Federations (CEF) took place in July 2010 for two days and gathered 25 representatives of European federations or networks.

The first day focused on: involvement with the EMA; Rare Disease Day; collaboration with European Reference Networks and the Clinical Trials Charter. The second day was dedicated to a capacity building session entitled: "How to apply for EU funds in the field of rare diseases".



In 2010, EURORDIS launched a pilot grant project to assist European Rare Disease Federations, finance their network meetings (Board meetings, network meetings, conferences, etc.) Through

this pilot programme, EURORDIS granted a total of 15.000 € to six European federations for six network meetings.

## RARE! TOGETHER

The wiki-like guide <http://raretogether.eurordis.org> has been redesigned and updated. This website was created as part of the Rare!Together mentoring project, which aims to help in the creation, operation and management of European

rare disease federations. This knowledge base will become a good practice reference handbook and a toolkit designed for patient organisations planning to set up their European federation, as well as to existing European federations.

## RARE DISEASE ONLINE COMMUNITIES

EURORDIS, in partnership with NORD, has created the Rare Disease Online Communities project to offer an online social network for patients and families to connect, support one another and share vital experience on aspects of living with a rare disease. The Online Patient Community Portal [www.rarediseasecommunities.org](http://www.rarediseasecommunities.org) is a multilingual platform in 5 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. Two online patient communities were launched in 2010 for the autoinflammatory diseases Cryopyrin Associated Periodic Syndromes (CAPS) and the rare cancer Von Hippel Lindau disease (VHL).

During 2010 the website received over 9,000 unique visits from 95 countries which led to 150 patients or caregivers signing-up to participate

in the service. In addition these communities saw international partnerships emerge from 15 individual patient groups who formally partook in the setup and governance of each of these communities.

Three training workshops took place in order to further involve patient organisations in the governance and management of future online communities. EURORDIS was also present at medical conferences, such as the World Auto-Inflammation Congress, in order to raise awareness of its efforts in this domain with medical professionals.

EURORDIS presented the tool at prestigious global conferences, such as Health 2.0 (Paris, April 2010), where it was awarded the audience prize of "Best Start-Up Idea of the Conference" and which merited an invitation to the Health 2.0 Conference in San Francisco in October 2010.

## MAILING LISTS

In addition, EURORDIS continued the maintenance of 30 Mailing lists (listservs) for patient advocacy and patient support through a partnership with Médicalistes. In 2010, there are 7 advocacy and governance mailing lists and 23 disease specific

mailing lists. The most active mailing lists are for Behçet's, Tuberous Sclerosis Complex and EUROPLAN.



## EURORDIS WEBSITE

The website [www.eurordis.org](http://www.eurordis.org) recorded approximately 17 500 visitors a month and 14 900 websites linked to it. There was a greater turnover of information and new sections tailored

to meet the needs of the evolving situation of rare disease policy in Europe. In particular, a section on the activities carried out to promote National Plans was added.

>>>[WWW.EURORDIS.ORG](http://WWW.EURORDIS.ORG)



## RARE DISEASE BLOG

More contributions from opinion leaders and patient advocates to the EURORDIS/NORD sponsored blogging platform [www.rarediseaseblogs.net](http://www.rarediseaseblogs.net), featuring commentary on rare disease policy

and orphan drugs. Content from the blog was incorporated into the EURORDIS website by developing a real time linkup.

## ELECTRONIC NEWSLETTER

The e-Newsletter design and layout changed in January 2010 in accordance with the new graphic design and layout of the EURORDIS website. The revamping of the e-Newsletter also includes new features: RSS feed, PDF download, multimedia (photos, embedded videos and links to the EURORDIS YouTube Channel). The sign-up

process to the e-Newsletter was optimised. In 2010, 10 issues of the e-Newsletter were produced in English, French, Spanish, German, Italian and Portuguese. The subscription rate went up 10 % in 2010 compared to 2009, with a total of 7862 subscribers.

## PHOTO & VIDEO CONTEST

The contest attracted 272 photo entries and 36 video entries.

Photo submissions were received from 34 different countries, and showcased over 80 different rare diseases.

Video submissions were received from more than 25 different countries, and showcased more than 21 different rare diseases.

The entries are visible on Rare Disease Day's Flickr photo wall and YouTube Channel. The winning photo features two women living with SMA (Spinal Muscular Atrophy, Type IIa) in Germany. The winning video, entitled "Garret's story", features the daily life of an Epidermolysis Bullosa patient in the USA.

## EUROPEAN NETWORK OF RARE DISEASE HELP LINES

Organisation of a meeting in Krakow to review the efforts made by help lines to adhere to the criteria to become a member of the European Network of Rare Diseases Help Lines. A governance scheme was agreed upon and an action plan was developed with 15 helplines from 9 countries.

Production of fact sheets for patient advocates on National Help Lines and the European Network of Help Lines to promote the creation of more national helplines and foster public support for

existing ones, in the context of National Plans on Rare Diseases.

The activities of the European Network of Rare Diseases Help Lines have been suspended since August 2010 due to lack of operating funds. Activities will continue to be suspended in 2011 and the job position is on hold, until EURORDIS can secure adequate funding from public or private sources from 2012 onwards.



*Rare Disease Day*



## RARE DISEASE DAY

2010

Rare Disease Day 2010 took place on 28 February 2010. The campaign has been implemented in 46 countries, of which 27 European countries (newcomers Latvia, Lithuania and Slovenia.) EURORDIS and 22 Rare Disease National Alliances, together with 24 patient groups acting as country organisers, mobilised hundreds of patient organisations throughout 5 continents around the slogan: "Patients and Researchers: Partners for Life!"

EURORDIS organised a workshop bringing rare disease stakeholders together to discuss the future rare disease research agenda in Europe. The event entitled «Bridging Patients and Researchers to Build the Future Agenda for Rare Disease Research in Europe» took place on March 1st, 2010 at the Centre de Presse International in Brussels. It was co-organised with E-RARE in partnership with the European Commission, Orphanet and the EC-funded project EUROPLAN. 100 participants from 15 countries attended, including European and national research authorities, researchers, patient organisations and industry.

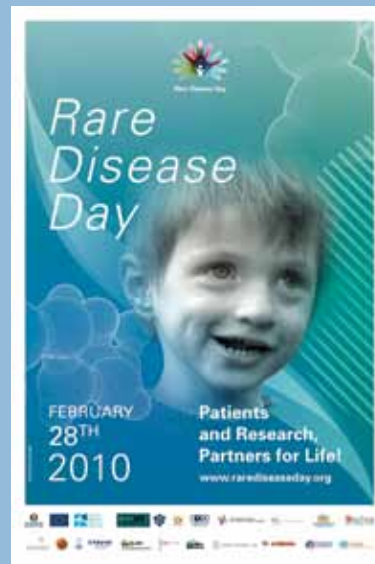
In addition, patient organisations were invited to nominate a scientist, who has helped advance research into their disease, to the Research Hall of Fame. EURORDIS received 46 nominations, which were visible on the Rare Disease Day website for 10 months.

The RDD 2010 website ([www.rarediseaseday.org](http://www.rarediseaseday.org)) received 39 000 visits from 156 countries in two months, and included 46 country pages with a calendar of events organised in each country. The website contained a downloadable tool kit with an Information Pack, logo, poster and other materials accessible to organisers; a section for patients to upload photos and videos; press releases for journalists and links to 4 social media platforms (Facebook, YouTube, Flickr, and Twitter) especially created for the day.

More than 10 000 fans joined RDD Facebook and 700 people followed the campaign on Twitter.

Other interesting features this year included: the Photo and Video Contest with 272 and 36 entries respectively; a news media feed which displayed media clippings on a loop and 'Friends of Rare Disease Day' and a new dedicated section where all Rare Disease Day supporters could register and describe their contribution to the campaign. In total 189 individuals, patient organisations, industry, health professionals, research and public authorities signed up as Friends of Rare Disease Day.

800 posters were printed and sent out to National Alliances, European Federations and member patient organisations and almost 2000 news articles were collected through a media monitoring service.



# HEALTH POLICY & HEALTH CARE SERVICES 2010



**PUSHING FOR THE ADOPTION AND ACTIVELY  
SUPPORTING THE IMPLEMENTATION OF COHERENT  
AND COORDINATED RARE DISEASE STRATEGIES AT  
THE EUROPEAN AND NATIONAL LEVEL.  
CONDUCTING SURVEYS AND MANAGING PROJECTS  
THAT AIM AT GIVING PATIENTS A VOICE IN THE  
HEALTH CARE POLICY THAT AFFECTS THEM.  
PROMOTING THE IMPLEMENTATION OF SERVICES  
ADAPTED TO THE SITUATION AND SPECIAL NEEDS OF  
PEOPLE LIVING WITH RARE DISEASES.**

## **PATIENTS' CONSENSUS ON PREFERRED POLICY SCENARIO FOR RARE DISEASES PROJECT – POLKA**

Continuation of the POLKA project, which started in 2008. The central idea of this project is to foster the opinion of patient representatives on future European policies for rare diseases or to collect their views on existing ones. In short: "Shaping the future by empowering patients with adequate advocacy tools". The project rests on 3 pillars: Deliberative patients' debates; European Reference Networks and Centres of Expertise for rare diseases and 5th European Conference on Rare Diseases.

### **DELIBERATIVE PATIENTS' DEBATES – PLAY DECIDE**

Creation of six new Play Decide sessions on the following topics of particular importance to rare disease patients: neonatal 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 games have been translated into 21 languages (this is the first time EURORDIS has decided to develop materials in most European languages). We also contributed to the overhaul of the dedicated website: [www.playdecide.eu/getinvolved/projects/](http://www.playdecide.eu/getinvolved/projects/)

In 2010, 114 sessions have been played, involving 633 participants (most of whom are patients or patient representatives) from Brazil, Canada and 16 European countries. Hungary, Denmark and Poland are leading. All together they have organised half of the sessions played until now. An intensive communication campaign has been launched to invite many more to organise Play Decide sessions, as the ultimate goal at the end of the POLKA project (August 2011) is to organise at least 500 sessions.



## EUROPEAN REFERENCE NETWORKS AND CENTRES OF EXPERTISE FOR RARE DISEASES

Foster the involvement of patient representatives in shaping the policy of European Reference Networks of Centres of Expertise, and continuous advice to the European Commission on its policy regarding these networks.

To analyse the implication of patient organisations in existing European Reference Networks (ERN), **EURORDIS** is involved as a partner in some ERN funded by the DG SanCo or as a member of their Advisory Board. Interviews have been conducted with ERN academic leaders and patient organisation representatives. On this basis, guidelines on good practices for the collaboration between ERN and patient groups and a template Work Package for future collaboration have been developed and adopted by the **EURORDIS** Council of European Rare Diseases Federations and Networks.

To analyse the involvement of patient organisations in the activities of Centres of Expertise in France, Denmark and United Kingdom, a Delphi method is being used, consisting of an online survey of patients and health care professionals in selected centres in these countries. Results will be presented and discussed in face to face meetings in 2011. This will help analyse where satisfaction diverges or converges, and propose ideas to improve the situation, when needed.

In 2010, **EURORDIS** has been a partner in the European Network on Rare Paediatric Neurological Diseases (nEUroped), in the European Registry and Network for Intoxication-type Metabolic Diseases (E-IMD) and in the European Registry for Wolfram syndrome, Alstrom syndrome and Bardet Biedl syndrome (EURO-WABB).

## 5<sup>TH</sup> EUROPEAN CONFERENCE ON RARE DISEASES, ECRD KRAKOW 2010: "FROM POLICIES TO EFFECTIVE SERVICES FOR PATIENTS"



The 5<sup>th</sup> European Conference on Rare Diseases ECRD 2010 was held in Krakow from 13 to 15 May 2010 and was a great success:

- Increased participation from Eastern and Central Europe
- Full implementation of ECRD 2007 participants' main expectations
- Rich conference programme: 71 speakers, 24 sessions, 58 posters
- Involvement of Polish organisations and Polish Ministry of Health

A total of 545 participants attended the event (as compared to 320 in 2005, 420 in 2007), including 71 speakers, 42 session chairs and 38 full fellowship recipients. The participants were from 41 countries (as compared to 24 in 2005, 35 in 2007), including 36 European countries, and 28 from EU/EEA. The ECRD offered 38 fellowships to allow patient representatives from new Member States or Central and Eastern Europe to participate.

### Eight main themes were proposed:

1. National Plans and Strategies for Rare Diseases
2. Centres of Expertise and European Reference Networks
3. Science from the Bench to the Bed Side
4. Information and Medical Education
5. The EU Committee of Experts on Rare Diseases
6. Policy Scenarii for Rare Diseases (POLKA project)
7. Rare Diseases in Central / Eastern Europe
8. Services to Patients, Families and Carers (posters)

A supplement on the Orphanet Journal of Rare Diseases was published with a selection of abstracts from speakers' presentations and posters.

The **EURORDIS** Annual General Assembly took place on the occasion of the ECRD.

### COUNTRY VISITS TO ESTONIA

On 24 March 2010, EURORDIS met with 20 patient representatives, policy makers and health care professionals in Tallinn. The purpose was to learn more on the reality of rare diseases in Estonia

and to envisage future working relations, as well as the creation of an Estonian Alliance for Rare Diseases.



### EUROPEAN NETWORK OF RESPITE CARE SERVICES

This activity is a continuation of the work that was done during the Rare Disease Patient Solidarity (RAPSODY) project on increasing the quantity and quality of Respite Care Services (RCS) that exist around Europe.

More services have been added to the online tool. There are now 15 RCS featured on the site which are searchable by patients looking for services in their area.

The Best Practice Guidelines have been updated and modified to reflect the range of different types of respite services (residential, domiciliary and emergency) that exist today. All listed Services are being invited to comment or modify the text by means of a wiki that has been set up on the network's website.

### THERAPEUTIC RECREATION PROGRAMMES

This activity is a continuation of the work that was done during the Rare Disease Patient Solidarity (RAPSODY) project to increase the quantity and quality of Therapeutic Recreation Programmes (TRP) that exist around Europe today. EURORDIS continues to identify TRP around Europe which it

then adds to the online cartography listing. There are now 40 TRP listed on the site. Best Practice Recommendations have been consolidated into a working paper open for comments.



# RESEARCH, DRUGS & THERAPIES 2010

CONTRIBUTING TO THE PROMOTION AND  
MAINTENANCE OF RARE DISEASES AS A PRIORITY  
IN EU RESEARCH POLICY AND FUNDING SCHEMES.  
REPRESENTING THE NEEDS OF PATIENTS IN  
EUROPEAN RESEARCH NETWORKS AND EMPOWERING  
PATIENTS IN CLINICAL RESEARCH ACTIVITIES.  
REPRESENTING PATIENTS IN THE DRUG REGULATORY  
PROCESS AND WORKING WITH INDUSTRY TO SPEED  
UP THE DEVELOPMENT AND AVAILABILITY OF  
TREATMENTS.  
PROMOTING TRANSPARENT AND QUALITY  
INFORMATION ON CLINICAL TRIALS AND MEDICINES  
FOR PATIENTS.



## 2010: A RARE DISEASE DAY DEDICATED TO RESEARCH

A conference, entitled « Bridging Patients and Researchers to Build the Future Agenda for Rare Disease Research in Europe », was held on 01 March, 2010 in Brussels. Throughout the day, prestigious speakers, panellists and participants made the case for rare disease research to be included in public funding schemes and to remain high in the European research agenda. This conference was attended by 106 attendees from 15 countries.

### SURVEY ON “EUROPEAN RARE DISEASE PATIENT GROUPS IN RESEARCH: ROLE AND PRIORITIES”

This meeting was also the occasion to present the EURORDIS Survey on “European Rare Disease Patient Groups in Research: Current Role and Priorities for the Future” conducted in collaboration with a team of researchers from the “Centre de sociologie de l’innovation” (Ecole des Mines, Paris, France). The survey brought to the

forefront, in what ways and to what extent, rare disease patient organisations support research. It also examined priority areas and bottlenecks encountered, from the patient’s perspective.



## EURORDIS REFERENCE PAPER ON “WHY INVEST IN RARE DISEASE RESEARCH”

The Reference Paper arguing in favour of investing in rare disease research was finalised. Further consultations were launched within EURORDIS and with external stakeholders. Relevant arguments were collected in favour of targeted and increased investment in rare disease research, at national and EU level.

This Reference Paper will be followed by a second

EURORDIS Reference Paper on priorities in rare disease research in view of the forthcoming adoption of the EU 8th Research Framework Programme 2014-2020, as well as in support of national research strategies within the national rare disease plans, or of international coordination strategies within the International Consortium for Rare Disease Research

## INTERNATIONAL CONSORTIUM FOR RARE DISEASE RESEARCH (IRDiRC)

Participation to the first EU-US Bilateral Workshop on Rare Diseases and Orphan Products “Fostering Transatlantic Cooperation on Research into Rare Diseases” (Reykjavik, 27-28 October 2010).

Active involvement in the activities of the Steering Committee created in Reykjavik to set up an International Consortium for Rare

Disease Research (IRDiRC), led by the European Commission and the US National Institutes of Health. It is expected that this International Consortium will be launched officially in April. EURORDIS will participate in working groups and is actively contributing to policy proposals.

## EURORDIS INVOLVEMENT IN BIOBANKING ACTIVITIES: THE EUROBIOBANK NETWORK ([WWW.EUROBIOBANK.ORG](http://WWW.EUROBIOBANK.ORG)) AND THE BIOBANKING AND BIOMOLECULAR RESOURCES RESEARCH INFRASTRUCTURE (BBMRI)

A total of over 440,000 rare disease samples (DNA, Cell and Tissue) were available across the EuroBioBank Network in 2010. An indicator of the network's dynamism is the number of samples exchanged: 22,058 samples collected and 6,657 samples distributed. 10 publications acknowledging EuroBioBank have resulted from scientific work performed with samples from the network.

In 2010, the EuroBioBank network, as partner of the TREAT-NMD Network of Excellence (Translational Research in Europe – Assessment and Treatment of Neuromuscular diseases [WWW.treat-nmd.eu](http://WWW.treat-nmd.eu)), organised five specialised training sessions in muscle cell cultures at the Friedrich-

Baur Institut/MTCC in Munich, a member of EuroBioBank.

In addition, as coordinator of the EuroBioBank network, EURORDIS pursues its collaboration with the Biobanking and Biomolecular Resources Research Infrastructure (BBMRI), in particular, with the inclusion of EuroBioBank in the BBMRI prototype under preparation.

Thanks to its experience of patient involvement in biobanks, EURORDIS actively contributed to the elaboration of the BBMRI Stakeholders' Forum document on “Basic Principles for Patient Participation in BBMRI” which was officially presented to the BBMRI management in June 2010.



## COLLABORATION WITH RARE DISEASE RESEARCH NETWORKS

Patient representation and collaboration of **EURORDIS** in the following rare disease research networks supported by the European Commission:

### CLINIGENE

The European network for the Advancement of Clinical Gene Transfer and Therapy is a Network of Excellence funded by the EU 6th Research Framework Programme. The overall objective of the network is to integrate multidisciplinary research and development in gene therapy. **EURORDIS** is member of the Ethical Review Board.

### ECRIN

The European Clinical Research Infrastructures Network is a network of excellence gathering the main public clinical research centres in order to share good practices and methodologies to facilitate international clinical trials in all fields. **ECRIN** and **EURORDIS** have been collaborating since the onset of this network more than five years ago. In past years, they collaborated on capacity building sessions for rare disease patient advocates in France, Spain, Italy and Denmark and on the possible implementation of the **EURORDIS** Charter on Clinical Trials to engage patient advocates in the design and follow up of academic clinical research. As a patient representative, **EURORDIS** has been invited to participate in an **ECRIN** project as an Associated Partner for the next five years. The project is under review.

### E-RARE

E-Rare is the Era-Net project that has been established to develop synergies between eight national research programmes on rare diseases in the European Union, and to set up a common research policy on rare diseases. **EURORDIS** is a member of the External Advisory Board. In 2010, E-Rare and **EURORDIS** jointly organised the « Bridging Patients and Researchers to Build the Future Agenda for Rare Disease Research in Europe » Conference in Brussels on the occasion of Rare Disease Day.

### NEUROPED

The European Network on Rare Paediatric Neurological Diseases (nEUroped) project aims to develop a network of communication and information sharing across the field of rare nervous system disorders in children, characterised by paroxysmal attacks. **EURORDIS** participates as a full network partner.

### TREAT-NMD

TREAT-NMD is a major initiative in the neuromuscular field, creating the infrastructure and the tools to ensure that the most promising new therapies reach patients as quickly as possible. As a partner of the TREAT-NMD Network of Excellence, **EURORDIS** represents the EuroBioBank network, which ensures the biobanking activities for the project and contributes to its activities on patient registries.

## PATIENT REPRESENTATIVES AT THE EUROPEAN MEDICINES AGENCY

Patient representation in the following European Medicines Agency (EMA) Committees:

- Committee for Orphan Medical Products (COMP): 123 orphan drugs designated, 4 marketed in 2010
- Paediatric Committee (PDCO)
- Committee for Advanced Therapies (CAT)
- Patients' and Consumers' Working Party

A total of 519 dossiers were examined by our patient representatives as part of the scientific committees they belong to. These include: 157 dossiers for orphan drug designations, 46 for classification or certifications by the CAT and 326 for paediatric investigation plans by the PDCO.

Continuation of three Task Forces involving a total of 34 patient representatives and volunteers in the areas of Orphan Drugs; Paediatric Drugs and Drug Information, Transparency and Access; and organisation of one meeting for each Task Force. Continuation of the Therapeutic Action Group (TAG) composed of all **EURORDIS** representatives

in the above-mentioned scientific committees and working party at the EMA.

The Drug Information, Transparency and Access (DITA) Task Force met twice in 2010 and exchanged information on: Fabrazyme and Replagal shortages; the transparency of information released by the EMA; the EMA consultation on the ethics of clinical trials in third countries; the situation of Revlimid in Poland; the off-label use of medicines, and the Code of Practices guiding relations between industry and patient organisations; the Pharmacovigilance Working Party at the EMA, Scientific Advice and finally, compassionate use.

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.

Regarding the Fabrazyme shortage, **EURORDIS** liaised with the EMA and Fabry disease patients, published a Questions and Answers document and advised patient organisations in the regulatory process.

## EURORDIS SUMMER SCHOOL FOR ADVANCED PATIENT ADVOCATES

The third EURORDIS Summer School held in Barcelona in September 2010 aimed at building on



the capacities of the participants acquired during the previous training sessions. This 4-day advanced training course gathered 34 fellows from

15 countries and demonstrated, via small group sessions and patients' testimonies, 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. In addition, the subject of Health Technology Assessment was introduced for the first time.

Based on the experience gained from the EURORDIS Summer School, we are developing a comprehensive E-Learning tool covering topics such as clinical trial design, statistics and ethics.

All the above experience will be further developed to include additional topics and to target different disease groups through an Innovative Medicines Initiative project. If accepted, this project will start in 2012 and last for five years.

## CHARTER FOR CLINICAL TRIALS ON RARE DISEASES

The general aim of the EURORDIS Charter for Clinical Trials in 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.

## DRUG INFORMATION ASSOCIATION PATIENT FELLOWSHIP PROGRAMME



Foster patient representation at the Drug Information Association (DIA) 21st annual EuroMeeting in Monaco March 8-10, 2010. For the fifth consecutive year, the DIA Patient Fellowship programme enabled 30 patient representatives from 15 different countries to participate in this important conference. A special booth dedicated to the patient fellows was set up to increase their visibility.

## 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 work involved in the Clinical Added Value of Orphan Drugs (CAVOD).

In 2010, EURORDIS has significantly increased its activities on HTA, focusing on acquiring the knowledge base, training rare disease patient representatives, gathering their experience in HTA procedures and promoting the involvement of patients into HTA procedures across Europe.

For the second year, five patient representatives from the EURORDIS staff and members were identified and selected to participate in four days of training

at the University of Hall in Tyrol, Austria.

In addition, patient representatives were identified and selected to participate in a two-day training session at the DIA HTA Forum, held on 4-5 November, 2010 in Paris.

EURORDIS was also co-organiser of the EPPOSI Workshop on "Patient Engagement in Health Technology Assessment" on 17 November 2011 in Brussels. EURORDIS representatives acted as chairpersons and speakers, shared their experience and contributed to shape future policy in this area. Fellowships were granted to 15 patient representatives, most of them members of EURORDIS, enabling them to take part in this workshop.



## THE EUNETHTA JOINT ACTION

In 2010 EURORDIS was appointed as one of four patient representative organisations at the EUnetHTA Stakeholders' Forum, which held its first meeting in June. 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.

## COMPASSIONATE USE AND OFF-LABEL USE

A survey was conducted amongst 26 pharmaceutical companies regarding their experience of compassionate use and off-label use during the last three years, and also amongst patient organisations. Results will be analysed and published in 2011. This survey will serve as background work for the EURORDIS Round Table of Companies Workshop on Compassionate Use & Early Access Programme in 2011.

On off-label use, a questionnaire was proposed to participants at the Summer School 2010 and a survey will be launched in 2011. A Questions and Answers document on off-label use was prepared and distributed to participants at EUROPLAN national conferences.

## EURORDIS ROUND TABLE OF COMPANIES (ERTC) 2010 WORKSHOPS

2010

### ➤ *"Patients' Access to OMPs, Innovative Pricing Schemes and National Measures in a Global Financial & Economic Crisis Environment"*

13 December 2010, Paris: 86 attendees from 19 countries

Attracting a large audience, the topic of this workshop was timely in a global financial and economic crisis environment, with additional pressure on national healthcare budgets. In a context of promotion of national plans and strategies on rare diseases in all the European Member States, the development of new and more appropriate measures to improve access to Orphan Medicinal Products has become a priority.

### ➤ *"Retooling the EU Orphan Drug Regulation and US Orphan Drug Act for Better and Faster Development of Rare Disease Therapies"*

18 June 2010, Barcelona: 61 attendees from 16 countries

A consensus was reached about using all the potential and available tools offered by the current EU and US regulations, rather than starting a revision process for which the outcome would be rather uncertain. Several ways of improving the orphan drug regulation were evoked and solutions proposed to accelerate the development of orphan drugs on both sides of the Atlantic.

### ➤ **In 2010, the ERTC comprises:**

- 35 members
- 5 new members (Bayer Schering, Clovis Oncology, Inc., InterMune Inc., Synageva BioPharma Corp. and ViroPharma Limited)
- 2 upgrades (BioMarin Europe Ltd. from € 5,000 to €10,000 and Pfizer PIO from € 10,000 to €25,000)

# GOVERNANCE, ORGANISATION & FUNDING 2010

## NEW EURORDIS STRATEGY 2010-2015

Members of EURORDIS unanimously adopted by secret ballot the new Strategy 2010-2015 at the General Assembly in May 2010 in Krakow.

This Strategy is the result of an 18-months strategic review process which has involved in-depth analysis and innovative thinking of the Board, staff, volunteers, partners and stakeholders and a broad consultation to all Members of EURORDIS through a survey to set forth priorities and a

debate at the General Assembly 2009.

The Annual Work Programmes (Action Plan, Budget, Governance, Organisation, Planning) for the period 2010 to 2015 are based on this Strategy based on regular strategic re-assessment and discussion to define or adjust the orientation in the short, medium and long term.

## BOARD OF DIRECTORS

Two new Board members were elected at the General Assembly 2010. The President and the General Secretary were re-elected. The Board is composed of 12 members from 8 European countries.

Mr. Jean Elie and Mr Anders Olauson stepped down after ten years of hard volunteer work on the Board of EURORDIS. Mr Olauson was President in 2000 and Mr Elie was Treasurer from 2000 until May 2010.

## STAFF TEAM ORGANISATION

The team comprises 28 people, 24,5 full-time equivalent (FTE) as of December 2010. The team is composed of paid staff, two seconded staff from AFM-Telethon and INSERM, 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 2010:

- Creation of a 'Volunteers and NGO Partners Coordinator' permanent position, held by Ariane Weinman
- Creation of an 'Events Manager' permanent position, held by Sharon Ashton
- Creation of an 'Online Patient Communities Coordinator' permanent position, held by Robert Pleticha
- Creation of a 'Communications and Development Director' permanent position, held by Jill Bonjean
- Creation of a 'Human Resources Consultant' short-term position, held by Beilla Baouz
- Creation of a 'Compassionate Use Coordinator' short-term position, held by Blanca Vera Gargallo
- Return of Nathacha Appanah, Communications Assistant, from parental leave
- Position of Executive Assistant to the CEO, now held by Veronique George
- Position of Web Content Manager, now held by Justine Evans
- Upgraded position of Accounting and Administrative Manager, held by Elisa Latxague
- No replacement of Health Policy Manager position (Help Lines and Specialised Social Services).



## VOLUNTEERS

**EURORDIS counted with 62 volunteer positions in 2010.**

EURORDIS' volunteer base plays an important and active role in our activities and projects. They are active in advocacy activities at the European level and are instrumental in catalysing the participation of all stakeholders on rare disease strategies at the national level.

We are proud to count 12 volunteers representing EURORDIS and the interests of patients in EU high-level Committees of the European Medicines Agency and the European Commission:

**\* EMA Committee for Orphan Medicinal Products:** Two EURORDIS representatives out of three patient representatives: Ms 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.

**\* EMA Committee for Paediatric Drugs:** One EURORDIS' representative out of three patient representatives: Dr Tsveta Schyns (European Network for Research on Alternating Hemiplegia, ENRAH, Belgium).

**\* EMA Committee for Advanced Therapies:** One EURORDIS' representative out of two patient representatives: Dr. Michele Lipucci di Paola (Associazione Veneta Lotta Talassemia, Italy) EURORDIS alternate patient representative to Dr. Fabrizia Bignami (Therapeutic Development Director) who is EURORDIS full member patient representative.

**\* EMA Patients' and Consumers Working Party:** The Co-Chair, Ms. Lise Murphy (Marfan Syndrome, Sweden) is a volunteer EURORDIS' representative; Francois Houÿez (Health Policy Director) is the permanent representative of EURORDIS in this Working Party.

**\* EU Committee of Experts on Rare Diseases:**

The eight seats (four full members and four alternates) for patients are held by EURORDIS' representatives. Mr Le Cam is the only staff member, other representatives are volunteers, all being patients or parents of patients: Ms Dorica Dan (Romanian Prader Willi Association and Rare Diseases Romania), Mr Jan Geissler (Leukämie-Online e.V, Germany), Mr Torben Grønnebaek (Wilson Disease and Rare Disorders Denmark), Mr Alastair Kent (Genetic Alliance UK and Rare Disease UK), Ms Christel Nourissier, (EURORDIS and Rare Disease Alliance, France) Ms Bianca Pizzera (International Patient Organisation for Primary Immunodeficiencies, IPOPI, Italy) and Mr Gabor Pogany (William Syndrome and Rare Diseases Hungary).

**\* Advisors to National Plans and Strategies on Rare Diseases:**

Six advisors appointed by EURORDIS based on a Call for Expression of Interest and appointed by the Board, were in charge of coordinating 15 national conferences on National Plans on Rare Diseases within the framework of EUROPLAN (please refer to the Advocacy section). In addition a large number of volunteers in each of the 15 countries where EUROPLAN conferences were organised, were involved in the organisation of the conferences and were instrumental in bringing all stakeholders together.

**\* Volunteers contribute to enhancing EURORDIS' participation in international conferences**

Volunteers are also involved in international conferences to voice the rare disease patients' views in a multi-stakeholders framework.

**\* Volunteer translators**

There are 5 volunteers ensuring the translation and proofreading of various key documents and website content on a regular basis. ✓

EURORDIS' organisation to support volunteers involved in committees and in other advocacy activities.

EURORDIS has established different volunteer groups, coordinated by EURORDIS staff. There are three internal task forces: the Orphan Drugs Task Force, the Paediatric Drugs Task Force and the DITA Task Force (Drug, Information, Transparency and Access). These groups are aimed at supporting the work performed by others EURORDIS' volunteers in EU committees as well as developing initiatives in favour of rare disease patients in different fields (therapeutics, public health, social services and others)

\* The Therapeutic Action Group (TAG): This group is composed of the five EURORDIS volunteers and the three staff members who represent EURORDIS in EMA scientific committees. They have monthly conference calls to discuss on the work performed in their respective committees and to share relevant information. The TAG is coordinated by Dr. Maria Mavris, Drug Development Manager.

\* The Policy Action Group (PAG): Composed by all the members of EURORDIS representatives on the EUCERD (seven volunteers and our CEO, Yann Le Cam), Flaminia Macchia, European Public Affairs Director and Ariane Weinman, Volunteers and NGO Partners Coordinator. The group organises regular conference calls to better follow up the work of the EUCERD.

\* The European Public Affairs Committee (EPAC): This internal committee plays an active and key role in EURORDIS' governance and advocacy activities. It is composed of 30 members: 18 volunteers (Board members, TAG members and full members representing patients on the EUCERD), as well as 12 staff members. Each EPAC member has a specific mandate to represent EURORDIS in institutions and in conferences.

The EPAC members communicate regularly by email and discuss important issues such as cross-border healthcare and information to patients. Each EPAC member is entitled to vote for the adoption of a EURORDIS' position on specific issues.

## NEW HOME FOR EURORDIS AT THE RARE DISEASES PLATFORM IN PARIS

In August 2010, EURORDIS settled into newly renovated dwellings on the grounds of the Hospital Broussais in Paris within the Plateforme Maladies Rares (Rare Diseases Platform).

After anticipating this move for several years, it finally became a reality. In addition to allowing us to better welcome our members and partners at national and European levels, these new facilities provide an even more harmonious and collaborative work environment within the Platform.



The French Minister of Health, Roselyne Bachelot-Narquin, inaugurated the new offices of the Rare Diseases Platform on 21 October 2010.

The Plateforme Maladies Rares, created in 2001, is unprecedented in Europe as it unites key players in the field of rare diseases under one roof: rare disease patient organisations (Alliance Maladies Rares, EURORDIS), rare disease information services (Orphanet, MRIS) and major players in the field of rare disease research funding (GIS-Maladies Rares and the AFM).





## FINANCE AND OPERATIONS

- Accounting and monthly financial reporting in a timely manner including cash flow and risk analysis detailed report
- Monthly meeting with managers to update the Budget and the Year-end Financial Forecast
- Operating Grant Steering Committees organised

every two months with review of progress on implementation, deliverables and budget

- Quarterly collection of activity indicators
- Management of human resources related activities such as recruitments, regulations, etc.

## CONTRACT GRANTS

### New

Operating Grant for year 2010 ("EURORDIS\_FY2010"), single beneficiary, DG Health and Consumers, 12 months.

### Renewed

- Web2.0, The French Pharmaceutical Companies Association (LeeM), 12 months.
- Rare!Together, Medtronic Foundation, 24 months.
- Summer School Session 2010, Drug Information Association Philanthropy Programme, event advocacy and core activities, AFM-Téléthon, 48 months 2010-2013.

### Ongoing

- 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.
- e-Newsletter, Medtronic Foundation, 12 months.



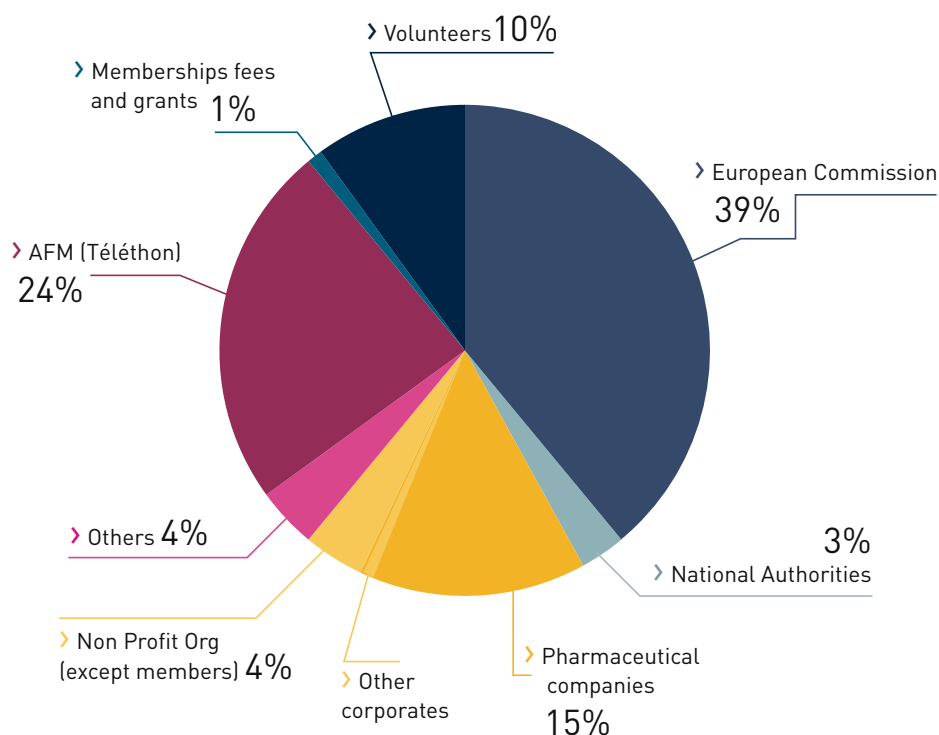
# REVENUE AND EXPENSES

2010 (in thousand of euros, provisions excluded)



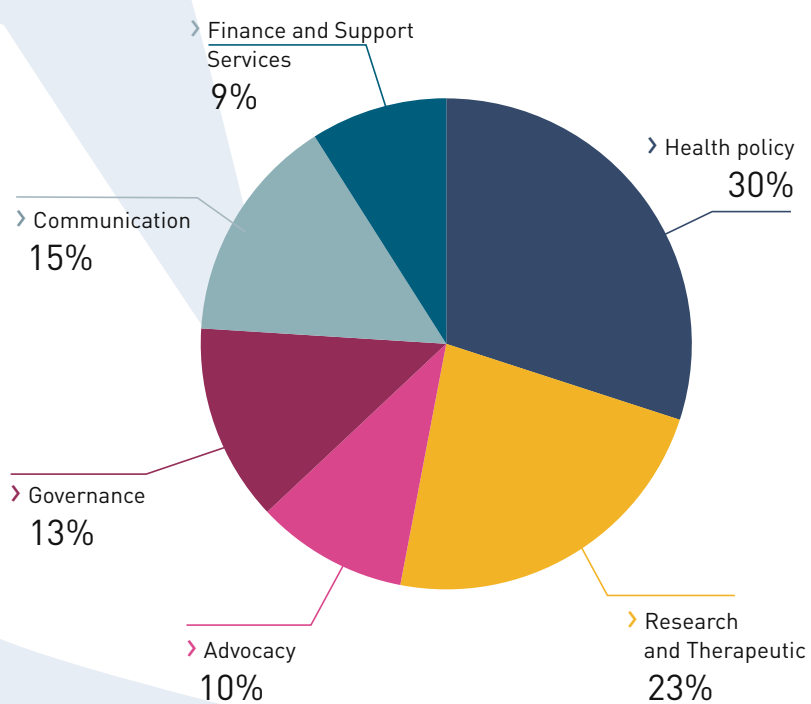
**EURORDIS**  
Rare Diseases Europe

› REVENUE BY ORIGIN 2010 = 3,553 k€



AFM (Téléthon)	843
Membership fees and grants	41
Volunteers	376
European Commission	1380
National Authorities	95
Pharmaceutical Companies	541
Other corporates	6
Non Profit Org (except members)	131
Other	140

› EXPENSES BY DESTINATION 2010 = 3,417 k€



Health Policy	1007
Research and Therapeutic	800
Advocacy	351
Governance	428
Communication	523
Finance and Support Services	308

# BOARD OF DIRECTORS

MAY 2010-MAY 2011



PRESIDENT		
Mr. Terkel Andersen *	<ul style="list-style-type: none"> <li>• President</li> </ul>	Danish Haemophilia Society Denmark
DIRECTORS		
Ms Karen Aiach *	<ul style="list-style-type: none"> <li>• Treasurer</li> </ul>	Alliance SANFILIPPO France
Ms Avril Daly		Genetic & Rare Disorders Organisation Ireland
Ms Dorica Dan		Romanian Prader Willi Association Romania
Mr. John Dart		DEBRA International UK
Ms Renza Galluppi		Federazione Italiana Malattie Rare - UNIAMO Italy
Mr. Torben Grønnebæk		Rare Disorders Denmark Denmark
Mr. Flavio Minelli *	<ul style="list-style-type: none"> <li>• Officer</li> </ul>	Unione Italiana Ittiosi Italy
Ms Béatrice de Montleau		Associations Française contre les Myopathies France
Dr. Mirando Mrsic		The Croatian Society of Patients with Rare Diseases Croatia
Ms Christel Nourissier *	<ul style="list-style-type: none"> <li>• General Secretary</li> </ul>	Prader Willi France France
Ms Rosa Sánchez de Vega*	<ul style="list-style-type: none"> <li>• Vice-President</li> </ul>	FEDER – Federación Española de Enfermedades Raras Spain

\*Member of the Board of Officers



<b>NORWAY (2)</b>	FRAMBU - RESOURCE CENTRE FOR RARE DISORDERS
	MORBUS ADDISON ASSOCIATION NORWAY
	NORDIC HPTH ORGANISATION
	NORSK FORENING FOR EHLERS-DANLOS SYNDROM
	NORSK FORENING FOR TUBEROS SKLEROSE / NORWEGIAN ASSOCIATION OF TUBEROUS SCLEROSIS
<b>POLAND</b>	DEBRA POLSKA
	MATIO-FUNDACJI POMOCY RODZINOM I CHORYM NA MUKOWISCYDOZĘ
	POLISH PKU AND RD ASSOCIATION "ARS VIVENDI"
	THE DINA RADZIWILLOWA CHILD'S HEART FOUNDATION
<b>PORTUGAL</b>	ALIANÇA PORTUGUESA DE ASSOCIAÇÕES DAS DOENÇAS RARAS
	APLL - ASSOCIAÇÃO PORTUGUESA DE LEUCEMIAS E LINFOMAS
	ASSOCIAÇÃO NACIONAL DE FIBROSE QUISTICA
	ASSOCIAÇÃO PORTUGUESA DE DOENTES NEUROMUSCULARES
	FEDRA - FEDERAÇÃO PORTUGUESA DE DOENÇAS RARAS
	LIGUA NACIONAL PARA O ESTUDO E APOIO DA DEFICIENCIA MENTAL
	NUCLEO DE EPIDERMOLISE BOLHOSA
	RARISSIMAS - ASSOCIACAO NACIONAL DE DEFICIENCIAS MENTAIS E RARAS
<b>ROMANIA</b>	ASOCIATIA ROMANA SPINA BIFIDA SI HIDROCEFALIE
	ASOCIATIA WERDNIG HOFFMAN AWH
	ASOCIATIA WILLIAMS SYNDROME
	ROMANIAN NATIONAL ALLIANCE FOR RARE DISEASES
	ROMANIAN PRADER WILLI ASSOCIATION
<b>RUSSIAN FEDERATION</b>	ALLREGIONAL PUBLIC ORGANISATION "HELP TO CYSTIC FIBROSIS PATIENTS"
	HUNTER SYNDROME ASSOCIATION
	INTERREGIONAL PUBLIC ORGANISATION "ASSISTANCE TO THE INVALIDS FROM CHILDHOOD, SUFFERING FROM GAUCHER DISEASE,AND THEIR FAMILIES
	NATIONAL ASSOCIATION OF RARE DISEASES PATIENTS "GENETICS"
<b>SERBIA</b>	CHILDHOOD CANCER PARENT ORGANISATION "ZVONCICA"
<b>SLOVAKIA</b>	DEBRA SR
	SLOVAK CYSTIC FIBROSIS ASSOCIATION
<b>SLOVENIA</b>	FOUNDATION OF CHILD NEUROLOGY
<b>SPAIN</b>	ALIANZA ESPAÑOLA DE FAMILIAS DE VON HIPPEL LINDAU
	ASOCIACIÓ CATALANA DE LAS NEUROFIBROMATOSIS
	ASOCIACIÓ D'AFECTATS DE SIRINGOMIÈLIA
	ASOCIACIÓN ANDALUZA CONTRA LA FIBROSIS QUÍSTICA
	ASOCIACIÓN ANDALUZA DE PACIENTES CON SÍNDROME DE TOURETTE Y TRASTORNOS ASOCIADOS
	ASOCIACIÓN BALEAR DE AFECTADOS POR LA TRIGONITIS Y LA CISTITIS INTERSTICIAL
	ASOCIACIÓN DE AFECTADOS DE NEUROFIBROMATOSIS
	ASOCIACIÓN DE DEFICIENCIAS DE CRECIMIENTO Y DESARROLLO
	ASOCIACION DE ENFERMEDADES RARAS DE TOTANA D'GENES
	ASOCIACIÓN DE EPIDERMOLISIS BULLOSA DE ESPAÑA - ASOCIACIÓN "PIEL DE MARIPOSA" (AEBE)
	ASOCIACIÓN DE ESCLERODERMIA CASTELLON
	ASOCIACIÓN DE HEMOGLOBINURIA PAROXÍSTICA NOCTURNA
	ASOCIACIÓN DE HUESOS DE CRISTAL DE ESPAÑA
	ASOCIACIÓN DE LAS MUCOPOLISACARIDOSIS Y SÍNDROMES RELACIONADOS
	ASOCIACIÓN DE NEVUS GIGANTE CONGÉNITO
	ASOCIACIÓN DE PACIENTES DE LA ENFERMEDAD DE HUNTINGTON
	ASOCIACIÓN DEL SÍNDROME DE LESCH-NYHAN ESPAÑA
	ASOCIACION ESPAÑOLA DEL SINDROME CDG, DEFECTOS CONGENITOS DE LA GLICOSILACION
	ASOCIACIÓN ESPAÑOLA DE ANGIOEDEMA FAMILIAR POR DEFICIT DE C1
	ASOCIACIÓN ESPAÑOLA DE ANIRIDIA
	ASOCIACIÓN ESPAÑOLA DE ENFERMOS DE GLUCOGENOSIS

# CONFERENCES

## 2010

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- Second EPPOSI Workshop: "A European Strategy for Chronic Conditions", Brussels, Belgium, 9 December  
[Frank Brunsmann: The EURORDIS View on Electronic Health Records \(EHR\): Draft EHR checklist](#)

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- Patient Partner Final Workshop: Patients Partnering in Clinical Trials, Brussels, Belgium, 7-8 December  
[François Houjéz and Rob Camp represented EURORDIS](#)

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- EMA - TOPRA Joint Meeting: "What does the future for European regulatory affairs hold?", London, UK, 6-7 December  
[Maria Mavris, panelist in the session: "What will be the Future Role of the Regulatory Agencies?"](#)

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- Rare Neurological Diseases of Childhood: "We Treat the Child to Treat the Adult", European Parliament, Brussels, Belgium, 2 December  
[Tsveta Schyns represented EURORDIS](#)

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- World Orphan Drugs Congress, Geneva, Switzerland, November 29 through December 1st  
[Yann Le Cam: "Building an integrated, comprehensive and long-term strategy to address unmet patients' needs in Europe"](#)

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[Christel Nourissier: "Leading the way : The National Plans for Rare Diseases 1 and 2 In France"](#)

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- 11th EPPOSI Workshop on Partnering for Rare Diseases Therapy Development: «Working together to define Research, Regulation and Realities for the EU Rare Disease Community», Prague, Czech Republic, 29-30 November  
[Lesley Greene: Co-Chair of the Workshop representing patients](#)  
[Co-chair of Session 2: Regulation – The EU and Beyond: Developing Future Regulatory Policies for Better Rare Disease Therapies](#)

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[Yann Le Cam: speaker in the session: "Developing new tools for improving access: CAVOD, structured coordination between Member States and sui generis provision"](#)

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- European Network of Centres for Pharmacoepidemiology and Pharmacovigilance (ENCePP) Information Day, London, UK, 26 November  
[François Houjéz and Rob Camp represented EURORDIS](#)

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- nEUroped Extended Network Meeting, Brussels, Belgium, 21-23 November  
[Anna Kole: Integration of Patients - nEUroped](#)

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- Joint Consensus Meeting – European Network of Centres of Expertise (ENCE): Cystic Fibrosis - Lymphangiomyomatosis (LAM) - Lung Transplantation, Germany, Frankfurt, 17 – 18 November

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[Anna Kole: European Reference Networks of Centres of Expertise for Rare Diseases: Win-win of possible stakeholders](#)

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- EPPOSI Workshop: Patients' Engagement in HTA, Brussels, Belgium, 17 November  
[Yann Le Cam: "Added value of engaging patients in HTA – The patients' perspective"](#)

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[Yann Le Cam, co-chair of the session: "How should we engage with patients to ensure that HTA is truly patient-centred? When and how should patients engage in the HTA process?"](#)

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- Friends of Europe: "An Innovative Healthcare Agenda for Europe" Brussels, Belgium, 16 November  
[Yann Le Cam represented EURORDIS](#)

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- DIA 2nd Health Technology Assessment (HTA) Conference "Building a new system to get effective treatment to European patients", Paris, France, 4-5 November  
[Yann Le Cam, session chair of the panel discussion on: "Building a new system to get effective orphan treatment to EU patients – where do we go from here? And what can we learn that will be applicable to more "common condition"?"](#)

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- DIA Orphan Drugs conference «Future Direction for Orphan Drugs in Europe», Paris, France, 3 November  
[Lesley Greene, co-chair of the session: "Opportunities and Challenges in Orphan Drug Research and Development"](#)

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[Lesley Greene and Fabrizia Bignami: "Addressing Unmet Medical Needs"](#)

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[Yann Le Cam co-chair of the session: "Market access and HTA: Particular challenges and opportunities for orphan drugs generating the level of evidence needed for decision-making on the value of orphan drugs in the therapeutic strategy of a rare disease."](#)

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- 2nd Eastern European Conference on Prader Willi Syndrome, Zalau, Romania, 29-30 October  
[Christel Nourissier and Rob Pleticha represented EURORDIS](#)

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- Finnish Rare Disease Symposium, Helsinki, 29- 30 October  
[Taina Hickey: "The Voice of 12 000 Patients: Patients' Needs and Expectations: Access to Health Services", Eurordis-Care3 survey](#)

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- First EU-NIH Workshop: “Trans-Atlantic cooperation on research into rare diseases”, Reykjavik, Iceland, 27 – 28 October

Béatrice de Montleau and Valentina Bottarelli: “Linking Information Resources: Patients and Families”

- European Haemophilia Consortium conference, Lisbon, Portugal, 22-24 October

François Houÿez: “The voice of patients living with a rare disease at the European level”

- Conference on Rare Diseases, Mariazell, Austria, 15 – 16 October

Christel Nourissier: “Getting together to become stronger: The example of the European Organisation for Rare Diseases”

- Friends of Europe, “The State of Europe”, Brussels, Belgium, 14 October

Yann Le Cam represented EURORDIS

- “Personalized medicines between scientific perspectives and the need for clinical governance”, Parma, Italy, 12 October

Michele Lipucci di Paola, Former Vice-President of EURORDIS, Panelist

- First International South Caucasian Conference on Rare Diseases and Orphan Drugs Yerevan, Armenia, 7-8 October

Yann Le Cam : “Overview of rare diseases as a public health issue in European policy”

Dorica Dan: “The Romanian approach on rare diseases in Europe”

Professor Josep Torrent-Farnell, MD, PhD : “10 years of experience with orphan drugs”

- 2010 – Health 2.0 Conference, San Francisco, United States, 7-8 October

Denis Costello presented the EURORDIS Online Patients’ Communities

- Second European Conference on Rett Syndrome, Edinburgh, Scotland, 7 - 10 October

Gérard Nguyen represented EURORDIS

- Press Conference at the occasion of the establishment of the “Fondation Imagine” bringing together 32 centres of expertise for rare diseases at the Necker Hospital in Paris, France, 5 October

Christel Nourissier, General Secretary, represented EURORDIS

- Conference on Orphan Drugs and Cancers hosted by MEP Brepoels, European Parliament, Brussels, Belgium, 5 October

Yann Le Cam: “Sustainable and improved access to orphan drugs: why and how Member States should collaborate in EU?”

- III Foresight Training Course on “Benefit/Risk Assessment of Medicines to achieve shared objectives: from Research to Reality”, organised by Gianni Benzi Pharmacological Research Foundation, Krakow, Poland, 1-3 September

François Houÿez: “Benefit/Risk in the perspective of the European Road Map and National Agencies proposals”

François Houÿez: “Benefit/Risk assessment and HTA: HTA and patients’ rights”

- European Commission – DG Research Workshop: «Encouraging new ways of doing research». Brussels, Belgium, 16 July

Yann Le Cam represented EURORDIS

- “Fifth Eastern European Conference for Rare Diseases and Orphan Drugs “Rare Diseases in the Focus of Personalized Medicine” &

The First All-Russian Conference for Rare Diseases and Rarely Used Medical Technologies “Lifeline”, Saint-Petersburg, Russia, 2-4 July

Paloma Tejada: Presentation of EURORDIS

Vladimir Tomov (President of the National Alliance of People with RD in Bulgaria, member of the CNA): Presentation of the EURORDIS Council of National Alliances

- Forum des Associations – Fondation Groupama, Paris, France, 21 June

Denis Costello: Presentation of the Online Patients’ Communities (with a testimony of Mr. Rivière, Muckle Wells Syndrome and CINCA)

Paloma Tejada: Presentation of EURORDIS and its communication tools

- «Les maladies rares sont-elles encore une priorité en France?» (Are rare diseases still a priority in France?), organised by the LEEM (French Drug Manufacturers Federation), Maison de la Recherche, Paris, France, 15 June

Yann Le Cam panelist in the Round Table: “L’évaluation : Autorisation de mise sur le marché et Amélioration du service médical rendu » (Evaluation : Marketing Autorisation and Clinical Added Value of Medicines)

- DIA Management Board meeting, Washington DC, 12 June

Maria Mavris: Presentation at the DIA Management Board meeting: EURORDIS’ involvement at the EMA, the DIA Patient Fellowship Programme and the Summer School supported by the DIA Philanthropy programme.

- RIME: Réunion d’Information des Membres – Alliance Maladies Rares, Paris, France, 11 June

François Faurisson presented the outcomes of the EURORDIS survey on the involvement of patients’ organisations in research on rare diseases

François Houÿez presented the Play Decide Games

- First EUnetHTA Stakeholder Forum meeting, Dublin, Ireland, 9 June

François Houÿez represented EURORDIS

- BBMRI Stakeholder’s Forum - Information and Discussion Meeting : “The European Biobanking Research Infrastructure: A Step Closer”, Brussels, Belgium, 9 June

Fabrizia Bigmani is a member of the Programme Committee and was panelist in this Forum to discuss patients’ participation in BBMRI

- Genetic Interest Group Conference 2010 – “Giving Patients a Voice”, London, UK, 8 June

Lesley Greene, CLIMB & EURORDIS co-chaired the session on “Patient Information” in the workshop: “What could a national strategy for Rare Diseases look like?”

# CONFERENCES

## 2010 (2)

- “Together for Rare Diseases: Celebrating 10 Years of Success of the EU Regulation on Orphan Medicines” - Reception at the European Parliament, hosted by Ms Françoise Grossetête, MEP (EPP) – European Parliament, Brussels, Belgium, 1 June

Yann Le Cam: “Positive impact of the OMP Regulation for rare disease patients and patients’ expectations for the future”

- “26th Convegno Nazionale AS.IT.O.I. (Associazione Italiana Osteogeneis Imperfecta), Rome, Italy, 28 – 30 May  
Flavio Minelli: “EURORDIS: il valore della Alleanza di Associazioni in Europa”

- Bulgarian national media Seminar organised by geneticists (Prof. Ivo Kremenski) on «individual medicines, opportunities and perspectives», 21 – 22 May  
Christel Nourissier represented EURORDIS

- “Innovation in healthcare: from research to market – SMEs in focus” - Co-organised by the European Commission DG Research and DG Enterprise and Industry, Brussels, Belgium, 20-21 May  
Michele Lipucci di Paola: “Role of Patients in EU Centralised Procedure for ATMP (Advanced Therapy Medicinal Products)”

- NORD Corporate Council, Washington D.C., USA, 18 May  
Yann Le Cam: Presentation on the current European policy environment for rare diseases and orphan drugs; EURORDIS in brief and its strategy 2010 – 2015; the EURORDIS Round Table of Companies and the partnership NORD – EURORDIS

- European Medicines Agency: “10 Years of the Orphan Regulation in Europe”, London, UK, 3-4 May  
Yann Le Cam: “Is the Orphan Regulation addressing patient needs?”

Panel discussion on “the next 10 years”

- Patients’ view: Dr Fabrizia Bignami

- Health 2.0 Conference (organised by Denise Sielber) – Paris, France, 6-7 April  
Denis Costello made a presentation on the EURORDIS Online Patient Communities

- 2nd Pan-European Conference on Haemoglobinopathies: “Patients and health professionals together for optimal care” in partnership with the Charité-Universitätsmedizin Berlin on the occasion of its 300th anniversary, Berlin, Germany, 13-14 March  
Michele Lipucci di Paola, panelist in the “Joint Workshop – European Developments in the areas of Expert Centres and Registries”

- 22nd Annual DIA EuroMeeting, Monaco, 8-10 March  
Maria Mavris: “Transparency from clinical trials: Are we patient-focused?”

- Rare Disease Day 2010 - European Workshop: “Bridging Patients and Researchers to Build the Future Agenda for Rare Disease Research in Europe”, Centre de Presse Internationale (IPC), Brussels, Belgium, 1 March  
Fabrizia Bignami: “Role of Patient Groups in Research and their Priorities for the Future”

- Belgium Rare Disease Day 2010 organised by RADIORG. BE: “Patients and Researchers, Partners for Life!”, Brussels, Belgium, 27 February  
Yann Le Cam: “La reconnaissance d’un médicament orphelin”

- International Congress on « Rare Diseases and Orphan Drugs”, Istituto Superiore di Sanità, Rome, Italy, 22 – 25 February  
Terkel Andersen : “How to Promote Research Into Rare Diseases - EURORDIS’ Position”

- Press Conference for the Rare Disease Day 2010, organised by AFM, and Plateforme Maladies (Eurordis) , « Construire des ponts entre malades et chercheurs », Paris, France, 18 February  
Speech by Yann Le Cam: Presentation of EURORDIS, the Rare Disease Day 2010: “Bridging patients and researchers”, the results from the survey sent to EURORDIS’ members, the link with AFM, the French National Plan for RD and researchers

- Xth anniversary of the Alliance Maladies Rares, Paris, France, 15 February  
Christel Nourissier represented EURORDIS

- “Act against Europe’s most common birth defects – The right advice at the right time can reduce neural tube defects now” – Launch of the report – European Parliament, Brussels, Belgium, 27 January  
Flaminia Macchia represented EURORDIS



# ACKNOWLEDGEMENTS

## 2010

EURORDIS would like to thank the following organisations and companies for their highly valued support in 2010

### AFM - Téléthon

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



### INSERM

EURORDIS is grateful to the INSERM for the secondment of the Clinical Research Advisor of EURORDIS



### EUROPEAN COMMISSION

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

- Patients' Consensus on Preferred Policy Scenarios for Rare Diseases (POLKA)
- The European Project for Rare Diseases National Plans Development (EUROPLAN)
- The Operating Grant for Rare Disease Associations (EURORDIS FY2010)
- The European Network on Rare Paediatric Neurological Diseases (nEUroped)

EURORDIS is grateful to the TREAT-NMD project, funded by the European Commission (DG Research) under the Sixth Framework Program, for its support to the EuroBioBank Network



# ACKNOWLEDGEMENTS

## 2010

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### THE E-NEWSLETTER and RARE TOGETHER PROJECT



EURORDIS is grateful to The Medtronic Foundation for their financial support for the EURORDIS E-Newsletter and the Rare!Together Project

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### ON LINE PATIENT COMMUNITIES



EURORDIS is grateful to the LeeM, NORD, Alexion and Celgene for their financial support to the Online Patient Communities project

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### POLKA PROJECT CO-FUNDING

Biotherapies for Life™

CSL Behring



NOVARTIS

sigma-tau

and In-kind contributions from:



National Commissioning Group  
NHS UK NHS



Fundació Doctor Robert Universitat Autònoma  
de Barcelona Casa Convalescència

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### SUMMER SCHOOL FOR PATIENTS ADVOCATES



The DIA (Drug Information Association) Philanthropy Programme



and In-kind contributions from:  
Fundació Doctor Robert Universitat Autònoma de Barcelona Casa  
Convalescència

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### UNRESTRICTED FINANCIAL SUPPORT



## EUROPEAN CONFERENCE ON RARE DISEASES - ECRD 2010 KRAKOW

EURORDIS is grateful to the organisations listed below for their financial support, including fellowships for patient representatives:

		
		
		
	and In-kind contributions from the Medical School of the Jagiellonian University	
		

## ROUND TABLE OF COMPANIES

EURORDIS is grateful to the organisations listed below for their financial support through the EURORDIS Round Table of Companies

- Ruby Members

		
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- Emerald Members

# ACKNOWLEDGEMENTS

## 2010

### ROUND TABLE OF COMPANIES

- Sapphire members

		
		 *
		
		
		
		 *

\* Small size companies with a not-for-profit status for which a fee waiver has been granted for 2010



# WORK PROGRAMME 2011





# OUR ACTION PLAN FOR 2011



## 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 of and access to rare disease care
- Accessing cross-border healthcare and making patient mobility possible
- Promoting research and bridging patients and researchers
- Starting to address the issues of genetic testing, genetic counselling and newborn screening

### ADVOCACY ACTIONS

• SUPPORT IMPLEMENTATION OF THE NEW EU POLICY FRAMEWORK ADOPTED IN 2008 (COMMISSION COMMUNICATION ON RARE DISEASES) AND IN 2009 (COUNCIL RECOMMENDATIONS ON ACTIONS IN THE FIELD OF RARE DISEASES) THROUGH:

• Participation of eight EURORDIS' patient representatives in the EU Committee of Experts on Rare Diseases (EUCERD) created in December 2010

• Support the development of a Policy Road Map and of a Joint Action on Rare Diseases structuring EUCERD work towards achievements on key themes

• Participation of these eight patient representatives and additional EURORDIS' patient representatives in EUCERD Working Groups particularly on the key themes of National Strategies / Action Plans & Indicators, Research Policy, Centres of Expertise & European Reference Networks, Standardisation & Databases & Registries, Access to Rare Disease Therapies, Social Specialised Services and Integration of Rare Diseases into Social Policies and Services

• Support these patient representatives with a EURORDIS Policy Action Group (PAG), staff, volunteers, information sharing and reports

• Contribute with concrete policy proposals to implement strategy foreseen in this policy framework and take action to lead European collaboration in some areas

• Communication through the Newsletter, the Website, Conferences

• PROMOTE DEVELOPMENT OF NATIONAL STRATEGIES AND ACTION PLANS ON RARE DISEASES IN ALL MEMBER STATES BY 2013, THROUGH:

• Active participation in the EUROPLAN project:

methodology recommendations for the development of plans, identification and promotion of best measures for national policies, development of policy indicators, dissemination of information

• Dissemination of outcomes of the 15 national conferences (Romania, Bulgaria, Hungary, Croatia, Poland, France, Germany, Netherlands, Italy, Spain, Greece, UK, Denmark, Sweden, Ireland,) organised by National Alliances with EURORDIS in the framework of the EUROPLAN project to promote national plans on rare diseases. Dissemination of their summaries with key recommendations.

• Take initiative within the EUCERD to promote national plans by 2013

• Active EURORDIS support in several Member States and EU Presidency (2011 Hungary & Poland, 2012 Denmark & Cyprus, 2013 Ireland & Lithuania)

• Communication through the Newsletter, the Website, Conferences

• 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

• Through position papers and advocacy actions to European Commission, European Parliament, Council and EU Presidency

• ADVOCATE ON THE DIRECTIVE ON PATIENTS' RIGHTS IN CROSS-BORDER HEALTHCARE TO PROVIDE A SUSTAINABLE LEGAL AND FINANCIAL INSTRUMENT FOR EUROPEAN REFERENCE NETWORKS AND



PROMOTE POLICY MEASURES ADDRESSING THE SPECIFIC NEEDS OF RARE DISEASE PATIENTS AND FAMILIES

- CONTRIBUTE TO THE DRAFTING OF THE EU POLICY AND ACTION PLAN ON ORGAN DONATION AND TRANSPLANTATION

- PROMOTE RARE DISEASES AS AN INTERNATIONAL PUBLIC HEALTH PRIORITY THROUGH:

- International spreading of Rare Disease Day
- Coordination of a reference international position paper
- Drawing of strategy directed towards the World Health Organisation
- Support to the initiative of an International Rare Disease Research Consortium

- Support to the International Conference for Rare Diseases and Orphan Drugs (ICORD) as a tool to expand the rare disease movement at the international level and to liaise with international bodies

- ADVOCACY IN SUPPORT OF RARE DISEASE RESEARCH. A HIGH PRIORITY IN THE STRATEGY 2010-2015. BASED ON THE 2010 SURVEY ON THE INVOLVEMENT OF PATIENT ORGANISATIONS IN

RESEARCH, RARE DISEASE DAY 2010, CONFERENCE IN BRUSSELS IN MARCH 2010, THE REFERENCE PAPER AND ADVOCACY ACTIONS; EURORDIS WILL TAKE ACTIVITIES IN RESEARCH POLICY TO A NEW LEVEL IN 2011 WITH SEVERAL ACTIONS: (SEE ALSO SECTION ON SHAPING RESEARCH POLICY)

- Promote a more favourable research policy framework for rare diseases: Dissemination of the first reference paper "Why Research Policy on Rare Diseases?" and drafting, consultation, dissemination of a second reference paper "Which Strategy & Priorities for Rare Diseases Research?"

- Promote the creation of a Working Group on Research in the EU Committee of Experts on Rare Diseases and play a front line role in promoting better policy

- Build capacities of its members to advocate on research policy

- Be involved in planning meetings, scientific working groups and drafting of documents on the goals, governance, organisation and policies of the new EU-US initiative of an International Rare Disease Research Consortium

## INFORMATION & NETWORKING

### COMMUNITY BUILDING

- PRIORITY RECRUITMENT OF MEMBERS IN NEW AND FUTURE EU MEMBER STATES, RARE CANCER GROUPS AND EUROPEAN FEDERATIONS

- ORGANISE EURORDIS MEMBERSHIP MEETING 2011 AMSTERDAM ON 13-14 MAY

- BUILD CAPACITIES OF THE EUROPEAN NETWORK OF NATIONAL ALLIANCES BASED ON NATIONAL PLANS, RARE DISEASE DAY, ACCESS TO MEDICINES, PLAY DECIDE GAMES

- ORGANISE TWO WORKSHOPS OF THE COUNCIL OF NATIONAL ALLIANCES

- BUILD CAPACITIES OF THE EUROPEAN NETWORK OF DISEASE-SPECIFIC EUROPEAN / INTERNATIONAL FEDERATIONS & NETWORKS THROUGH SHARING PRACTICES WITH A WIKI GUIDE AND MENTORING OF NEW EUROPEAN FEDERATIONS & NETWORKS ((RARE!TOGETHER PROJECT); DEVELOPING EUROPEAN REFERENCE NETWORKS; JOINING IN THE RARE DISEASE DAY; BUILDING CAPACITIES ON DRUG DEVELOPMENT AND ACCESS; PROMOTING PATIENT'S PREFERRED POLICY SCENARIOS AND ON LINE PATIENT COMMUNITIES

- ORGANISE ONE WORKSHOP OF THE COUNCIL OF EUROPEAN FEDERATIONS AND A ONE-DAY CAPACITY BUILDING SESSION

- INCREASE OUTREACH TO PATIENT GROUPS IN CENTRAL & EASTERN EUROPE, RUSSIA AND THE CAUCASUS:

- Give them priority in Patient Fellowship Programmes for EURORDIS Membership Meeting

2011 Amsterdam, EPPOSI Workshops , DIA EuroMeeting 2011 Geneva

- Make special efforts to involve them in our activities such as Play Decide Games available in 22 languages

- Endorse, promote and take part in national conferences across Central & Eastern Europe upon request by our members

- Support Russia with the 1st All Russian Conference on Rare Diseases & Orphan Drugs organised in Moscow organised by the Russian alliance of "Patients with Rare Diseases", the 2nd All Russian Conference on Rare Diseases & Orphan Drugs in Saint Petersburg organised by Genetics and the Conference on Rehabilitation Services organised in Saint Petersburg by GAOORDI

- Support the 2nd South Caucasian Conference on Rare Diseases & Orphan Drugs

- Possibly make EURORDIS Website, e-Newsletter and main documents available in Polish and Russian

- Country visits

- 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 Cyprus, Switzerland, Austria

## OUR ACTION PLAN FOR 2011



### EXPAND THE EURORDIS-NORD SOCIAL NETWORKS OF ON LINE PATIENT COMMUNITIES AND ADVOCATE COMMUNITIES :

- Launch 40 to 50 On Line Patient Communities in conjunction with NORD and the existing patient groups for these rare diseases
- Adapt tool to specific communities in conjunction with relevant patient groups
- Experiment methodology for operational support to create a community, recruit train and support moderators, promote a community
- Promote the service at large and in each on line 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

- Strengthen overall partnership with NORD
- Strengthen overall Corporate Partnership in Social Media and adopt a Code of Conduct
- Update, revise with moderators and adopt an upgraded version of the EURORDIS Guidelines on Good Practices on Social Media
- STRENGTHEN THE IDENTIFICATION, RECRUITMENT AND SUPPORT TO VOLUNTEERS TO BE INCREASINGLY INVOLVED IN EURORDIS ACTIVITIES; PARTICIPATING IN NGO'S PARTNERSHIPS AND REPRESENTING PATIENTS IN COMMISSIONS AND EMA WORKING GROUPS AND COMMITTEES
- SUPPORT EURORDIS' VOLUNTEERS INVOLVEMENT WITH SHORT BRIEFING MATERIAL ON KEY TOPICS



## INFORMING & RAISING AWARENESS

### ORGANISE RARE DISEASE DAY 2011 AROUND THE THEME "RARE DISEASES AND HEALTH INEQUALITIES" (RARE BUT EQUAL)

- Info Pack, poster and slogan, dedicated website, patient testimonies of health inequalities, social media networks, letters to MEPs
- Expand in more countries in Europe and at international level

### ORGANISE A ONE-DAY RARE DISEASE DAY CONFERENCE IN BRUSSELS ON MONDAY 28 FEBRUARY ON "HEALTH INEQUALITIES AND RARE DISEASES" AIMED AT PROMOTING RARE DISEASES AS A PRIORITY IN THE 3RD EU PUBLIC HEALTH PROGRAMME

- IMPROVE CONTENT QUALITY AND NAVIGATION ON EURORDIS NEW WEBSITE:
- Promote EURORDIS' new Web 2.0 Platform to continue to address all stakeholders while developing rare disease awareness amongst the general public

- Further development of content and facilitate access via three focal points on homepage: Satellite – (Rare Disease Day, Rare!Together),

Core – eurordis.org including specialised services, social media – (Online Patient Communities, Facebook, Twitter, Flickr, Rare Disease Blog),

- Within core website consolidate issues of strategic importance (e.g. EUROPLAN, CAVOD, patient's trainings),
- Stimulate the International Rare Disease Blog supported by EURORDIS-NORD through eurordis.org and social media,
- Draft specifications for technical backend maintenance of eurordis.org and rarediseaseday.org,
- Maintain quality, updated information in six languages in all sections and possibly expand to two new European languages (Polish and/or Russian).

### PUBLISH 10 ISSUES OF THE E-NEWSLETTER FOCUSING ON PRIORITIES 2011

- ORGANISE A VIDEO & PHOTO CONTEST 2011, INDEPENDENTLY FROM RARE DISEASE DAY



- IMPROVE ACCESS TO AND QUALITY OF INFORMATION THROUGH RARE DISEASE HELP LINES:

- Disseminate Policy Fact Sheets and guideline on the creation and development of national help lines and the European Network of Rare Disease Help Lines

- Put on hold the governance and membership base of the European Network of Rare Disease Help Lines

- Put on hold the shared tools: common software for the collection of data on enquiries, online data base to aggregate and analyse enquiries, annual caller profile analysis

- Put on hold the application for an EU-wide unique 116 number

## INFORMATION SERVICES TO PATIENTS

- IMPROVE ACCESS TO QUALITY INFORMATION SOURCES ON THE WEB FOR PATIENTS, FAMILIES AND HEALTHCARE PROFESSIONALS:

- Update and deepen the review of 20 web services providing essential information on rare diseases and related research, treatments, drugs, services

- Create a new search service on EURORDIS website to provide user-friendly access

- Brand this new service and scale up a communication launch in 2S2011

- Provide search-by-disease in the EURORDIS' website members listing using the EURORDIS Contact Database

## HEALTH POLICY & HEALTHCARE SERVICES PROMOTING RARE DISEASE HEALTH POLICY DEVELOPMENT

- DISSEMINATE THE REPORT OF THE 5TH EUROPEAN CONFERENCE ON RARE DISEASES IN POLAND – ECRD 2010 KRAKOW ON 13-15 MAY “FROM POLICIES TO EFFECTIVE SERVICES TO PATIENTS”

- PLAN THE 6TH EUROPEAN CONFERENCE ON RARE DISEASES IN BELGIUM – ECRD 2012 BRUSSELS. SELECT THE PROGRAMME COMMITTEE, PROMOTE THE CONFERENCE, DEVELOP THE PROGRAMME

- EMPOWER THE NATIONAL ALLIANCES FOR THEIR ACTION IN SUPPORT OF NATIONAL PLANS ON RARE DISEASES BY:

- Involving them in all aspects of EUROPLAN (methodology, recommendations, good practices, indicators, national conferences) through a working group of six volunteer advisors and support of the European Policy Advisor

- Involving them in the new EUROPLAN II project / Joint Action for period 2012-2014

- Their direct participation in the EU Committee of Experts on Rare Diseases and related Working Groups on National Policy and Indicators

- Dedicate the next two EURORDIS Membership Meetings 2011 & 2013 to capacity building on national plans and make it a central theme of ECRD 2012 Brussels

- Organise the EURORDIS Membership Meeting 2011 Amsterdam “The Patients’ Voice in National Plans” on information and 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 steering committees

- Produce Policy Fact Sheets on important policies and services expected by patients and families, derived from the Commission Communication, Council Recommendation and National Plans. Create a hard copy folder for broad dissemination to patient groups and possibly translate them into 7 languages

- Develop new ways to exchange information, experience, good measures, concerns between rare disease national alliances and in particular the patient advocates involved in the promotion or implementation of national plans – such as regular conference calls

- Promote exchange of information and sharing of experience through the newsletter and the participation to national conferences or other meetings and through the website by expanding the web sections on EU and national policies and making them more user-friendly

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

- FINALISE ALL DELIVERABLES OF THE POLKA PROJECT WITHIN BUDGET AND ON TIME BY AUGUST 2011 – FINAL REPORT DUE END OCTOBER 2011

- PROMOTE INTENSIVELY THE PLAY DECIDE GAMES IN ORDER TO STIMULATE CIVIL SOCIETY DEBATES AND CITIZENS' INFORMED OPINION ON:

- Stem cell research
- Pre-implantation genetic diagnosis
- Neonatal screening
- Cross-border healthcare
- Is there any upper limit for spending on a single patient: the case of Orphan Drugs
- Diagnosis, information to the patient and genetic counselling

- PROMOTE POLICY ON EUROPEAN REFERENCE NETWORKS & CENTRES OF EXPERTISE, EXPERT NETWORKS AND HEALTHCARE PATHWAYS:

- Disseminate the Declaration of Common Principles on Centres of Expertise & European Reference Networks

- Identification of Patients' Preferred Policy Scenarios on centres of expertise based on Play Decide Games

- Improve Good Practices Guidelines for involvement of patient organisations in Centres of Expertise & European Reference Networks, based on the Delphi method used in the POLKA project

- Progressively withdraw from direct participation in disease specific European Reference Networks (as a Partner in the nEUroped project and as an Advisor in several ERN) and collaborate with European/International Federations and Networks to take over or play an active role. EURORDIS will coordinate this participation across.

- Further develop Good Practices Guidelines for formal involvement of European/International Federations & Networks of patients in European Reference Networks based on a template Work Package for ERN projects

- Organise a one day-workshop at the Membership Meeting 2011 Amsterdam

- Create a dedicated Website section on Centres of Expertise & European Reference Networks

- PROMOTE POLICY ON NEWBORN SCREENING, GENE TESTING, PRE-IMPLANTATION DIAGNOSIS:

- Promote awareness and citizens debates on gene testing and on newborn screening through the promotion of four Play Decide Games, identify the patient preferred policy scenario

- Participate as a sub-contractor in the project "Newborn Screening" lead by ISS to collect and analyse patient-related information

- Promote the importance of post-screening follow-up and support to families after neonatal screening and diagnosis

- Organise a workshop on neonatal screening in the EURORDIS Membership Meeting 2011 Amsterdam to build the capacities of our members on newborn screening policy and stimulate feedback from patient organisations, patients and families on real life experience of services provided at Member State level

- Create a dedicated website section and disseminate information, including better promotion of information available from EuroGeneTest, European Society of Human Genetics and International Society of Neonatal Screening.

- DEVELOP FIRST ACTIONS ON QUALITY OF LIFE OF PEOPLE LIVING WITH RARE DISEASES:

- Integrate the issues on quality of life of people living with rare diseases in the Rare Disease Day 2011 Info Pack and Rare Disease Day EURORDIS event in Brussels on Health Inequalities.

- Collaborate with the EU project BURQOL to develop indicators for quality of life

- Promote EU and national policies for social research and quality of life studies

- Promote activities at EUCERD level and in the Joint Action on Rare Diseases (2012-2014) on the "Integration of people living with rare diseases into existing Social Policies and Services" such as social guidelines, information and training of social service providers to address complex needs

- Conduct preliminary research and 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 possible start in 2012 and implementation and 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 drafting of detailed common guidelines through a wiki tool
- Promote the creation of new services through the Policy Fact Sheet
- Provide updated online information resources

• **STIMULATE THE DEVELOPMENT OF AND IMPROVE ACCESS TO THERAPEUTIC RECREATION PROGRAMMES:**

- Share experiences through the dissemination of

## SUPPORT SERVICES TO PATIENTS

current guidelines and their improvement through a wiki tool

- Promote the creation of new services through an Advocacy Fact Sheet
- Provide updated online information resources
- Promote activities at EUCERD level and in the Joint Action on Rare Diseases on Specialised Social Services such as Therapeutic Recreational Programmes and Respite Care Services including adapted housing and related services

## RESEARCH DRUGS & THERAPIES

### SHAPING RESEARCH POLICY

• **PROMOTE RARE DISEASE RESEARCH AS A POLICY AND BUDGET PRIORITY AT EU AND NATIONAL LEVELS WITHIN AN INTEGRATED APPROACH BY:**

- Disseminating the Reference Paper on "Why Rare Diseases should be a Policy and Budget Priority?"
- Developing a Reference Paper with proposals on rare disease research for the 8th Framework Programme and national plans
- Policy Fact Sheets

- Organising workshop session on research in the context of the Membership Meeting 2011 Amsterdam

- Participating in the ERA-Net project E-RARE involving Member States

- Participating in policy activities and activities related to national plans

• **CONTRIBUTE TO THE DRAFTING OF THE REVISED EU DIRECTIVE ON CLINICAL TRIALS**

## PROMOTING DRUG DEVELOPMENT & ACCESS TO TREATMENTS

• **EXPAND ACTIVITIES ON DRUG DEVELOPMENT, INFORMATION AND ACCESS:**

- Participate in the EMA Committee for Orphan Drugs (COMP) with two representatives and two permanent observers

- Participate in the 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 the EMA Patients & Consumers Working Party with one representative and one alternate

- Participate in the EMA Working Group on Clinical Trials in Third Countries

- Link-in patients' expertise for Protocol Assistance, Risk Management, Crisis Communication, Pharmacovigilance and Public Information

- Support EURORDIS' patient representatives with a Therapeutic Advisory Group

- Review all orphan drug designation applications (ODD), orphan protocol assistance (PA), review of designation criteria at the time of marketing authorisation and reports on significant benefit, all paediatric investigation plans (PIP) for rare diseases, paediatric research waivers and deferrals, all gene, cell therapy and tissue engineering applications

- Review and validate all public information on rare disease therapies disseminated by EMA at the time of Designation (PSOs) and Marketing Authorisation (EPARs, Package Leaflets, Significant Benefit Public Reports)

• **CAPACITY BUILDING OF OUR MEMBERS AND VOLUNTEERS ON CLINICAL TRIALS, DRUG DEVELOPMENT, EU REGULATORY AFFAIRS AND HEALTH TECHNOLOGY ASSESSMENT:**

- Organise EURORDIS Summer School 2011 in Barcelona in June to train 30 to 50 new patient

## OUR ACTION PLAN FOR 2011

advocates (priority given to those from national alliances, European federations, Central & Eastern Europe and patient groups from targeted disease areas)

- Launch e-learning platform in English with 4 sections and 20 modules with concepts, basic knowledge, glossary, case studies, self-evaluation questionnaire. Brand the service and scale up its communication launch in 1S2011

- Take part in the DIA EuroMeeting 2011 Geneva in March with speakers, a DIA Patient Fellowship Programme for about 40 fellows and a booth stand

- Empowerment of our volunteers: Information and monthly reports; call for volunteers; further development of three Task Forces on Orphan Drugs, Paediatric Drugs and Drug Information Transparency & Access; organise one workshop for each Task Force

### • IMPROVE ACCESS TO ORPHAN DRUGS:

- Promote the Common Scientific Assessment of the Clinical Added Value of Orphan Drugs through collaboration between Member States and Common Assessment Reports;

- Promote Benefit-Risk-Effectiveness Management Plans on Orphan Drugs

- Contribute to the debates on new approaches

to Risk & Benefits Assessments, in order to take more into consideration patient values

- Promote Conditional Pricing and other innovative approaches on Pricing & Reimbursement

- Participate in the EU Stakeholders Forum on Corporate Responsibility in Pharmaceuticals for a pilot on orphan drug pricing in 2011-2012

- Promote Early Access Management Plans on Orphan Drugs

- Interact with the EUnetHTA and its Stakeholders Forum

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

### • PROMOTE THE DIALOGUE WITH PHARMACEUTICAL AND BIOTECH COMPANIES INVOLVED IN RARE DISEASE THERAPY DEVELOPMENT:

- EURORDIS Round Table of Companies: consolidate membership and organise two workshops in Barcelona in June and in Paris in November / December 2011

- Deepen direct dialogue with international big pharmaceutical companies

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

## SUPPORTING CLINICAL RESEARCH

### • PROMOTE GOOD PRACTICES FOR CLINICAL RESEARCH ON RARE DISEASES:

- Promote adoption of the EURORDIS Clinical Trials Charter by sponsors

- Support patient representatives in the implementation of the Charter, including possible establishment of a Community Advisory Board and support by a Mentor

- Collaborate with Patient Partner EU Project

- Collaborate with ECRIN Project

- Contribute to the revision of the EU Directive on Clinical Research

### • EXPLORE THE ENVIRONMENT TO PROMOTE RARE DISEASE PATIENT REPORTED OUTCOMES ON ADVERSE EFFECTS OF ORPHAN DRUGS, EXPERIENCE IN CLINICAL TRIALS AND ON OFF-LABEL USES OF MEDICINES IN RARE INDICATIONS

- MAINTAIN EURORDIS' INVOLVEMENT IN THE EPPOSI BOARD AND IN DIFFERENT THEMATIC PROGRAMMES AND WORKSHOPS SUCH AS THE RARE DISEASE THERAPY WORKSHOP, THE CHRONIC DISEASE WORKSHOP, THE WORKSHOP ON HTA AND PATIENTS' INVOLVEMENT

- PARTNER OR ADVISE IN THE EUROPEAN NETWORK OF EXCELLENCE AND INFRASTRUCTURE PROJECTS:

- Treat-NMD

- CliniGene

- ECRIN

- E-Rare

### • SUPPORT RARE DISEASE BIOLOGICAL RESOURCE BANKS AND EUROPEAN NETWORKING: MAINTENANCE OF THE EUROBIOBANK NETWORK, ONLINE SERVICES AND SHARED TOOLS; COLLABORATION WITHIN THE TREAT-NMD PROJECT; PROMOTION OF POLICY RECOMMENDATIONS AND LIAISING WITH THE 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 inter-operability

- Contribute to the analysis of the European legal framework and solutions

- Start elaboration of preferred policy scenarios

## CROSS-CUTTING PRIORITIES

- **IMPLEMENT EURORDIS STRATEGY 2010-2015:**
  - Develop 5-year Action Plans for each Operational Unit
  - Develop common Road Maps for the Council of National Alliances and the Council of European Federations to align EURORDIS 5-year Strategy based on common short-term, medium-terms and long-term objectives and measurable results
  - Anticipate planning of next ECRDs and Membership Meetings
- **DEVELOP EURORDIS STRATEGIC PARTNERSHIPS:**
  - Consolidate Strategic Partnership with AFM-Téléthon (new collaborative agreement 2010-2012)
  - Expand common actions with NORD (USA) based on Strategic Partnership Memorandum signed in 2009
  - Consolidate partnership with the members of the Round Table of Companies
  - Develop a partnership with CORD (Canada)
  - Develop a strategic partnership with ORPHANET
  - Develop a strategic partnership with the European Society of Human Genetics, European Academy of Paediatrics, International Society for Neonatal Screening, the European Society for Medical Oncology, the Standing Committee of European Doctors and other essential learned societies and academic partners
- **CONTINUE THIRD YEAR PILOT ON COLLECTION OF EURORDIS INDICATORS ON ACTIVITY AND RESULTS**
- **INCREASE EC SUPPORT TO EURORDIS' RARE DISEASE ACTIVITIES THROUGH:**
  - Operations Grant
  - Conference Grant
- Patient Driven Public Health Project in DG SanCo Work Programmes (2010-2011) as project leader or partner
- Patient Driven Health Research Project in DG Research FP 7 (2010-2011) as project leader or partner
- An IMI project in a consortium led by European Patient Forum (EPF) with other European umbrella patient organisations
- **DEVELOP AND DIVERSIFY PRIVATE FUNDING:**
  - Maintain overall giving levels from industry donors
  - Engage corporate donors and foundations, beyond the pharmaceutical industry, in supporting EURORDIS' actions
  - Implement Gala Dinner in Brussels for February 2012 "Hope and Solidarity with Rare Disease Patients in Europe"
  - Launch individual giving program in 2012
- **REVISE AND ADOPT NEW PROCEDURES IN THE FIELD OF FINANCE, HUMAN RESOURCES AND OFFICE SUPPORT SERVICES**
- **ADOPT A "RÈGLEMENT INTERIEUR" (INTERNAL REGULATIONS) FOR HUMAN RESOURCES**
- **MAKE EURORDIS CONTACT DATABASE MANAGEMENT FULLY OPERATIONAL**
- **ENHANCE IT SUPPORT: EQUIPMENT, SERVICES, INTRANET SHARING, VIRTUAL OFFICE OPEN TO VOLUNTEERS**

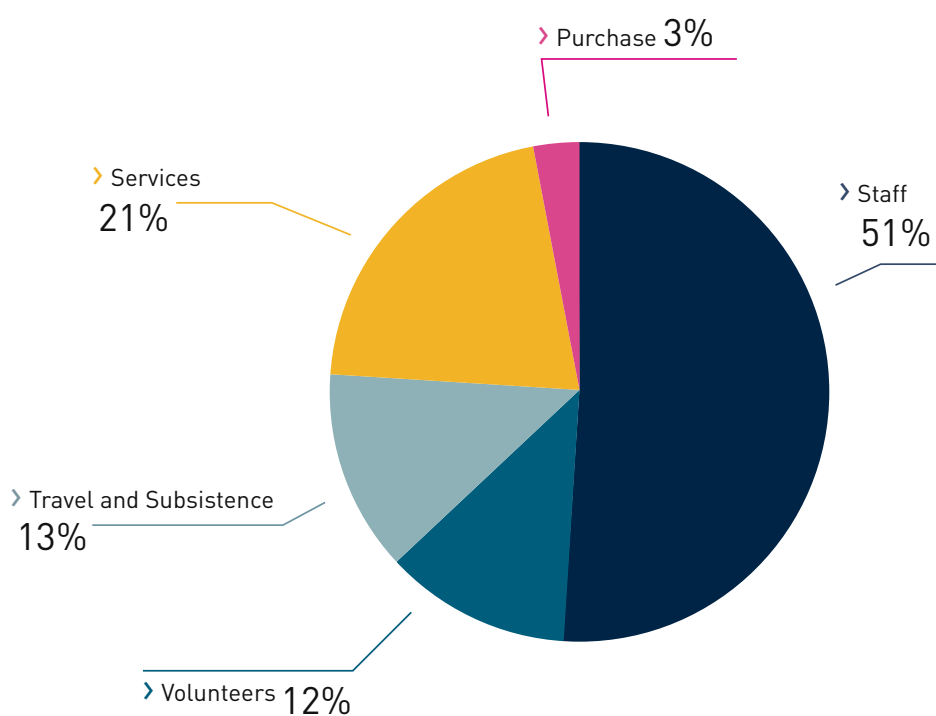




# BUDGET

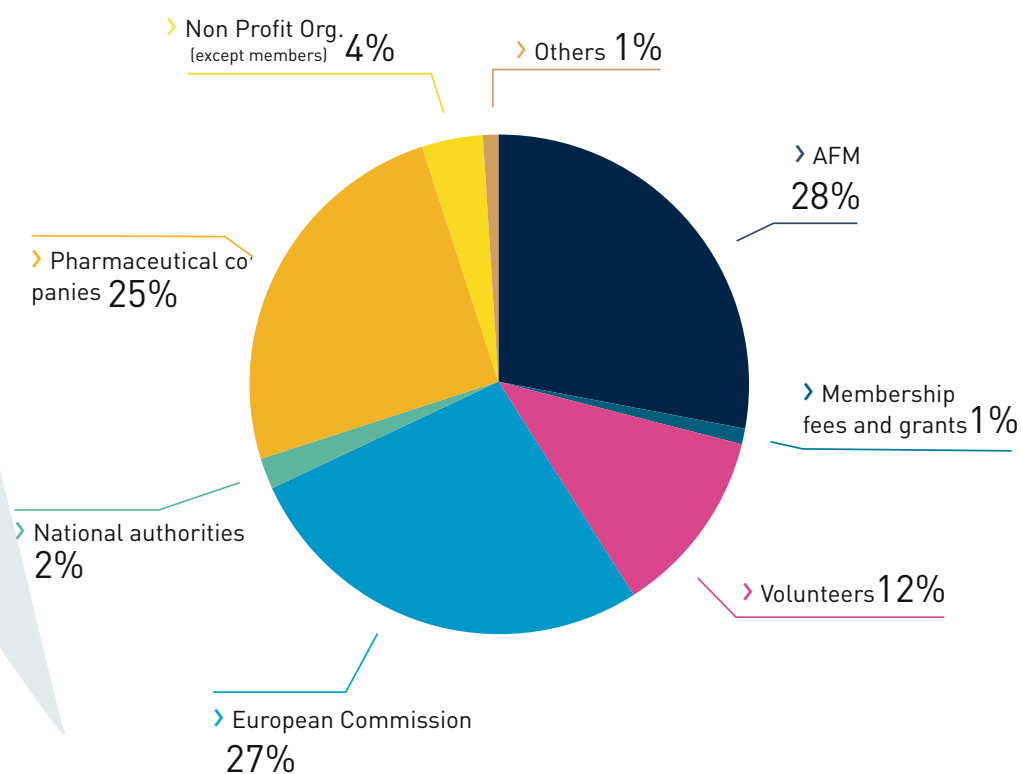
2011 (in thousand of euros, provisions excluded)

› EXPENSES BY TYPE 2011 = 3,145 k€



■ Staff	1606
■ Volunteers	376
■ Travel and Subsisten	422
■ Services	672
■ Purchase	69

› REVENUE BY ORIGIN 2011 = 3,139 k€

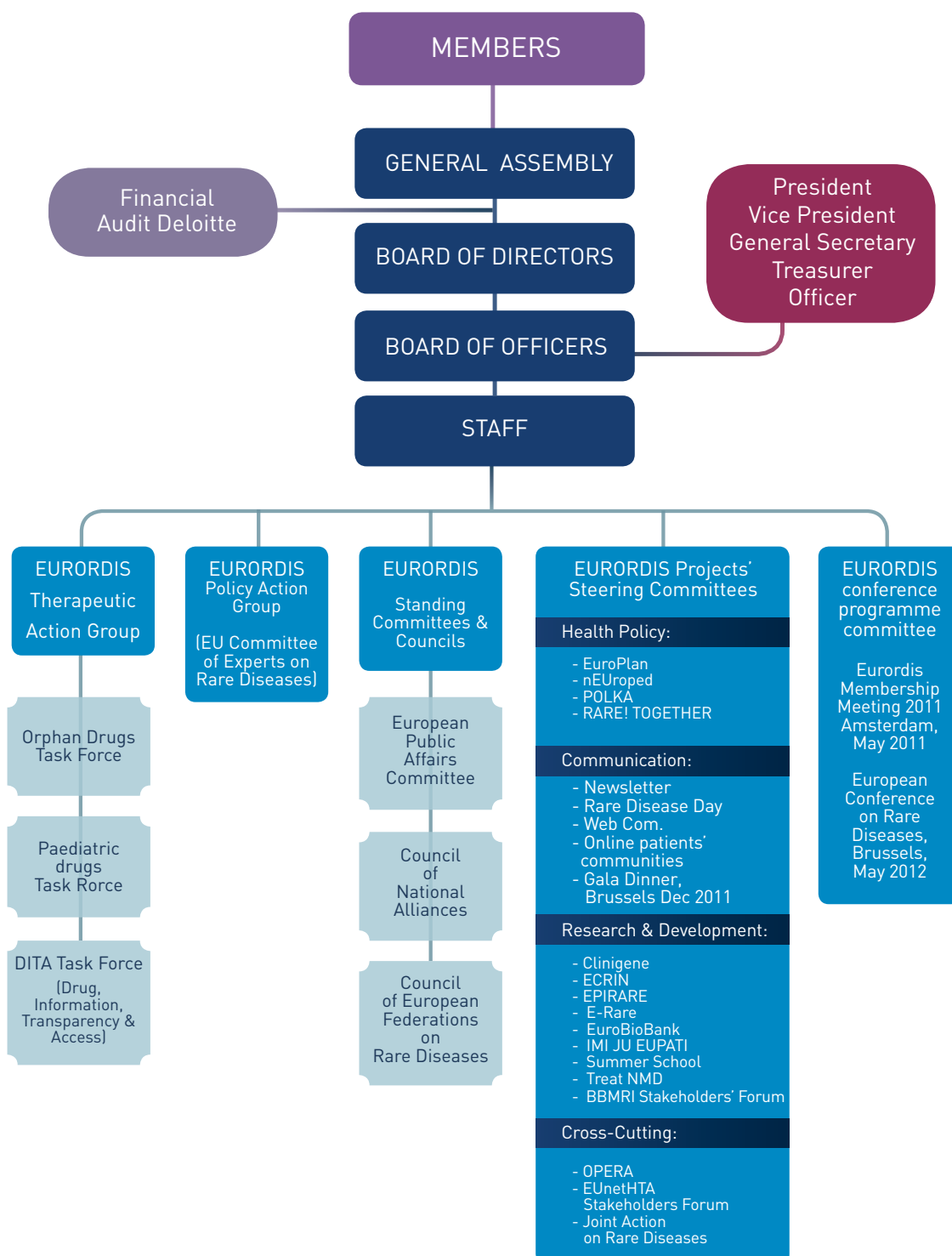


■ AFM	864
■ Membership fees and	35
■ Volunteers	376
■ European commission	864
■ National authorities	71
■ Pharmaceutical compa	777
■ Non profit Org. (ece	135
■ Others	17

# EURORDIS GOVERNANCE CHART 2011



**EURORDIS**  
Rare Diseases Europe

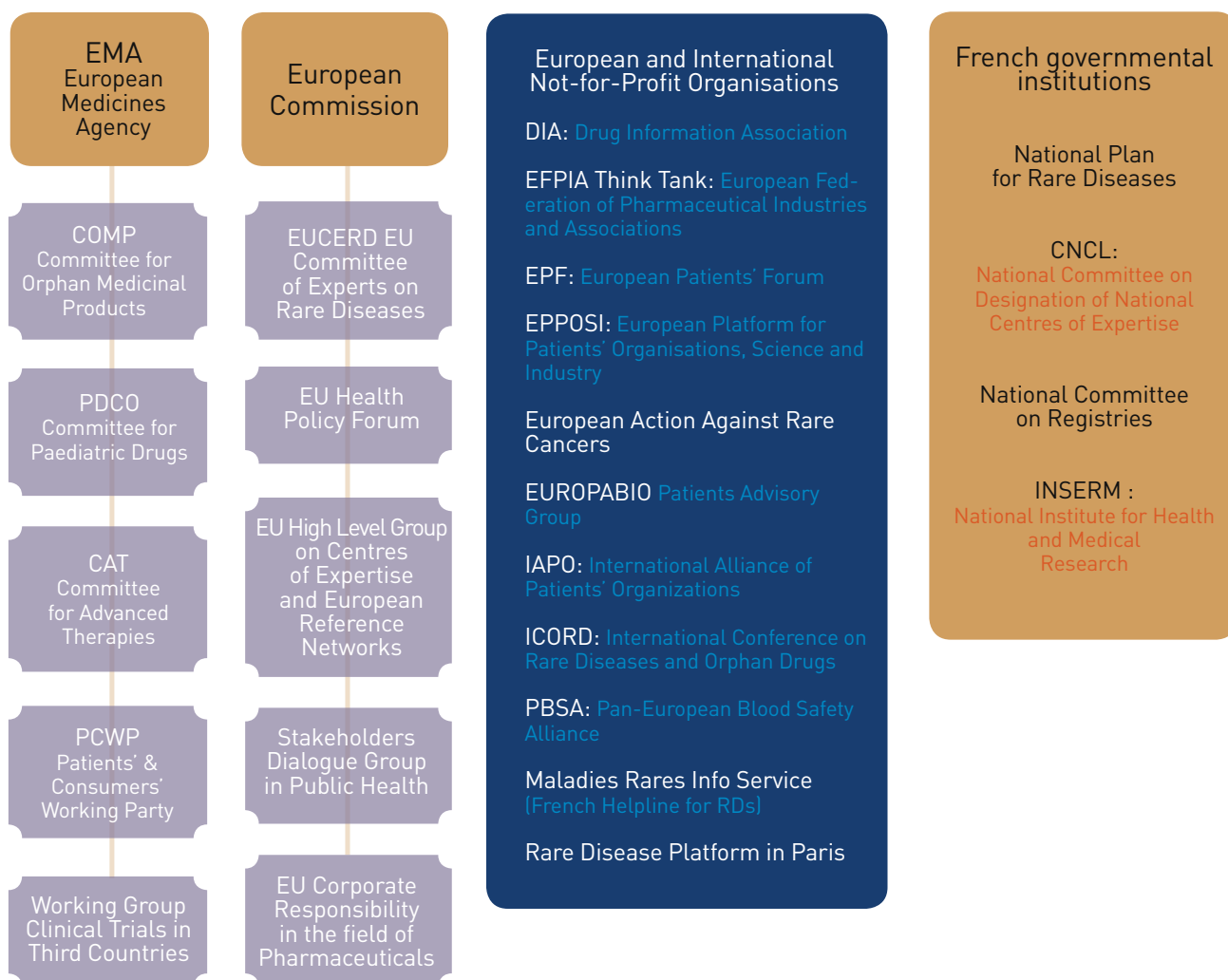


# EURODIS' REPRESENTATION IN EXTERNAL INSTITUTIONS AND ORGANISATIONS IN 2011



→ Governmental Institutions

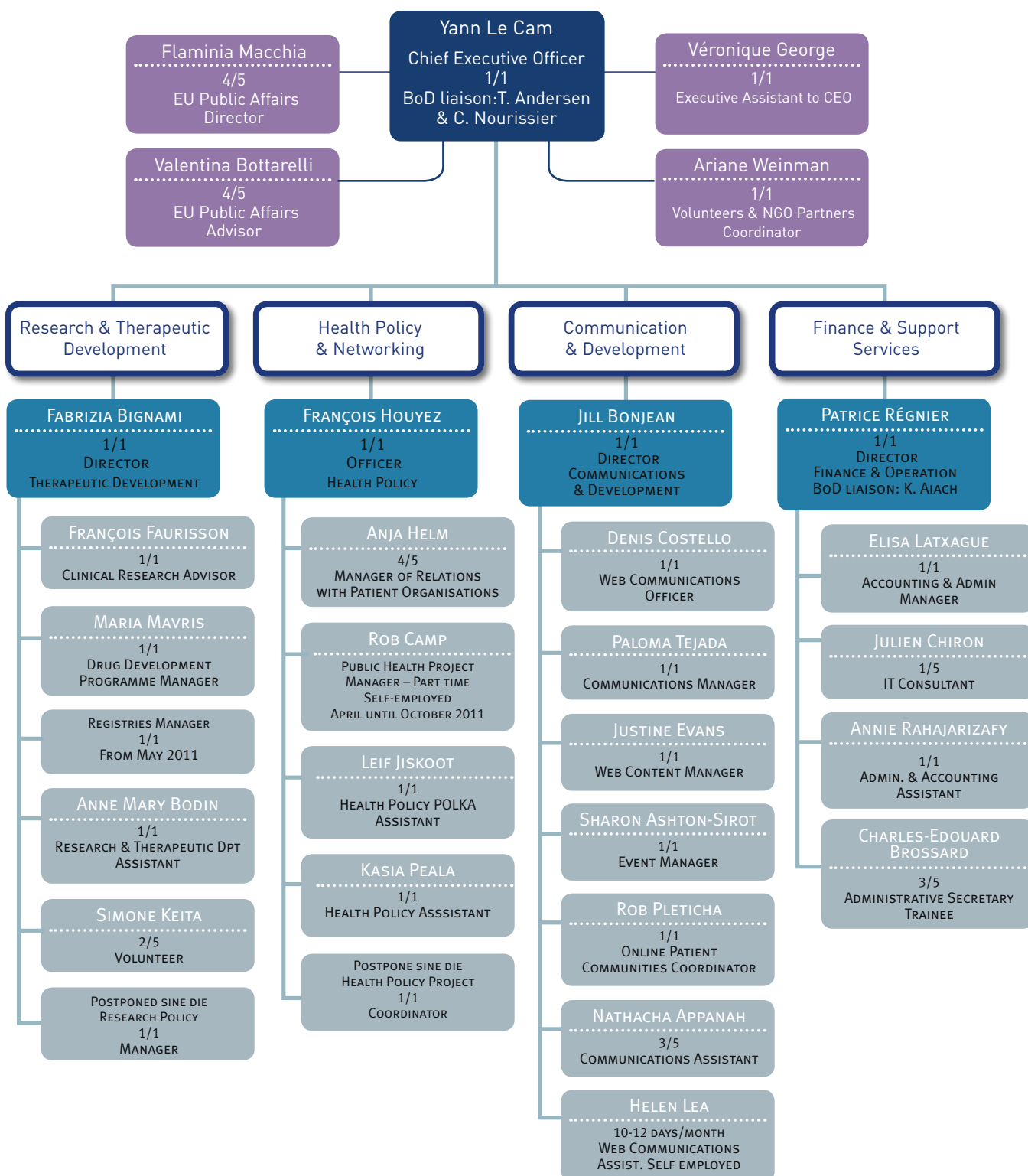
→ Non-Governmental Organisations



# EURORDIS TEAM CHART 2011



**EURORDIS**  
Rare Diseases Europe







**EURORDIS**  
Rare Diseases Europe

Comité éditorial  
Eurordis

Conception et réalisation

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