



EURORDIS
Rare Diseases Europe

ACTIVITY REPORT

2015

& WORKPLAN 2016

European Organisation
for Rare Diseases

The Voice of Rare Disease
Patients in Europe

eurordis.org





TABLE OF CONTENTS

02	FOREWORD	
04	EURORDIS IN BRIEF	
05	STRATEGIC APPROACH 2015–2020	
08	ACTIVITY REPORT 2015	
	HIGHLIGHTS 2015	09
	ADVOCACY	10
	INFORMATION & NETWORKING	24
	HEALTH & SOCIAL POLICY	34
	RESEARCH, DRUGS & THERAPIES	36
	GOVERNANCE, ORGANISATION & FUNDING	45
	REVENUES & EXPENSES	48
	BOARD OF DIRECTORS	50
	MEMBERS	51
	CONFERENCES	57
	ACKNOWLEDGEMENTS	61
65	WORKPLAN 2016	
	ACTION PLAN 2016	66
	GOVERNANCE CHART 2016	79
	TEAM CHART 2016	81
	BUDGET 2016	82
84	ACRONYMS & DEFINITIONS	

FOREWORD

BY THE PRESIDENT AND THE CHIEF EXECUTIVE OFFICER

EURORDIS has reached 705 members in 63 countries (26 EU countries)*, including **32 national alliances in Europe and 52 European federations**. In 2015 alone, 65 new members joined EURORDIS. A membership re-assessment process to examine whether members still fulfil our strict criteria is also now in place. These progressions further enhance EURORDIS' mission to build a strong pan-European community of patient organisations and the 30 million people living with a rare disease in Europe, in order to be their voice at the European level and to fight against the impact of rare diseases on their lives.

The **EURORDIS Membership Meeting (EMM 2015 Madrid)** was held in Madrid on 28–30 May 2015. The event attracted 250 participants and included 4 capacity-building workshops.

The **European Network of National Alliances** was further strengthened with new national rare disease alliances created in Serbia and Macedonia. Two meetings of the Council of National Alliances were held, one in Madrid in May 2015 and a second one in

Paris in October, back to back with the Council for European Federations, which was dedicated to the launch of European Reference Networks for Rare Diseases.

2015 also saw the official launch of **Rare Diseases International** with over 80 participants at the inaugural meeting adopting the "Joint Declaration: Rare Diseases as an International Public Health Priority" and promoting EURORDIS' initiatives to provide support to rare disease patient groups at an international level.

The **EURORDIS website**, with comprehensive information in **7 languages**, was developed further in 2015. The EURORDIS website has over **300,000 real visits per year** and is continually updated. It hosts the annual EURORDIS Photo Contest, which has seen an increase in its success rate with **376 photos received from 42 countries**.

Rare Disease Day continues to grow internationally with **over 80 countries participating in 2015**. The media outreach was strong, with hundreds of news articles collected and over 1000 events organised all over the world. The **new RDD website** developed in 2014 was updated in 2015 and had over 150,000 visits in January and Febru-

ary alone. The **Rare Disease Day video** was the most successful to date with 1.5 million views on EURORDIS social media and was translated into 27 languages. **Rare Disease Day has become a truly international event and an essential force to raise public awareness and promote national rare disease policies**.

The EURORDIS online patients' community network **RareConnect** experienced further expansion in 2015. By the end of 2015, 83 disease-specific online communities had been successfully created from international partnerships with **691 different patient organisations and the engagement of 276 volunteer moderators**. RareConnect was rebranded in 2015, with a new logo, visual identity and new features such as the RareConnect discussion groups. Partnerships between RareConnect and national alliances was further explored, illustrated by the addition of new RareConnect staff members hosted by the national alliances of Croatia and Serbia.

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

In 2015, we also accelerated activities in the field of **rare cancers**, namely through the **EURORDIS Policy Action Group (PAG) on Rare Cancers**, which includes the 2 patient representatives on the **Commission Expert Group on Cancer Control**. The PAG on Rare Cancers prepared a paper on the commonalities and differences between rare diseases and rare cancers for the purpose of advocating for synergy in policies, and prepared for the Joint Action on Rare Cancers.

One top advocacy priority for EURORDIS in 2015–2016 is to position the perspective of patients at the heart of the development of European Reference Networks (ERNs). Throughout 2015, EURORDIS continued paving the way for the next steps in ERN implementation to take place in 2016. Support of the implementation of the EU policy on European Reference Networks, the optimal uptake of this policy for rare diseases, **the orientation of ERNs toward improving patients' health outcomes**, and the



Yann Le Cam / Chief Executive Officer

*Correct at time of print

preparedness of our members and stakeholders to be involved in ERN development were all operational priorities. In addition, **EURORDIS undertakes a deep running structuration of its membership around the ERN grouping of rare diseases through the formation of European Patient Advocacy Groups (ePAGs).**

Patients' access to orphan medicines across Europe, which has deteriorated due to the economic crisis and resulting impact on healthcare budgets, is another top advocacy priority. In 2015, EURORDIS sustained its advocacy work in favour of European-level initiatives aimed at improving access for patients, including: Medicines Adaptive Pathways to Patients, the Mechanism of Co-ordinated Access to Orphan Medicinal Products (MOCA), European parallel scientific advice by EMA and HTA agencies and future common HTA assessment reports. **Earlier, wider, equitable, sustainable patient access to medicines continued to be a top advocacy priority for 2015–2016.** EURORDIS will continue to **promote European collaboration between Member States on pricing and structured market access** that is based on value, volume and post-marketing authorisation data generation, also exploring managed entry agreement, fair pricing, differential pricing and pricing based on outcomes.

EURORDIS continued to strengthen training for patient advocates by developing the EURORDIS Summer School and online training resources. For the first time, the EURORDIS Summer School welcomed researchers as participants, as well as patient advocates. The Summer School is a 5-day capacity-building programme for patient advocates in the areas of medicines development, clinical trials and aspects of the EU regulatory process. 43 participants attended representing 20 countries and 25 diseases.

In 2015, **EURORDIS continued its active support of the participation of patient representatives in the committees and working parties of the European Medicines Agency (COMP, PDCO, CAT, PCWP, SAWP, CHMP),** culminating in **293 days of meetings and 541 scientific dossiers** examined for scientific advice/protocol assistance, paediatric studies and overall clinical development up to marketing authorisation. EURORDIS is promoting the concept of patient engagement all along the life cycle of a product and has created the role of Patient Engagement Manager within the team.

In 2015, **EURORDIS continued to grow its activities in health technology assessment (HTA).** EURORDIS is one of four patient representative organisations at the EUnetHTA Stakeholders Forum and is contributing to a vision for the future of HTA in EU as co-chair of the EUnetHTA Stakeholder Forum. EURORDIS is promoting adaptiveness to rarity as well as the involvement of patients in HTA procedures and is coordinating the identification and involvement of patients in the SEED project.

EURORDIS continued to grow its activities, volunteer base, budget and staff to take on the advocacy challenges and expand its programmes to members and patients. We believe **our organic growth reflects the positive resonance that our actions have within the rare disease community.** However, **funding of our core activities remains a critical issue when it comes to sustaining our development. EURORDIS will continue to diversify and consolidate its resources in 2016.**



Terkel Andersen / President

In 2015, EURORDIS was proud to rely on **366 volunteers**, including **88 volunteer patient advocates, 2 office support volunteers and 276 volunteer RareConnect moderators.** EURORDIS volunteers have a unique insight into the complexity of different rare diseases across Europe and reinforce EURORDIS as a grassroots movement.

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



EURORDIS IN BRIEF

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

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

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

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

KEY FIGURES 2015



EURORDIS' MISSION

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





STRATEGIC APPROACH 2015–2020

The EURORDIS Strategy 2015–2020 was presented at the 2015 Annual General Assembly in Madrid. EURORDIS Members mandated the EURORDIS Board of Directors to approve the final Strategy which was adopted in November 2015.



EURORDIS in 2020 has consolidated its position as the organisation of reference for rare diseases both in EU and in Europe for its legitimate membership base and its credible European patient voice:

- EURORDIS has created a global patient voice for rare diseases to promote the cause as an international public health challenge and is recognised as an actor in international processes that have an impact on patients living with rare diseases;
- National Alliances, European Federations, EURORDIS and Rare Diseases International have aligned a structured strategic approach based on Common Goals;
- EURORDIS enables acting at national, European, international levels, partnering with all stakeholders, and in all strategic areas of public health, healthcare, research, social, human and patient rights, so to have a patient-centric 360° view;
- EURORDIS is combining unity and diversity; EURORDIS has structured its membership base in European Patient

Advocacy Groups per rare disease groupings based on common goals and democratic processes so to enable patient engagement in areas of strong common interest such as European Reference Networks, Registries & Data Collection, European Research projects, R&D and Assessment of therapies, Disease Management and Good Diagnostic & Care Practices, Screening & Genetic testing and associated ethical issues, social services;

- European Patient Advocacy Groups per rare disease grouping are empowering our members while being inclusive of and more supportive of the rarest diseases;
- EURORDIS' European Patient Advocacy Groups per policy area are enabling greater engagement of our members and partnering with relevant stakeholders.



EURORDIS in 2020 is facilitating the effective implementation of European legislations (regulations such as those on orphan medicines, paediatric use of medicines, advanced therapies, transparency; directives such as Patient's Right to Cross Border Health Care, Clinical Trials, Data Protection) and policy strategies (e.g. Commission Communication & Council Recommendation on Action in Rare Diseases, Commission Communication on Orphan Medicines, Communication on Cancer Control) at European and national levels (e.g. National Plans on Rare Diseases) in more policy areas – research, public health, healthcare, social, digital, rights – for the benefit of patients and families:

- EURORDIS is encouraging, supporting and taking legal action when needed in order to defend patients' rights;
- EURORDIS is promoting a better regulatory and policy environment for PLWRDs to sustain rare diseases as a policy priority; to push forward access to diagnostic, treatments, care, cross-border care; to prevent genetic discrimination and promote patients' rights;
- EURORDIS is producing more patient-generated knowledge through the EURORDIS Rare Barometer Programme and promoting patient-centered policy;
- EURORDIS has developed a foresight vision to address rare diseases in the next decade, toward 2030.



EURORDIS in 2020 is empowering its member patient organisations and volunteers through more and enriched information, education and capacity building, all working to reinforce their autonomy:

- EURORDIS is also empowering the existing processes by enabling PLWRDs to be represented and rare disease patient advocates to be engaged in a larger number of innovative research & development, assessments, decision-making bodies, scientific opinion-making committees and projects relevant to fulfil its mission;
- Furthermore, EURORDIS is empowering rare disease patient advocates and all stakeholders in the rare disease community in the interest of PLWRDs;
- EURORDIS is providing a platform enabling direct matchmaking, networking, sharing, collaborative learning and collaborative design of innovative strategies;
- EURORDIS in 2020 is developing direct services to PLWRDs for their high value to our members and to patients & families;
- EURORDIS has developed RareConnect as a strong global social network of online communities of PLWRDs; RareConnect is developed in partnership with patient organisations and stakeholders; RareConnect is an agile platform offering multilingual, multifunction services enabling support, empowerment, co-production of knowledge;
- EURORDIS has catalysed a comprehensive information system of web-based service & back office for national helpline services to improve access to existing sources of quality information;
- EURORDIS has developed services to facilitate actual patients' rights and real access to cross-border healthcare;
- EURORDIS in 2020 is raising public awareness & societal support to the cause of rare diseases, mostly through its members within communication framework created by EURORDIS eg Rare Disease Day and European Year on Rare Diseases; EURORDIS has reached out to PLWRDs in EU if not in Europe and is recognised by them; EURORDIS' members and PLWRDs are engaged with EURORDIS in some key advocacy & citizen actions; PLWRDs are increasingly supporting EURORDIS as individual donors.



EURORDIS in 2020 is more sustainable in terms of governance and of human, financial and organisational resources; EURORDIS' resources have grown through a diversification of public and private funding (corporate, foundations, events, donors, fee-based services); EURORDIS has reinforced its volunteer base and long-term leadership capacities; EURORDIS has consolidated its multi-cultural multi-skilled staff and established a human resource management; EURORDIS is innovating advanced quality governance:

- EURORDIS is becoming a movement, its organisation is multi-centric, flexible, responsive, web-based;
- EURORDIS is working through partnerships, alliances and consortiums.

COMMUNITY STRUCTURE OVERVIEW

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

STRATEGIC PARTNERSHIPS

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



ACTIVITY REPORT ²⁰¹⁵

HIGHLIGHTS 2015	09
ADVOCACY	10
INFORMATION & NETWORKING	24
HEALTH & SOCIAL POLICY	34
RESEARCH, DRUGS & THERAPIES	36
GOVERNANCE, ORGANISATION & FUNDING	45
REVENUES & EXPENSES	48
BOARD OF DIRECTORS	50
MEMBERS	51
CONFERENCES	57
ACKNOWLEDGEMENTS	61

HIGHLIGHTS 2015

- | EURORDIS has **695 member patient organisations**, located throughout 63 different countries, including 526 full members in 26 EU countries.
- | The **EURORDIS Membership Meeting 2015 Madrid** attracted **250 participants from 43 countries** including 27 patient representatives that received fellowships from EURORDIS.
- | **Rare Disease Day 2015** was organised in over 80 countries worldwide, with as many partners and over 1000 related events taking place. The Rare Disease Day video was translated into 27 languages and gained over 1.5 million views on EURORDIS social media only, our most successful video to date.
- | **RareConnect** expanded to **83 global communities** in collaboration with 680 patient organisations and 280 volunteer moderators. RareConnect opened up the possibility for any person living with a rare disease to initiate a discussion group. Since launching this new feature in September 2015, RareConnect now hosts **over 200 discussion groups** covering both specific rare diseases as well as topics which go across diseases, for example, access to medicines.
- | **Rare Diseases International** was officially launched during an inaugural meeting in Madrid in May with over 80 participants. The Joint Declaration: Rare Diseases as an International Public Health Priority was also adopted in 2015. This new initiative is supported by EURORDIS to help develop a legitimate network and strong rare disease voice at an international level.
- | The **Rare Barometer Programme** was created in 2015 as a permanent programme of quantitative and qualitative studies to collect patient experiences and expectations. With this programme EURORDIS wants to enhance its advocacy capacity and promote **patient–perspective-based policy**.
- | EURORDIS actively participated in the **Commission Expert Group on Rare Diseases** and the **Commission Expert Group on Cancer Control**. The major achievement of the Commission Expert Group on Rare Diseases was the adoption of the Addendum to the EUCERD Recommendations of January 2013 on European Reference Networks, which proposes a grouping of all rare diseases into 21 thematic networks in order to ultimately ensure that all rare diseases will be covered by a future European Reference Network.
- | In 2015, the rare disease community, saw the **first steps in implementation of European Reference Networks (ERN)**, following the development and establishment of the ERN legislative framework. EURORDIS continued to promote clinical excellence and improvement of patient health outcomes, while enabling patients to be central to the implementation of ERNs.
- | EURORDIS created the consortium **Partnership for the Assessment of Clinical Excellence in European Reference Networks (PACE-ERN)** together with the European Hospital and Healthcare Federation (HOPE) and Accreditation Canada International, which was contracted in 2015 by the European Commission to develop the **ERN Manual and Tools for applicants and evaluators in preparation** for the 2016 call.
- | EURORDIS was nominated as an official partner in the **EU Joint Action on Rare Cancers** and took part in the ongoing negotiations. The Joint Action is expected to start in 2016. EURORDIS continues to be an active partner in the **EU Joint Action on Rare Diseases RD-ACTION**.
- | The **8th EURORDIS Summer School** for patient advocates was held in Barcelona with a new format that combines training for both expert patients and researchers on medicines development. **43 participants attended representing 20 countries and 25 diseases**. A new partnership with COST financed the participation of 11 researchers.
- | **541 dossiers on orphan drugs, advanced therapies and paediatric investigation plans** were reviewed as part of EURORDIS' participation in the **European Medicine Agency's Scientific Committees**. These include: 270 dossiers for orphan drug designations, 61 classifications or certifications by the CAT and 210 Paediatric Investigation Plans by the PDCO.
- | EURORDIS continued to grow its activities in **health technology assessment (HTA)** as one of four patient representative organisations in the EUnetHTA Stakeholders Forum and by contributing to the future of HTA in EU as co-chair of the EUnetHTA Stakeholder Forum.
- | EURORDIS was proud to rely on **366 volunteers** in 2015, including 88 volunteer patient advocates, 2 office support volunteers and 276 volunteer RareConnect moderators, from a wide variety of diseases and countries, and all adhering to the EURORDIS Charter for Volunteers.



ADVOCACY

Sustaining Rare Diseases as an EU Public Health Priority

COMMISSION EXPERT GROUP ON RARE DISEASES

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

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

- EURORDIS
- the European Network of National Alliances for Rare Diseases
- the Network of European Federations of Rare Diseases
- EGAN (the Patients Network for Medical Research and Health)

Out of the current eight representatives, four members were already on the EUCERD.

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

Three meetings were held at the European Commission in Luxembourg throughout 2015. The work of the Commission Expert Group on Rare Diseases builds on the achievements of the EUCERD (2010–2013). Since 2010, key recommendations on major issues for people living with rare diseases have been adopted by consensus by all the members (representatives of EU Member States, EEA countries, academia, industry and patient organisations):

- Quality Criteria for Centres of Expertise for Rare Diseases in Member States (2011)
- European Reference Networks for Rare Diseases (2013) & Addendum (2015, related to grouping of rare diseases and patient involvement)
- Cross-border genetic testing of rare diseases in the European Union (2015)
- Rare disease patient registration and data collection (2013)
- Ways to Improve Codification for Rare Diseases in Health Information Systems (2014)
- Improving Informed Decisions Based on the Clinical Added Value of Orphan Medicinal Products (CAVOMP) Information Flow (2012)
- Core indicators for rare disease national plans and strategies (2013)

In 2015, discussions took place on the future recommendation on the integration of rare diseases into mainstream social services and policies, due for adoption in 2016.

The EUCERD Joint Action working for rare diseases has supported the work of both the EUCERD and the Expert Group from March 2012 until November 2015.

The major achievement in 2015 of the Commission Expert Group was the adoption of an addendum (10 June) to the EUCERD Recommendations of January 2013 on European Reference Networks which focuses on two strategic transformative approaches: the grouping of rare diseases and the involvement of patients. The recommendation proposes a grouping of all rare diseases into 21 thematic networks in order to ultimately ensure that all rare diseases will be covered by a future European Reference Network (ERN).

In addition, it emphasises the key role that patients and their representatives should play in the decision and opinion-making process in rare disease ERNs and their necessary meaningful involvement in structural and clinical network activities.

EURORDIS spearheaded the two approaches and played an active role in the various discussions to voice the rare disease patients' perspective. The stakeholders involved in the EUCERD Joint Action prepared the groundwork for this recommendation to be discussed by the members of the Expert Group on Rare Diseases. Over the past three years, several workshops and discussions took place on the development and shaping of future ERNs for rare diseases.

All reports and recommendations produced by the European Commission Expert Group on Rare Diseases in 2015 are available on the website: http://ec.europa.eu/health/rare_diseases/expert_group/index_en.htm

COMMISSION EXPERT GROUP ON CANCER CONTROL

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

Two volunteer representatives of EURORDIS have been nominated to represent patients affected by rare cancers in the Commission Expert Group on Cancer Control. One of them also sits on the European Commission Expert Group on Rare Diseases. EURORDIS considers that it is important to create a bridge between the two groups as several health policy issues related to rare cancers are common to these two groups: national rare disease /

cancer plans, registries, European Reference Networks, quality criteria for Centres of Excellence, research policy, creating evidence and data in small populations, research policy and regulation, genetic testing and counselling, etc.

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

Two meetings of the Commission Expert Group on Cancer Control took place on 2–4 March and 15–16 September 2015. The major topics discussed in 2015 comprised:

- Ongoing European Joint Action CANCON (European Guide on Quality Improvement in Comprehensive Cancer Control)
- National Cancer Plans
- Disseminating the 4th European Code Against Cancer
- Rare Cancers
- Research
- Cancer information systems, registries
- Palliative care

RARE CANCERS: MAPPING OUT THE SIMILARITIES AND DIFFERENCES BETWEEN RARE CANCERS AND RARE DISEASES 2015–2016

EURORDIS' mission is to be the voice of all rare disease patients, including patients affected by rare cancers. As of end of 2015, EURORDIS counted amongst its members 63 rare cancer patient organisations and rare disease patient organisations concerned by cancers.

On 21 November 2015, the EURORDIS Board approved the document "Mapping out the similarities and differences between rare cancers and rare diseases 2015–2016", which has been published in the EURORDIS member news and in Orphanews (27 February 2016).



Rationale: Patients (and those who care for them) who are affected by rare diseases, rare cancers, and also by rare diseases which may give rise to cancers, have expressed their need to map out the similarities and differences that exist between rare diseases and rare cancers on main issues from the concept of rarity to research, registries, access to specialised medical centres and doctors for timely diagnosis, adequate treatments, clinical trials, access to psychosocial aid.

This document serves to better identify what priority advocacy actions need to be carried out together and where to join forces in order to improve access to equal and timely diagnosis as well as appropriate care, information and support.

This mapping, initiated in November 2014, has been drafted by the two EURORDIS volunteer patient advocates on the Commission Expert Group on Cancer Control – Jan Geissler and Kathy Oliver – with the support of the

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

The third EU health programme is implemented by means of annual work plans which set out priority areas for funding. EURORDIS worked throughout 2015 in view of the adoption of the Annual Work Programme 2016 (due in early 2016) to advocate for funding in key areas for rare disease patients, notably those under Objective 4 – “Facilitating access to better and safer healthcare for Union citizens”.

These areas include notably European Reference Networks (ERNs) and patient registres. In view of the launch of the 2016 call for establishing ERNs, EURORDIS advocated to secure financial support for the functioning of ERNs and for patient registries that will need to be enshrined in ERNs-to-be and connected with the European Platform for RD registration that the EC is setting up.

THE EU RESEARCH AND INNOVATION FRAMEWORK PROGRAMME 2014–2020: “HORIZON 2020”

In late 2013, the new programme for research and innovation of the EU, covering the period 2014–2020, was adopted, along with the first Work Programmes for 2014–2015. The topics of the Work Programme 2014–2015 for Health, Demographic Change and Wellbeing are characterised by an overarching, non-disease specific approach. However, a special focus on rare diseases is maintained, in line with IRDiRC priorities and the regulation establishing Horizon 2020.

Further to the two topics launched in 2014, two additional rare disease specific topics were announced and published at the end of 2015 (although the call will not open before late July 2016):

- “New therapies for rare diseases”, that followed the extremely popular call (421 applications) launched in 2014, that highlighted a widespread interest in research for new rare disease therapies and the lack of sufficient EC funding (only 10 out of more than 400 applications could be funded).
- “Diagnostic characterisation of rare diseases”, a topic that aims to develop research on molecular diagnoses for a large number of undiagnosed rare diseases.

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

In early 2015, EURORDIS submitted its contribution to the European Commission public consultations on the priorities for the next Work Programme for Health, Demographic Change and Wellbeing 2016–2017, as well as for the Work Programme for Science in and for Society 2016–2017. EURORDIS also shared rare disease research priorities with patient representatives at the Horizon 2020 Advisory Group for Health, assisting the European Commission in the development of Horizon 2020 Work Programmes.



volunteers on the Policy Action Group on Rare Cancers, including Catherine Vergely and Frédéric Arnold for paediatric cancers, and with Ariane Weinman.

The co-chairs of Rare Cancers Europe were consulted for advice: Prof Paolo Casali, European Society of Medical Oncology (ESMO); Markus Wartenberg, Sarcoma Patients EuroNet (SPAEN); and Susanna Leto, Novartis.

The member organisations of EURORDIS concerned by rare cancers were consulted in Spring 2015 and provided their inputs.

EUROPEAN JOINT ACTION ON RARE CANCERS

EURORDIS has been nominated as an official partner in this EU Joint Action on Rare Cancers (JARC) led by the Fondazione IRCCS Istituto Nazionale dei Tumori, Milan. EURORDIS participated in the ongoing negotiations. The start of the JARC is expected for Q3 or Q4 2016.

THE EU HEALTH PROGRAMME 2014–2020: “HEALTH FOR GROWTH”

On 26 February 2014, the European Parliament adopted the Commission proposal for the establishment of the third Health Programme 2014–2020: “Health for Growth”. The EU health programme is the main instrument the European Commission uses to implement EU health strategy.

RARE DISEASE DAY EURORDIS POLICY EVENT

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

As with previous years, EURORDIS hosted a policy event in Brussels on the occasion of Rare Disease Day. The event was entitled: **"Rare but Real: Talking Rare Diseases"** with attendance from multiple representatives such as the Commissioner of Health and Food Safety, Vytenis Andriukaitis. The event included inspiring testimonies given

by patients and family members and for the first time enabled participants to actively take part in the event online. There were 150 attendees on site and participants from 30 different countries tuned in to event online. Over 800 tweets were exchanged during the event using the hashtag #RareEU2015. The patients' organisations are playing an instrumental role in shaping rare disease policy and advocating for patients' rights at national and European levels, an effort that often generates results in the long term. To date, 19 EU Member States have a rare disease national plan/strategy as compared to only 4 in 2008.



Rare Diseases: An International Public Health Priority

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RARE DISEASES INTERNATIONAL

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

Throughout 2014, contact was made with a number of prominent international rare disease organisations in order to follow up on their interest in creating the Rare Diseases International network. RDI was officially launched at its first annual meeting in May 2015 in Madrid. The RDI Preformation Group (PFG), an interim governance structure of RDI made up of 10 international patient organisations, continued to pilot the initiative

throughout the year, approving membership applications and developing key documents of the initiative, including the process for the first official elections of the Council of RDI (permanent governing body replacing the PFG), which will take place at the second RDI meeting in Edinburgh in May 2016.



**RARE
DISEASES
INTERNATIONAL**

A EURORDIS INITIATIVE

OFFICIAL LAUNCH OF RARE DISEASES INTERNATIONAL – 28 MAY 2015

The first annual meeting of Rare Diseases International took place on May 28, 2015 in Madrid, Spain, alongside the EURORDIS Membership Meeting. Over 60 patient representatives from 30 countries gathered for the official launch and inaugural meeting and to adopt the principles of a Joint Declaration aimed at advocating for rare diseases to be an international public health priority.

The **Joint Declaration: Rare Diseases as an International Public Health Priority**, was approved in principle by all organisations present at the first annual meeting in Madrid in May 2015. The Joint Declaration has been a work-in-progress for several years, was updated and finalised using information received through an international survey.

“NORD is pleased to join Rare Diseases International and to collaborate with leading patient advocacy groups from around the world to help make rare diseases an important global public health priority.”

Peter Saltonstall, President and CEO NORD



INTERNATIONAL PARTNERSHIPS WITH PATIENT ORGANISATIONS

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

The MoUs are part of the ongoing effort to stress the international dimension of the rare disease movement

and the global benefits to be gained by collaboration and enhanced cooperation with partners. The MoUs specify the common objective to strengthen the united international voice of rare disease patients and promotes a global approach through participation in the annual awareness-raising event Rare Disease Day, international patient forum RareConnect, Rare Diseases International, and other possible joint actions such as common position papers and collaboration in diverse areas including research, diagnostics, information, treatment, and quality of life.

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

EURORDIS–NORD STRATEGIC PARTNERSHIP

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



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

The two organisations continued to play a key role in promoting rare diseases as a public health priority in 2015 with notable areas of collaboration being: Rare Disease Day 2015 (NORD is EURORDIS' official partner for RDD in the USA); being actively involved in the RDI Pre-Formation Group and attending the inaugural meeting in May in Madrid. EURORDIS also participated in NORD's Rare Diseases and Orphan Products Breakthrough Summit on 21–22 October.

EURORDIS–CORD PARTNERSHIP

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



In 2015, CORD and EURORDIS continued their close collaboration: CORD actively participated in Rare Disease Day 2015 (CORD is EURORDIS' official partner for RDD in Canada); was a member of the RDI Pre-Formation Group; and attended the RDI the inaugural meeting in May in Madrid.

There was also regular discussion on HTA, value and pricing of rare disease therapies.

EURORDIS–JPA PARTNERSHIP

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



In 2015, JPA and EURORDIS continued their collaboration. JPA actively participated in Rare Disease Day 2015; was a member of the RDI Pre-Formation Group; and attended the RDI the inaugural meeting in May in Madrid.



EURORDIS–RVA PARTNERSHIP

EURORDIS and Rare Voices Australia (RVA) signed a memorandum of understanding in early 2015. This partnership symbolises a continued effort to stress the international dimension of the rare disease movement and the global benefits to be gained from collaboration in this field. RVA and EURORDIS have agreed that the following are key common objectives:

- RVA coordinates Rare Disease Day in Australia as a EURORDIS official partner and RVA will participate in the development of the Rare Diseases International initiative.
- To address the major expectations that patient communities in Australia and the EU have in common including to: promote rare disease research policy; to enable earlier and better diagnosis of diseases; to promote the development and availability of safe and effective orphan medicinal products and treatments; to provide methods for improving patients' quality of life; and to provide easier and wider access to quality information on rare diseases.
- To support the development of an Australian national strategy or plan for rare diseases, the topic of the recent successful RVA Rare Disease Summit.



EURORDIS–RPU PARTNERSHIP

EURORDIS and the Russian Patients' Union signed a memorandum of understanding in May 2015. RPU and EURORDIS agreed to the following common objectives:



- To strengthen the common European and international voice of people living with a rare disease and address rare diseases as a common challenge, promote rare diseases as an international public health priority and develop worldwide actions, frameworks and rules; and
- To address the major expectations that patient communities have in common in Russia and EU, such as promoting rare disease research policy and funding, enabling earlier and better diagnosis of diseases, promoting equal access to the best standard of care and promoting the development/ availability of safe and effective drugs and treatments.

RPU and EURORDIS have agreed to enhance collaboration through: the continued participation of the RPU in the CNA and in Rare Disease Day; sharing of experiences relevant to the Russian national strategy on rare diseases; and participation in the development and activities of Rare Diseases International, among other activities.



RUSSIAN PATIENTS
ASSOCIATION
Health for all

Improving Access to Medicines

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Access to orphan medicines is defined by the number of eligible patients who, in a given jurisdiction, can be treated by an orphan medicine and who do not participate in a clinical trial where the orphan medicine in question is tested, at a given point in time.

Access can be defined prior to the marketing authorisation (e.g. via compassionate use programmes, named patient compassionate use, or roll-over studies...), or post the marketing authorisation (via commercial availability, off-label use, financial assistance programmes, humanitarian access, or via a generic benefiting from compulsory licensing...).

At the pre-authorisation phase, obstacles come, inter alia, from the large diversity of compassionate use schemes between countries (some do not have a regulatory scheme), and/or the willingness of the company to initiate one, or the lack of information on these programmes.

At the authorisation phase, many initiatives to make the evaluation of medicines more efficient exist, see the Commission Expert Group on Safe and Timely Access to Medicines for Patients ("STAMP") at http://ec.europa.eu/health/documents/pharmaceutical-committee/stamp/index_en.htm.

At the post-authorisation phase, obstacles come, inter alia, from the delays in deciding if the medicine should be reimbursed/covered and for whom, following the technology assessment (HTA) or in negotiating a price, from difficulties in importing the medicine in countries where the holder of the marketing authorisation has decided not to launch the product, from the organisation of care for complex medicines (for example those that

need surgery and an implantable device to deliver the product), from shortages that can occur at any time.

For each of these difficulties, EURORDIS contributes to finding solutions:

PRE-AUTHORISATION: COMPASSIONATE USE

EURORDIS is working with the European Medicines Agency (EMA) to list all contact points within National Competent Authorities who are responsible for the organisation of compassionate use programmes (CUP) at the national level.

In parallel, EURORDIS obtained the agreement from the Heads of Medicines Agencies (HMA) to provide access to information on CUPs to patients in a single location linked to HMA and EMA. This was further discussed at the HMA Management Group which agreed that the national policies as regards to compassionate use programmes in the Member States should be made more transparent and patients should be facilitated in finding relevant information.

EURORDIS views on compassionate use programmes were presented at the "Early Access Programmes Conference", 13–14 Oct. 2015, London.

AT THE AUTHORISATION PHASE : CONDITIONAL APPROVAL AND PRIME

EURORDIS responded to the EMA consultation on "**Guidelines for conditional marketing authorisation**", proposing that for discussions on the renewal of the

conditional authorisation, with or without an oral explanation with the marketing authorisation holder, patients should be consulted and invited in a CHMP meeting, particularly when the opinion is likely to be negative, or if key questions can benefit from the dialogue with patients, e.g. difficulties in relation to the recruitment or the retention of patients in clinical studies requested by the conditional approval.

Another important EMA initiative is a new scheme called PRIME for PRiority MEDicines. This is for medicines of major therapeutic interest, for unmet needs in devastating diseases, where eligible candidates will benefit from a special environment at the EMA, with early appointment of a rapporteur and iterative scientific advice from the very early stage, even before clinical trials start, to the evaluation. EURORDIS responded to the public consultation, and is now working with the EMA to see how relevant patients' organisations can be consulted and/or involved.

POST-AUTHORISATION: PRICING AND REIMBURSEMENT

"Disparities in access to market-authorized orphan medicines exist between as well as within the European Member States. This situation translates into varying delays from the marketing authorisation date to the pricing and reimbursement decisions amongst Member States. Different EU initiatives on how to improve access to treatments based on patients' health outcomes are currently being explored and how to coordinate these at the European level is being examined. In 2015, disparities and delays in access to orphan medicines across Europe and within individual Member States have continued to be a major concern and a priority area for action for EURORDIS, in light of their potentially dramatic consequences for people living with a rare disease.

At the time of writing, EURORDIS estimates that a third of patients across Europe still have no access to the orphan medicines they need and that another third have access only after waiting years, as the orphan medicines they need may be introduced first in larger European markets and only later in others. Even more recently, a new cause for preoccupation is the noted increase in decisions by public authorities to restrict the availability of certain orphan medicines on the sole basis of their prices, perceived as too high in comparison to the medicines' determined value.



TO ADDRESS THE ACCESS ISSUE, EURORDIS HAS INVESTED MUCH EFFORT IN 2015 ON OPENING UP A MORE CONSTRUCTIVE DIALOGUE WITH ALL STAKEHOLDERS

— both public and private — focusing on key principles such as the need for a more affordable and sustainable economic model that safeguards ways and means to reward innovation, the need to put the therapeutic value of a product above its "value for money" intended in a narrow sense, or still the need for strong European collaboration mechanisms all along the life cycle of medicines.

TO PREPARE THE REIMBURSEMENT DECISION: THE HTA MOMENTUM

Two important European initiatives representing an effort to align different methods and policies vis-à-vis the decision to reimburse medicines or not generate high expectations in the patients' community. One is more a political and strategic collaboration (referred to as the "HTA Network"), the other is more a scientific and technical cooperation (referred to as "EUnetHTA", the European Network of HTA agencies).

THE HTA NETWORK

The HTA Network is a voluntary network, set up by Directive 2011/24 (article 15) gathering all Member States, Norway and Iceland. It also associates, as observers, stakeholders representing industry, payers, providers and patients. In 2015, the HTA Network adopted its strategy for the years to come, and a reflection paper on the reuse of joint work in national HTA activities.

As EURORDIS is an observer in the HTA Network, it could contribute in writing and in the meetings (HTA Network meetings 23 March and 29 October) to the adoption both of the strategy and the reflection paper.

EUNETHTA

EUnetHTA is a joint action, the current joint action is the second one and ends in March 2016. During 2015, EURORDIS contributed to the reflection for the next phase, and together with other patients' and consumers' organisations, proposed a platform for the involvement of patients in EUnetHTA activities for the next joint action to come (2016–2019).

All along 2015, Eurordis played a key role in identifying **patients as experts for HTA Early Dialogues** with developers of new health technologies (medicines, medical devices, complex medical interventions...) organised or by EUnetHTA itself, or by the coordinator of the SEED project (Shaping European Early Dialogues, coordinated by the Haute Autorité de Santé, France).

This experience was shared at the HTAi International Conference, 15–17 June in Oslo.

(On this, see "Promoting Patients' Involvement and Rare Diseases Health Technology Assessment (HTA)").

In 2015, EUnetHTA made public the work produced by the European collaboration and actually reused at the national level, in an effort to reduce duplication and share part of the joint work.

THE EURORDIS-EPF "CALL TO PAYERS"

In May 2015, EURORDIS and the European Patients' Forum (EPF) joined forces to launch a call to all national authorities competent for the pricing and reimbursement of medicines in Europe, asking them to step up their collaboration on pricing with a view to ultimately improving patients' access to medicines. Our call focused on two proposals: one being to establish a common "table for price negotiation" with a core group of EU Member States, the other being to scale up markedly all previously existing pilots for early dialogue between payers and companies developing medicines.

Our "Call to Payers" was translated in 6 different languages and widely disseminated after its release – to policy-makers (European Commission, European Parliament), regulators (EMA), industry (European federations like

EFPIA, EuropaBio and EUCOPE) but also to our entire membership.

Looking back, our "Call to Payers" has been successful inasmuch as it has generated support on the part of several EU Member States, and helped to "move the lines" between stakeholders on this important debate. EURORDIS is proud to have put the interests of patients living with a rare disease front and centre and to have spoken up about the tensions between profits and solidarity, proposing concrete solutions in the process.

The "Call to Payers" will continue to bear implications for our work in 2016 and beyond as it is seriously supported by the Health Commissioner and considered in the EU Presidencies.

MECHANISM OF COORDINATED ACCESS TO ORPHAN MEDICINAL PRODUCTS (MOCA)

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

In 2015, EURORDIS continued to take an active part in the work of the "Mechanism of Coordinated Access to Orphan Medicinal Products" (MoCA), established in 2010 as part of the European Commission's Platform on Access to Medicines in Europe and currently placed under the umbrella of the Medicines Evaluation Committee (MEDEV).

As of 2015, MoCA has developed its body of work with about 5 different pilots, each of them centred around a specific orphan medicinal product submitted by a company. To support productive discussions within the platform, EURORDIS has provided its contribution to continuously improving the structure of the regular exchanges of views by involving more systematically relevant expert patients from our network in each single pilot so as to channel the patients' voice and our ideas on aspects as diverse as relevant endpoints in clinical trials, patient outcomes and their measurement, etc.

EURORDIS has also contributed to the evolving reflection on the structure and ambition for MoCA, which was to result in 2016 in a refreshed set of Terms of Reference likely to increase the momentum of this important initiative.

COLLABORATIVE EFFORTS ON EQUITY OF ACCESS AND SUSTAINABLE APPROACHES TO THE FINANCING OF INNOVATIVE PHARMACEUTICALS

As in 2014, EURORDIS has continued in 2015 to play an active role in a number of multi-stakeholder platforms bringing together public authorities, patient organisations and the private sector to discuss today's major challenges

in access and in ensuring the sustainability of European healthcare systems, particularly with the foreseen entry on the market of many new, innovative medicines.

We continued to participate in regular roundtables convened by FIPRA International in Brussels and chaired by former UK Health Minister and MEP John Bowis and by the Chair of Belgium's National Health and Disability Insurance Board, Jo De Cock. In continuation of the work started in 2014, these meetings have focused primarily on aspects related to equity of access and more sustainable solutions for healthcare systems to bear the cost of innovative medicines. However, discussions have progressively opened up to touch upon other related topics such as outcomes-based approaches to pricing and reimbursement and patient access to personalised medicine.

The work of the FIPRA roundtables is set to continue well into 2016 and to lead to a number of stakeholder-endorsed statements that should help inform the greater political debate on those important issues.

PATIENT PROGRESSIVE ACCESS

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

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

In 2015, EURORDIS became partner in the IMI project on Medicines Adaptive pathways to Patients, ADAPT-SMART. The overall scope of ADAPT-SMART is to establish an enabling platform for the coordination of MAPPs-related activities within IMI2 and engaging a dialogue with relevant stakeholders. This is enabled by the overall objectives of ADAPT-SMART, which are to (i) identify relevant MAPPs activities, creating a MAPPs repository of knowledge and opportunities; (ii) identify the scientific challenges and opportunities related to MAPPs implementation; (iii) support new IMI2 research and innovation actions by facilitating the inclusion of MAPPs enablers (tools and methodologies) to address/exploit the identified challenges and opportunities and (iv) develop a comprehensive scientific research plan to substantiate and support MAPPs implementation.

In 2015 EURORDIS co-led the activity that will lead next year to the definition of the Selection Criteria for a product to enter this new development path. In 2015, EURORDIS also participated in the redaction of the Glossary and took part to the preliminary around the future solutions to manage and reduce the uncertainties that patients face when they enter MAPPs. Future activities will focus on the definition of the optimal and seamless pathway to develop medicines under the MAPPs framework. The effort from EURORDIS, along with all partners of the project, will be pursued for the next two years.

LEADING FROM THE FRONT

In 2015, EURORDIS has made a clear choice of not only contributing concrete ideas at the policy or technical levels in all of the forums and platforms in which we have been involved, and which are listed here above, but also to elevate whenever possible the debate to the highest level of decision-making, as our strong belief is that the time for action and change is now.

In that very spirit, in October and November, EURORDIS engaged with the Executive Boards of two major European industry federations, EFPIA and EUCOPE. These two meetings were the opportunity to deliver once again our views on issues related to access, pricing and reimbursement of medicines to an audience composed of senior leaders of major pharmaceutical companies (up to CEO level). EURORDIS also presented innovative and stimulating thoughts as to what tomorrow's economic and scientific model could, or should, look like, inviting the industry to define which future solutions it could potentially accept to explore hand-in-hand with public authorities and patients.

Feedback received from these two interventions indicates that they were successful in nurturing further promising discussions in 2016.

AFTER THE REIMBURSEMENT DECISION, THE ACTUAL ACCESS TO MEDICINES NEEDS TO BE MONITORED

Since the adoption of a **Common Position on Medicine Supply Shortages** by EURORDIS and 45 patients', consumers' and healthcare professionals' organisations in 2013, important progress were made to remedy part of the causes that explain shortages of medicines. Possible causes are numerous; shortages can be due to manufacturing issues or to economic causes.

Patients with rare diseases are particularly affected by shortages. However, the extent of the problem is difficult to quantify and the consequences for their health are difficult to evaluate, given the difficulties to obtain valid public health data on shortages.

EURORDIS addressed these consequences and the challenge to obtain clear information on the shortages at the DIA EuroMeeting in April 2015 in Paris, and at the second workshop on shortages at the European Medicines Agency in October 2015.



For shortages related to manufacturing issues, important progresses were made both by the industry and the regulators. For example, a proposal made by EURORDIS is now being implemented by all pharmaceutical companies: the idea was to monitor more closely medicines that can be the cause of major public health threats when their manufacturing is interrupted. For these medicines, EURORDIS proposed that holders of the marketing authorisation should develop a "Supply Shortage Risk Assessment Plan" for each product at risk, for example when there is only one manufacturing site in the world. ISPE (the International Society for Pharmaceutical Engineering) and PDA (Parenteral Drug Association) created a Drug Shortage Assessment and Prevention Plan which corresponds to what EURORDIS had proposed.

The next steps consist in discussing with all parties involved to analyse shortages due to economic causes, and identify possible solutions. Contacts were made with the Permanent Representatives of the next to come European Presidencies, to include this important topic on the agenda of European Health Ministers.

Patients' Involvement in the Benefit/ Risk Evaluation of Medicines

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EURORDIS has long called for the involvement of patients in benefit/risk evaluation of medicines. Since 2011, EURORDIS has played a frontline advocacy role in promoting such involvement by working closely with the Patient and Consumer Working Party of the Committee for Medicinal Products for Human Use (CHMP), starting patient involvement in several scientific advisory groups of the CHMP and training patients for such activities.

For many years, patients have been consulted by the CHMP mostly in writing, or via invitations to Scientific Advice Meetings in the frame of the marketing authorisation evaluation. In 2014, this advocacy activity has culminated with the first participation of patients in a CHMP oral explanation with the marketing authorisation applicant to discuss the marketing authorisation of a new treatment for a rare disease.

To date, patients were invited by the CHMP on four occasions and the pilot has been prolonged for one year to generate more experience, both for the CHMP and for the patients.

A key factor for the success of this pilot is the demonstration that patients can contribute to the scientific discussion with no influence from the pharmaceutical company, so that CHMP members can be convinced the opinion of the patients to be sincere and authentic.

These initiative at the European level is seconded by others at the National level. Eurordis was invited to a meeting of the Medicines Board at the Dutch agency MEB/CBG in October 2015 when the Dutch Ministry of Health announced the appointment of a patient as member of the Medicines Board for the first time.

Modalities of patients' involvement in the benefit/risk evaluation were the subject of an exciting conference organised by CIRS, the for Centre for Innovation in Regulatory Science, also in October 2015, where EURORDIS was invited to present its views on when and how patients could best interact with regulators.

Improving Access to Quality Care

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MEASURING DIFFICULTIES PATIENTS HAVE IN ACCESSING CARE "ACCESS CAMPAIGN"

Since 2012, EURORDIS started to investigate the consequences of health budgets cuts for patients living with rare diseases in Europe. In 2014, a large survey was launched targeting patients who had difficulties in accessing care. A high response rate, with 1,844 responses from 31 European countries that were analysed in 2015.

The main difficulty for patients with rare diseases is still to find the right specialist for their diseases, 33.7% of respondents reported difficulties finding the specialised doctor they need (622/1,844) and this was explained by different factors, including long waiting times, the lack of a specialist or the distance, even in countries that benefit from a policy for Centres of Expertise.

Unsurprisingly, the second type of cause is of financial nature: or the care is poorly reimbursed, or the advance payment is too high and the reimbursement process takes too long. For 28.8% of the respondents, too expensive and not well reimbursed care or advance payment explained the difficulties they had.

For 43.2% the situation worsened over the last two years, as opposed to 8.9% who saw improvements.

National reports will be written and communication on the results will take place in 2016.

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

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

EURORDIS has engaged in extensive advocacy work with respect to the cross-border Directive from the very beginning of process. Following the approval of the Directive, EURORDIS has accompanied the transposition phase of the Directive into National Laws encouraging National Alliances throughout 2013 to engage in discussion with their health authorities for the national transposition of the Directive. EURORDIS is currently following the implementation of the Directive by contributing to the development of European Reference Networks and being involved in the EUnetHTA Stakeholders Forum, an EU initiative to improve coordination and harmonise the assessment tools used by the main HTA agencies in

Europe, as prescribed by the Cross-Border Health Care Directive.

In 2015, EURORDIS contributed to the **European Patient Forum's Position Statement** that followed the first Implementation Report of the Directive made by the European Commission. In the wake of these results and based on EPF's own findings from their national

workshop on the subject, a position statement shed light on the shortcomings of the implementation in many Member States, on the low awareness among EU citizens of their rights and on what needs doing more urgently from the patient perspective. EURORDIS participated to the strategic discussions with EPF and provided specific contributions on the sections of the Statement related to Rare Diseases and European Reference Networks.

EUROPEAN REFERENCE NETWORKS (ERN)

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

EURORDIS has put forward a **patient-centred vision** of the overall system to be achieved at an EU level, aimed at establishing a **limited number of ERN that cover all rare diseases and that are gathered according to therapeutic**

area. These ERN will pragmatically stem from existing **networks of experts and patient groups** and will widen in a **step-wise approach** to the most mature centres wishing to establish a reference network at the European level. EURORDIS advocates that the overarching goal is to reach a global system where no rare disease patient will be left out, so that each patient can 'find a home'. This should also include undiagnosed patients by the end of the process.

EURORDIS is advocating that ERNs should aim at **improving patients' health outcomes based on clinical excellence and European collaborative approaches.**

In 2015, the rare disease community, saw the first steps in implementation of European Reference Networks (ERN), following the development and establishment of the legislative framework that enables ERN to be founded. EURORDIS continue to enable patients to be central to the implementation of ERNs, specifically in:

- Leading the development of the European Commission's technical proposal for the Assessment Manual & Technical Toolbox;
- Developing and establishing a thematic grouping approach for rare disease ERNs, ensuring all RD "have a home" under an ERN;

- Creating the concept with our members and the clinical community of structuring patient organisations under the thematic grouping of ERNs to optimise their contribution and ensure patients are at the heart of the decision-making process in ERNs; this led to the preparatory work for the establishment (in early 2016) of ePAGs, European Patient Advocacy Groups for each ERN disease grouping;
- Developing the concept of ERNs into tangible set of potential clinical services provided in a virtual environment.

Building on our role in RD-ACTION, EURORDIS has supported both the patient and clinical community to get ready for the first call for European Reference Networks, specifically by:

- Collaborating between the rare disease and eHealth community (especially the community built around the eHealth projects epSOS and EXPAND) on the understanding of interoperability and data sharing needs for ERNs. A "Task Force on Interoperable data-sharing within the framework of the operations of ERNs" has been created to formalise this collaboration and its launch was presented to the eHealth Network and the Expert Group of Rare Diseases in November 2015. Two EURORDIS members are represented in the TF. The TF aims to contribute to defining the specifications of the IT Platform in support of ERNs and to deliver, by the time of the launch of the first wave of ERNs, a European Interoperability Roadmap for data sharing in the framework of operations of ERNs.
- Presenting and supporting the Rare Disease workshop at the 2nd Conference on ERNs organised by the European Commission in Lisbon (October 2015), on

the needs of our community and on the formation of patient involvement in the decision and opinion making processes in ERNs;

- Engaging with international consortia for undiagnosed conditions to raise awareness of ERNs and stimulate discussions on models for undiagnosed patients in ERNs;
- Reviewing funding opportunities for ERNs through European funding mechanisms, specifically for a sustainable business model of their digital services through the VALUeHealth project.

EURORDIS continues to build awareness of European Reference Networks with the rare disease community, patient organisations and clinical leads, through:

- The organisation of two webinars on European Reference Network webinars to develop the concept of thematic grouped ERNs for rare diseases, raise awareness of their role and function, and the process developed to assess applications;
- Capacity-building sessions on ERNs among EURORDIS' member patient organisations, National Alliances

and European Federations in specific CNA and CEF workshops, and at the EURORDIS Membership Meeting (May 2015) on ERNs;

- The creation and the preparatory work for Theme 4: Game changes in healthcare – European Reference

Networks for the European Conference on Rare Diseases 2016;

- The development of ERN specific webpages on the EURORDIS website, the dissemination of information through eurordis.org and EURORDIS eNews.

Powering our collective advocacy capacities

RARER BAROMETER EURORDIS SURVEY PROGRAMME

Rare Barometer was created and launched in 2015 to facilitate and streamline the inclusion of patient perspectives in EURORDIS policy and decision-making processes so to promote patient-perspective-based Policy. This unique programme creates a long-term opportunity to conduct studies (through validated qualitative and quantitative methods) to collect patient experiences and expectations.

The consultation of patients through this programme will support advocacy and policy-making activities at EURORDIS and for EURORDIS members.

In order to carry out quantitative surveys, EURORDIS has created Rare Barometer Voices, a group of people living with a rare disease who participate in EURORDIS surveys and studies. The objective is to transform opinions and experiences about topics that directly affect people living with a rare disease into figures and facts that can be shared with a wider public. The initiative covers 48 countries from the European continent. The registration page and surveys are available in 23 languages so that people can express themselves in their own language.

Patients, parents, siblings or other family members,

patient representatives can register to share their experiences and thoughts. All of the information shared is fully confidential.

Participant are sent an email to participate in surveys. They are free to decide which surveys they want to participate in. The survey data are then collated and analysed to provide data on a European and also national level, as well as according to specific diseases. Finally, participants are sent the results of the survey by email so they can get insight into what other people in similar situations like them feel and think.



CAMPAIGN FOR A EUROPEAN YEAR FOR RARE DISEASES IN 2019

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

In 2015, advocacy for the designation of the European Year for Rare Diseases in 2019 continued. EURORDIS enquired relevant Commission services about the procedure for an official designation, the approach of the current European Commission and its President, who has a last say on decisions regarding European Years' designations.



The advocacy plan was revamped in line with the latest information on the procedure and approach of the Juncker's Commission. EURORDIS contacted MEPs who showed support for the initiative. A written question was submitted by Françoise Grossetête MEP to the European Commission asking for future plans of the Commission to designate European years (no official designation was decided for 2016). The process was presented to the CNA meeting in October 2015.

In parallel, the objectives of the European Year and material have been reviewed in view of strengthening and supporting the advocacy and communication campaign.

The development of a detailed Roadmap was started, based on a benchmark study of previous European Years' campaigns.

ADVOCACY TOWARDS OPTIMAL ENVIRONMENT FOR CLINICAL TRIALS IN THE RD FIELD

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

In 2015, EURORDIS has been active taking part to initiatives developed in different contexts:

Clinical trials in children

The EURORDIS Therapeutic Action Group (TAG) composed of the volunteer patients' representatives on the EMA scientific Committees and Patients' and Consumers Working Party, answered the queries addressed by the EC in March 2015 to the members of the Commission Expert Group on Rare Diseases, on the issue of clinical trials in children given that many trials are delayed or not feasible.

The TAG made several proposals, emphasising that the recruitment of children in clinical trials should be discussed in several working groups, involving the EC, the EMA, the healthcare professionals and of course the patients and their carers (often the parents). For instance, the EC could:

- Set up a working group at the EMA PDCO (Paediatric Drugs Committee) specifically dedicated to the issue of the involvement of children in clinical trials;
- Develop closer links with the IRDiRC Working Group on "Small Population Clinical Trials" in which the EMA and EURORDIS are both involved;
- In order to address societal issues interrelated with the involvement of children in clinical trials, this topic could be addressed in the Commission Expert Groups on Rare Diseases and on Cancer Control (in which EURORDIS is a member).

Clinical Trials in Small Populations

Clinical research and trials in rare diseases face evident obstacles: very low disease prevalence, small and heterogeneous patient populations, difficulty to recruit such patients, disease severity, etc. While traditional randomised controlled study designs are not suited, there is a need to develop novel and rigorous controlled study designs along with relevant analyses that assess treatment efficacy in heterogeneous small populations.

Across 2015, EURORDIS actively contributed to the discussions and the work carried out in the framework of the IRDiRC's Task Force on Clinical Trials in Small Populations. The Task Force worked to develop a "Preparatory Document for Joint Workshop on Small Population Clinical Trials Challenges in the Field of Rare Diseases", in view of a workshop co-organised with the European Medicines Agency in early 2016.

Participation in research projects on small populations CT designs (see section on Research)

The 7th Research Framework Programmes of the European Commission (FP7) co-funds research projects specifically addressing the design of small population clinical trials.

In particular, ASTERIX (Advances in Small Trials dEsign for Regulatory Innovation and eXcellence) aims to optimise methodology for clinical trials in small populations to achieve more reliable and cost-efficient clinical development of treatments for rare diseases.

The project InSPiRe (Innovative methodology for small populations' research) focuses on the development of novel methods for the design and analysis of clinical trials in rare diseases or small populations defined.

In both projects, EURORDIS is member of the Independent Scientific Advisory Committees oversees the scientific direction and progress of the projects.

DATA PROTECTION

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

Following long-lasting negotiations, the European Parliament and the Council of the EU adopted a compromise agreement on the Data Protection Regulation that will replace the previous legislation. In the new Regulation, Data Protection is regarded as a fundamental right of the individual. Principles of "lawfulness, fairness and transparency" must be respected when collecting personal data. Stricter rules apply to "sensitive data", which include "genetic data" and "data concerning health". However, the new rules recognise and regulate the possibility of processing personal data for scientific research and health-related purposes, with a number of safeguards and derogations.

Altogether, the new provisions seem to reflect a balanced approach "between facilitating the safe and secure use of patient data for health research and the rights and interests of all individuals", in line with the position issued by EURORDIS in 2013. Also, the final text of the Regulation de facto recognises the distinction between the inherently different uses of data, whether for public interest purposes e.g. for health research, or for being held by IT providers, social media, search engines, etc.

EMA CONSULTATIONS WITH PATIENTS IN 2015

Among other important **EMA consultations**, EURORDIS contributed to the **EMA consultation on the Access Policy and Clinical Trials Transparency**, the revision of the **Access Policy to Eudravigilance**, the concept paper for the guideline on the Assessment of Clinical Safety and efficacy in the preparation of Community **Herbal Monographs**, on **Priority Medicines** (PRIME, guidelines for conditional marketing authorisation, on a reflection paper on a proposal to enhance early dialogue to facilitate accelerated assessment of priority medicines) and on **Conditional Authorisation**.



INFORMATION & NETWORKING

Membership Base – 695 members

65 new members joined EURORDIS in 2015. New countries represented: Nepal, China, Morocco.

At the end of 2015, EURORDIS had 695 members in 63 countries, 37 of which are European countries, 26 being members of the European Union.



EURORDIS Membership Meeting – Madrid 2015

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

The EMM 2015 Madrid took place on 28–30 May 2015 at the Rafael Atocha Hotel, Madrid. The programme committee of EMM 2015 was made up of 11 members, including

EURORDIS Directors and staff and representatives of the Spanish National Alliance FEDER. The event attracted 250 participants from 43 countries and comprised: 3 satellite workshops (Council of National Alliances, launch of Rare Diseases International, RareConnect workshop); the EURORDIS General Assembly followed a conference focusing on European Reference Networks; and 4 capacity-building workshops on topics including "Research", "Centres of Expertise & European Reference Networks", "Access to Orphan Medicinal Products and Care" and "Social Services". 27 fellowships were provided to patient representatives from 15 countries.



Council of National Alliances

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

EURORDIS supports a network of 38 national alliances, 32 of which constitute the CNA. Two new national alliances joined the CNA in 2015, representing Macedonia and Serbia.

The CNA's main activities in 2015 were:

- a) The work on Centres of Expertise (CoE) and European Reference Networks (ERN). Focusing notably on the need for National Centres of Expertise to be officially recognised by Member States and the support of Member States to the leaders of the ERN
- b) The preparation and coordination of Rare Disease Day 2016
- c) By the end of 2015, 26 National Alliances within Europe had signed the Common Goals & Mutual Commitments document.

In 2015, two CNA Workshops took place in Madrid (May) and in Paris (October), and were again held partly in

common with the Council of European Federations (CEF), in order to allow for cross-cutting discussions on common issues. The first workshop gathered 40 participants and focused on topics such as: Rare Disease Day 2016; best practices in governance on National Plans; the key role of National Alliances to be ready for the call for European Reference Networks (ERN) at the end 2015; presentation of the new RareConnect Governance Model; presentation of recent activities of the Spanish Alliance for Rare Diseases (FEDER).

The second CNA meeting took place in Paris in October 2015. On the second day the meeting was held together with the CEF and gathered 51 participants. Topics included: Centers of Expertise: national actions and Member States process for ERN; European collaboration between National Competent Authorities on Pricing & Reimbursement; a presentation of ACHSEs' ongoing projects; the European Year of Rare Diseases; and the Rare Barometer Programme plan.

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

Council of European Federations & Networks

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



Representatives of European Rare Disease Federations gathered in Paris in October 2015 to discuss issues that are important across Europe and across diseases. For the fourth time, part of the meeting took place in conjunction with the CNA (Council of National Alliances) to discuss cross cutting issues and share experiences.

The second part of the meeting focused on a discussion of the EURORDIS Membership Criteria for European Federations, the RARE-Bestpractices project, Rare Diseases International and the Innovcare Project. The second day of the meeting was fully dedicated to a training on European Reference Networks (ERN).

EURORDIS continued for the sixth year the program "Support to European Rare Disease Federations". The smallest and/or youngest organisations often have great difficulties in financing their network meetings (Board meetings, Network meetings, conferences etc). EURORDIS has granted a total of 23 000 € to 13 European RD Federations to help them organise their different meetings.

Rare!Together

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Rare!Together aims at helping in the creation, operation and management of European rare disease federations, in particular through the website Raretogether.eurordis.org. This website a good practice reference handbook

and toolkit for existing European Federations and for patient organisations planning to set up their European Federation.

European Network of Rare Disease Help Lines

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

In 2015, 13 help lines from 9 countries participated in the activities: Bulgaria (ICRDOD), Croatia (Croatian help Line for rare Diseases), France (Maladies Rares Info Services, AFM-Téléthon), Italy (Coordinating Centre for Rare Diseases Veneto Region, and Telefono Verde Malattie Rare), Portugal (Linha Rara), Romania (NORO, Myasthenia Gravis Romania), Spain (SIO-Feder and ENERCA), Switzerland (Info Maladies Rares) and Hungary (Information Centre for the Rare Disease Patients, a new help line).

In addition to its annual meeting, the network conducted its 8th Caller Profile Analysis in October, based on all enquiries received. The network also reviewed methods used by help lines to evaluate the satisfaction of enquirers.

The information was gathered by each help line over the month of October 2015. It is based on the analysis of a total of 1,714 calls, emails, letters or visits. A total of 68 paid or volunteer staff responded, it took in average 23.5 minutes to respond to each enquiry. Most of enquiries were made by telephone (63%), and the enquirer was more often a patient (33.8%), or a relative (32.2%), then a healthcare professional (20.9%). Enquirers were looking for information on the disease (25.1%), or on specialised centres (15.4%), or on social care (13.5%), for the main purposes of the enquiries. For enquiries where a disease was specified, there was a large representation

of neurological diseases, or of diseases with a dominant impact on cognitive functions (29%).

The second largest group was represented by neuro-muscular and skeletal diseases (17%), followed by diseases

due to inborn errors of metabolism (including endocrine disorders 6%) and skin diseases (6%). Enquiries by undiagnosed patients represented 11% of the enquiries related to information on disease.



RareConnect – Rare Disease Online Communities

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RareConnect.org is a EURORDIS initiative which provides a platform for rare disease patients and patient organizations to develop online communities and conversations across continents and languages. Its goal is to provide a safe, accurate and lively online platform that helps meet the needs of patients and families living with a Rare Disease, in that it allows them to connect with others, access quality information and actively participate in community-driven knowledge generation which can complement and enhance more and better research on

rare diseases. Launched in 2010, RareConnect is now home to 83 disease-specific communities created in partnership with 680 patient groups and managed with the support of 280 volunteer moderators.



RARECONNECT DISCUSSION GROUPS

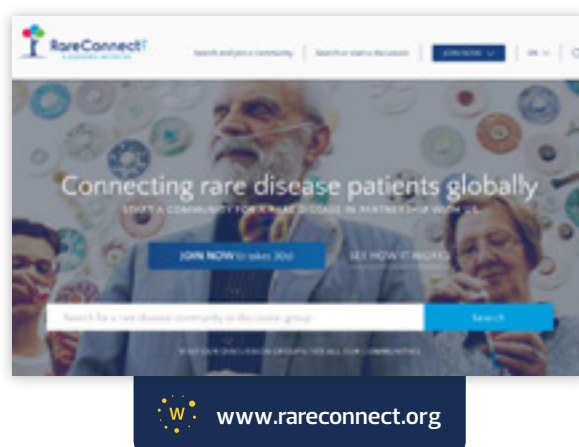
Perhaps the biggest evolution since the launch of RareConnect, during 2015 the platform opened up the possibility for any person living with a rare disease to initiate a discussion group. Since launching this new feature in September 2015, RareConnect now hosts over 200 discussion groups covering both specific rare diseases as well as topics which go across diseases e.g. access to medicines or wheelchairs. Patient Organisations are welcome to apply to be listed as a resource in discussion groups where they have information or services of direct interest to members.

2015 saw RareConnect get a makeover with a new logo and governance structure reflecting a deeper involvement by rare disease National Alliances, partner patient organisations and moderators in its Governance. This move was formalised by the setting up of a Members and Partners Advisory Committee whose remit is to provide feedback and advice on the current performance and future evolutions of the platform.

During 2015 the RareConnect support team grew to include 3 community managers based in Zagreb (Croatia) and Belgrade (Serbia) as well as a Web Technology Manager to assist the platform's evolution into mobile technology and day-to-day technical support and maintenance.

The global reach of RareConnect continues to grow with traffic during 2015 nearing 700,000 visitors from 225

countries. Subscribed members surpassed 20,000 amounting to a 51% increase compared to 2014.



EURORDIS Website

The EURORDIS website outlines the events and activities of EURORDIS and provides information relating to the role of patient organisations in the development of rare disease and orphan medicines policy. For European and international visitors, the website information is translated into 7 languages (English, French, German, Italian, Portuguese, Russian and Spanish). The website boasts over 400,000 visits annually.

In 2015, new sections were added to the website, providing information to people living with a rare disease and their families including European Reference Networks and international Activities. The Photo Contest section was redesigned for the 2015 edition.

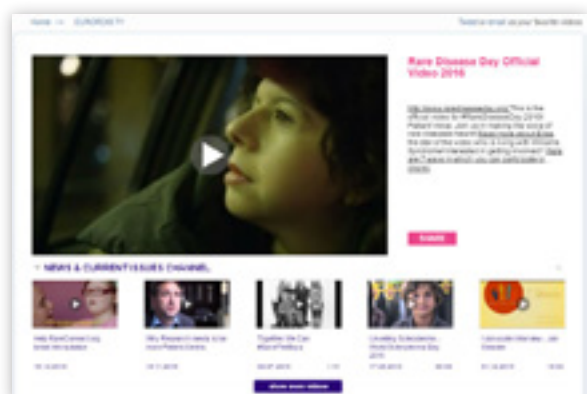
In order to better serve our wide range of stakeholders in our communication and to better meet patients' and patient organisation needs in terms of information on the website, we commenced a study on our communications strategy at the end of 2015 in order to look into the potential of future changes to the EURORDIS website.



EURORDIS TV

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

EURORDIS TV reunites many of the videos accessed free on internet which concern rare diseases or the rare disease community. 11 channels help users navigate. Included in the selection are EURORDIS produced videos from events or for communications' purposes. A new channel, grouping all EURORDIS webinars was added for easier navigation.



EURORDIS Photo Contest 2015

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Initiated in 2006, the EURORDIS Photo Contest is an annual event that raises awareness of rare diseases among members of the general public and members of the rare disease community. It helps to build awareness for rare diseases and breaks the isolation of patients by sharing their story via photography.

The EURORDIS Photo Contest 2015 was open to public voting via social media (#RareButReal2015). Close to 400 photos were entered the competition, sent by participants from 42 countries. This year an Instagram Prize was awarded by Ami Vitale, a professional National Geographic photographer. This expert in photography joins fashion photographer Rick Guidotti in the team of judges.

EURORDIS PHOTO CONTEST 2015 PUBLIC WINNER

The winning photo, as voted for by the public, is of Lucas from Spain who is living with osteogenesis imperfecta. After recently fracturing his arm, Lucas still did not miss the annual parade of the historic Cantabrian Wars!



EURORDIS PHOTO CONTEST 2015 EXPERT WINNER

The Expert's Choice award goes to a photo of Luca from Switzerland who has spinal muscular atrophy. It is entitled 'Easy Rider'.



EURORDIS PHOTO CONTEST 2015 INSTAGRAM WINNER

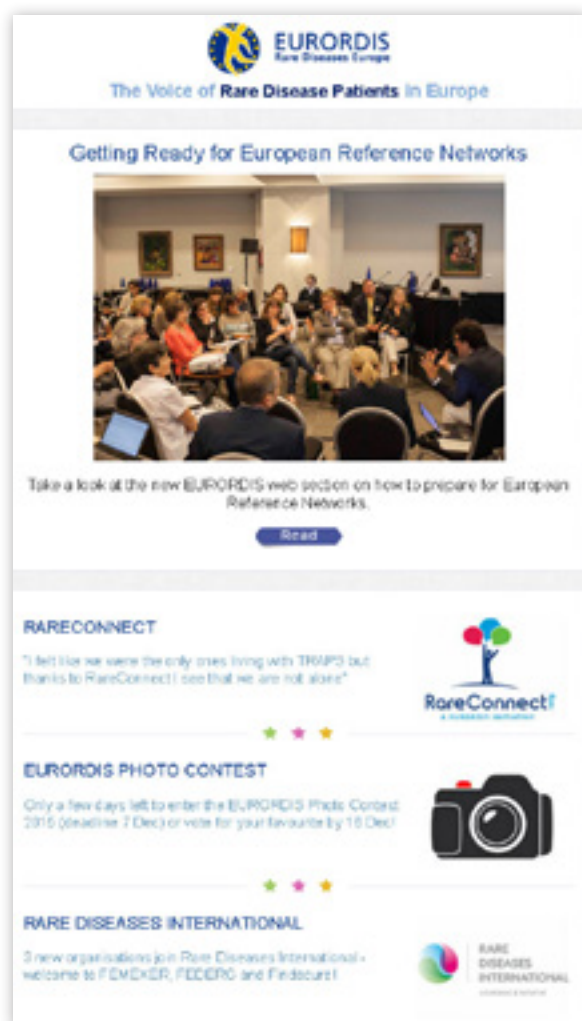
The winner of the Instagram Award is a photo of Eli and horse Lucy from the USA. Eli is 10 and has a rare genetic disease called Fanconi anaemia.

Newsletters – EURORDIS eNews and Member News

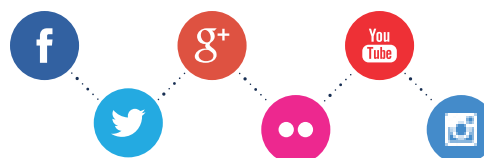
The EURORDIS eNews is bi-monthly news report in 7 languages that communicates breaking news of interest to patient advocates, people living with a rare disease and their families and policy makers. Each eNews features a lead article (devoted to important news in the rare disease community or EURORDIS activity) in addition to short news sections on topics including new RareConnect communities, member events and EURORDIS TV content. Content is also made available via EURORDIS Facebook and Twitter.

In 2015, the Member News was launched with monthly issues in 7 languages, recapping important information for EURORDIS member patient organisations. Sent via email, the news is organised in 3 parts: Information, Feedback and Action.

Both newsletters sent by EURORDIS are constructed and disseminated in-house by EURORDIS staff or automatic programs created by EURORDIS staff. The only element that is outsourced is the weekly translations; the eNews is available in all 7 languages of the website (English, French, German, Italian, Portuguese, Russian and Spanish). In 2015, over 8,000 subscribers received 29 issues of the eNews and our patient organisation members received 6 Member News.



EURORDIS Social Media



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

In 2015, EURORDIS launched an Instagram account.

EURORDIS continued in 2015 to build its social media communities on Facebook, Twitter, Google+ and with the photo contest opened a new Instagram account. Followers of the accounts stay in touch with the latest information on EURORDIS activities as well as updated news from EURORDIS staff, including Yann Le Cam, who travel to conferences and events representing EURORDIS.

EURORDIS Policy Fact Sheets

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EURORDIS has created specific fact sheets to help rare disease patients and organisations advocate at a National level on relevant issues and manage a patient organisation. The EURORDIS policy fact sheets provide comprehensive, validated information on specific topics relevant to rare diseases. All these documents are available in printed form and online on Eurordis.org in English and are widely distributed to members at relevant events.

In 2015, a fact sheet for patient organisations declaring in-kind support in their financial statements was produced and made available on the website. This includes volunteer support.



Rare Disease Day 2015

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

28 February 2015 marked the 8th edition of the Rare Disease Day Campaign with the slogan 'Day-by-day, Hand-in-hand'. The campaign saw active participation in over 80 countries, including all 28 EU countries. EURORDIS and 32 rare disease national alliances acting as country organisers, together with other rare disease patient groups, mobilised thousands of patient organisations across 5 continents. New countries holding an event in 2015 included Bolivia, Estonia and Madagascar.

Around the world thousands of events were staged on or around Rare Disease Day by patient organisations and other partners. EURORDIS organised a Policy Event in Brussels entitled 'Rare but Real: Talking Rare Diseases': (for further information refer to the 'Rare Disease Day Policy Event' in the Advocacy section of this report).

Rare Disease Day 2015 saw the launch of a mobile site for smart phone users. The number of smart phone users reached a peak of 60% on Rare Disease Day. Participation by the public in the "Tell Your Story" feature as well as "Raise and Join Hands" increased on the website. The official video for Rare Disease Day went viral on EURORDIS social media. Translated in 27 languages, the video reached millions around the world, building awareness for people living with a rare disease and their families. EURORDIS thanks Carlo Hintermann for his beautiful video creation using Chinese shadows.





EURORDIS Black Pearl Evening

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The EURORDIS Black Pearl Evening serves to raise and strengthen the rare disease cause throughout Europe by recognising individuals, patient organisations, policy makers and companies that together are improving the lives of people affected by rare diseases. The aim of this evening is to celebrate Rare Disease Day and to foster solidarity and hope for rare disease patients throughout Europe. It also provides an excellent opportunity for leaders and key contributors in the rare disease domain to help promote the cause and network.

The fourth EURORDIS Black Pearl Evening, held on the occasion of Rare Disease Day 2015, took place in Brussels on 24 February 2015.

Over two hundred people attended the event – Brussels community members, national and international officials, researchers, professionals striving to make treatments available to rare disease patients, individuals living with a rare disease and their families and friends. They joined together to recognise the rare disease cause and the recipients of the EURORDIS Awards 2015.



EURORDIS Awards

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The EURORDIS Awards are designed to recognise the outstanding commitment and achievements of patient advocacy groups, volunteers, scientists, companies, media and policy makers who have contributed directly or indirectly to reducing the impact of rare diseases on people's lives. These prestigious awards were judged by the EURORDIS Board of Directors based on over 150 nominations received from EURORDIS members, non-member patient groups, volunteers, staff and the general public with the aim of promoting leadership and the

highest achievements in favour of people living with rare diseases.

2015 marked the third EURORDIS Awards for outstanding accomplishments in the field of rare diseases, which were presented in Brussels on the occasion of Rare Disease Day. HRH Princess Astrid of Belgium was present for the awards ceremony, as was Rare Disease Day Ambassador Sean Hepburn Ferrer, the eldest son of the late Audrey Hepburn, who passed away from a rare cancer.

The 2015 EURORDIS honorees were as follows:

■ LIFETIME ACHIEVEMENT AWARD

Ms Abbey Meyers is an outstanding rare disease patient advocate. Ms Meyers was drawn into the world of political advocacy, fundraising and organisation development when it became painfully apparent through her experience as the mother of a child with Tourette syndrome that patients with rare diseases were being neglected in favour of more common diseases that affected larger patient populations. Ms Meyers founded the National Organization for Rare Disorders (NORD) in the USA.

■ EUROPEAN RARE DISEASE LEADERSHIP AWARD

Professor Josep Torrent-Farnell is Professor of Clinical Pharmacology and Therapeutics at the Autonomous University of Barcelona and a strong advocate for the patient voice. He became the first Chair of the Committee for Orphan Medicinal Products (COMP) at the European Medicines Agency and with his Vice-Chair, Yann Le Cam, introduced many aspects of patient involvement in the EMA that remain today.

■ POLICY MAKER AWARD

Mrs Glenis Willmott is Labour Member of the European Parliament for the East Midlands (UK) and has demonstrated outstanding dedication and commitment in addressing the needs of patients in the European Union. She played an instrumental role in the passing of key legislation through her work as Rapporteur for the Regulation on Clinical Trials on medicinal products for human use and as Shadow Rapporteur for the regulation establishing the Health for Growth Programme.

■ VOLUNTEER AWARD

Ms Rosa Sánchez de Vega is a dedicated rare disease patient advocate, Co-founder of the Spanish Alliance for Rare Diseases (FEDER) and President of the European Federation of Aniridia. As a rare disease patient herself,

and the mother of a son with the same condition, Rosa Sánchez de Vega has successfully managed to channel her difficulties with Aniridia into a positive force for change.

■ MEDIA AWARD

Mr Peter O'Donnell is a prominent writer and editor in the rare disease field currently working as Associate Editor of European Voice. He has worked as an editor and speechwriter for numerous clients in the corporate, political and academic world and has frequently chaired EU-level policy debates on EU affairs. This has made him very well placed to be able to write and report forthrightly on the various complicated issues surrounding rare diseases.

■ PATIENT ORGANISATION AWARD

Children with SMA is a non-profit foundation which has undertaken the incredibly difficult mission of supporting those affected by, or involved with, Spinal Muscular Atrophy (SMA) in Ukraine.

■ SCIENTIFIC AWARD

Professor Kate Bushby is a Professor of Neuromuscular Genetics. Her commitment to research in rare diseases has been demonstrated through her impressive publication list, clinical activities and involvement in policy actions. She has played a leading role in the European and national rare disease policy area, acting as vice Chair on the European Union Committee of Experts on Rare Diseases from 2010 to 2013 and still acts in the capacity of invited expert on the new Commission Expert Group on Rare Diseases.

■ COMPANY AWARD

Pfizer's Rare Disease Research Unit is dedicated to developing new medicines across the spectrum of rare diseases. Pfizer has demonstrated commitment to, and passion for, the rare disease cause with 22 approved products to treat rare diseases worldwide including 4 in Europe.



Specialised Social Services and Social Policies

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In 2015 EURORDIS continued its focus on social services and policies, mainly through its involvement in Work Package 6 of the EUCERD Joint Action Working for Rare Diseases (EJA) and in the new EU-funded project INNOVCare (Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions), with the support of the newly created Social Policy Advisory Group.

This last year of the EJA was mostly focused on the elaboration of the draft Commission Expert Group Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies, in which EURORDIS was highly involved.



EURORDIS supported the drafting process, based on a robust methodology, including literature review, analysis of important EU and national policy documents (including National Plans/Strategies for Rare Diseases and Reports from EUROPLAN National Conferences), organisation of multi-stakeholder discussions and consultations to members of the Commission Expert Group on Rare Diseases, patient representatives, social services providers and other relevant stakeholders.

The EURORDIS Membership Meeting 2015 Madrid provided the occasion for EURORDIS members to exchange on the draft of the Commission Expert Group Recommendations to Support the Incorporation of Rare Diseases into Social Policies and Services and to get to know interesting social policies and innovative care pathways models promoted in Croatia, France, Spain and Sweden.

Also within the scope of the EJA, EURORDIS continued to map specialised social services in cooperation with national alliances and European federations, reaching over 95 services from 23 countries by the end of the EJA (September). The map of these services, in addition to the definitions, fact sheets, testimonies and the Guiding Principles for Specialised Social Services can be found on the EURORDIS website in the designated section entitled 'Services to Patients: Specialised Social Services' and have been widely disseminated in 2015 through the eNews and EURORDIS social networks.

The new EU-funded INNOVCare project, in which EURORDIS is highly involved, was launched in October to address the social needs of people living with a rare disease and the gaps in the coordination between medical, social and support services in EU Member States.

INNOVCare proposes and tests an innovative care pathway aiming at linking health services to social, support and employment services that people living with a rare disease use on a daily basis, ensuring the transfer of information and expertise between care providers. The pathway also centralises the coordination of care through a resource centre for rare diseases and regional case managers, in an effort to relieve the burden of care for patients and their families.

In the last trimester of 2015, EURORDIS took the first steps into the implementation of its INNOVCare tasks:

- Communication: internal and external communication on the project; organisation of ad-hoc advisory groups with policy makers; organisation of the final conference of the project;
- State-of-the-art analysis of the social needs of people with rare diseases and of social care in Member States: study on the needs of patients/families in their social and daily life; study of care pathways in a selection of Member States; comparative analysis between patients' feedback and existing care systems to identify good practices and gaps;
- Facilitation of the creation of a European Network of Resource Centres for Rare Diseases and organisation of meetings for exchange of good practices;
- Representation of people living with a rare disease and their families in coordination with EURORDIS' members and with EURORDIS Social Policy Advisory Group;
- Ensuring the link between INNOVCare, the rare disease community, key EU rare disease projects and relevant.

INNOVCare was presented to and discussed with EURORDIS members during the meetings of the Council

of National Alliances and of the Council of European Federations in October.

During this last year, EURORDIS also continued to strengthen the cooperation with key organisations working on social policy, in order to exchange important information and good practices: the Social Platform and the International Federation of Social Workers Europe.

EURORDIS further contributed to increase awareness of rare diseases amongst social workers and to draw attention to the need for training social services providers

by organising workshops at the 2nd Joint Nordic Conference: Courage in Social Work (Helsinki, June) and at the Conference of the International Federation of Social Workers Europe (Edinburgh, September) dedicated to "Empowering clients with rare, complex diseases to be 'in the driving seat': user focused training for social workers". The *Guiding Principles on Training for Social Services Providers* and on *Examples of Training Programmes for Social Services Providers* were distributed during these workshops.

SOCIAL POLICY ADVISORY GROUP (SPAG)

The Social Policy Advisory Group (SPAG) was created in May 2015 to closely follow EURORDIS' activities to promote the integration of rare diseases into social services and policies.

The 13 volunteer patient representatives in the SPAG provide grassroots experience on the social challenges

experienced by patients and families and provide advice in relation to social policy, social care and related issues (such as holistic care, social services, social innovation, disability, special education, psychological support). This helps to guarantee the formulation of patient-centric approaches to the different social challenges throughout the work of EURORDIS.



RESEARCH, DRUGS & THERAPIES

In 2015, EURORDIS focused its works in the field of rare disease research by putting efforts in strengthening the ties within the research communities to aim to a better integration of the different initiatives, spanning from the Research Infrastructures (ERIC) to its role within the ERA-

Net E-Rare-3. EURORDIS participated also to several European Commission-funded projects so to improve diagnostic and therapeutic possibilities for rare diseases patients.

Shaping and Supporting Research Policy

INTERNATIONAL RARE DISEASE RESEARCH CONSORTIUM – IRDiRC



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

EURORDIS continues to be involved in the International Consortium for Rare Disease Research (IRDiRC) with its presence in its Executive Committee (Béatrice de Montleau, EURORDIS patient representative and Yann Le Cam, CEO of EURORDIS) and in the Therapies Scientific Committee (TSC) (Yann Le Cam, CEO of EURORDIS, Chair of the TSC and Virginie Hivert, EURORDIS Therapeutic Development Director). In addition, since this year, EURORDIS is now also represented in the Interdisciplinary Scientific Committee (ISC) by Gema Chicano, member of the Board of Directors of EURORDIS.

2015 has been a pivotal year for IRDiRC. Governance chart and organization of the work have been revamped so to move from a 'brainstorming' activity to an 'implementation' phase.

In particular, the TSC has published a set of recommendations which have been constituent for this transition, (<http://www.irdirc.org/tsc-recommendations/>) to guide policies and funding strategies so as to reach its goal of 200 new therapies by 2020, based on IRDiRC Policies & Guidelines which were adopted in April 2013. They focus on the improvement of guidelines for the clinical development of orphan drugs; the alignment of scientific and regulatory guidance and the enhancement of the continuous data collection and assessment all along the life cycle of therapy.

Previously each Scientific Committee was working with 4 working groups. The TSC recommendations were built on the brainstorming outcomes of its 4 WGs and the Scientific Committee itself. Following the drafting of the TSC recommendations, key topics have been identified where specific efforts were needed so to obtain concrete achievements. Task forces have therefore been created to tackle these issues.

In particular, the taskforce on Patient-Centred Outcomes

Measures (PCOM) has led to a workshop with top experts in the field held in Paris on 30th November 2015 and post-workshop report and its recommendations are now available (<http://www.irdirc.org/activities/current-activities/tf-pcom/>).

A second taskforce is focusing on Small Population Clinical Trials (<http://www.irdirc.org/activities/current-activities/tf-spct/>) and a third one on data mining & repurposing (<http://www.irdirc.org/activities/current-activities/data-mining-repurposing/>).

Two more taskforces are foreseen for the forthcoming steps: 'Patient Engagement in Rare Disease Research and Health Product Development' and 'Rare Disease Clinical Research Network'.

Another major change in the IRDiRC functioning, largely promoted by EURORDIS, has been the creation of an Operating Committee with conference calls being held monthly between the three Scientific Committee Chairs, the Chair and Vice-Chair of the Executive Committee and the Scientific Secretariat of the Consortium.

PROJECT GENETICS CLINIC OF THE FUTURE (GCOF)

This project is funded under the Horizon 2020 programme. The GCOF kick-off meeting took place recently in Brussels in March 2015. Technology used for next-generation sequencing (NGS) of DNA has rapidly developed over the past five years so that the cost and time it takes to read an individual's DNA is now dramatically lower. Consequently, new genome technologies like NGS are steadily being adopted for use in the diagnosis of genetic disorders. The idea behind the GCOF Project is to map the opportunities and challenges that surround the clinical implementation of these technologies so that the needs, interests and concerns of patients and other relevant stakeholders are taken into account in the future. Complex questions on issues such as data sharing and informed consent need to be addressed due to the confidential nature of personal data produced by such technologies. Through the GCOF Project, the hope is to be able to understand how to harness the potential of these technologies for health care while respecting fundamental ethical and regulatory frameworks. To ensure that the expectations and needs of society, and in particular patients, are met in the broad implementation of these technologies, the GCOF will engage all relevant stakeholders (patient

representatives, genomics researchers, clinical geneticist, bioinformaticians, policy makers, ethics experts etc.) in dialogue and consequently produce recommendations on how to incorporate technologies such as NGS into the diagnosis journey. Within this project, EURORDIS will collaborate with experts from other project partners to carry out a survey on patient perspectives. The results of this survey will be published as a white paper, which will include recommendations for new approaches to the collection, storage and distribution of clinical data.

PROJECT ASTERIX

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

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

Tsveta Schyns, EURORDIS Therapeutic Action Group member, is a member of the Advisory Board of project. The board will provide expert advice on key open project decisions, general philosophy and direction of ASTERIX. It will also be instrumental in dissemination and exploitation of project results, as well as for the quality of deliverables and overall project status. The members of the Advisory Board met in January 2015 in Amsterdam. The main recommendations to the members of the consortium encompassed the enhancement of an early engagement of stakeholders such as EMA, patient organisations, doctors/clinic/nurses, payers and industry and the need of a roadmap highlighting the strategy to achieve the goals through the deliverables of the projects.

Later in the year, Kerry Lesson Beevers, EURORDIS Therapeutic Action Group member, was interviewed, along with other members of the Patients Think Tank about the content of the registries to be useful for clinical trials, and around outcomes measures. Kerry also participated to the face-to-face meeting of the Patient Think Tank followed by the general assembly in Barcelona in October 2015.

PROJECT INSPIRE

Project InSPiRe (Innovative methodology for small populations research – FP7) focuses on the development of novel methods for the design and analysis of clinical trials in rare diseases or small populations defined, for example, by a rare genetic marker. Tsveta Schyns is a member of the Independent Scientific Advisory Committee of the project, which oversees its scientific direction and progress of the project. This comprises international clinical and non-clinical experts and patient group representatives. The committee met in May 2015 in Paris. The main conclusion is that the members noted and agreed that the project is progressing to the right direction, according to the work plan, to achieve its objectives. It was also mentioned that the methodological advances, although directed towards trials in small populations, could apply to clinical trials more generally.

E-RARE-3

In February 2015, in Paris, the kick-off meeting of the E-Rare-3 (on-going until the end 2019 – 5 years) was held in Paris. E-Rare-3 project has 25 partners (research funding national agencies) from 17 countries and is led by the Agence Nationale de la Recherche (France).

The project scope is to pursue and expand the activities in accelerating the development of new diagnostics and therapeutics for patients living with rare diseases. Like its predecessors, E-Rare 3 will launch open calls to fund research that addresses research gaps. In particular, it will tackle the understanding of disease mechanisms and natural history of rare diseases with the aim to develop new diagnostic tools and treatments. In this new, third phase of E-Rare the implication of EURORDIS could be enlarged by its involvement in the tasks specifically dedicated to the engagement of patients' organisations in research.

The participation of EURORDIS in E-Rare-3 opens the possibility for patients' organisations to foster their engagement in funding of research on rare disease at the transnational level and to develop an innovative funding schema with patient organisations.

By being involved in co-financing of selected projects in the framework of E-Rare 3, patient organisations will be able to:

- Reach out and access international research projects for a specific rare disease/group of rare diseases;
- Navigate through, and integrate in, the complicate space of research funding;
- Leverage significant funds for research dedicated to a specific rare disease (each research project is funded by several funding bodies; the average cost of E-Rare funded project is around 750K€);
- Finance excellent research, even in the absence of resources to administer the competition and the launch of a call;
- Alleviate the need for infrastructures where research should be performed (other partners will provide the infrastructural support);
- Foster the participation of relevant stakeholders in a specific disease area (even for the patient organisations that do not have enough budget to fund research).

EURORDIS also participated in September 2015 in Call Steering Committee meeting in Istanbul that aimed to define the topic of the Joint Transnational Call 2016 that was launched in December 2015. The call focused on Clinical research for new therapeutic uses of already existing molecules (repurposing) in rare diseases. This topic was listed as priority #1 for the Joint Transnational Call 2016.

In May 2015, during the EURORDIS Membership Meeting, a workshop was held with patient advocates involved in research to design the foundations of the innovative co-funding model in accordance with the Joint Transnational Call rules and in collaboration with the E-Rare-3 project partners.

OTHER RESEARCH INITIATIVES

EURORDIS representatives voiced the experience, expectations and needs of rare diseases patients by participating in several meetings, workshops and annual assemblies. For instance, a working meeting was attended in Bologna in March 2015 that aimed to shape the perimeter of a common services on Rare Diseases within the biomedical research initiatives.

This was followed by a second meeting in Luxembourg in September 2015 where challenges and opportunities were discussed by each infrastructure's representatives. EURORDIS also presented the need of the rare diseases patient communities towards a better organisation of the translational research at the EATRIS Annual conference in May 2015 in Amsterdam. As a follow up, EURORDIS is part of the EATRIS Patient Advisory Committee. This EATRIS-PAC is coordinated by EGAN, an EURORDIS member.

Promoting the Development of Effective Rare Disease Research Infrastructures

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

This was achieved by supporting the involvement of patients and their representatives at both the governance and clinical research levels of several international projects and consortia.

TREAT-NMD

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

COMMUNITY ADVISORY BOARDS

For the implementation of its Charter for Clinical Trials, EURORDIS supports the creation of Community Advisory Boards (CABs) where patients and developers of medicines, sponsors of clinical trials meet to discuss the development plan and the practical aspects of the trials.

At a time where Patient Engagement is more and more in sight, these C.A.B will certainly develop in the near future. One has already been functioning with patients from tuberous sclerosis, and a new one held its first meeting in 2015 with patients representing systemic sclerosis, with interstitial lung disease (SSC_ILD). These groups are now actively involved in 3 clinical trials and a compassionate use programme.

BIOBANKS

Patient organisations are aware of the need for more rapid progress in rare disease research and of the difficulties that researchers and clinicians face in obtaining human

biological material to conduct research on rare diseases, and EURORDIS has represented this patient-driven view.

As the former coordinator of EuroBiobank, EURORDIS continued its strong commitment in promoting and actively participating in the governance of the first network of biobanks dedicated to rare diseases. EURORDIS continued to raise the importance of supporting biobanks for rare disease research to European and national level policy makers, liaising with the Biobanking and Biomolecular Resources Research Infrastructure (BBMRI) Consortium, most recently joining the BBMRI Rare Disease Working Group.

EURORDIS also presented its position on consent as collaboration during the round table on "ELSI / How to involve patient and citizens" during the HandsOn Biobanking conference organised in Milan in August 2015.

GENOMICS DATA

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



In 2015, EURORDIS was a key partner in the drafting of the scientific paper entitled, 'You should at least ask'. The expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research. The publication is foreseen in early 2016. Within the myriad articles about participants' opinions of genomics research, the views of people with a rare disease are unknown. It is important to understand if their opinions differ from the general public by dint of having a rare disease and vulnerabilities inherent in this. Here we document rare diseases patients' attitudes to participation in genomics research, particularly around large-scale, international data and biosample sharing.

This work is unique in exploring the views of people with a range of rare disorders from many different countries. The authors work within an international, multidisciplinary consortium, RD-Connect, which has developed an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for RD research. Focus groups were conducted with 52 RD patients from 16 countries. Using a scenario-based approach, participants were encouraged to raise topics relevant to their own experiences, rather than these being determined by the researcher. Issues include wide data sharing, and consent for new uses of historic samples and for children. Focus group members are positively disposed towards research and towards allowing data and biosamples to be shared internationally. Expressions of trust and attitudes to risk are often affected by the nature of the RD which they have experience of, as well as regulatory and cultural practices in their home country. Participants are concerned about data security and misuse. There is an acute recognition of the vulnerability inherent in having a RD and the possibility that open knowledge of this could lead to discrimination.

Meanwhile, EURORDIS conducted a Delphi-like exercise to address the views of people living with a rare disease. This work is unique in exploring the views of people with a range of rare disorders from many different countries. The authors work within an international, multidisciplinary consortium, RD-Connect, which has developed an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for RD research. Focus groups were conducted with 52 RD patients from 16 countries. Using a scenario-based approach, participants were encouraged to raise topics relevant to their own experiences, rather than these being determined by the researcher. Issues include wide data sharing, and consent for new uses of historic samples and for children. Focus group members are positively disposed towards research and towards allowing data and biosamples to be shared internationally. Expressions of trust and attitudes to risk are often affected by the nature of the RD which they have experience of, as well as regulatory and cultural practices

in their home country. Participants are concerned about data security and misuse. There is an acute recognition of the vulnerability inherent in having a RD and the possibility that open knowledge of this could lead to discrimination.

OTHER RARE DISEASE INFRASTRUCTURE ACTIVITIES

EURORDIS also contribute to the CORBEL project, CORBEL (acronym for COordinated Research Infrastructures Building Enduring Life-science Services), aiming at establish a collaborative and sustained framework of shared services between the Biological and Medical Research Infrastructures (RI). This will transform biomedical research in Europe – from discovery of basic biological mechanisms to applied medical translation – through provision of harmonized accession processes, unified ethical and legal support, joint data management, and coordinated user access to advanced research instruments, facilities and samples. Individually, the services offered by the RIs are critical to their own user communities; collectively, through CORBEL, this common access will be transformative across the life sciences from generation of knowledge at the bench to the treatment of patients at bedside. EURORDIS was invited to be a member of the Medical Infrastructures / Users Forum (MIUF) coordinated by ECRIN. The MIUF will play a major role in driving and coordinating activities inside WP3, but will also contribute to other work packages (particularly management and communication, and ensure links with WP on access, ELSI, innovation, and training). It will also play a major role beyond the project, as an interface between scientific communities, funding schemes, and the pan-European research infrastructures. EURORDIS was also invited to be a member of the Multi-stakeholder taskforce on access to individual participant data (IPD) in clinical trials. This initiative, supported by the Horizon2020 programme is major step towards the integration of services offered by the European Research Infrastructures.

PARTNERSHIPS WITH THE RARE DISEASE RESEARCH COMMUNITY: EUROPEAN SOCIETY OF HUMAN GENETICS

In June 2015, EURORDIS participated in the European Society of Human Genetics (ESHG) annual meeting in Glasgow, during which the ESHG and EURORDIS entered into a new partnership through the signature of a Memorandum of Understanding. The two organisations will align and support one another on a range of activities including conferences (such as the annual ESHG Conference and the European Conference on Rare Diseases & Orphan Products, the latter of which is organised by EURORDIS and partners), policy actions and initiatives of common interest.



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

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In January 2013, EURORDIS joined, as one of 15 partners, the RARE-Bestpractices project, a 4 year (2013–2016) project funded by DG Research under FP7–HEALTH–2012 INNOVATION–1. This project is developing a sustainable platform for sharing best practices for the management of rare diseases, supporting the collection of standardised and validated data and the efficient exchange of knowledge and reliable information on rare diseases. RARE-Bestpractices is aimed at collecting, evaluating and disseminating best practices and agreeing on a methodology suitable to develop and update best practice guidelines. The project also creates a forum for exchanging information, sharing lessons learnt and facilitating collaborations.



EURORDIS acts as a transversal partner in the different work packages, ensuring the involvement and capacity-building of patient organisations across Europe regarding the project. EURORDIS is especially involved in the work packages that focus on dissemination and networking with relevant activities in the field of rare diseases and on defining a common methodology for the production of guidelines on clinical management of rare diseases.

In the context of the RARE-Bestpractices project, a Patient Advisory Council (RBP-PAC) was created to advise on project activities, to guarantee a patient-centric approach throughout the project and to consolidate the expertise of patient representatives on the subject of clinical guidelines. A meeting of the RBP-PAC was organised during the EURORDIS Membership Meeting in Madrid, in May 2015. Several patient representatives could also attend the two trainings organised through the project in February and in December 2015, on the topic of Health Care Guidelines on rare diseases: quality assessment.

A capacity building session was held during the EURORDIS Council of European Federations in October 2015, in order to introduce the tools developed by the project and to involve patient representatives further into the project.

Additionally, in the perspective of the elaboration of two pilot guidelines that will test the methodology developed by the project (on sickle cell diseases and on catastrophic antiphospholipid antibody syndrome) and their respective patient versions (to be released in 2016), EURORDIS has established contacts with relevant patient organisations and identified one patient representative for each panel of experts.

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

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In parallel to the marketing authorisation process, patients are now consulted when HTA bodies assess new health technologies (medicines, and also medical devices, connected devices, complex surgery, diagnostic tests etc.), at least at the European level. Since September 2014, patients have been invited to participate in early dialogues between HTA experts and industry (through the SEED¹ project), and as experts in the review of joint assessments led by EUnetHTA².

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

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

EURORDIS is one of four patient representative organisations at the EUnetHTA Stakeholders Forum. EUnetHTA is an EU initiative to improve coordination and harmonise the assessment tools used by the main HTA agencies in Europe. It is funded by the European Commission and Member States. EUnetHTA is a joint

action between the European Commission and Member States; it is now in the second joint action (JA2).

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

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

EURORDIS's Director of Treatment Information and Access, François Houjé, is the current co-chair of the Forum. Visit Eunetha.eu to learn more.



¹ Shaping European Early Dialogues (<http://www.earlydialogues.eu/has>)

² European Network of HTA Agencies (<http://www.eunetha.eu>)

EARLY DIALOGUES BETWEEN DEVELOPERS OF NEW HEALTH TECHNOLOGIES AND HTA EXPERTS: THE SEED PROJECT³

The French HTA agency, Haute Autorité de Santé, is coordinating the SEED project that involves thirteen other HTA bodies from eleven Member States. The SEED project will end mid-2015. It offers an opportunity for industry to meet with HTA experts at an early stage to discuss their development plans.

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

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

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

POLITICAL AND STRATEGIC ACTIVITIES: THE EUROPEAN HTA NETWORK AT THE EUROPEAN COMMISSION

Reflection on Long-Term HTA Collaboration in Europe

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

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

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

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

Support to Patient Representatives at the European Medicines Agency

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

Dedicated expert patient representatives contributed to the examination and scientific evaluation of more than 540 dossiers in 2015 through the work of the scientific committees they belong to, as well as to the activities of several ad hoc working groups all along the year.

The Therapeutic Action Group (TAG), composed of EURORDIS and non-EURORDIS patient representatives in the above-mentioned scientific committees and working party at the EMA, continued their work and maintained communication internally with monthly conference calls and emails.

In addition to these permanent activities at the EMA, patient representatives are also invited on a sporadic

basis to attend the scientific committees and the Scientific Advice Working Party (SAWP) as experts for their disease.

In 2015, 25 patient representatives attended meetings of the SAWP for protocol assistance. Protocol assistance is a version of scientific advice specific to orphan medicinal products and is a way for the company developing the medicine to obtain scientific and regulatory advice on the manufacture of a medicine, as well as on pre-clinical and clinical tests being performed. Patients are involved in order to provide first-hand input on the most relevant outcome measures and endpoints for clinical trials. The process of scientific advice/protocol assistance is recommended to avoid major objections (regarding the design of clinical trials) during evaluation of the marketing authorisation

application.

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



³ <http://www.earlydialogues.eu>

Applications responding to the European Commission's Call for Expression of Interest were submitted for the COMP for Lesley Greene (extension of mandate) and Julian Isla Gomez (new application) in 2014. Lesley Greene was successful and started her new mandate in July 2015. She has been re-elected Vice-Chair of the Committee in October 2015. Julian Isla was ranked first on the reserve list and is attending the COMP meetings as an observer with the financial support of EURORDIS.

Applications submitted in response to Call for Expression of Interest for CAT membership have been submitted in September 2015 and are under review by the Commission. After an internal Call for Expression of Interest within its Members, EURORDIS has nominated Michele Lipucci di Paola (current member) and Christos Sotirelis (expert patient)

In order to improve its capacity to involve patients all along the life cycle of the therapeutic development, EURORDIS has created a position of Patient Engagement Manager. From September 2015, Elisa Ferrer is working closely with EMA, Patient Relations Team and SAWP Secretariat so to analyse Protocol Assistance dossiers, identify and suggest patients from the EURORDIS network or beyond. Patients that have been trained to the R&D processes (e.g. EMA training day, EURORDIS Summer School, EUPATI) are given priority.

The Patients' and Consumers' Working Party, of which Eurordis is a member, is a unique forum where all scientific committees of the Agency meet with patients and consumers.

In 2015, PCWP members worked in particular on the EMA policy on draft functional specifications for the EU portal

and EU database, on guidelines for conditional marketing authorisation, on a reflection paper on a proposal to enhance early dialogue to facilitate accelerated assessment of priority medicines, EU Medicines Agencies Network Strategy to 2020, Concept paper in the revision of the "Guideline on the assessment of clinical safety and efficacy in the preparation of Community herbal monographs".

The Working Party established five new topic groups:

- 1 Measuring the impact of patient involvement and, exploring how to measure the benefit/value of patient input on regulatory outcomes, exploring the impact that involvement in EMA activities has on empowerment of PCOs
- 2 Acknowledge and promote visibility of patient input in the Agency's activities
- 3 Training: explore synergies with existing training initiatives, explore methods to further enhance support for patients involved in EMA activities
- 4 Social media: explore how PCOs use social media to communicate with their members and the wider community, brainstorm on issues for discussion within a workshop on social media in 2006
- 5 Involvement of young people / children

Identify existing youth groups within eligible organisations; look to create, within the umbrella of the PCWP, a "young persons' network" with young participants Eurordis is directly involved in 1), 3) 4) and 5).

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

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

Two DITA Task Force meeting were held, in Paris, one in April and one in November 2015 to discuss methods to evaluate the cost-effectiveness and economic aspects of medicines.

In November, two researchers were invited: Zachary Fitzpatrick, Fulbright Scholar from the US, residing in Paris for the academic year and Institut Pasteur, working on gene therapy and ethical aspects of research, and Mark Nuijten, health economist at Business Valuation Erasmus University Rotterdam.

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

- The EMA policy on draft functional specifications for the EU portal and EU database
- Guidelines for conditional marketing authorisation,
- A proposal to enhance early dialogue to facilitate accelerated assessment of priority medicines (PRIME)
- The EU Medicines Agencies Network Strategy to 2020
- A Concept paper in the revision of the "Guideline on the assessment of clinical safety and efficacy in the preparation of Community herbal monographs"
- Review of EMA documents for the public or contributions to EMA consultations: European Public Assessment Reports for the Public (9), Package Leaflets (19), and a patient card for patients treated with bisphosphonates.

DITA TASK FORCE AND HTA

DITA Task Force members also responded to the EUnetHTA consultation on "Patient and Social Issues" and responded on the "Opinion of the Expert Panel on effective ways of investing in Health" Preliminary Report

on Access to Health Services in the European Union.

DITA task force new research "Acting on the Treatment Information you have"

With respect to off-label use of medicine, a new research has been proposed, to explore how patients are actually taking their medicines, how they understand the black symbol for additional monitoring which is printed on some package leaflets, how they see the use of off-label products. The questionnaire was adapted from a pilot one used in 2012. The research was successfully submitted to an Institutional Review Board (INSERM) and the data collection period is planned for early 2016.

OFF-LABEL USE OF MEDICINES

Discussions on off-label use with industry took place in January 2015 at a meeting with EUCOPE, European Confederation of Pharmaceutical Entrepreneurs, where

EURORDIS presented the Good Off-Label Use Practices (GOLUP), a concept created by Prof Marc Doms and presented at the European Conference on Rare Diseases in 2014.

A second meeting at EUCOPE in May 2015 was an opportunity to exchange views on a strategy for more and better medicines.

Finally, several volunteers of the DITA attended the Drug Information Association EuroMeeting in Paris, 13-15 April 2015

SOCIAL MEDIA AND PHARMACEUTICALS

In March, representatives of the DITA task force were invited to share their opinions on social media and pharmaceuticals at the Belgium Regulatory Affairs Society. In particular, the role of social media as a platform to exchange information on medicines was discussed.

New European Projects in the Field of Pharmacovigilance

WEB-RADR

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



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

More on Web-RADR: Web-radr.eu

SCOPE

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

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

More on SCOPE: Scopejointaction.eu

EURORDIS Capacity Building & Training for Patient Advocates

In 2015, EURORDIS continued its capacity-building & training activities for patient advocates by: holding the highly successful EURORDIS Summer School for the 8th consecutive year; expanding its online e-learning tools; and continuing involvement in the DIA Patient Fellowship Programme. In 2015, collaboration continued with two training projects: the European Patients' Academy on Therapeutic Innovation (EUPATI) and the European Clinical Research Infrastructure Network – Integrating Activity (ECRIN-IA).

EURORDIS EXPRESS SUMMER SCHOOL

The EURORDIS Summer School was initiated in 2008 as part of our continued commitment to empowering people living with rare diseases. This four and a half day course provides training in aspects of medicines development and EU regulatory processes where patient representatives can be involved. A new format was developed for the Summer School in 2015 that combines training for both expert patients and researchers on medicines development.

The 8th EURORDIS Summer School session was held in Barcelona in June 2015 and once again aimed at training patient representatives in clinical trials, medicines



development and EU regulatory processes, HTA and market access. 43 participants attended representing 20 countries and 25 diseases selected based on a call for candidates.

The 2015 Summer School provided participants with a fundamental understanding of the processes involved in medicines development, the time required and the different stages of clinical trials. Coupled with formal presentations of the topics, the participants were divided into small groups and encouraged to share their experiences and knowledge in the context of documents provided to complement the lectures.

This year in partnership with a COST Action BM1207 financed the participation of 11 researchers. European Clinical Research Infrastructure Network project (ECRIN), ten (10) participants representing Nutrition and Medical Devices were also included in the EURORDIS Summer School. The COST Action financing will continue in 2016.

ONLINE LEARNING

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

All of the above experiences will be further developed to include additional topics and to target different disease groups through the European Patients' Academy on Therapeutic Innovation (EUPATI), a 5-year project funded by the Innovative Medicines Initiative (IMI). This patient-led academy will provide scientifically reliable, objective, comprehensive information to patients on pharmaceutical research and development.

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

EURORDIS Round Table of Companies (ERTC) 2015 Workshops

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

As is the case every year, two workshops were held in 2015:

'RARE DISEASES: GOING GLOBAL' 25 FEBRUARY 2015, BRUSSELS: 89 ATTENDEES FROM 13 COUNTRIES

The 22nd workshop of the EURORDIS Round Table of Companies held in Brussels on the occasion of Rare Disease Day 2015, focused on the theme "Rare Diseases: Going Global!". In the current climate, the development of innovative rare disease therapies is mostly global in nature due to the rarity of patients and experts, as well as to the fact that companies and investors have an increasingly international approach to the market. The meeting's objective was to discuss the opportunities that 'going international' presents, as well as to look at possible approaches for a long-term, structured, progressive internationalisation that involves patients, patient organisations, researchers, industry and competent authorities. The discussion focused on current strategic initiatives that have the potential to structure this internationalisation and how they can be synergised to optimise results.

'SHARING RARE DISEASE PATIENT DATA: TRANSLATING PRINCIPLES INTO ACTION' 29 SEPTEMBER 2015, BARCELONA: OVER 100 ATTENDEES FROM 13 COUNTRIES

The 23rd Workshop of the EURORDIS Round Table of Companies Workshop brought together over 100 participants to discuss how to make clinical research more patient-centred through patient-reported outcomes and patient-centred outcome measures. Making clinical studies more patient-centred means that they are designed to be as relevant as possible to patients and therefore the most efficient at generating evidence to prove that a therapy will improve lives. The ERTC workshop served as a forum for discussion between all stakeholders present (regulators, pharmaceutical and biotech companies, patient representatives and research bodies) with the aim of reaching a common understanding of patient-reported outcomes (PROs) and patient centered-outcome measures (PCOMs). It is aligned with the work done by the International Rare Diseases Research Consortium (IRDiRC), which has issued recommendations highlighting the importance of PCOMs and PROs.

In 2015, the ERTC comprised:

- 59 members
- 10 new members: Cytokinetics, Horizon Pharma, HRA Pharma, Mapi, OpenApp, Roche, Santhera, Sarepta, UCB, Ultragenyx

GOVERNANCE, ORGANISATION & FUNDING

Board of Directors



During the General Assembly held in Madrid on 29 May 2015, EURORDIS full members voted on four vacant positions in the Board of Directors, re-electing the UNIAMO, Italy, represented by Simona Bellagambi and Genetic & Rare Disorders Organisation (GRDO), Ireland, represented by Avril Daly. The Board also welcomed Françoise Salama as the new representative of the AFM-Téléthon, France and Nick Sireau, representing the AKU Society, UK.

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

The AKU Society (represented by Nick Sireau) resigned from the EURORDIS Board of Directors in November 2015 for personal reasons. The position will be up for election at the upcoming General Assembly in May 2016

Staff Organisation



The team comprised 41 people, 32 full-time equivalent (FTE) as of December 2015. The team is composed of paid staff, one office volunteer, one consultant and trainees. Most staff members are based in the Paris office located in the Rare Disease Platform. EURORDIS' Public Affairs Directors, Public Affairs Junior Manager and the Research and Healthcare Director are based in the Brussels office. The EURORDIS RareConnect team managing the online patient communities network is based in Barcelona alongside the Patient Engagement Manager. The Rare Diseases International Senior Manager is based in Geneva. The Chief Executive Officer shares his time between the Paris and Brussels offices.

The following are the main changes in human resources in 2015:

- Flaminia Macchia, European & International Public Affairs Director, has left EURORDIS
- Jean-Louis Roux, Public Affairs Director, has joined EURORDIS
- Carmen Lasheras Ruiz, RareConnect Project Manager, has joined EURORDIS

- Sandra Courbier, Surveys and Social Research Manager, Rare Barometer Leader, has joined EURORDIS
- Elisa Ferrer, Patient Engagement Manager, has joined EURORDIS
- Jennifer Steele, Public Affairs Junior Manager, has joined EURORDIS
- Laura Amorini, Web Technology Manager, has joined EURORDIS
- Tihana Kreso, RareConnect Manager, has joined EURORDIS
- Igor Ban, RareConnect Manager, has joined EURORDIS
- Sandra Pavlovic, RareConnect Manager, has joined EURORDIS
- Anna Kole, Research Infrastructure Project Manager, has left EURORDIS
- Virginie Bros-Facer, Research Infrastructure Project Manager, has joined EURORDIS
- Juliette Sénécat, Health & Social Projects Manager, has returned from parental leave



Volunteers

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

These volunteers are called the EURORDIS volunteer patient advocates.

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

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



OUR REPRESENTATIVES ON THE EMA SCIENTIFIC COMMITTEES

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

The EURORDIS patients' representatives and staff on EMA scientific committees and working party, together with other patients' representatives nominated on COMP (Dr Pauline Evers and Mr Mario Ricciardi) form the Therapeutic Action Group (TAG). The TAG holds monthly conference calls to update on the work of each EMA committee and take actions where necessary.

OUR REPRESENTATIVES ON THE COMMISSION EXPERT GROUP ON RARE DISEASES

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

- Ms Dorica Dan, Chair of RONARD, the Romanian National Alliance for Rare Diseases – European Network of National Alliances for Rare Diseases, full member
- Mr Jan Geissler, Vice President of the Leukemia Patient Advocates Foundation – Network of European Federations of Rare Diseases, full member
- Mr Alastair Kent, Director of the Genetic Alliance UK and Chair of Rare Disease UK & EGAN, full member
- Mr Yann Le Cam, Chief Executive Officer of EURORDIS, full member
- Ms Amanda Bok, Chief Executive Officer of the European Haemophilia Consortium – Network of European Federations of Rare Diseases, alternate
- Ms Lene Jensen, Chief Executive Officer of Rare Disorders Denmark – European Network of National Alliances for Rare Diseases, alternate
- Mr Flavio Minelli, co-founder and Board member of the Italian Ichthyosis Union (UNITI), member of EGAN, alternate
- Mr Christopher Natchtigäller, member of EURORDIS and former President of ACHSE, the German Alliance for Rare Diseases, alternate

Two observers from EURORDIS on the Expert Group are in charge of supporting the group of patients' representatives:

- Ms Flaminia Macchia, Public Affairs Director, from January to end June 2015
- Ms Valentina Bottarelli, Public Affairs Director, from July 2015 to present
- Ms Ariane Weinman, European Public Affairs Manager

The patients' representatives and the observers form the EURORDIS Policy Action Group (PAG).

OUR REPRESENTATIVES ON THE COMMISSION EXPERT GROUP ON CANCER CONTROL

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

- Mr Jan Geissler, EURORDIS / Vice President of the Leukemia Patient Advocates Foundation, full member
- Ms Kathy Oliver, EURORDIS / Chair and Co-Director of the International Brain Tumour Alliance (IBTA), alternate

The work of the two EURORDIS volunteer representatives is supported by the EURORDIS Policy Action Group on Rare Cancers (PAG-RC) composed of other four patient representatives from different rare cancer and paediatric cancer organisations (Dawn Green, Eric Low, Catherine Vergely, Drakoulis Yannoukakos) and one staff (Ariane Weinman).

Finance and Support Services

Finance and support services' activities in 2015 included:

- Accounting and monthly financial reporting in a timely manner including cash flow and risk analysis detailed report.
- Monthly meetings with managers to update the budget and the year-end financial forecast.
- Operating Grant Steering Committees organised every three months to review progress on implementation, deliverables and budget.
- Management of human resources activities, such as recruitment.
- Management of office support: IT infrastructure, contact database, office supplies.
- Management of legal and fiscal matters.

Contract Grants

NEW

RD-Action: Joint Action to expand and consolidate the achievements of the former EUCERD JA, DG Sanco, 2015–2018

E-RARE 3: For the extension and strengthening of the transnational cooperation on rare disease research funding organisations, Horizon 2020, 2015–2019

InnovCare: Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions, DG Employment and Social Innovation (EaSI), 2015–2018

Adapt-SMART: An enabling platform for the coordination of Medicines Adaptive Pathways to Patients (MAPPs) activities, Innovative Medicines Initiative (IMI), 2015–2017

PACE-ERN: Partnership for Assessment of Clinical Excellence in European Reference Network (PACE-ERN) Consortium, Call for Tender to develop a European Reference Network (ERN) manual and toolbox, Health Programme, 2015–2016

RENEWED

Framework Partnership Agreement 2015–2017 (Operating Grant), single beneficiary, DG Health and Consumers, 2015–2017

Specific Grant Agreement (Operating Grant) for year 2015 (SGA FY2015), single beneficiary, DG Health and Consumers, 12 months

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

ONGOING

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

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

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

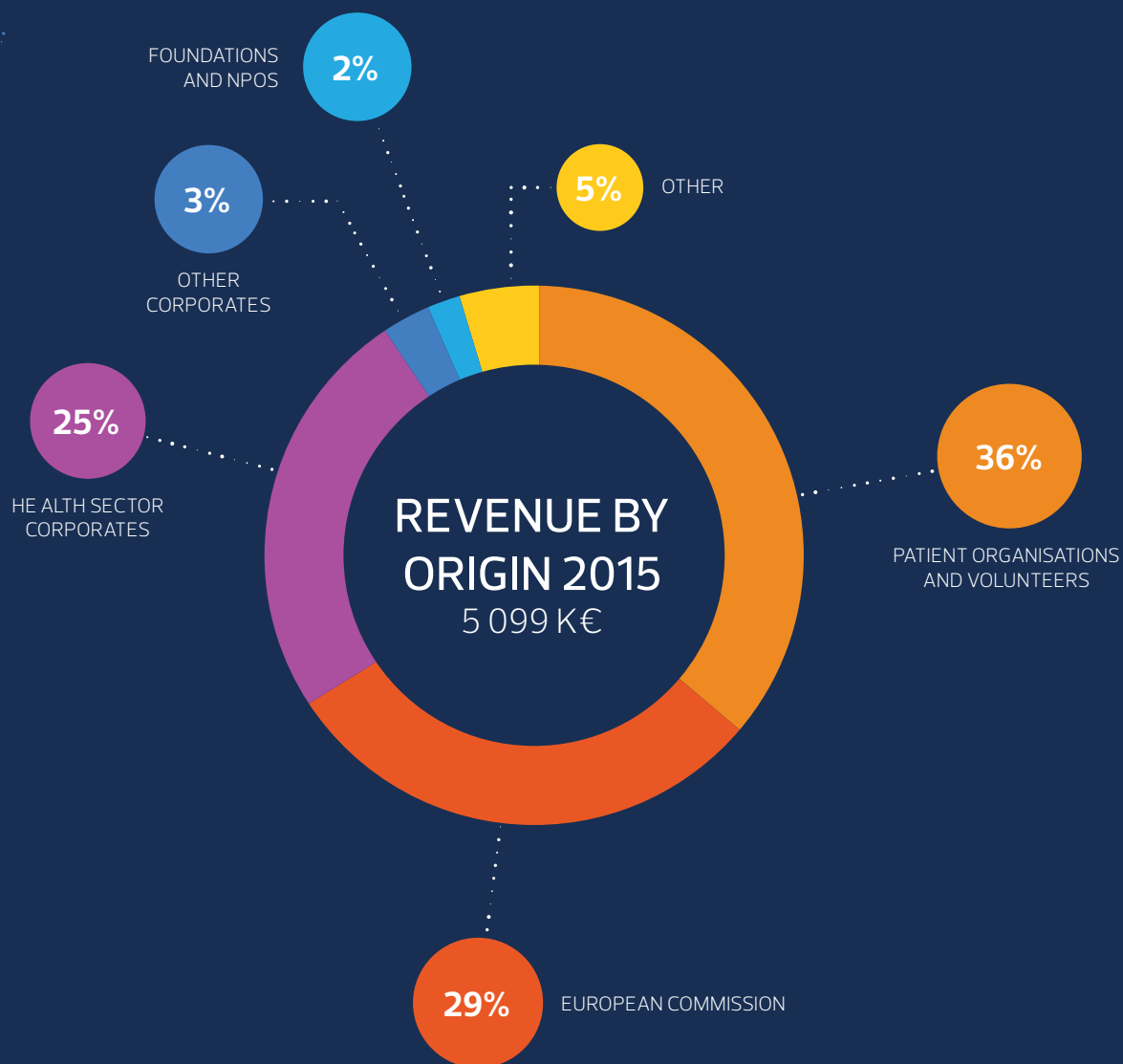
RARE-Bestpractices, a platform for sharing best practices for the management of rare diseases, DG Health and Consumers, 2013–2016.

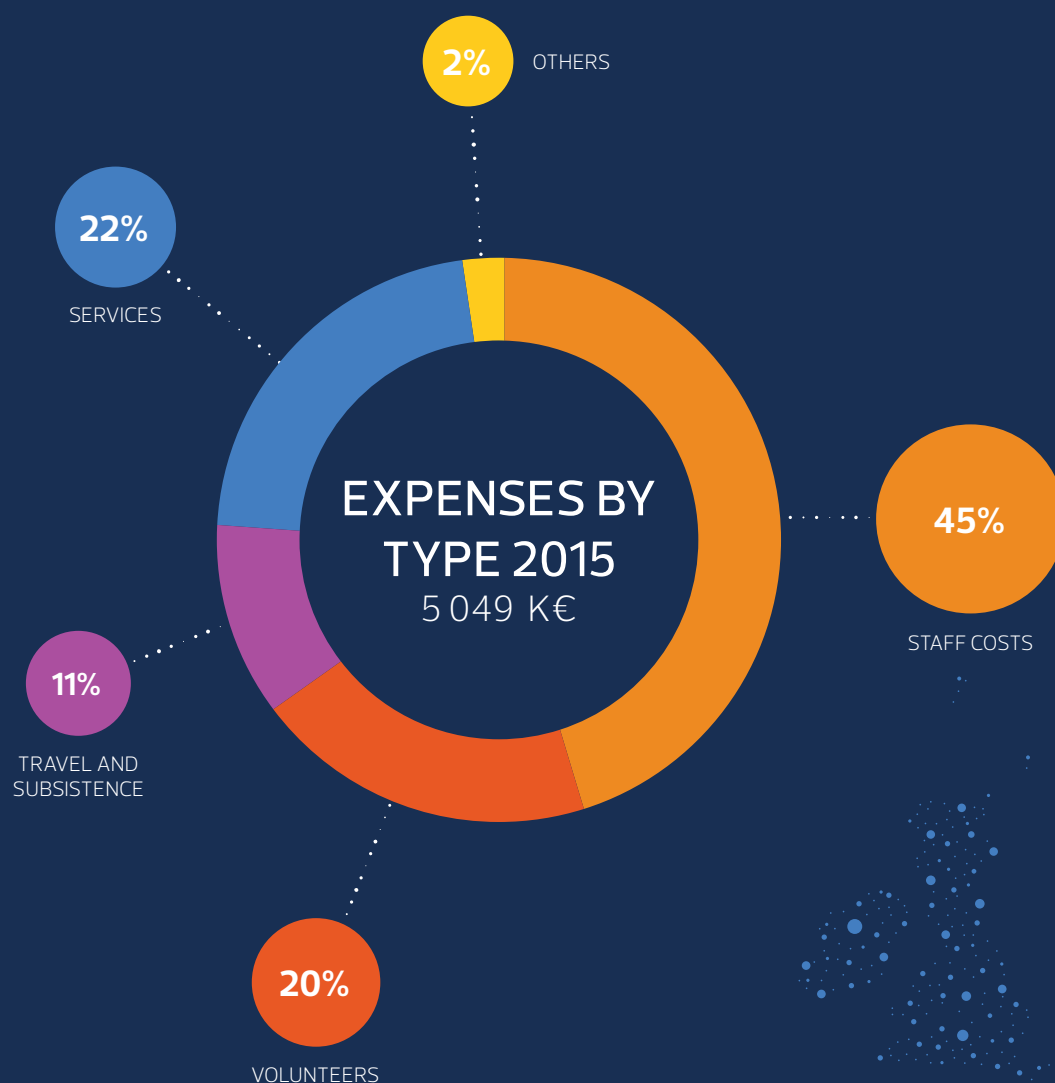
Web-Radr, Recognising Adverse Drug Reactions, IMI-JU, 2014–2017.

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

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

REVENUE & EXPENSES 2015







BOARD OF DIRECTORS MAY 2015 – MAY 2016

PRESIDENT		
Mr Terkel Andersen	Danish Haemophilia Society	Denmark
DIRECTORS		
Ms Simona Bellagambi	UNIAMO – Federazione Italiana Malattie Rare	Italy
Ms Avril Daly Vice-President	Genetic & Rare Disorders Organisation	Ireland
Ms Dorica Dan Officer	Romanian Prader Willi Association	Romania
Mr John Dart General Secretary	DEBRA International	UK
Ms Birthe Byskov Holm	Rare Diseases Denmark	Denmark
Ms Anne-Sophie Lapointe	Alliance Maladies Rares	France
Ms Françoise Salama	AFM-Téléthon	France
* Mr Nick Sireau	AKU Society	UK
Mr Dimitrios Synodinos Treasurer	PESPA – Greek Alliance for Rare Diseases	Greece
Ms Gema Chicano Saura	FEDER – Federación Española de Enfermedades Raras	Spain
Ms Geske Wehr	Selbsthilfe Ichthyose e.V.	Germany

* The AKU Society (represented by Nick Sireau) resigned from the EURORDIS Board of Directors in November 2015 for personal reasons. The position will be up for election at the upcoming General Assembly in May 2016.



CONFERENCES & WORKSHOPS 2015

| **"What can be done by stakeholders to increase patient's access to authorized Orphan Medicinal products?", Seminar in honor of Kerstin Westermarck, Medicinal Products Agency, Uppsala, Sweden, 16 December**

Lesley Greene: "WHY have orphan drugs? The Personal Experience of an unmet medical need – Diagnosis of an Orphan Disease"

| **ECRIN Workshop: "Conducting Independent, Multinational Clinical Trials in Europe", Paris, France, 7–8 December**

Nancy Hamilton: "EURORDIS Summer School"

| **European Medicines Agency workshop: "Demonstrating significant benefit of orphan medicines concepts, methodology, and impact on access", London, UK, 7 December**

Yann Le Cam: "The Patient's Perspectives on Significant Benefit"

Virginie Hivert participated as EURORDIS' representatives

| **ACHSE (German National Alliance for Rare Diseases): "Internationale Vernetzung" (International Networking), Berlin, Germany, 27– 28 November**

Anja Helm: "Opportunities and challenges of international networking"

| **2nd Conference on Sanfilippo Syndrome and Related Diseases, Geneva, Switzerland, 26–28 November**

Paloma Tejada: "The role of patient organisations in shaping rare disease policy"

| **RARE 2015 Conference, Montpellier, France 26–27 November**

Anne-Sophie Lapointe: "Quelles sont les responsabilités de la société vis-à-vis des personnes atteintes de maladies rares ?" (What are the society's responsibilities towards people living with rare diseases?)

| **EFPIA Board Meeting, Brussels, Belgium, 17 November**

Yann Le Cam: "Cross-Border Collaboration in the Area of Pharmaceutical Pricing"

Jean-Louis Roux participated as EURORDIS' representative

| **6th Annual World Orphan Drug Congress, Geneva, Switzerland, 11–13 November**

Sharon Ashton, Jill Bonjean, Mathieu Boudes and Paloma Tejada attended as EURORDIS' representatives

| **Retina Conference 2015, workshop on Patient Relevant Clinical Endpoints (Retina), Dublin, Ireland, 5 November**

Virginie Hivert: "EURORDIS work on patient relevant outcome measurements"

| 4th European Congress on Rett Syndrome, Rome, Italy, 30 October to 1st November

Béatrice de Montleau: "Supporting Research for Rare Diseases in Europe"

| "HTA without border: From collaboration in science to decision making" co-organised by the European Commission and French HTA agency (HAS), Paris, France, 30 October

Yann Le Cam: "Patients @ Early Dialogues (HTA/Regulatory) "

Jean-Louis Roux participated as EURORDIS' representatives

| Health Collaboration Summit 2015 (co-organised by EFPIA), Brussels, Belgium, 28–29 October

Mathieu Boudes: "Engaging patients all along the life cycle of medicines"

| Clinical trials for rare diseases: "Exploring synergies between ECRIN and Orphanet networks", Paris, France, 27 October

Yann Le Cam: "The international initiatives: Introduction to IRDiRC"

| European Biotechnology Week: Makers's Fair , Rome, Italy, 17–18 October

Simona Bellagambi represented EURORDIS

| 8th Annual Scientific Conference on Rare Diseases organised by PESPA, the Greek Alliance for Rare Diseases: "Training Patients and Doctors regarding Rare Diseases: The Necessity of Working Together", Athens, Greece, 16–17 October

Zoe Alahouzou, presentation on EURORDIS

| Xth International Conference on Rare Diseases and Orphan Drugs (ICORD), Mexico city, 15–16 October

Paloma Tejada: "Rare diseases international: la alianza mundial de pacientes con enfermedades raras"

"Global Strategies for Rare Diseases: The Patients' perspective"

Gema Chicano: "Patients' voices: International cooperation within patient groups: Why, Who and How"

| RIME Alliance Maladies Rares, Information meeting with members of the French National Alliance for Rare Diseases, Paris, France, 14 October

Denis Costello: Presentation of Rare Connect

| Lunch debate: "Cross Border Healthcare – Achievements and Challenges in EU Legislation" – organised by EUCOPE (the European Confederation of Pharmaceutical Entrepreneurs), European Parliament, Brussels, Belgium, 13 October

Yann Le Cam, Keynote speaker

Valentina Bottarelli participated as EURORDIS' representative

| EUROPABIO multi-stakeholder conference: "From Science to Patients: Enabling Personalised medicine through Healthcare Biotechnology", Brussels, Belgium, 12 October

Yann Le Cam, Panelist: "What can 'end users' of personalised medicine do to integrate personalised medicine in routine activities? How can successes be replicated in the future?"

| II European Reference Networks Conference: "from planning to implementation", Lisbon, Portugal, 8–9 October

PACE-ERN Consortium: **Matt Johnson** and **Louise Clément:** "Manual for Technical Assessment of the ERNs"

Yann Le Cam: "The patients' views" (within the Round Table: "Network organisational challenges and experiences: issues, solutions and lessons learned")

| Multiple System Atrophy Symposium and Fundraising Dinner, Antwerp, Belgium 2 October

Marta Campabadal: Presentation of RareConnect

| Annual Joint Conference by DIA, EFGCP and EMA on Better Medicines for Children, London, UK, 1–2 October

Kerry Leeson-Beevers, Panellist in session: "Looking Back at the 10 Years of the Paediatric Legislation – the Ways Forward".

| "50 Years of EU Pharma legislation: Achievements and future perspectives", organised by the European Commission, Brussels, Belgium, 28 September

Yann Le Cam, panellist in the session: "Risk regulation – What is the appropriate level?"

Virginie Hivert, participant

| Forum ORPHANET / French National Alliance for Rare Diseases / Foundation Groupama for Health: "Therapeutic education, e-health, telemedicine: impact of new information and communication technologies on rare diseases", Paris, France 28 September

Nancy Hamilton: e-learning applied to health: international experience feedback

| European Cancer Congress ECC 2015, Vienna, Austria, 25–29 September

Dorica Dan represented EURORDIS

| Annual PHA European Conference, Castelldefels, Spain, 17–20 September

Marta Campabadal: Presentation about Crowdfunding

| 5th annual Orphan Drugs Summit, Copenhagen, Denmark, 17–18 September

Birthe Holm: "The voice of patients living with rare diseases in Europe"

| Final conference of the EUCERD Joint Action “Working for Rare Diseases”, Luxembourg, 15 September

Valentina Bottarelli: “Achievements of EUROPLAN National Conferences and Good Practices”.

Dorica Dan and **Raquel Castro:** “Specialised Social Services and Integration of Rare Diseases into Social Services and Policies”

| European IPF (Idiopathic Pulmonary Fibrosis) Patient Associations Meeting, Brussels, Belgium, 14 September

Anja Helm: Presentation of EURORDIS' activities and its services offered to European Federations of rare disease patients

| Cambridge Rare Disease Summit 2015, Cambridge, UK, 14 September

Denis Costello: Presentation of RareConnect, an initiative of EURORDIS

Eva Bearryman also represented EURORDIS

| Findacure: “Crowdfunding for Rare Disease Patient Groups”, London, UK, 14 September

Eva Bearryman represented EURORDIS

| Conference of the International Federation of Social Workers Europe, Edinburgh, 6–9 September

Workshop on “Empowering clients with rare, complex diseases to be ‘in the driving seat’: user focused training for social workers”

Dorica Dan (EURORDIS), **Ian Johnston** (International Federation of Social Workers), **Lisen Julie Mohr** (Frambu, Norway)

| Presentation of MonitoRARE – first report on the situation of people with rare diseases in Italy – at the Italian Parliament, Rome, Italy, 23 July

Terkel Andersen represented EURORDIS

| “Unveiling Scleroderma: Hidden Truth”, event at the European Parliament organised by FESCA (Federation of European Scleroderma Associations) in aid of World Scleroderma Day, Brussels, Belgium, 30 June

Eva Bearryman represented EURORDIS

| 6th International R&D Dating (RIR): “From Rare Diseases to Personalized Medicine”, Collège de France, Paris, France, 30 June

Virginie Hivert and **Ariane Weinman** represented EURORDIS

| DIA TOPRA Workshop: “Adaptive Pathways”, Brussels, Belgium, 30 June

Yann Le Cam: “Patient Initiatives to Support Adaptive Licensing”

| Comunicación & Social Media: Construir una identidad online y ser activo en el mundo digital, Santander, Spain, 30 June

Marta Campabadal presentation of RareConnect, creation of online patient communities for rare diseases

| Second International Conference on Rare and Undiagnosed Diseases, Hungarian National Science Foundation, Budapest, Hungary, 26–27 June

Simona Bellagambi presented the EURORDIS's position on the development of an Undiagnosed Condition Network

| MHRA – Medicines & Healthcare products Regulatory Agency: “Pathway to Innovation from Research to Patients”, Wellcome Trust London, UK, 11 June

Lesley Greene: “Patient Perspective on involvement in regulatory decision making and Drug Development”

| 2nd Joint Nordic Conference: “Courage in Social Work”, Helsinki, Finland, 10–11 June

Session on “Power and Influence in Social Work”

“Empowering clients with rare, complex diseases to be ‘in the driving seat’: user focused training for social workers in Scandinavia”

Raquel Castro (EURORDIS), **Ian Johnston** (International Federation of Social Workers), **Lisen Julie Mohr** (Frambu, Norway), **Anders Olason** (Ågrenska, Sweden)

| Tecnologia i lleis per Associacions, Barcelona, Spain, 10 June

Marta Campabadal, presentation on RareConnect

| European Society of Human Genetics Annual Meeting, Glasgow, United Kingdom, 6–9 June

Mathieu Boudes and **Virginie Hivert** were in charge of the EURORDIS booth

Sandra Courbier also represented EURORDIS

| European Alliance for Personalised Medicines: “Smaller Member and Regions Together (SMART): STEPs in the Right Direction to a Brave New Healthier Europe”, Brussels, Belgium, 2 June

Session: Medical Adaptive Pathways to Patients—Setting the Stage with Stakeholders

Yann Le Cam, Panellist: “Medical Adaptive Pathways to Patients (MAPPS) – Taking Steps Forward”

| EATRIS 2nd Conference: “Building Bridges in Translational Medicines”, Amsterdam, the Netherlands, 27–28 May

Mathieu Boudes: “How could EATRIS contribute to better patient outcomes? The rare disease patients' perspective”

| INFARMED, I.P. Annual Conference, Lisbon, Portugal 20 May 2015

Yann Le Cam: “Access vs Sustainability of drugs: Information and Communication”

| INNERMed – Inherited Neurometabolic Diseases Information Network – Zagreb, Croatia, 11–12 May

Vlasta Zmazek: Introduction to EURORDIS

1st International Conference on the Diagnosis, Management and Treatment of Hypoparathyroidism, Florence, Italy, 7–8 May

Chairs Terkel Andersen & Renza Barbon Galluppi, UNIAMO:
"ADVOCACY: Patient's Role in promoting Care for Rare Diseases"

World Orphan Drugs Congress USA 2015, Washington, D.C., 23–24 April

Yann Le Cam, Panellist: "Panel: Rare disease community working together for orphan drug development"

Findacure's Patient Group Workshop, London, UK, 17 April

Lara Chappell: "How to create awareness days and use existing international campaigns to benefit patient groups"

Drug Information Association (DIA) Annual EuroMeeting, Paris, France, 13–15 April

Yann Le Cam:
"Regulatory Evidence Standards: Are we maximising value generation? The Patients' perspective"
"Medicines Adaptive Pathways to Patients (MAPPs) – Holding hands across the lifecycle"

François Houjé:

"Shortage consequences: their impact on patients, health care professionals and regulators"
"Solving and preventing shortages / Communication between different stakeholders"
"Looking at the future shortages of medicinal products"
"Patient involvement in the development dialogue with regulatory authorities and HTAs"

BBMRI-ERIC (Biobanking and Biomolecular Resources Research Infrastructure): "Exploring a Common Service for Rare Diseases", Bologna, Italy, 25–26 March

Mathieu Boudes: "EURORDIS and its involvement in Biobanking activities, needs and expectations"

"Rare Cancers: Exploiting the Potential of European Reference Networks", hosted by Alojz Peterle MEP, Brussels, Belgium, 24 March

Flaminia Macchia and Ariane Weinman represented EURORDIS

EMA (European Medicines Agency) 20 years anniversary: 'Science, Medicines, Health: Patients at the heart of future innovation', 18 March

Yann Le Cam: "The Patients' Perspectives"

First European Patient–Expert School of patient organisations related to imprinting disorders, supported by COST– European Network for Human Congenital Imprinting Disorders, Bremen, Germany, 12 – 13 March

Anja Helm: "Support of patients' organisations and their networking by EURORDIS"

Inauguration de la plateforme RaDiCo et de ses cohortes (launch of the French national platform RaDiCo – rare diseases cohorts), Paris, France, 4 March

Virginie Hivert represented EURORDIS

Interview of Rosa Sanchez de Vega, former EURORDIS Vice-President and FEDER Chair, by Antonio Armas, Director of Gestión Radio, Madrid, Spain, 19 February

Rosa Sanchez de Vega: Presentation of EURORDIS and European policies in the field of rare diseases

VII Congreso Internacional de Medicamentos Huérfanos y Enfermedades Raras, Sevilla, Spain, 12–14 February

Gema Chicano: « Preguntas y Respuestas sobre privacidad y seguridad desde las asociaciones de pacientes »

International Childhood Cancer Awareness Day (ICCD 2015) : 'Balancing Personal Data Protection and Research Progress: The Case of Childhood Cancer', hosted by MEP Glenis Willmott, at the European Parliament, Brussels, Belgium, 3 February

Flaminia Macchia represented EURORDIS

2nd European Conference on Clinical Research, Paris, France, 2–4 February

Virginie Hivert: "Which of the current drivers for change will be positively influencing patient participation to clinical trials: the view of patient associations"

Findacure: How to Identify Rare Disease Patients, London, UK, 30 January

Marta Campabadal represented EURORDIS.

European Forum for Good Clinical Practice Annual Conference 2015 "How do we improve health without betraying confidentiality within current and upcoming EU Regulations?", Brussels, Belgium, 27 – 28 January

Flaminia Macchia represented EURORDIS.

ACKNOWLEDGEMENTS

EURORDIS would like to thank the following organisations and companies for their financial support in 2015:

Patient Organisations and Public Entities

• • • • •

AFM – Téléthon

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



CNSA

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



European Commission DG Health and Food Safety

- The Operating Grant for year 2015 (EURORDIS_FY2015)
- The EUCERD Joint Action Working for Rare Diseases (EJA)
- RD-ACTION – Rare Diseases Joint Action, Data and policies for Rare Diseases



Co-funded by
the Health Programme
of the European Union

European Commission DG Research and Innovation

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



European Commission Employment and Social Innovation (EaSI) Programme

InnovCare (Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions) project to develop and test an innovative patient-centred approach for social care provision to complex conditions



Pharmaceutical and Biotechnology Sector Companies

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EURORDIS has diversified its pharmaceutical and biotechnology sector companies' sponsorship from 43 to 51 different companies in 2015. EURORDIS believes that diversification of funding is a key success factor to minimise potential conflict of interest with donors. Companies have supported EURORDIS primarily through the EURORDIS Round Table of Companies¹, the EURORDIS Membership Meeting 2015 Madrid, the RareConnect Online Patients Communities², Rare Diseases International³ and the EURORDIS Black Pearl Evening⁴. The breakdown of each company's donations by project is detailed on the EURORDIS website on the "Corporate" tab of the "Financial Information" section.

THE TOP FIVE DONORS

1 SHIRE



2 CELGENE



3 PFIZER



4 GSK



5 BIOMARIN



GENZYME, A SANOFI COMPANY



NOVARTIS



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

² <http://www.rareconnect.org>

³ <http://www.rarediseasesinternational.org>

⁴ <http://www.blackpearl.eurordis.org>







OTHER PHARMACEUTICAL COMPANIES (LISTED ALPHABETICALLY)



OTHER HEALTH SECTOR CORPORATES

Cydan	For EURORDIS Round Table of Companies contribution	
CLINIGEN	For EURORDIS Round Table of Companies contribution and Black Pearl Evening 2015 contribution	
Mapi	For EURORDIS Round Table of Companies contribution	
OpenApp	For EURORDIS Round Table of Companies contribution	
Marc Krueger & Associates, Inc.	For EURORDIS Round Table of Companies contribution and Black Pearl Evening 2015 contribution	

OTHER CONTRIBUTORS

AIPM	Association of International Pharmaceutical Manufacturers for co-funding the Russian version of EURORDIS Website and eNews	
Arès Life Sciences	For Black Pearl Evening 2015 contribution	
Burson-Marsteller	For on-going in-kind contribution of communications advice to build awareness of rare diseases	
DLA Piper UK LLP	For in-kind contribution of legal services	
EFPIA	For co-funding the Innovative Medicines Initiative-Joint Undertaking (IMI-JU) project: European Patients' Academy on Therapeutic Innovation (EUPATI)	
Enfin Bref Production	For in-kind production of the Rare Disease Day 2015 video	
EveryLife Foundation for Rare Diseases	For Black Pearl Evening 2015 contribution	
Mediaplanet	For in-kind contribution of communications services in the context of Rare Disease Day 2015	
Publicis Lifebrands International	For in-kind communications work on the Rare Disease Day International campaign	
SIRIMAGE	In-kind filming services of patient testimonials	

EURORDIS thanks the following members of Rare Diseases International for their voluntary contributions

- Alliance Maladies Rares
- Debra Internatioanl
- Genetic Alliance Australia Ltd
- Hong Kong Alliance for Rare Diseases
- IPOPI
- Retina International





ACTION PLAN 2016	66
GOVERNANCE CHART 2016	79
TEAM CHART 2016	81
BUDGET 2016	82

WORKPLAN 2016



OUR 2016 ACTION PLAN

Advocacy

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ADVOCACY ISSUES

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

ADVOCACY ACTIONS

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

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

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

2 The Commission Expert Group on Cancer Control:

Participation of one EURORDIS patient representative, one alternate and one observer to represent rare cancers in the Commission Expert Group on Cancer Control

3 The EURORDIS Policy Action Group (PAG):

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

- The PAG teams up with other volunteers and EURORDIS staff to provide the patient view on issues dealt with by the two Commission Expert Groups such as: National strategies; national plans; Centres of Expertise and European Reference Networks; Cross-border healthcare; Codification and classification of rare diseases; Guidelines on diagnostics and care;

Gene Testing and Counselling; Information provision to patients and professionals; Access to rare disease therapies and orphan medicinal products; Integration of rare diseases into social policies and services

- The PAG coordinates the patient perspectives within the two expert groups

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

4 2nd Joint Action on Rare Diseases – RD Action: Promoting Implementation of Recommendations on Policy, Information and Data for Rare Diseases (2015–2018)

EURORDIS is involved in two work packages:

- Work Package 2 – Dissemination: As leader of this work package, EURORDIS in collaboration with JA partners, is in charge of the development of the JA dissemination plan. EURORDIS will disseminate EU policy outcomes, monitor progress, promote new strategies and innovative approaches, to integrate all main stakeholders at EU and national levels through: the organisation of the European Conference on Rare Diseases and Orphan Medicinal products (ECRD 2016 Edinburgh) and the coordination of the organisation of up to 30 multi-stakeholder National workshops on specific rare disease policy, organised locally by the national alliances.
 - Work Package 6: EURORDIS will be part of the Consultative Group of RD Action. The participation in the Consultative Group will directly contribute to the development of policy recommendations by the Commission Expert Group.
- 5 EURORDIS will use its channels for disseminating policy outcomes to members through EMM 2016 Edinburgh, internal dissemination, EURORDIS Website, eNews and social media
- 6 Joint Action on Rare Cancers: Participation in ongoing negotiations in the Joint Action on Rare Cancers and involvement in it from 2016

■ Advocate to improve the regulatory process for orphan medicinal products

- Develop EURORDIS position for the public consultation on the Commission Communication on the implementation of the Orphan Medicinal Product Regulation, including specific comments on significant benefit
- Contribute to the review of current initiatives in the regulatory field as undertaken by the Safe and Timely Access to Medicines for Patients (STAMP)
- Contribute to new approaches for the engagement of patients in the benefit/risk evaluation, taking into consideration patient preferences (responding to the IMI2 call on Patient Preferences Elicitation and collaborating with EMA)
- Promote advanced approaches, policies and pilots: EMA scientific advice involving patient representatives and medical experts, progressive

patient access, Medicines Adaptive Pathways to patients (IMI), compassionate use and early access programmes, shortages, and off-label use of medicines in rare indications

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

- Support the initiatives of MEPs for the re-submission of the revision of the Transparency Directive 89/105/EEC, for greater transparency on the decision making for the reimbursement/coverage decision of medicines
- Develop and adopt EURORDIS position paper on 'Improving Access to Orphan Medicinal Products in Europe and its sustainability for society'
- Promote and take an active part in the dialogue on Medicine Alternative Development Pathways (MAPPs) with EFPIA and EMA and contribute actively through IMI ADAPT-SMART project.
- Contribute to developing at EU level approaches on methodology and criteria for the Health Technology Assessment of the Effectiveness or Relative Effectiveness appropriate for evaluation of the value of Orphan Drugs (EUnetHTA)
- Promote new approaches, policies and pilots: very early dialogue/ scoping / de-Risking before proof of concept, participation of Payers in the scientific advice of EMA and HTA.
- Support the Mechanism of Coordinated Access (MoCA) to Orphan Drugs, developed within the EU Stakeholders Forum on Corporate Responsibility in Pharmaceuticals, promoting pan-European collaboration between EU Member States on Orphan Drugs based on a common approach to the value of new medicines, pricing, volume and post-Marketing Authorisation data generation. Contribute to the early dialogue between Payers and Companies through MOCA pilots involving patient representatives.
- Promote a type of negotiation with National Competent Authorities for Pricing and Reimbursement
- Promote reflections on Joint Procurement, Joint Purchasing, Managed Entry Agreement, Differential Pricing, Parallel Trade.
- Promote these innovative approaches in Member States and national plans on rare diseases and increase direct cooperation with leaders from the rare disease therapies sector
- Prepare and widely disseminate the annual EURORDIS Access Campaign report based on the ongoing survey
- In emergency situations, take action to support patients' access to therapies

■ Advocate to improve access to medical devices for patients with rare diseases

- Analyse the impact of the European legislation on medical devices

- Promote the importance of involving patients in the evaluation process by notifying bodies

■ Promote the sustainability of rare diseases as a policy and budget priority in the EU programmes for the period 2015– 2020:

- As a research priority in the Horizon 2020 work programme 2018–2019 and in the Innovative Medicines Initiatives
- As a public health priority in the 3rd EU Public Health Programme 'Health for Growth' & Annual Work Programmes and advocate on the rare disease community's priorities
- EU Directive on Patient's Rights to Cross-border Healthcare:
- Collecting patient feedback, monitoring the implementation, disseminating information and promoting access to national contact points
- Actions related to the European Reference Networks including advocating for funding and participation in the Task Force on the interoperable data sharing in the framework of the operation of ERNs

■ Advocate in support of rare disease research:

- Engage EURORDIS in European research infrastructures and research, policy debates
- Accelerate the establishment of the Joint Research Centre European Platform of rare disease registries
- Promote research objectives in Executive and Scientific Committees and Task Forces of IRDiRC (see page ...)
- Elaborate and promote the objective of Rare Disease Clinical Research Networks, articulated with ERNs
- Define the patient position through the RDConnect project and the Rare Barometer programme; contribute to the debate and adoption of the Data Regulation.

■ Contribute to the reflection on patient access and an adequate economic model for advanced therapies, i.e. gene therapy, and cell therapies

■ Advocate to improve quality and access to rare disease diagnosis:

- Contribute to the work carried out within RD Action on genetic testing, counselling, screening and next generation sequencing
- Elaborate EURORDIS' position
- Define EURORDIS' position on companion diagnostics and precision medicine
- Monitor the finalisation of the in vitro diagnostics legislation

■ Monitor the adoption of the CEGRD recommendations on integration of rare diseases into social services and policy (see ...):

- Continue supporting the integration of rare diseases into social services and policy through RD–Action
- Map the emerging topics and areas of work and relevant stakeholders within the area of social services to define future action

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

- Rare Diseases International (RDI), a network of rare disease patient organisations: organisation of an RDI meeting back to back with ECRD 2016 Edinburgh and election of the RDI governance group, the Council of RDI; further recruitment of members and promotion of the initiative; involvement in ICORD conference.
- The promotion of the joint declaration 'Rare Diseases: an International Public Health Challenge' by major rare disease patient organisations
- Elaboration of a strategy to integrate into the UN System: ECOSOC, WHO, and Council of Europe and the establishment of the UN Committee on RDs at the UN Headquarters in New York in 2016, involving RDI and international rare disease stakeholders
- Signing of MoUs with international patient organisations
- The international development of Rare Disease Day
- Continued active participation in the International Rare Disease Research Consortium

■ Cross-cutting advocacy activities:

- European Year for Rare Diseases 2019: Continue advocacy campaign to build a coalition of policy leaders and stakeholders supporting the EYRD 2019, establishing an Activity Road Map
- Foresight Study 2030: Advocate to secure funding for the Foresight Study Rare Diseases 2030 to identify long-term policy scenarios and engage in public debate with stakeholders

■ Gathering patient knowledge for evidence-based advocacy:

- EURORDIS Rare Barometer programme: The EURORDIS Rare Barometer programme is a patient derived knowledge survey programme launched in 2015. Specific topics covered in 2016 include perspectives of rare disease patients about genome sequencing; social and daily life challenges faced by people living with a rare disease and their families; priorities in terms of awareness. In addition, the Rare Barometer programme will develop quality of life measurement in collaboration with RareConnect. A new tool comprising of a contact database of rare disease patients and patients representatives (launched as Rare Barometer Voices) will enable EURORDIS to carry out quantitative studies on a regular basis.

Information & Networking

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COMMUNITY BUILDING

- **Maintain the EURORDIS Membership over 650 members and ensure regular interaction**
- **Maintain implementation of process of regular membership reassessment, as established in 2013**
- **Recruitment of members at large, particularly in all EU member states, acceding and candidate EU Member States, rare cancer groups and European Federations**
- **Organise EURORDIS Membership Meeting 2016 Edinburgh on 26 May, including General Assembly and 4 capacity-building workshops**
- **Build capacities of the European network of 32 national alliances through:**
 - Promoting greater convergence and collaboration between national alliances, as well as between national alliances and EURORDIS, through implementation of the 'Common Goals & Mutual Commitments between National Alliances in Europe and EURORDIS: An agenda between 2014 & 2020'
 - Sharing information, experience, guidance and common actions in national plans, Rare Disease Day, cross-border healthcare, access to medicines and evaluation of Centres of Expertise
 - Organising two workshops of the Council of National Alliances
 - Organising regular webinars on specific topics
 - Maintaining direct interaction or visits between EURORDIS and national alliances
 - Encouraging contact between patient organisations in same countries and the creation of new national rare disease alliances where there are none yet
- **Build capacities of the European network of 50 disease-specific European / International Federations & Networks through:**
 - Sharing information, experience, good practices and guidance – integration of RareTogether! Website in Council of European Federations (CEF) section – between European Federations and common actions in European Reference Networks, Rare Disease Day, drug development & interaction with EMA & access to medicines & patients reporting of adverse events, cross-border healthcare, social policy & services, and Online Patient Communities
 - Providing regular advice to European Federations & Networks
 - Organising a two day workshop of the Council of European Federations, including a one day training course
 - Expanding the EURORDIS Programme to Support

European Federations & Networks with money for their governance meetings, membership meetings, first European conferences on their disease

- **Structure EURORDIS Membership into 20–25 rare disease groupings aligned with the rare disease grouping for European Reference Networks enabling relevant members in each group to elect their representatives in a EURORDIS Patient Advocacy Group (EPAG). These EPAGs will be the prime contact to develop ERN applications and engage into the ERN activities. The objective for 2016 is to establish around 10 EPAGs and the others progressively over the next two years.**
- **Outreach to patient groups in Central and Eastern Europe, Balkans, Russia and Caucasia, support of their actions to raise public awareness and promote policy on rare diseases and promote the creation of national alliances:**
 - Maintain and expand EURORDIS Website, eNews, Member News and main documents available in Russian; develop RareConnect in Russian and Serbo-Croatian.
 - Provide official EURORDIS endorsement/ promote and take part in national conferences across Central & Eastern Europe on request of our members
 - Support Russian and Caucasian endeavours in raising awareness of rare diseases in their regions through support of their national conferences or EURORDIS–EUROPLAN conferences on national RD strategies as well as Rare Disease Day
 - Provide access to EURORDIS Patient Advocate Fellowships Programmes for the EURORDIS Rare Disease Day Event, the EURORDIS Membership Meeting 2016 Edinburgh and to the EURORDIS Summer School 2016 Barcelona
- **Support EURORDIS volunteers' involvement through:**
 - Strengthening the identification, recruitment and support of volunteers to be increasingly involved in EURORDIS activities based on the EURORDIS Charter of Volunteers, participating in NGO partnerships and representing patients in the European Commission and EMA working groups and committees
 - Creating four EURORDIS Thematic Working Groups of volunteers on the following issues: national plans, social services, registries, best practice clinical guidelines
 - Briefing on key topics, access to shared reference documents and public presentations
 - Maintaining and expanding the volunteer section on the EURORDIS website
- **Expand RareConnect, the global social network of online patients communities:**
 - Implement the RareConnect Action Plan 2014–2016

and Strategy for 2020

- Adjust governance and management structure to new development phase in particular with the participation of more national alliances
- Maintain and support the 80 existing online patient communities in conjunction with the over 660 patient groups and over 250 volunteer moderators involved
- Launch 20–25 new online patient communities, adapting the tool to the needs of each specific community in conjunction with relevant existing patient groups for these rare diseases
- Successfully integrate 4 new community managers into the team based in the Western Balkan region together with the Croatian and Serbian National Alliances to support the scaling up of RareConnect's outreach and support activities
- Increase outreach to patient organisations in Italy, Germany, Balkan region, Russia, Belarussia and Ukraine based on the linguistic competency of the new team of community managers in the Balkans
- Add additional languages to RareConnect such as Serbo-Croatian
- Launch 5–10 Featured RareConnect Discussion Groups. Featured Discussion Groups will moderated online conversations on issues relating to advocacy or other strategic EURORDIS activities where EURORDIS volunteers or staff will play a role in driving the content such as Rare Diseases International, social services, best practices and others
- Develop RareConnect as a platform to perform surveys in conjunction with the EURORDIS Rare Barometer programme
- Continue to implement technical and user-experience improvements to the platform including an improved mobile interface which will enable visitors to participate on a more regular basis and accomplish priority tasks from their mobile device
- Launch a new feature within RareConnect which will allow members of RareConnect communities to view on a map the location of other members (based on informed consent)
- Develop the capacity of RareConnect to attract financial support from its community of members
- Develop a communication plan for RareConnect based on social media campaigns in Italian, German, Serbo-Croatian, and Russian

INFORMING & RAISING AWARENESS

■ Organise Rare Disease Day 2016 around the theme “Patient Voice” and with the slogan “Join us in making the voice of rare diseases heard” under the new 4 year overarching theme of “Patient Involvement”

- Develop partnership with marketing and communications agency PUBLICIS LifeBrands International, as established in 2015. PUBLICIS works with EURORDIS to:
- Produce new Rare Disease Day logo options with

full style guide for use by all participating patient organisations

- Updating look of RareDiseaseDay.org with completely new and improved design and user experience
- Produce Rare Disease Day 2016 video concept and support production of video. Following the wide success of previous years, the video will again be translated into 25+ languages to be disseminated through social media and eurordis.org
- Produce Rare Disease Day poster concept and creative design based on video concept
- Continue to expand the Join and Raise Hands campaign and tie it in to social media campaign
- News about the worldwide campaign disseminated in mass emailing: A digest of social media and country pages information disseminated in the weeks leading up to 29 February

■ Organise RDD 2016 events in Brussels:

- The EURORDIS Awards ceremony 2016 with a new format: EURORDIS has crafted a new format for 2016 Awards with an official Awards Ceremony to be live video-streamed before the Black Pearl Evening.
- The opening session of the EURORDIS Round Table of Companies on access to rare disease therapies will be live streamed to the public to mark the occasion of RDD 2016. Speakers include the EU Commissioner for Health and MEPs

■ Plan Rare Disease Day 2017: marketing and communication plan, info pack, video, poster & slogan, website, patient testimonies, social media and Rare Disease Day Event in Brussels

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

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

■ Maintain content quality and information architecture of EURORDIS Website:

- Centered on target audiences: patient organisations and advocates, patient and families, other stakeholders (secondary)
- Maintain navigation and user-friendliness of website

- Maintain quality, updated information in seven languages (EN, FR, DE, ES, IT, PT, RU) in all sections
- Further develop content and facilitate access via three focal points: the core EURORDIS.org website, its satellites (eNews, Rare Disease Day, RareConnect, Help Lines) and EURORDIS Social media (Facebook, Twitter, Flickr, YouTube)
- Continue to strengthen the EURORDIS social media channels by increasing interactive content by working with the fundraising team, the Rare Barometer Programme and the therapeutic team to disseminate the most up-to-date and relevant information possible to patient advocates
- Develop new web sections on key advocacy priorities, EURORDIS programmes and projects
- Maintain social media and patient empowerment webinar series to moderators and patient groups

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

- Make use of pre-existing recorded material, edit and upload to EURORDIS TV
- Edit/ produce videos using content recording at EMM on a range of various advocacy topics including European Reference Networks and 'What does Rare Disease Day mean to you?', for example
- Promote EURORDIS TV through "Video of the week" feature included in EURORDIS eNews and on homepage of EURORDIS.ORG
- Develop search feature on EURORDIS TV to allow users to find videos related to a specific search term

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

■ Publish monthly Member News (sent to all EURORDIS member organisations only, new as of 2015)

■ Present Photo Contest 2015 at EURORDIS membership meeting 2016 Edinburgh with continued and increased presence on Instagram

INFORMATION SERVICES TO PATIENTS

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

- Develop the European Network of Rare Disease Help Lines:
 - Governance & business meeting, develop membership base
 - Explore new financial tools for the help lines (Structural Funds)
 - Develop new trainings for help lines respondents ("How to take a call")
 - Submit and publish an article on the Caller Profile Analysis in a scientific journal
 - Organise the annual caller profile analysis
 - Engage and train help lines on reporting adverse events of medicines used in all rare disease treatments (whether orphan or not, and off-label)
 - Link to national plans
 - Link to ERNs

Health Policy & Healthcare Services

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PROMOTING RARE DISEASE HEALTH POLICY DEVELOPMENT

■ Organise the 8th European Conference on Rare Diseases and Orphan Products— ECRD 2016 Edinburgh on 26 – 28 May, 2016:

- Prepare the Programme and the Programme Committee:
 1. Coordinate the multiple partnerships with DIA, Genetic Alliance UK, EMA (COMP), FDA (OOPD), The Scottish Government, HOPE, Orphanet, ESHG, EFPIA–EuropaBio and EUCOPE
 2. Coordinate the multiple partnerships with

associate partners comprising over 20 rare disease National Alliances, research institutions, projects and industry trade associations

3. Create Programme Committee and Develop Programme with 7 parallel themes.
4. Promote ECRD & registrations in six languages (EN, FR, ES, DE, RU, IT). Plan Call for Posters, renew EURORDIS Patient Advocates Fellowship Programme, organise logistics of sessions selected to be simultaneously translated, develop communication around the event, outreach to medical journals to encourage higher attendance from healthcare professionals

- Plan satellite workshops for partners eg. Orphanet, IRDiRC ExCom, etc.

Support the national alliances in their action in advancing national rare disease policy

EURORDIS Policy Fact Sheets for Patient Advocates: Development of new policy fact sheets on: (possible subjects depending on priority advocacy actions in 2016)

Targeted communication

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

PUTTING RARE DISEASE PATIENTS AT THE HEART OF THE HEALTHCARE SYSTEM

Support the implementation of the policy on European Reference Networks (ERN) and Centres of Expertise (CoE) toward a patient-centric approach aiming at clinical excellence and best possible patient health outcomes.

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

- Promote the long-term EURORDIS strategic vision on ERN
- Work with EURORDIS' partners under the Rare Disease Joint Action to develop and shape emerging policy areas for ERNs, including for eHealth, IT, data sharing, clinical guideline development, clinical outcome and research
- Connecting healthcare and research under ERNs and research networks or research infrastructure across the translational research pathway, including undiagnosed disease networks
- Disseminate the Declaration of Common Principles on CoE & ERN

Build the capacities of the EURORDIS membership and their readiness to support European Reference Networks

- Continue to take action to help prepare rare disease patient groups for the launch of the Call for Proposals for ERN
- Support the implementation of grouping rare diseases, to take optimal advantage of the limited number of future ERNs while ensuring that on in the long term every rare disease patient has a home, a healthcare pathway

- Initiate EURORDIS Patient Advocacy Groups (EPAG) actions to support collaboration between European federations, patient organisations or online communities and ERNs in key areas such as: governance of ERNs, disease registries, biological repositories, clinical trials, treatment protocol trials, standards of diagnosis & care, information to patients and outreach to patients

- Organise a series of workshops at the ECRD 2016 Edinburgh dedicated to ERN & CoE

- Develop EURORDIS website section and produce updated fact sheet on ERN

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

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

- Produce the final Assessment Manual and Toolbox for Applicants and for Independent Assessment Body(s) and the Operational Criteria
- Support and train the Independent Assessment Body(s) to implement the assessment model, manual and criteria prior to the first call for applications

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

- Partnering in the EU project RARE-BestPractices, a platform for sharing best practices for management of rare diseases, in order to:
 1. Build capacities of rare disease patient organisations and of people living with a rare disease on the importance, use and benefits of best practice guidelines. Do this through training activities targeted at patient advocates and dissemination of information and project outcomes (WP1)
 2. Establish a EURORDIS working group on best clinical practices
 3. Contribute to the development and the implementation of a core methodology on best practice guidelines for rare diseases (WP3)
 4. Ensure a strong interaction and coordination of the RARE-BestPractices project with other initiatives such as EUROPLAN II, RD-Connect and IRDiRC and contribute to the dissemination of the RARE-BestPractices project outcomes (WP1,6):
- How to better use scientific progress in biomedical research to translate results into tangible benefits

for rare disease patients and to offer high quality information to patients and healthcare professionals;

- How to promote and support a consistent level of healthcare services for rare disease patients in the EU while implementing the EU Directive on Patients' Rights in Cross-border Healthcare

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

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

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

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

INTEGRATING RARE DISEASES INTO SOCIAL SERVICES AND SOCIAL POLICY

Voicing social and daily life challenges faced by people living with a rare disease and their families

- Performing a survey and a qualitative study on social and daily life challenges faced by people living with a rare disease and their families, via EURORDIS Rare Barometer programme and in coordination with the INNOVCare project
- Elaboration of the EURORDIS position paper on the social challenges faced by people living with a rare disease/their relatives based on the results of the Rare Barometer survey and in with the support of EURORDIS Social Policy Advisory Group

Promote integration of rare diseases into social services and social policy:

- EURORDIS continues to support the elaboration of the CEGRD recommendations on integration of rare diseases into social services and policies, initiated during the EUCERD Joint Action (EJA) (2012-2015) and followed up within the RD-Action (adoption expected for the early 2016). The recommendations will be presented at ECRD 2016 Edinburgh and broadly disseminated via EURORDIS website, social media and events
- Promote integrated care and patient-centred services, namely via: 1) the INNOVCare project, which proposes the implementation and evaluation of the socio-economic impact and cost-benefit of a care pathway including resource centres and case managers; 2) the publication of a rare disease chapter within a Handbook of Integrated Care (spring 2016), elaborated in cooperation with EURORDIS Social Policy Advisory Group
- Study of social care pathways in a selection of EU Member States and comparative analysis between patients'/families' needs and the existing care systems to identify gaps/good practices and highlight points for improvement in regards to the integration of people with a rare disease into social services and policies
- Support the elaboration of good practices for social care in rare diseases via the European Network of Resource Centres for Rare Diseases to be created within the INNOVCare project
- Continue to disseminate the guiding principles and case studies developed within the EJA. This includes the map of existing specialised social services; case studies on experienced services; guiding principles for specialised social services and for training of specialised social service providers
- Promote training for social services providers by organising: 1) presentations/workshops at international social work conferences; 2) webinars for patients (how to talk to your social worker) and professionals, in cooperation with the International Federation of Social Workers Europe and with resource centres

- Dissemination of information on social innovation opportunities for rare diseases and for patient organisations, via presentations at EMM (tbc) and ECRD 2016 Edinburgh
 - Follow-up projects on social research, social innovation and integrated care and dissemination of relevant findings
 - Advocate for the promotion of EU and national policies for social research and quality of life studies
 - Networking and establishment of partnerships with other organisations and professional associations active in the social field at the EU level
- Stimulate the development and the improvement of access to specialised social services:**
- Maintain and continue to disseminate EURORDIS website sections containing the map of social services and information on respite care services, therapeutic recreation programmes, adapted housing and resource centres
 - Facilitate the exchange of experiences and good practices between resource centres and case managers for rare diseases, via the European Network of Resource Centres and INNOVcare project
 - Advocate for respite care services, therapeutic recreation programmes, adapted housing services and resource centres through the dissemination of policy fact sheets
 - Encourage EURORDIS staff, volunteers and members to volunteer at summer camps

Research, Drugs & Therapies

INVOLVEMENT WITH RESEARCH PUBLICATIONS

■ Member of the Editorial Board of OrphaNews Europe

■ Member of the Editorial Board of the Journal Expert Opinion on Orphan Drugs

■ Link with Orphanet Journal of Rare Diseases

SHAPING AND PROMOTING RESEARCH POLICY SUPPORTING CLINICAL RESEARCH

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

- Dissemination of EURORDIS policy fact sheets and reference papers on research
- Participation in and support of the development of the International Rare Disease Research Consortium (IRDiRC), of which EURORDIS is a member of the Executive Committee, a member and Chair of the Therapies Scientific Committee and involved in several Task Forces (Patient-Centered Outcome Measures, Patient Engagement, Repurposing, etc.). Take part in all meetings and activities
- Increased participation in the ERA-Net project E-Rare involving National Funding agencies
- Participation in research policy activities related to national plans for rare diseases (EUROPLAN)

■ Promote patient rare disease registries and data collection

- Engage in the development of the EU platform on Rare Disease Registries at the Commission Joint Research Centre through its Advisory Board, based on the EPIRARE Book and EURORDIS 10 key

principles, EUCERD Recommendation on registries and patient's preferred policy scenario on Registries, while making the link with the servicing of European Reference Networks, as well as planning of healthcare services & social services, clinical practices, medicine development

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

- Engage EURORDIS in the Horizon 2020 project aiming at streamlining the services offered by the biomedical European research infrastructures.
- RD-Connect, an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research. EURORDIS ensures the involvement of patient organisations in capacity building on registries, biobanking and omics and directly contributes to developing registry, biobanking and omics infrastructures and their integration. EURORDIS ensures a strong interaction and coordination of the RD-CONNECT network with other initiatives within and beyond Europe, as well as the dissemination of the RD-CONNECT project outcomes at the international level (WP1, WP7 and WP8).
- BBMRI Consortium, ensuring patient representation in its governance
- EuroBioBank, promoting specific agreements between patient organisations and EuroBioBank by providing information, template agreements and advice

SUPPORTING CLINICAL RESEARCH

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

- Participate as Partner in the IMI Consortium co-led by TI Pharma, AstraZeneca and BMS, scientifically supported by the EMA 'Accelerated Development of Appropriate Patient Therapies: a Sustainable Multi-stakeholder Approach from Research to Treatment', a support action involving EURORDIS, European Patient Forum, EUnetHTA, NICE, HAS, ZIN, CASMI, TI Pharma / Esher and NEWDIGS. EURORDIS, along with EPF is representing the patient perspectives in the project and is leader of the task 2.03 aiming at establishing the entry criteria to the Medicine Adaptive Pathway to Patients

- Bring these innovative policy approaches and activities to the IRDIRC

Promote adoption of the EURORDIS Charter for Collaboration between Patient Organisations and Sponsors of Rare Disease Clinical Trials; facilitate the implementation of the Charter with the support of a Mentor

Support specific actions in Rare Cancers:

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

GAINING KNOWLEDGE ON RARE DISEASE TREATMENTS

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

- Maintain relationship with the International Society of Pharmaco-economics and Outcomes research and co-chair its Patient-Centred Special Interest Group

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

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

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

- Launch new survey 'Tell us how you take your treatment', including off-label use in conjunction with the EURORDIS Rare Barometer Programme
- Prepare possible extension of the survey on off-label use in rare diseases to all other EU Member States
- Continue the activities in parallel to the workplan of the PCWP
- Liaise with EMA and heads of medical agencies for more transparency of compassionate use programmes. Support the creation of a compassionate use facilitation group. Propose guidelines to companies and Member States for the organisation of compassionate use programmes. Publish a Q&A on the subject
- Maintain pages on EURORDIS website containing information on compassionate use programmes with links to the competent department in national agencies
- Maintain a sub-group of volunteers on specific needs for blind and vision-impaired patients
- Create online video tutorials on how to search information on main websites (EU portal on CT, database on suspected adverse drug reactions, EudraPHARM)

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

PROMOTING DRUG DEVELOPMENT et ACCESS TO TREATMENTS

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

- Participate in the EMA Committee for Orphan Medicinal Products (COMP) with one representative member (and vice-Chair of the Committee) and two observers
- Contribute to the EMA Pharmacovigilance and Risk Assessment Committee (PRAC) as external experts for rare disease therapies
- Participate in the EMA Paediatric Committee (PDCO) with one representative member and one alternate
- Participate in the EMA Committee for Advanced Therapies (CAT) with one representative member and one observer
- Explore methods for patients to contribute to or witness the benefit/risk evaluation in the EMA Committee for Human Medicinal Products (CHMP), contribute to the procedure in place for oral explanation with the applicant, contribute to other consultancy methods (writing procedures, questions from the rapporteurs...) and propose other modalities
- Participate in EMA Human Scientific Committees' Working Party with Patients' and Consumers' Organisations (more commonly known as the Patients' and Consumers' Working Party or PCWP), with one representative member and one alternate. Mentoring patients on the CHMP procedure for oral explanations used for risk/ benefit evaluation for oral explanation with the applicant / authorisation holder
- Participate in EMA user testing group of the European Web Portal on Clinical Trials
- Launch an analysis of the European Database for Suspected Adverse Drug Reactions (ADR) (EudraVigilance) together with the EMA, including orphan medicinal products for which patients are reporting suspected ADRs
- Consultation on new EMA intranet for delegates
- Identify and support patient experts for protocol assistance/scientific advice at SAWP (Scientific Advice Working Party), and for Scientific Advisory Groups of CHMP
- Support EURORDIS patient representatives in EMA Scientific Committees and Working Parties with the EURORDIS Therapeutic Action Group (TAG) via monthly conference calls and sharing information, agendas, reports, providing mutual support and by discussing main issues. The TAG also includes EMA patient representatives which are not representing EURORDIS on these Committees and Working Parties, no matter whether they are EURORDIS members or not.
- Participation of patients in EMA multi-HTA parallel scientific advice and other HTA early dialogues as proposed by EUnetHTA Joint Action
- Participation of patients to the pilots of the MoCA initiative (Mechanisms of Coordinated Access)
- Review all orphan drug designation applications and protocol assistance dossiers. Review of designation criteria at the time of marketing authorisation and reports on significant benefit, paediatric investigation plans for rare diseases, including waivers and deferrals and advanced therapy (gene, cell and tissue engineering) applications
- Review and validate all public Information on orphan medicinal products disseminated by EMA at the time of designation (PSOs) and marketing authorisation (European Public Assessment Reports summaries, Package Leaflets, Significant Benefit Public Reports)
- Take action to enhance collaboration between EMA and FDA on rare therapy development beyond orphan drug designation – Participation to monthly conference calls between FDA and EMA orphan offices.

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

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

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

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

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

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

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

- Organise EURORDIS ExPRESS Summer School 2016 in Barcelona in June with 25–30 new patient advocates representing a diversity of diseases and geographical locations
- Collaborate with COST to incorporate 10–15 researchers in this new format of the EURORDIS Summer School
- Collaborate with ECRIN to incorporate 10 non-rare disease patient representatives from the therapeutic indication areas of nutrition and medical devices, in addition to rare diseases
- Continue to improve and contribute to the Training Resources section of the EURORDIS website that currently provides access to all presentations from the Summer School in downloadable pdf form, as well as video recordings of the speakers. Online training tools include a preparatory section for the Summer School and the e-learning platform for continued evaluation of learning. This section has been entirely revamped in 2015 to be more user-friendly. Webinars are also available.
- Put emphasis on strengthening links with the Summer School/ExPRESS alumni
- Take part in the DIA EuroMeeting 2016 in April with speakers, session chairs, contribution to programme committee, a DIA Patient Fellowship Programme for approximately 40 fellows and a patient fellows booth
- Empower our volunteers via regular information and monthly therapeutic activity Reports, call for volunteers, support of Drug Information Transparency & Access (DITA) Task Force with more volunteers and organisation of one or two workshops of the DITA Task Force annually. Channels

to spread this information are going to be improved so that news are more accessible and/or more targeted according to the topics and the interested audience.

- Participate in the 'European Patients' Academy on Therapeutic Innovation' (EUPATI) from 2012 to 2016, an IMI-funded project gathering a consortium led by European Patients' Forum (EPF) with other European umbrella patient organisations
- Creation of material for training of the different audiences of the EUPATI project. Training material will be freely available on the EUPATI website.
- EURORDIS is also responsible for the design, organisation and delivery of the EUPATI face-to-face events (2 x 2 events of one week-duration)

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

- EURORDIS Round Table of Companies: consolidate membership and organise two workshops in 2016. The February ERTC will exceptionally have a new format as a Multi-stakeholder Symposium on "Improving Patient Access to Rare Disease Therapies: Value Determination, Appraisal, Pricing & Reimbursement" dedicated to Adaptive HTA, Value Determination and Assessment, link between value and price, new approach to price and national healthcare budget impact for patient access and affordability, sustainability for society, and attractiveness for investors. A second ERTC workshop will be held in September in Barcelona.
- Expand direct dialogue with pharmaceutical and biotech companies
- Strengthen the dialogue with EFPIA-EuropaBio Task Force on Orphan Drugs

Cross-Cutting Priorities

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Implement EURORDIS Strategy 2015–2020:

- Improve planning and anticipation of major EURORDIS activities such as ECRDs, Membership Meeting, Rare Disease Day, EURORDIS Round Table of Companies Workshops, RareConnect, EURORDIS Rare Barometer, major advocacy campaign and new projects
- Continue collection of EURORDIS Indicators and analysis

Develop EURORDIS strategic partnerships:

- Maintain strategic partnership with NORD (USA), renewed for 2014–2019
- Maintain the partnership with CORD (Canada), the Japanese Patients Association – JPA (Japan), Rare Voices Australia (RVA), the Russian Patients Union (RPU) and finalise partnership with the Chinese

Organisation for Rare Diseases, in addition to being open to new partnerships with other international patient organisations

- Develop strategic partnership with ORPHANET
- Maintain partnership with DIA-Europe
- Implement strategic partnership with the European Society for Human Genetics, EuroGentest, International Society of Social Workers, European Institute Women's Health, International Society For Pharmacoeconomics and Outcomes Research ISPOR, Health Technology Assessment International (HTAi), European Society of Medical Oncology (ESMO), European Hospital and Healthcare Federation (HOPE), etc
- Consolidate partnership with the members of the EURORDIS Round Table of Companies

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

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

| Increase EC support to EURORDIS activities through:

- | Application 2016 for Operating Grant Specific Grant Agreement 2017
- | Application for the 1st Joint Action of the Commission Expert Group on Rare Cancers
- | Study feasibility and apply for grant application to other programmes in other DGs in support of activities foreseen in EURORDIS Strategy 2016
- | Application for patient-driven health research project at IMI 2

| Develop and diversify private funding:

- | Maintain overall support level from industry donors spread between 45 companies and a variety of activities within the confines of the EURORDIS Policy of Relationship with Commercial Companies, EMA Policy on Prevention of Conflict of Interest and EAHC rules
- | Engage corporate and foundation donors beyond the pharmaceutical industry to support EURORDIS projects & actions. Priority: EURORDIS Membership Meeting; the website, eNews, eLearning, EURORDIS TV and InfoHub, all in seven languages; the RareConnect project and social media; support to European federations & networks; EURORDIS Rare Barometer pilot; and Rare Diseases International
- | Organise the EURORDIS Black Pearl Evening for Rare Disease Day 2016 in Brussels and plan event for 2017.
- | Plan the organisation of a Rare Elegance event, in celebration of Audrey Hepburn a tribute to elegance and generosity, linking to the rare disease that Audrey Hepburn suffered from and her charitable care for vulnerable people and children
- | Maintain the EURORDIS International Circle of Ambassadors, bringing together 6 – 8 community leaders from Europe and North America

- | Seek in-kind contributions from individuals and companies outside the health sector to help meet objectives for EURORDIS projects and actions, (ex: communications consulting, tools).

| Implement the Staff Strategy & Organisation & Evolution

- | Maintain organisation in 4 Units: Governance, Membership & European and International Public Affairs; Communications & Resource Development; Operations; Finance & Support Services
- | Create the position of Patient Advocacy Groups Manager
- | Create the position of Russian Outreach & Translations Manager
- | Create the position of Public Affairs Junior Manager
- | Create the position of Junior Events Manager
- | Create the position of Social Policy Senior Advisor
- | Maintain the internal coordination processes: operating grant steering committee meetings (4 per year), all staff Monday meetings (2 times a month), finance meeting (monthly), resource development meeting (quarterly & mini monthly), advocacy & strategy meetings (monthly), editorial meetings (monthly) and management meetings (monthly)

| Seek alternative human resources:

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

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

- | Maintain decentralised structure with offices in Paris (main office), Brussels (European public affairs), Barcelona (web communications & RareConnect), Zagreb and Belgrade (RareConnect Teams in Serbia and Croatia), Moscow (Russian outreach & translation) and a presence in London (EMA) and Geneva (Rare Diseases International), with integrated operations through work processes, IT standards/ intranet, voice, data and internet communication
- | Reinforce the Barcelona presence with a new contracted office space
- | IT support: equipment, services, virtual office open to volunteers
- | EURORDIS contact database management fully operational
- | EURORDIS funding database

GOVERNANCE CHART 2016

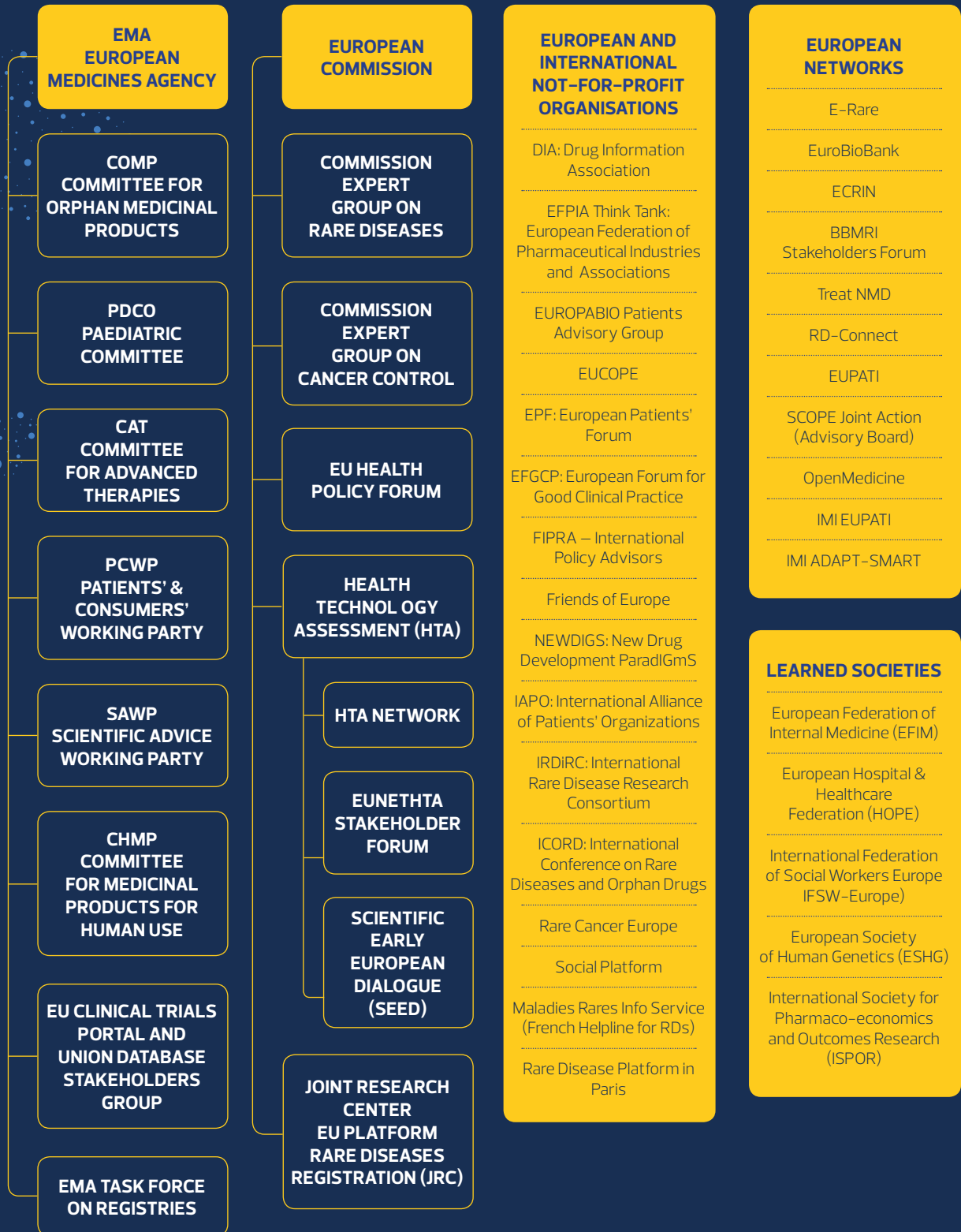


EURORDIS
Rare Diseases Europe



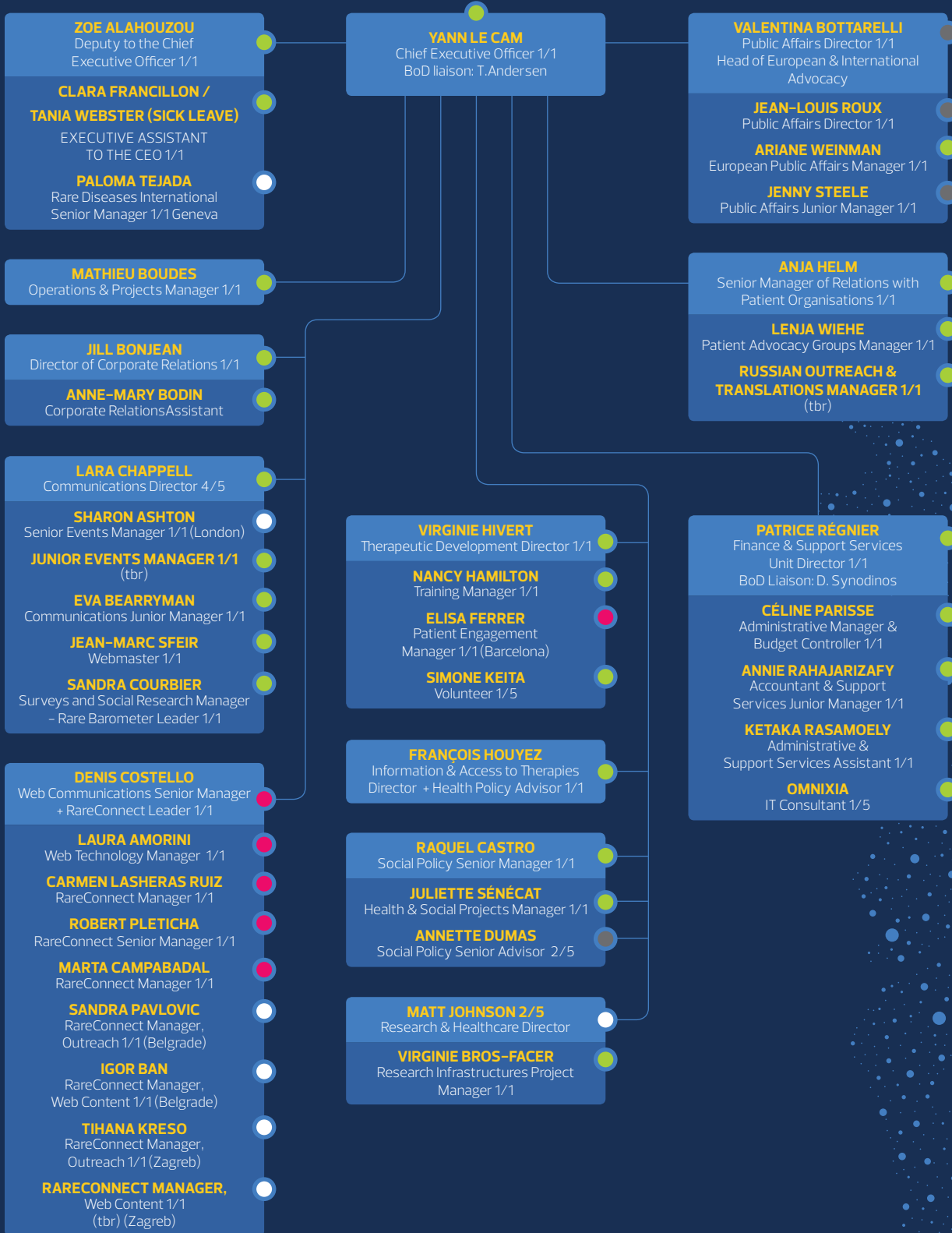
EXTERNAL REPRESENTATION

CHART²⁰¹⁶



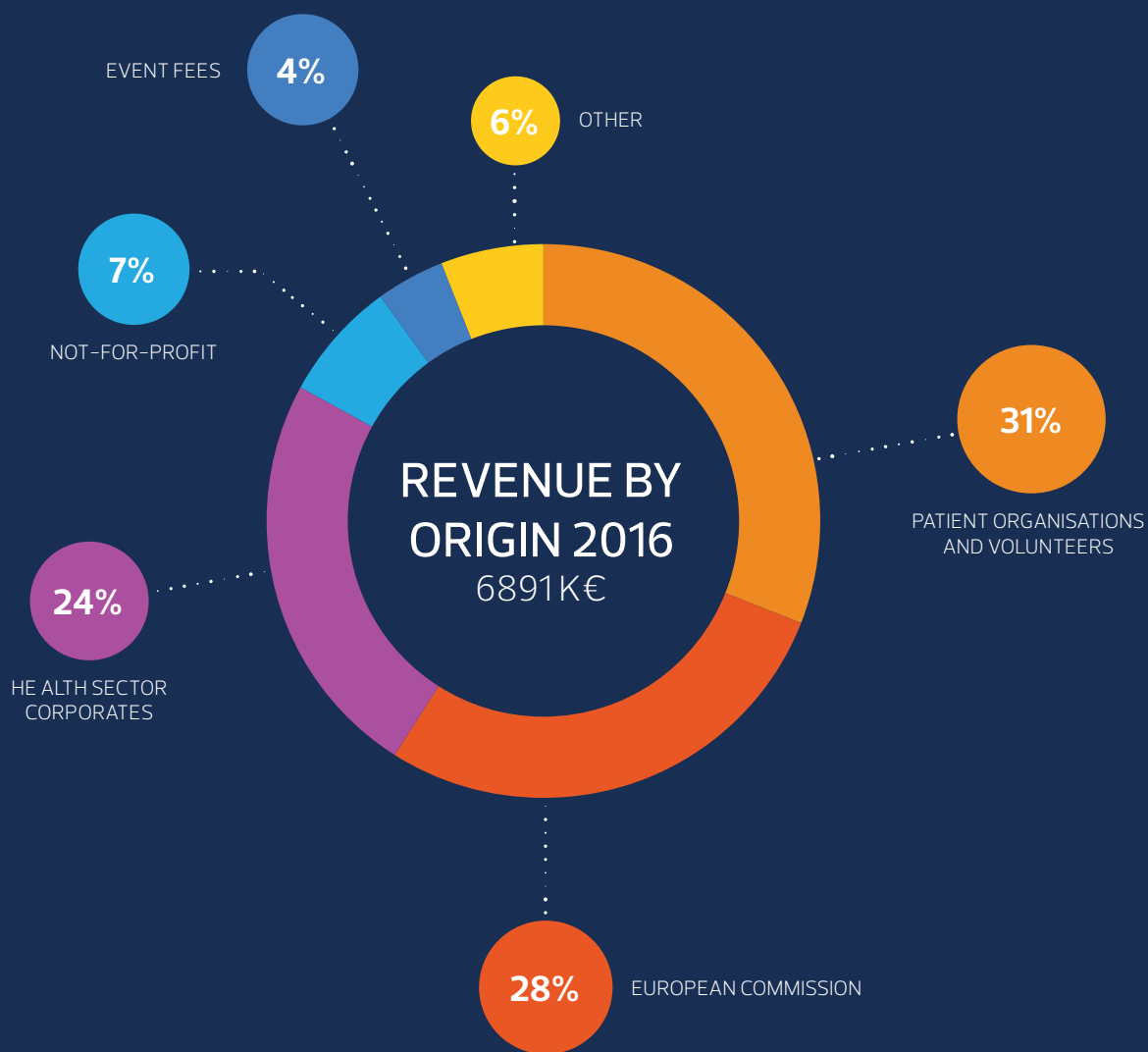
TEAM CHART 2016

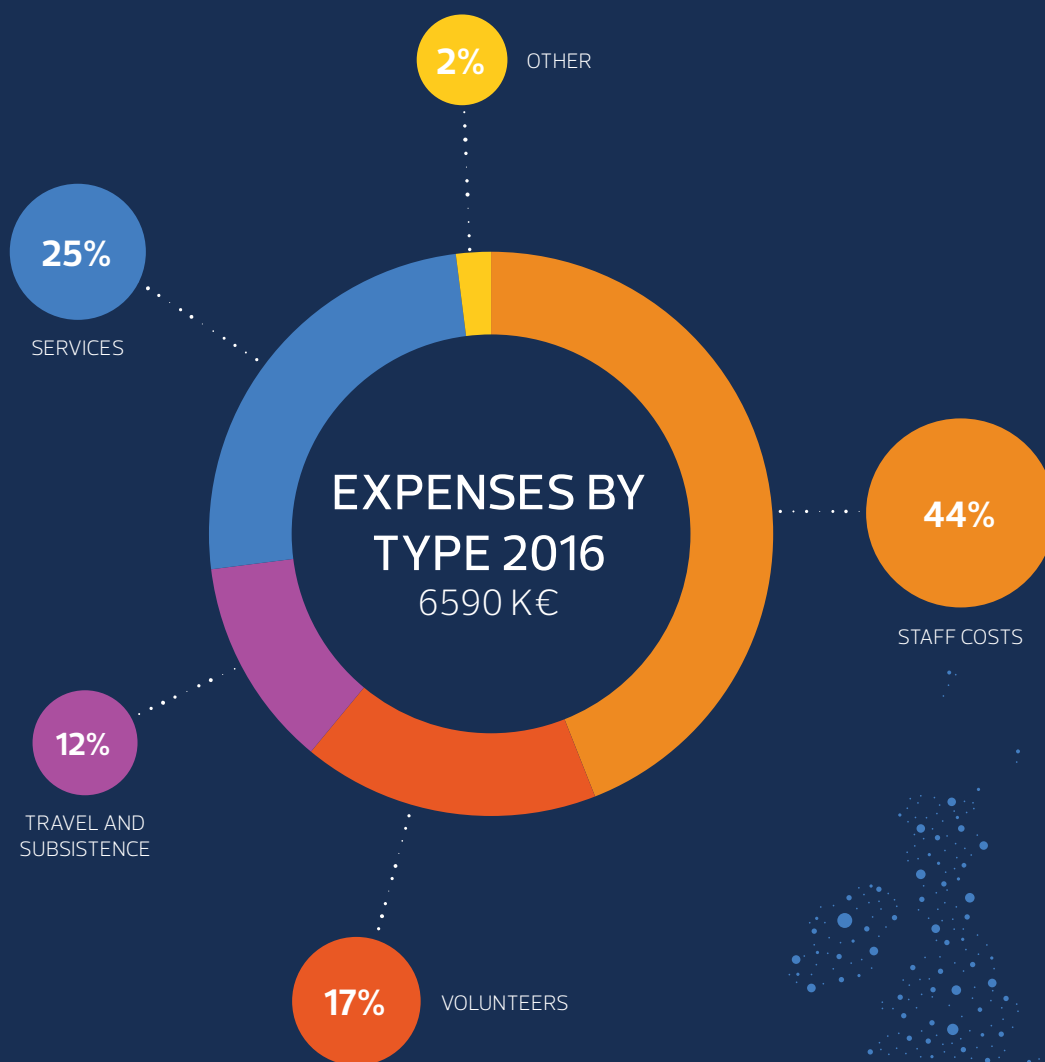
● Paris ● Brussels ● Barcelona ● Other



REVENUE & EXPENSES

2016





ACRONYMS & DEFINITIONS

EURORDIS INTERNAL COMMITTEES & TASK FORCES

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

PROJECTS OF EURORDIS OR IN WHICH EURORDIS IS INVOLVED

Adapt-Smart	An enabling platform for the coordination of Medicines Adaptive Pathways to Patients (MAPPs) activities, Innovative Medicines Initiative (IMI), 2015–2017
BBMRI Stakeholders' Forum	Biobanking and Biomolecular Resources Research Infrastructure
ECRIN	European Clinical Research Infrastructures Network
E-Rare	Network of ten partners – public bodies, ministries and research management organisations – from eight countries, responsible for the development and management of national/regional research programs on rare diseases
EunetHTA Forum	Support effective HTA collaboration in Europe that brings added value at the European, national and regional levels
EUROBIOBANK	European Network of DNA, cell and tissue banks for rare diseases
EUROPLAN	Fostering National Plans in Europe
EURORDIS Summer School (ESS)	4 day training on clinical trials for beginners. Since 2008, takes place each year in Barcelona, Spain.
EUPATI	Innovative Medicines Initiatives Joint Undertaking "Fostering Patient Awareness on Pharmaceutical Innovation"
EJA	Joint Action on Rare Diseases of the EU Committee of Experts on Rare Diseases: Funded by EC and by Member States, divided in work packages corresponding to specific activities, e.g. continuity of Europlan (Work Package 4); developing guidelines for social services dedicated to RDs (Work Package 6)
EYRD	European Year for Rare Diseases
GCOF	Genetic Clinics of the Future: To map the opportunities and challenges that surround the clinical implementation of next generation sequencing technologies, Horizon 2020, 2015–2017
InnovCare	Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions, DG Employment and Social Innovation (EaSI), 2015–2018
IRDiRC	International Rare Disease Research Consortium
Rare! Together	Project to promote European disease-specific federations
RDD	Rare Disease Day
RDI	Rare Diseases International
SCOPE	The Strengthening Collaboration for Operating Pharmacovigilance in Europe (SCOPE) Joint Action
TREAT-NMD	Translational Research in Europe – Assessment and Treatment of Neuromuscular diseases
Web-RADR	Development of tools for patients and healthcare professionals to report suspected adverse drug reactions to national EU regulators, Innovative Medicines Initiative (IMI), 2014–2017
RD-Action	Joint Action to expand and consolidate the achievements of the former EUCERD JA, DG Sanco, 2015–2018

EURORDIS & EUROPEAN REGULATORY NETWORK

CAT	Committee for Advanced Therapies – Michele Lipucci di Paola represents Eurordis
CHMP	Committee for Human Medicinal Products
COMP	Committee of Orphan Medicinal Products – Lesley Greene is Vice-Chair and Birthe Byskov Holm represents Eurordis as well – Maria Mavris is Observer

EMA	European Medicines Agency
HMA	Heads of Medicines Agencies
PCWP	Patients and Consumers Working Party – Richard Webst and François Houyez represent EURORDIS
PDCO	Paediatric Drugs Committee – Tsveta Schyns represents Eurordis
PRAC	Pharmacovigilance and Risk Assessment Committee
SAWP	Scientific Advice Working Party

EUROPEAN COMMISSION

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

EURORDIS & EUROPEAN COMMISSION

CEG-CC	Commission Expert Group on Cancer Control
Commission Experts Group on Rare Diseases (2013–2016) – CEG-RD	8 patients' representatives included 2 representatives of EURORDIS and 2 Observers
EUCERD (2010–2013)	EU Committee of Experts on Rare Diseases (8 Eurordis' representatives and 2 observers)
EU HPF	EU Health Policy Forum

EURORDIS & NON GOVERNMENTAL PARTNERS

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

MISCELLANEOUS

CAVOMP	Clinical Added Value of Orphan Medicinal Products
MOCA	Mechanism of Coordinated Access to orphan medicinal products
CoE	Centre of Expertise
ECRD	European Conference on Rare Diseases and Orphan Products
ERN	European Reference Network
PACE-ERN	Partnership for Assessment of Clinical Excellence in European Reference Network (PACE-ERN) Consortium
EUNRDHL	EU Network for Rare Diseases Helplines
HTA	Health Technology Assessment
MAPPS	Medicine Adaptive Pathways to Patients
MEP	Member of the European Parliament
MOCA	Mechanism of Coordinated Access to orphan medicinal products
EU MS	Member State (of the European Union)
ORPHANET	The online portal for rare diseases and orphan drugs
PLWRD	People Living with a Rare Disease
TRP	Therapeutic Recreation Programme



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