

EURORDIS  
**ACTIVITY REPORT** 2016  
& WORKPLAN 2017











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Acronyms & definitions





2017 marks the 20<sup>th</sup> anniversary of EURORDIS-Rare Diseases Europe.

The rare disease community, including all rare disease patient organisations and their partners, can be deservedly proud of its achievements over the last 20 years.

The landscape has changed drastically during this time. We have gone from near ignorance to the recognition of rare diseases as a public health priority in Europe. EU law now incentivises companies to develop orphan medicines. European expertise and innovative technologies that can potentially benefit people living with a rare disease have been developed. Most EU Member States now have national rare disease strategies and specialised centres of care. Rare disease research is stronger and recognised for its excellence.

**The rare disease patient community is bigger, better connected and highly skilled with great leaders at its helm.** Its spirit is collaborative across very different diseases, countries and stakeholders in Europe. Patients are now being taken seriously and listened to by policy makers, researchers and companies. **Patient centricity** has been the buzz term of the past decade.

We move forward with these changes. They are just the beginning. The needs of patients and families are huge and the community is calling for more action, now. We want to be ambitious, visionary. We will continue to always put **patients first**.

To encapsulate our vision for the next 10 to 20 years, EURORDIS is pleased to announce an updated vision for our organisation to enable **“better lives and cures for people living with a rare disease”** and an adjusted mission statement to **“work across borders and diseases to improve the lives of people living with a rare disease”**. This vision and mission reflect the progress that has been made over the last 20 years, but also our attitude to the challenges that lie ahead.

We have translated this into a new identity - we have changed our name from the European Organisation for Rare Diseases (EURORDIS) to **EURORDIS-Rare Diseases Europe** and have a brand new logo. Officially recognising our name to include Rare Diseases Europe, in use for several years now, is a further step to strengthen the identity of the rare disease movement, uniting with fellow national alliances around Europe (for example, Rare Diseases Denmark, Rare Diseases Hungary and Rare Diseases Sweden), as well as internationally with other patients groups like Rare Diseases International.

We now look to the future with great energy and drive. Building on our

collective expertise from experience and shared values, together we will continue to tackle the new challenges that lie ahead.

**Patient engagement** will be the buzz term of the next decade; patient engagement in research, in healthcare services, in medical and social care and in the lifecycle of medicinal products.

We strive to create the **transformational change** that rare disease patients and their families need by:

- + **Increasing awareness of rare diseases** among a larger public and key stakeholders through Rare Disease Day and other initiatives;
- + **Establishing rare diseases as a public health priority in the face of new challenges.** We must act with authenticity and determination to promote bold strategies to ensure that rare diseases are continuously recognised as a public health priority across Europe and around the world. This will ensure that the huge unmet needs of rare disease patients are addressed, and that all patients benefit from equal access to the new opportunities stemming from scientific, technological and organisational innovation (for example, the new European Reference Networks) in a sustainable and fair way.

# FOREWORD

by the President and the Chief  
Executive Officer



+ **Providing a cohesive voice** that represents all patients and diseases, making this voice international through initiatives such as Rare Diseases International and in partnership with international stakeholders through organisations including the NGO Committee for Rare Diseases (United Nations, New York);

+ **Supporting patient advocacy** based on **patients' knowledge** generated through the Rare Barometer survey programme so that the patient perspective is brought to the forefront during research, therapy development and policy making. EURORDIS will continue to act as a **knowledge broker** providing and encouraging the flow of rare disease patients' perspectives within and outside the rare disease community;

+ **Engaging patients** at all relevant levels to ensure the effective implementation of new policies and legislation relevant to patients' needs:

- **Research** (from basic research to ethics, and from research infrastructures to dissemination of research results);
- **Therapy development** (all along the critical points of the medicinal product lifecycle); and
- **Healthcare pathways** (from national centres of expertise to European Reference Networks, from patient registries to clinical trials, from best practices in diagnosis and patient cross-border mobility to accessing best care, and finally from patient therapeutic education to healthcare professional education).

+ **Empowering patient advocates and patient organisations** by:

- **Informing** them through relevant tools such as eurordis.org and EURORDIS webinars.
- **Connecting** them through networks such as RareConnect, the Council of National alliances and the Council of European Federations. We will support the growth of the European Patient Advocacy Groups and will also champion matchmaking tools that connect patients with researchers and clinicians. Patients will become better connected to the medical, scientific and policy communities.
- **Building the capacity** of patient advocates through workshops and trainings including the EURORDIS Summer School so that they are prepared to engage in research, therapy development, policy making and more. We will also continue to build the capacities of stakeholders including clinicians, researchers, companies and policy makers and help to better understand the perspectives of those living with a rare disease.

The challenges ahead are no small feat. But the rare disease community is not easily deterred. This is why we have come so far in the last 20 years.

We look forward to the next 20 years with pride, courage and hope for how we can enable **better lives and cures for people living with a rare disease**.

Rare but strong together. ”



**YANN LE CAM**

Chief Executive Officer



**TERKEL ANDERSEN**

President of the Board of Officers





# 20 Years of Achievements in the Rare Disease Community

*As EURORDIS-Rare Diseases Europe celebrates its 20<sup>th</sup> anniversary in 2017, we take stock of the achievements from the last two decades:*





## From ignorance to increased awareness

20 years ago, society was living in ignorance of rare diseases. A special Eurobarometer survey on European awareness of rare diseases published in 2001 found that Europeans had a relatively accurate understanding of what rare diseases are but detailed knowledge and awareness remained low.

Over the last two decades, we have raised awareness so that people better understand rare diseases and their impact through initiatives such as Rare Disease Day.

**Since Rare Disease Day started in 2008, tens of thousands of events have been held by thousands of patient organisations in over 100 countries.** Rare Disease Day brings together millions of patients, families, carers, medical professionals, policy makers and members of the general public in solidarity to raise awareness.

## Rare diseases now recognised as a public health priority

EURORDIS, national rare disease alliances and patient organisations have campaigned tirelessly to ensure that rare diseases are at the top of European and national policy makers' agendas. **Since 1999, the European Union has recognised rare diseases as a public health priority.**

EURORDIS actively contributed to the adoption of the 2008 Commission Communication on Rare Diseases, the

2009 Council Recommendation on an action in the field of rare diseases (and has promoted their implementation), and the 2011 EU Directive on patients' rights in cross-border healthcare.

In particular, EURORDIS has supported national alliances with the **development of national rare disease plans in most Member States.** For the next decade, we must act with authenticity and determination to promote bold strategies to ensure that rare diseases continue to be recognised as a public health priority across Europe and around the world. This will ensure that the huge unmet needs of rare disease patients are addressed, and that all patients benefit from equal access to the new opportunities stemming from scientific, technological and organisational innovation in a sustainable and fair way.

## An empowered rare disease community

Over the last 20 years, policy makers, researchers and companies have progressively recognised patients as disease experts. EURORDIS and rare disease patient groups have dedicated themselves to empowering patients to effectively engage in research, policy making and the development of therapies:

- + Tools and programmes such as the EURORDIS Summer School and online training, as well as the European Patients' Academy (EUPATI), have informed patients and built their capacities so that they are empowered in their roles in important policy-making forums.



To date, more than 300 participants from over 40 countries and representing more than 75 different diseases have been trained through the EURORDIS Summer School since it began in 2008.

- + Sources developed over the last 20 years like Orphanet, the online portal recognised as the primary directory of rare diseases and orphan medicines, are vital to ensure patients are **informed** to empower them in their advocacy activities.
- + Capacity building through dissemination of information via: eurordis.org (available in 7 languages), the EURORDIS eNews and Member News, webinars, workshops at the 20 annual EURORDIS Membership Meetings that have taken place all over Europe, and workshops through European Federations and National Alliances.
- + Patients are also **empowered by their connections** to the rest of the rare disease community through networks such as RareConnect.

### A strong, cohesive patient voice

We have progressed from only two existing National Alliances in 1997 when EURORDIS was created to a network of nearly 50 national rare disease alliances around the world, including the 34 alliances that are members of



the Council of National Alliances. Similarly, from 1997 when there were less than 20 existing European Federations to today, when there are 58 European Federations that are all members of the Council of European Federations. By connecting patients and organisations through this network, their voices become stronger and cohesive.

Over the last 20 years and through this network, animated by groups such as EURORDIS, the patient voice has grown in size and strength. **EURORDIS membership has gone from 187 patient organisations in 2000 to over 700 in 2017.**

During the last two decades, empowered and informed patients across the world have used their voice to advocate for improved research, development of therapies and policy making to decrease the impact rare diseases have on their lives.

This voice has been taken to the international level through Rare Diseases International and the NGO Committee for Rare Diseases.

Building on surveys conducted via the EurordisCare Programme, the Rare Barometer Survey Programme is also collecting the opinions of thousands of rare disease patients and family members to ensure that the rare disease patient voice is presented to and heard by policy makers. EURORDIS will continue to act as a knowledge broker providing and encouraging the flow of rare disease patients' perspectives within and outside the rare disease community.

EURORDIS represents the rare disease patient voice in larger coalitions including the European Patients' Forum and International Alliance of Patients' Organizations.

### Increased patient engagement in rare disease policy, research, healthcare and development of medicines

Over the last 20 years, patients have become further engaged in research and the entire process of developing and accessing care and therapies:

- + EURORDIS has developed committees and advisory groups that bring together patients to connect and integrate them into the governance and decision-making structures of bodies that produce or influence rare disease research, policy and develop therapies.
- + Patients have become an integral part of the medicines development decision-making process at the European Medicines Agency (EMA) through committees including: the Committee for Orphan Medicinal Products (where EURORDIS Chief Executive Officer Yann Le Cam was one of the first patient representatives to be appointed and also serve as vice-chair); the Committee for Advanced Therapies; the Paediatric Committee; and the Scientific Advice Working Party. **From 2007-2015,**





there were 743 patients and consumers involved in EMA activities. Since 2008, EURORDIS has facilitated the involvement of 178 patient experts in protocol assistance at the EMA.

- + Patient involvement in the entire lifecycle of medicines development has become mainstream in the last years, from research and development all the way through to post-marketing assessment processes. EURORDIS has supported the involvement of patients in the area of health technology assessment.
- + EURORDIS has empowered patients with the skills they need to participate in, for example, the former EU Committee of Experts on Rare Diseases, the European Commission Expert Group on Rare Diseases, the European Joint Actions on rare diseases (former EUCERD and current RD-Action) and the European Joint Action on Rare Cancers.
- + Patients have become more engaged in policy on healthcare pathways. For example, through the establishment of 23 European Reference Networks and the recently launched European Patient Advocacy Groups, which will enhance collaboration between patients, clinicians and policy makers.
- + Finally, patients have been integrated into the governance of important research projects and infrastructures that support rare disease research such as the International Rare Diseases Research Consortium (IRDiRC), E-Rare and RD-Connect.

## Less isolated rare disease patients and families

Because of a lack of information, small patient populations and limited medical expertise, living with a rare disease can be a very isolating experience.

As the rare disease community has grown in size and

strength over the last 20 years, people living with or affected by a rare disease have become less isolated.

Rare disease patients and their families are now connected across borders and languages.

Initiatives such as RareConnect and Rare Disease Day help to establish and develop these connections and to reassure patients that they are not alone, that they are part of a growing rare disease community. **There are now 23,000 RareConnect members while there have been 2.5 million visitors to rareconnect.org since April 2012.**

Rare disease helplines offer information and support to people affected by rare diseases. **There are over 50 helplines in Europe.**

In addition, over the past two decades, an increasing number of patient organisations have been created to provide information and create communities so that patients and their families are not isolated.

Disease-specific events organised by patient groups and international events including the European Conference on Rare Diseases & Orphan Products also play an important role in connecting patients and also providing them with the opportunity to meet researchers, clinicians and policy makers.

## An escalation in new medicines for rare diseases

- + In 1999, the landmark EU Regulation on Orphan Medicinal Products came into place, introducing for the first time incentives for companies looking to develop orphan medicines. This is just one of numerous regulations and policy documents that EURORDIS has contributed to that directly contribute to the development of therapies for rare diseases. It followed the US Orphan Drug Act of 1983.

**Since then, 126 medicines have been approved for rare diseases and over 1800 medicines have received an orphan designation, meaning the medicine benefits from market exclusivity.**

- + One of the overarching goals of the International Rare Diseases Research Consortium (IRDiRC) is to contribute to the development of 200 new rare disease treatments by 2020. This goal will be easily met this year, well ahead of time, thanks to the steady increase of orphan medicines that have received market authorisation in the EU and US. This is great progress but only meets the needs of a small proportion of the rare disease community. There remains a large unmet medical need to provide medicines for the rest of the over 6,000 identified rare diseases. Natural history studies should be carried out to increase knowledge of more diseases so that treatments can be developed.
- + By being actively involved in the development of



EU regulation around orphan medicines and in the designation procedure of orphan medicines at the EMA, EURORDIS has developed strong expertise in these areas over the last 20 years. The EURORDIS Round Table of Companies (ERTC) was set up in 2004 to bring together companies with a common interest in rare diseases and orphan drug development. Through the ERTC, this expertise has been shared with companies and gone towards reaching the end goal of more and innovative rare disease medicines available to patients. Over the last 13 years, ERTC members have benefitted from taking part in constructive dialogue, facilitated by EURORDIS, involving patients and regulators.

- + The availability of more medicines is a great success. The real challenge now is to improve access to those medicines. Initiatives such as the EURORDIS Access Campaign and Mechanism of Coordinated Access to Orphan Medicinal Products have already contributed to ensuring patients' access to new, innovative therapies. Improving access to medicines will be a focal point of EURORDIS' advocacy work for the next 10 years.



## Translation of scientific advancement into therapeutic innovation

Over the last 20 years, the rare disease community has contributed to creating a hospitable environment for the advancement of science that leads to more, innovative therapies and advanced therapies (a medicine for human use that is based on genes, cells or tissue engineering).

Patient groups all around the world including EURORDIS have influenced all stages of the process and relevant legislations to create this environment: from research, to development of medicines, through the regulatory process, and finally to ensuring patients' access to the new medicines:

20 years ago there were no funds allocated to rare disease research. Through the EU Framework Programmes for research, funds dedicated to rare disease research have exponentially increased from 64m€ (framework programme 5, 1998 – 2002), to 230m€ (framework programme 6, 2002 – 2006) and 530m€ (framework programme 7, 2007 – 2013). Over 100m€ has already been allocated to rare disease research from 2014 – 2015 under Horizon 2020, the EU Framework Programme for Research and Innovation 2014 – 2020. These funds allow for research and create the environment needed to advance science and develop therapies.

New challenges and opportunities are arising around new technologies such as genome sequencing and CRISPR. As well as the landmark 1999 EU Regulation on Orphan Medicinal Products, the 2007 EU regulation on advanced therapies has created ground-breaking opportunities for the treatment of diseases. In addition, the 2006 EU regulation on paediatric medicines facilitates the development and availability of medicines for children.

## Expert centres and a European network providing the best possible care

People living with a rare disease require specialised care and a high level of expertise from their healthcare providers. Rare disease patient populations are small and medical expertise is limited and scattered across borders. Rare disease patients can become lost and subject to marginalisation in classic healthcare systems designed for common diseases.

The rare disease community has advocated tirelessly over the last 20 years to bring about policies and regulations that have improved healthcare for rare disease patients across Europe:

- + Centres of Expertise (CEs) have been recognised as a solution to the challenge of providing effective healthcare to rare disease patients. The Council of the EU's Recommendation on an action in the field of rare diseases (2009) recommended that Member States adopt a national rare disease plan, including a series of measures to improve care and research, and within this identify/ support the creation of centres of expertise.
- + Since 2008, nearly all EU Member States have created national rare disease plans and all have established centres of expertise. EURORDIS and national alliances played a crucial role through EUROPLAN to promote patient-centred national rare disease plans.
- + The 2011 EU Directive on Patients' Rights in Cross-border Healthcare states that the European Commission should support the continued development of European Reference Networks (ERNs). The ERNs are networks of the aforementioned centres of expertise, healthcare providers and laboratories that are organised across the EU.



**At the end of 2016, the European Commission announced the first 23 European Reference Networks for rare diseases (ERNs).** This momentous step came after years of collaboration and efforts between rare disease patients, clinical experts, and policy makers in EU Member States, at the European Commission and the European Parliament to bring the ERNs to fruition.

EURORDIS played an integral role in the development of ERNs and in ensuring that patients are at the centre of the new networks. Through the creation of European Patient Advocacy Groups (ePAGs), EURORDIS has structured the rare disease community to ensure that patients are represented at the core of governance and development of ERNs.

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## Diagnosis of more rare diseases

The difficulty in obtaining a correct diagnosis is the first dramatic hurdle for rare disease patients and may take years or even decades to overcome. Some patients are never diagnosed. Late diagnoses delay the beginning of adapted treatments and can have severe irreversible, debilitating and life-threatening consequences.

Over the last 20 years, significant progress has been made in improving diagnosis of rare diseases:

- + One of the overarching goals of the International Rare Diseases Research Consortium (IRDiRC) is to make possible the diagnosis of as many rare diseases as possible by 2020. Progress in this field requires the characterisation of new rare diseases and the identification of their cause, mostly through discovery of new genes, and then developing clinical tests.

IRDiRC therefore monitors (i) the number of new rare diseases identified since 2010 (ii) the number of genes identified as linked to rare diseases and (iii) the number of diseases for which there is a clinical test available.

**More research and funding from targeted efforts by bodies like IRDiRC have contributed to improved diagnostic tools and the data from Orphanet shows that these three indicators are all steadily increasing.**

- + In 2009, 6,000 patients responded to a survey conducted via the EurordisCare Programme to share their experiences of diagnosis. Analysis of the results of the survey informed EURORDIS' active participation in European projects aimed at improving diagnosis, for example RD-Connect.
- + Through membership of the Commission Expert Group on Rare Diseases, EURORDIS contributed directly to

the Recommendation on Cross Border Genetic Testing of Rare Diseases in the EU, which recommended that "obtaining an accurate and timely diagnosis is a priority for all people with a potentially genetic RD; therefore, access to genetic testing [...] should be ensured ...".

- + There has also been an increase in the development of specific projects across Europe and beyond to improve diagnosis for undiagnosed and rare disease patients (for example, the Undiagnosed Diseases Network International, SWAN UK and the Wilhelm Foundation), while national alliances including the US National Organization for Rare Disorders, the Canadian Organization for Rare Disorders, ASrid Japan and Rare Voices Australia have made great efforts to advocate for the needs of undiagnosed patients. Through these projects, we have seen improved access to next generation sequencing.

More recently, these groups and EURORDIS came together to publish International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients.





# EURORDIS IN BRIEF

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 700 rare disease patient organisations from more than 60 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe.

By connecting patients, families and patient groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services.

**747**

member patient organisations



**65** countries  
(28 EU countries)

**41** National Alliances of RD Patients Organisations

**58** European Federations of specific rare diseases

 Founded in

**1997**



**40+**

Staff members, with offices in Paris, Brussels, Barcelona

Outreach to over

**1800**

patient groups

 Over

**440**

Volunteers

**+80**  
patient advocates

**+250**  
moderators

## Vision

EURORDIS' vision is to enable better lives and cures for people living with a rare disease.

## Mission

EURORDIS works across borders and diseases to improve the lives of people living with a rare disease.





# 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.*

# 1

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.



# 2

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.

# 3

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.

# 4

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.





**EURORDIS**  
RARE DISEASES EUROPE

# ACTIVITY REPORT

2016

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# HIGHLIGHTS 2016

EURORDIS has **733 member patient organisations**, located throughout 64 different countries, including 519 full members in 27 EU countries.



The **EURORDIS Membership Meeting 2016 Edinburgh** attracted **200 participants from 31 countries** and was held back to back with the **European Conference on Rare Diseases & Orphan Products (ECRD) 2016 Edinburgh** with **over 750 participants from 48 countries**. 40 fellowships were granted from 17 countries within the ECRD-EURORDIS Patient Advocate Fellowships Programme.



**Rare Disease Day 2016** was organised in over 84 countries worldwide, including all 28 EU countries. The new look for the **Rare Disease Day 2016 website** was more dynamic and engaging, with over 48 000 visits on Rare Disease Day alone. For the 5<sup>th</sup> year in a row EURORDIS produced a Rare Disease Day video which was translated into 34 languages (up from 27 in 2015).



2016 also saw the **official launch of the NGO Committee for Rare Diseases** which took place on 11 November 2016 at the United Nations headquarters in New York. **Over 100 participants from all continents** were present on the day coming from civil society, UN bodies, national governments, academia and industry. During the meeting, the coherence between the challenges of the rare diseases community and the Sustainable Development Goals Agenda was confirmed by members of United Nations agencies.



**Rare Diseases International** continued to expand reaching **43 members** and held its second meeting back to back with the EMM 2016 Edinburgh. The first RDI Council was elected by its members.



The **Rare Barometer Programme** which was created in 2015 as a permanent programme of quantitative and qualitative studies to collect patient experiences and expectations was further developed and promoted. Rare Barometer Voices, an online panel of people living with a rare disease who are willing to participate in EURORDIS' surveys, **recruited 4300 participants**.

EURORDIS has been the central actor in the development of ERNs with 2016 seeing the **creation of 24 European Reference Networks**. In early 2016, EURORDIS created European Patient Advocacy Groups (ePAGs) aligned to the scope of the different ERN applications, with 85 ePAG representatives democratically elected so far.



The **"ExPRESS 2016" 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. A much greater emphasis was placed on the new pre-training aspect of the Summer School Programme. **44 participants attended representing 13 countries and 28 diseases**.



**686 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**. A total of 235 EMA dossiers for public information, including 222 Public Summaries of Opinion of Orphan Designations (PSOs), 5 European Public Assessment Reports (EPARs) and 8 Package Leaflets (PLs), were reviewed by EURORDIS staff members in order to ensure the quality of the information disseminated by the Agency to the general public.



EURORDIS was appointed to the **Management Board of the European Medicines Agency (EMA)**. The EMA Board is made up of representatives of each of the 28 EU Member States, the European Commission, the European Parliament, two civil society organisations, and doctor and veterinarian organisations.



EURORDIS was proud to rely on **447 volunteers** including 89 dedicated volunteer patient advocates, 70 volunteer ePAG representatives, 1 for fund raising, 1 office support volunteer and 286 volunteer moderators of online communities of Rare Disease Patients, within the activity "RareConnect".





# 1. PATIENT ADVOCACY

## 1.1 Our Advocacy Goals within our Strategy Priorities for 2015-2020

In the course of 2016, EURORDIS advocacy activities were carried out to pursue its Advocacy Goals:

- + Promoting rare diseases as a sustainable public health priority in the EU programmes beyond public health: research, enterprise, digital, social areas
- + 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
- + Promote cross-border rare disease expertise and knowledge generation and sharing to improve quality of care diagnostic, medical care & social care at local level
- + Promote access to cross-border healthcare and making possible patient mobility
- + Promoting research and bridging patient's perspective and researcher activities
- + Addressing the new issues of genetic testing, genetic counselling & new-born screening
- + Voicing/expressing patient preferences in sharing of health and genetic data in rare diseases information systems and repositories

## 1.2 Our Advocacy Actions in 2016 to reach our goals

At the EU level, advocacy activities continue to be carried out in the broad framework of the support of the implementation of the EU strategy on RDs adopted in 2008 with the "Commission Communication on Rare Diseases: Europe's Challenges" and in 2009 with the "Council Recommendation on an Action in the Field of Rare Diseases". This broad strategy covers multiple policy

areas, as described below throughout Chapter 1. Similarly, advocacy activities utilise different tools, platforms and mechanisms that are instrumental to the policy advances with the rare disease community and stakeholders, i.e. Commission Expert Groups on both Rare Diseases and Cancer Control, Joint Actions on Rare Diseases and Rare Cancers, etc.

### 1.2.1 The European Commission Expert Group on Rare Diseases (CEG-RD) (2014-2017)

*The Commission Expert Group on Rare Diseases was set up by a **Commission Decision of July 2013**. 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**. The Expert Group provides advice and expertise to the Commission in formulating and implementing the Union's activities in the field of rare diseases and fosters 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)

Two meetings were held at the European Commission in Luxembourg. Throughout 2016, the CEG-RD accompanied the development of European Reference Networks for Rare Diseases (ERNs) with the support of the first and second EU Joint Actions for Rare Diseases.

Major topics discussed in 2016 included: Adoption and implementation of National Plans for Rare Diseases in EU Member States; Development of the European Platform on Rare Diseases Registration at the Joint Research Centre (JRC); Research programmes for rare diseases; Health Technology Assessment in the area of rare diseases.

The Commission Expert Group has ended its mandate at the end of 2016. We have concerns about the renewal of the group in the absence of clear signals by the Commission.

All reports and recommendations produced by the European Commission Expert Group on Rare Diseases in 2016 are available on the website:

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

### EURORDIS Policy Action Group (PAG)

The eight patient representatives in CEG-RD are supported by two observers from the EURORDIS staff and all together form the EURORDIS Policy Action Group (PAG). The PAG teams up with other volunteers and EURORDIS staff to provide the patients' view on issues dealt with by the CEG-RD, such as: National strategies and national plans; centres of expertise, European Reference Networks (ERNs); cross-border healthcare; codification and classification of rare diseases; access to rare disease therapies and orphan medicinal products; integration of rare diseases into social policies and services and others. The PAG also participates in the European Joint Action on Rare Diseases – RD-ACTION – through contributing to the Work Package "Rare Disease Policy".

### 1.2.2 The European Commission Expert Group on Cancer Control (CEG-CC) (2015-2018)

*In 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 CEG-CC assists the Commission in the preparation of legislative proposals and policy initiatives in the field of cancer. Two volunteer representatives of EURORDIS have been nominated to represent patients affected by rare cancers in the Commission Expert Group on Cancer Control. The EURORDIS patient representative is also a nominated member of CEG-RD to make the link between the two Expert Groups. 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.*

Several members of CEG-CC, including EURORDIS, are partners of the EU Joint Action on Rare Cancers, which started on 1<sup>st</sup> October 2016.

Major topics discussed in 2016 include: Ongoing Joint Action CANCON (European Guide on Quality Improvement in Comprehensive Cancer Control); European Cancer Information System (JRC); Screening, early detection and prevention; Cancer care: more appropriate utilisation of healthcare interventions.

### EURORDIS Policy Action Group on Rare Cancers (PAG-RC)

The work of the two EURORDIS volunteer representatives is supported by the EURORDIS Policy Action Group on Rare Cancers (PAG-RC) which was established in July 2014 following the EURORDIS call for expression of interest addressed to its members. It is made up of six patient representatives nominated by the EURORDIS Board of Directors and is supported by the EURORDIS Public Affairs staff. The PAG-RC follows up on the work carried out by the CEG-CC to provide the patients' views in the field of rare cancers and contributes to the making of relevant policies. Thus far it has produced the EURORDIS Table mapping out similarities and differences between rare diseases and rare cancers and has contributed to other initiatives and projects, such as the EU Joint Action on Rare Cancers (JARC).



### 1.2.3 2<sup>nd</sup> European Union Joint Action on Rare Diseases – RD-ACTION (2015-2018): Promoting the Implementation of Recommendations on Policy, Information and harmonised coding system for Rare Diseases

EURORDIS is involved in two work packages of RD-Action:

#### Work Package 2 - Dissemination:

The overarching goal of this work package of which EURORDIS is leader, is to disseminate rare diseases-related information and improve the two-way information flow between the national and European levels. The goals of the work package are to set up and maintain the Joint Action dissemination tools; to produce a twice-monthly newsletter of the rare disease community, Orphanews; to hold the **European Conference on Rare Diseases and Orphan Medicinal Products in May 2016, in Edinburgh**; to support national workshops aimed at disseminating at national level the JA activities and the Recommendations produced and adopted by the EUCERD/Commission Expert Group on Rare Diseases (CEGRD); to support national authorities in the consolidation of sustainable and resilient health systems.



#### Work Package 6: Rare Disease Policy

In this WP led by Newcastle University, EURORDIS is the main partner to propose policy priorities to the consultative group of Member states and to implement the activities participating to the Consultative Group of RD Action, selecting policy priority areas. Despite significant progress under the EJA towards implementing the Council Recommendation on an Action in the field of rare diseases (RD) and the Commission Communication ('Rare Diseases: Europe's Challenges'), many policy areas would benefit from additional pan-European collaboration. The topics within the scope of this work are broad, ranging from European Reference Networks to eHealth, from RD registration to the social integration of people living with RDs. The goals of this work package are to develop and implement a methodology to support the development of policies and recommendations in association with all relevant stakeholders; to provide information and policy support to the Commission Expert Group on Rare Diseases

and to produce the Report/Resource on the State of the Art of Rare Diseases Activities in Europe for which EURORDIS coordinates the contribution of National Alliances to the national sections.

### 1.2.4 European Union Joint Action on Rare Cancers (JARC) (2016-2019): Promoting EU Rare Cancer policy

The Joint Action on Rare Cancers is a multi-stakeholder collaboration between 18 Member States and the European Commission, coordinated by the Fondazione IRCCS Istituto Nazionale dei Tumori of Milan. There are 35 partners involved in the JARC including 8 Ministries of Health/Cancer Control Programme representatives and 27 universities, public health institutions, cancer registries, oncological institutes, patients' associations and other societies/organisations. The Joint Action on Rare Cancers aims to prioritise all rare cancers in the agenda of the EU and the Member States with regards to national cancer plans, harmonisation of practices and funding of research.

EURORDIS is involved in five Work Packages out of 10 in order to contribute to the objectives of the JARC:

- + Prioritise rare cancers (RCs) in the agenda of the EU and Member States;
- + Develop a position on prevalence and incidence of rare cancers which may help refine future national strategy on cancers or on rare diseases as well as orphan product status
- + Develop innovative and shared solutions, mainly to be implemented through the future European Reference Networks in the field of Rare Cancers, in the areas of quality of care, research, education and state of the art definition on prevention, diagnosis and treatment of rare cancers.

### 1.2.5 Advocate to improve the regulatory process for orphan medicinal products & paediatric use of medicines

In 2016, EURORDIS has advocated to improve the regulatory process for Orphan Medicinal Products in particular by working collectively and answering to two public consultations launched by the European Commission and by preparing its answer for a third one to be submitted in 2017.

The two first were the Public consultations on the notice from the Commission on aspects of the application of Article 3, 5 and 7 of Regulation (EC) N° 141/2000 on orphan medicinal products and the Public consultations on the concept of "similar medicinal product" in the context of the orphan legislation. The third one was the Targeted stakeholder consultation on the experience acquired with the Paediatric Regulation.

Our answer to the Public consultation on the notice from the Commission on aspects of the application of Article 3, 5 and 7 of Regulation (EC) N° 141/2000 on orphan medicinal products was putting emphasis on access to OMPs, the burning need for continuous generation of real-world evidence post-authorisation and the usefulness of Protocol assistance during the development of a product. This Notice was aiming to replace the Commission Communication of 2003.

Access is a challenge for patients, especially considering that the main incentive of the Orphan Regulation, the market exclusivity, is given for a fragmented EU market. Although a certain level of evidence is needed to define what is meaningful, the companies need to have some predictability that a product in development will retain its orphan status, or else we will see a de-incentivisation of the long term investment in the field of orphan medicinal products. EURORDIS is not asking for a lower threshold to the detriment of the quality of orphan therapies, but rather a realistic approach to a field with inherent difficulties and a patient oriented risk assessment. Our answer was insisting on a right trade-off between flexibility and evidence generation so to guarantee to patients access to medicines that will cover their needs. In particular, significant benefit should be assumed more easily and re-assessed after the product is placed on the market. Therefore we called on the Commission to consider taking actions to improve access to orphan medicinal products based on the spirit and text of the Regulation (EC) No 141/2000, including re-examining the interpretation of its Article 8.2 for potential review of criteria at five years i.e. 'conditional SB'.

The European Commission eventually issued the official text of the new Notice in November 2016.

In parallel with this process, the EC had launched another Public consultation, on the concept of "similar medicinal product" in the context of the orphan legislation. Although more technical than the Notice, this Consultation is of relevance for potential impact on future OMP development. One important aspect of this text proposal is that a more detailed assessment of the structural and functional characteristics of the product will translate into more products not being considered similar and therefore more therapeutic alternatives will be available for patients. In this context of in-depth evaluation of similarity, similar products will be "more strictly similar" and therefore proving clinical superiority would be more difficult, but those that prove it would be much better than the competition.

Regarding the Paediatric Regulation, the European Commission (EC) has launched a EU Public Consultation on the experience acquired with the Paediatric Regulation to get views and feedback from stakeholders, to support the Commission in drafting its second report on the Paediatric Regulation after nearly ten years of implementation.

Providing an adequate response to this EC consultation in order to reflect an acceptable consensus for all heterogeneous paediatric rare diseases has been a challenge. The EU Regulation on medicinal products for paediatric use is often perceived by various stakeholders, as a **complex and cumbersome regulation, difficult to apply**. Some stakeholders, including some patient groups, would like to amend this Regulation with a new voting at the European Parliament.

However, in light of numerous discussions with EMA, pharmaceutical industry, and patients' representatives, EURORDIS considers that there is no need for amendment at this stage, given also that "10 years" is not an enough long period to decide to change the legislation or not.

On 27 September 2016, EURORDIS organised its 24<sup>th</sup> Workshop of the EURORDIS Round Table of Companies (ERTC) on the theme: "Bringing Solutions to Young Rare Disease Patients: Let's discuss the paediatric regulation". Patients' representatives, industry's representatives as well

as EMA officials were invited to share their perspectives. The outcome of this ERTC workshop has enabled EURORDIS to shed the lights on various challenges and come up with a consensus.

EURORDIS is in favour of a better implementation of the Regulation to develop medicinal products for paediatric use.

## 1.2.6 Advocate to improve the patient access to rare disease therapies and promote a new business model sustainable for society

*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 after 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 alias, 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](http://ec.europa.eu/health/documents/pharmaceutical-committee/stamp/index_en.htm)).*

*At the post-authorisation phase, obstacles come, inter alias, 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.*

*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 below.*

*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. For each of these difficulties, EURORDIS contributes to finding solutions with its advocacy action:*



## Pre-authorisation: Compassionate use



Compassionate Use Programmes are established to regulate access to medicines prior to their marketing authorisation, to help treat patients who have no other options and who do not have the time to wait for the end of clinical trials and the authorisation process.

In 2016, following a long-lasting campaign to standardise schemes in EU Member States, EURORDIS' DITA Task Force proposed EURORDIS to adopt a position. The Position Paper is due for adoption by EURORDIS Board of Directors in March 2017: "[Early Access to Medicines in Europe: Compassionate use to become a reality](#)"

The Position Paper presents a range of policy proposals, including: supporting the adoption of the French approach (ATU programme) in all EU countries, including Compassionate Use in the "basket of benefits" as defined in the Cross Border Healthcare Directive; generalising the Medicines Adaptive Pathways to Patients and amending the EMA guidelines as requested by EC, so to expand the role of the European Medicines Agency.

The Position Paper proposes recommendations to all stakeholders: industry, European authorities, Member States and patients' organisations. For example, EURORDIS position is against the setting-up of an ad hoc ethics committee by the developer of a medicine that reviews requests for compassionate use and decides which patients can enter the programme. The Position Paper explains why EURORDIS is opposed to this approach.

### At the authorisation phase

**Conditional approval.** 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.

Volunteers of EURORDIS' Drug and Information Transparency and Access (DITA) Task Force attended the Drug Information Association (DIA) EuroMeeting in Hamburg, 6-8 April, with presentations on adaptive pathways and conditional authorisation.

**PRIME - PRiority Medicines** is an EMA initiative for medicines of major therapeutic interest, addressing unmet needs in devastating diseases. Under the PRIMA scheme, eligible candidates 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 of the product.

After responding in late 2015 to the public consultation on a "[Reflection paper on a proposal to enhance early dialogue](#)

[to facilitate accelerated assessment of priority medicines \(PRIME\)](#)", EURORDIS is now working with the EMA to see how relevant patients' organisations can be consulted and/or involved.

**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 supports the optimisation of 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.

In 2015, EURORDIS became partner in the IMI project on Medicines Adaptive Pathways to Patients, ADAPT-SMART, aiming to establish an enabling platform for the coordination of MAPPs-related activities within IMI2 and engaging a dialogue with relevant stakeholders. The overall objectives of ADAPT-SMART are to identify relevant MAPPs activities; to create a MAPPs repository of knowledge and opportunities; to identify the scientific challenges and opportunities related to MAPPs implementation; to facilitate the inclusion of MAPPs enablers (tools and methodologies) to address/exploit the identified challenges and opportunities; and to develop a comprehensive scientific research plan to support MAPPs implementation.

In 2016, EURORDIS continue to co-lead the [definition of the Selection Criteria for a product to enter this new development path](#). EURORDIS also took part to the discussion around the future solutions to manage and reduce the uncertainties that patients face when they enter MAPPs; and discussions on the definition of the optimal and seamless pathway to develop medicines under the MAPPs framework.

### Post-authorisation: pricing and reimbursement

#### Launching multi-stakeholder collaboration to address bottlenecks in access to orphan medicines in Europe

The EU Regulation on Orphan Medicinal Products is an example of successful legislation as it triggered innovation and led to 116 new rare disease therapies with marketing authorisation and 1,605 orphan products in development for diseases. Nevertheless, even today, nearly 20 years after the adoption of the EU Orphan Drug Regulation and the foundation of EURORDIS, access to orphan medicines across Europe cannot be considered as satisfactory, let alone optimal. With a third of patients not having access to the necessary orphan medicine and another third having access only after waiting years, there is clearly large room for improvement. More recently, some important medicines are not being made available because they are perceived to be too highly priced in comparison to the determined value. EURORDIS believes that European collaboration has to be scaled up to improve access to therapies for patients.

## Multi-Stakeholder Symposium on Improving Patient Access to Rare Disease Therapies

The first Multi-Stakeholder Symposium aimed at addressing a crucial bottleneck in making orphan medicinal products accessible across Europe by launching a method of collaborative negotiation with all the key partners involved.

On 24-25 February 2016, a combination of nearly 300 patient advocates, academics, policymakers, industry representatives, payers and HTA bodies came together in Brussels to discuss the current state of play and how to shape a more effective way to address value determination, appraisal, pricing and reimbursement of orphan medicines, all with the aim of improving patients' access to rare disease therapies throughout Europe.

By bringing together such a varied range of stakeholders, the Symposium created an opportunity for exchange to reach an understanding of the varied perspectives on issues and challenges surrounding patient access. Participants discussed various methods of appraisal, value determination and reimbursement and also took part in pricing simulation exercises.

Speakers included European Commissioner for Health, Vytenis Andriukaitis, and Members of the European Parliament, Philippe De Backer and Françoise Grossetête.



## A reflection paper to break the access deadlock



In the wake of our 1<sup>st</sup> Multi-Stakeholder Symposium on Improving Patient Access to Rare Disease Therapies in February 2016, EURORDIS identified the critical need to provide a synthesis of the immense number of initiatives, reflections and perspectives existing today on the topic of access, but also and most importantly to express our view and that of our members on which "step changes" may be necessary today in the currently accepted models of thought.

This collective effort kicked off in the second quarter of 2016 and, through a cycle of extensive consultations, two member webinars and one in-depth debate at the November 2016 meeting of the EURORDIS Council of National Alliances (CNA), it gave birth to a new Reflection Paper, "Breaking the Access Deadlock to Leave No One Behind", due for launch in early 2017.

## MoCA (Mechanisms of Coordinated Access)

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 GROW). MoCA is currently placed under the umbrella of the Medicines Evaluation Committee (MEDEV).

As of 2015, MoCA has developed its body of work with different pilots, each of them centred around a specific orphan medicinal product submitted by a company. Tripartite discussions with representatives of the company developing the product, payers from various countries and patient representatives (ad hoc patient experts + EURORDIS representative(s)) have been taking place on a regular basis (please see section 3.3.3). 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 its 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 resulted 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 previous years, EURORDIS has continued 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.

EURORDIS continued to participate in regular roundtables convened by FIPRA International in Brussels and chaired by former UK Health Minister, John Bowis OBE (former MEP) and by the Chair of Belgium's National Health and Disability Insurance Board, Jo De Cock. In continuation of the work started the years before, these meetings have focused primarily on aspects related to equity of access and led by the end of the year to the adoption of a new paper on "[Dynamic outcomes based approaches to pricing and reimbursement of innovative medicines](#)" – due to receive attention and discussion at the 2<sup>nd</sup> EURORDIS Multi-Stakeholder Symposium on Improving Patient Access to Rare Disease Therapies in February 2017. EURORDIS also contributed to the [Recommendations of the European Working Group for Value Assessment and Funding Processes in Rare Diseases \(ORPH-VAL\)](#), which published its position statement in the Orphanet journal of rare diseases to help improve the consistency of pricing and reimbursement decisions on orphan medicines in Europe.

#### **Engaging with the European Parliament**

In 2016, the issue of access to medicines profiled high on the European agenda. In a context of growing concerns around the prices of medicines and their potential impact on the sustainability of healthcare systems in Europe, the European Parliament launched a so-called "Own Initiative Report" to address the issue in a legislative setting (even though the report will not lead to binding legislative). The "[Report on EU option to improve access to medicines](#)" drafted by Soledad Cabezon Ruiz MEP sparked a lively discussion on the topic of access to medicines.

EURORDIS participated to the debate on improving access to medicines by developing a written position in response to the amendments that relate to rare diseases and orphan medicinal products and organising meetings with relevant MEPs, political groups and members of the European Parliament's Environment and Public Health Committee's Secretariat. EURORDIS, in its position, sheds light on the actual impact of the EU Regulation on Orphan Medicinal Products, the role of incentives and the need for a new economic model based on extended collaboration at the European level.

The Report is due for adoption by the European Parliament in early 2017.

#### **To prepare the reimbursement decision: the HTA momentum**

##### **Scientific and technical cooperation on HTA in Europe: the role of EUnetHTA**

2016 was a pivotal year for Health Technology Assessment (HTA) cooperation in Europe, with the transition from the second joint action (EUnetHTA Joint Action 2) to a third one (EUnetHTA Joint Action 3).

European bodies performing HTA started to collaborate with each other in 2006, as a European project, firstly defining what an HTA report should contain, then preparing tools to work jointly, including the development of guidelines, and then testing the pilots they had created.

With this third joint action, which will be the last one, EUnetHTA, will launch series of joint assessments for both pharmaceuticals, and non-pharmaceuticals such medical devices, in vitro diagnostic tests, and complex surgery, connected devices etc., a total of 80 assessments between 2017 and 2020. In addition, EUnetHTA proposes Early Dialogue meetings with the developers of new technologies at an early phase of development, sometimes in parallel with EMA Scientific Advice, to reduce the risks that inadequate data are submitted at a later stage, when HTA bodies review the product.

The third Joint Action is the last one to be funded by the European Commission can fund, and another mechanism needs to be in place beyond 2020 for the continuation of the work.

## The role of patients

The main activities where patients can be involved are: joint assessment of health technologies, and early dialogues.

A series of meetings took place with the European Commission, other patients' and consumers' organisations involved in EUnetHTA, and the coordinator of EUnetHTA. A platform of patients and consumers' organisations will help identify, contact, prepare, train, accompany and debrief the patients who will volunteer to participate in the EUnetHTA procedures,

EUnetHTA JA3 will test various methods of patient involvement in HTA activities and learn from this experience.

## Relative effectiveness of pharmaceuticals

Following a kick-off meeting in Amsterdam in March 2016, a workshop was conveyed in June 2016 to discuss how to assess the relative effectiveness of pharmaceuticals once they are authorised by the European Medicines Agency. There are different techniques to compare a new medicine with other available treatments (sometimes other than pharmaceuticals), and a consensus is needed to perform European evaluation of the so-called therapeutic added value: does the new treatment work in real life as in clinical trials? Does it add something to available treatments?

The different techniques include head-to-head comparisons, indirect comparisons, meta-analysis, reviews of real word data etc. but different HTA bodies will not necessarily accept them.

Another important aspect is the timing: ideally, an HTA report should be published shortly after the marketing authorisation, for the decision makers to decide promptly to cover/reimburse the new medicine, or not. For this, HTA experts and regulators will exchange information during the evaluation process at the EMA, and HTA experts will access the draft conclusions of the EU regulators and scientific evaluation, before they become public, so they can start evaluating the medicine in question with efficacy and safety data.

## Political and strategic guidance of HTA in Europe: the HTA Network

The HTA network, composed of representatives of the Member States and the European Commission was created by the Directive on Patients' Rights to Cross-border care. EURORDIS is one of the patients' and consumers' organisations consulted by the HTA Network on the future of HTA cooperation in Europe.

In 2016, the HTA Network adopted [its multi-annual programme and a reflection paper on the synergies between regulators and HTA](#), to which EURORDIS contributed. As the EUnetHTA Joint Action 3 is the last of its kind, the HTA Network and the Commission engaged a process to consult all relevant stakeholders on a possible legislative proposal to create a permanent HTA scientific secretariat after EUnetHTA JA3 has ended. A total of 249 responses were received, including [EURORDIS position](#) following internal discussions with members.

## Monitoring the actual access to medicines after the reimbursement decision

### Shortages of medicines

Since the adoption in 2013 of a [Common Position on Medicine Supply Shortages](#) by EURORDIS and 45 patients',

[consumers' and healthcare professionals' organisations](#), important progress was made to remedy part of the causes that explain shortages.

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. Discussion with parties involved have continued to analyse shortages due to economic causes and identify possible solutions.

On 17 November 2016, the Slovak government organised a conference in Bratislava in the context of the EU Presidency, where EURORDIS presented its analysis of the situation and the proposals made by the 45 organisations. In July 2016, the Council of Ministers discussed a reflection paper with the possibility to create a forum to discuss all causes of shortages, including the economic ones.

### Off-label

Following a first survey on off-label use of medicines in rare diseases, launched in 2012, in 2016, the DITA Task Force launched an even larger survey on treatment information on rare diseases ('Tell us how you take your treatment'), with a questionnaire translated in 13 languages. 1,965 responses were collected. The results of the survey were presented by EURORDIS at the workshop on off-label use, held at the European Parliament, on 27 September 2016 "Safeguarding patient safety and quality of care in Europe: Good practice for the off-label use of medicines" and at the European Medicines Agency (Information Day on Risk Management Planning and Post-Authorisation Studies, 7<sup>th</sup> November).

Based on the results, EURORDIS proposes an Off-label Facilitation Group for rare disease treatments at the EMA, that would provide with a scientific opinion when national authorities would question an off-label use of a medicine.

## 1.2.7 Advocate to improve Access to Care for rare disease patients



*EURORDIS addresses issues related to difficulties faced by rare disease patients in accessing treatments, including through the Access Campaign, relevant activities on off-label use and information around shortages. The [EURORDIS Access Campaign](#) includes a survey for gathering patient experiences regarding access difficulties. The online questionnaire is available in 19 European languages and is permanently available on line.*

The number of responses received for the EURORDIS Access Campaign exceeded the expectations: 1,961, of which 1,844 from Europe, to be compared to 600 that were expected. In 2016, results were posted on EURORDIS web site and presented on several occasions such as at the meeting of the Council of National Alliances and European Federations (3 November 2016, Paris) and the European Union Group of Experts on Rare Diseases (28 November 2016, Luxembourg).



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

*In the course of 2016, EURORDIS has continued its action of support and promotion of rare diseases as a priority at both the policy and financial level within relevant EU programmes and policy frameworks. Specifically, EURORDIS:*

- + Continued promoting rare diseases as a research priority in Horizon 2020, notably within the Work Programme for Health 2018-2020 and in the Innovative Medicines Initiatives;
- + Continued supporting rare diseases as a public health priority in the 3<sup>rd</sup> EU Public Health Programme 'Health for Growth' in view of the mid-term review of the Programme, to shape the orientations of the last Annual Work Programmes and the redefinition of financial instruments such as the Operating Grants and the Joint Actions.
- + Promoted and took an active part in the development of the European Joint Programme on Rare Diseases for integration and long-term support of rare disease research infrastructures supportive of European healthcare networks and clinical research.
- + Launched and promoted the concept of European Rare Disease Clinical Research Network, embedded in ERNs and in EU Research Infrastructures and initiate recommendations through IRDiRC.

### 1.2.9 Advocate for progress in Patient's Rights to 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 continued monitoring the implementation of the Cross Border Healthcare Directive (Dir. 2011/24/EU), by taking actions in support of the implementation of its multiple strands:

- + The feedback of patients living with rare diseases has continued to be collected within the framework of the **EURORDIS Access Campaign**.
- + In coordination with other members, EURORDIS supported the European Patient Forum in the monitoring of the implementation of patients' rights' across the EU and at national level. In particular, EURORDIS contributed to the development and adoption of the **EPF Position Statement** on the Directive on patients' rights in cross-border care. The Statement sheds 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 provided specific contributions on the sections of the Statement related to Rare Diseases and European Reference Networks.

- + Throughout 2016, EURORDIS continued to support the establishment of European Reference Networks (ERNs) for rare diseases, notably through the participation to the Joint Action on Rare Diseases (RD-Action), the development of a matchmaking tool, the support to establishment of clinical groupings that paved the way to the applications of 24 candidate ERNs. In parallel, EURORDIS launched the European Patient Advocacy Groups and advocated for their successful integration within each ERN to ensure a democratic process of patient representation. For more details, please see section below.

### 1.2.10 Advocate for the development of the ERNs form and functions and the integration of ERNs with wider health, social and research infrastructures.

*From the early preparatory work on the EU Directive on Patients' Rights in Cross-Border Healthcare, through to the approval of 24 European Reference Networks, EURORDIS has been the central actor in the development of European Reference Networks, turning the initial idea into reality. Our advocacy work to create a framework for ERNs where patients and clinicians are equal partners started in 2005 and today spans over a decade. EURORDIS delivered a patient-centred vision for ERNs into 24 concrete therapeutic thematic networks, ensuring all rare diseases have a home in their combined structure, and providing an optimal framework to meet the multisystem needs of rare disease patients.*

In 2016, EURORDIS positioned the rare disease patient community as the cornerstone to the successful establishment of 24 European Reference Networks. EURORDIS achieved this through a three-fold implementation strategy centred on the needs of the rare disease community, specifically: Engagement and capacity building; Technical and strategic preparation and; Operational implementation.

#### Engagement and capacity building

EURORDIS proactively engaged with the rare disease community to raise awareness of ERNs taking concrete steps to implement the EUCERD Addendum recommendation to place patients at the heart of the governance and decision making structures of RD ERNs.

EURORDIS focused on developing the capacity of the 'grass-roots' rare disease community, specifically developing their knowledge and understanding of the newly developing ERNs, and establishing a solid technical understanding of the following ERN areas:

- + Assessment standards, criteria and process of ERN applications;
- + Network structure and activities required under the EC ERN Delegated Acts and;
- + Potential clinical services ERNs will offer to patients. in the future.

EURORDIS created European Patient Advocacy Groups (ePAGs), aligned to the scope of the different ERN

applications, in early 2016 to support an ERN informed rare disease community able to meet the ambitious goals set out in the EUCERD Addendum's recommendation for ERNs. The establishment of the ePAGs and election of ePAG representatives enabled a uniform and democratic structure for patient involvement in ERNs network boards, clinical committees and working groups. EURORDIS supported their involvement in the development of ERN applications through a series of individual ePAG focus groups with Network Coordinators throughout the year (for further information please see section 2.1.6.)

### Technical and strategic preparation

EURORDIS led the Consortium for Partnership for the Assessment of European Reference Networks Clinical Excellence (PACE-ERN) in order to develop the technical proposal for the European Commission's ERN assessment manual for ERN applications. PACE-ERN Consortium was 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 successfully delivered:

- + The final Assessment Manual and Toolbox for Applicants and for Independent Assessment Body(s) and the Operational Criteria
- + The Independent Assessment Body(s) training to implement the assessment model, manual and criteria for the ERN applications

EURORDIS contributed to the development of the 'Possible Services to be provided by the future European Reference Networks' workshops, developing the ERN concepts into tangible services.

EURORDIS is a partner in RD Action (Joint Action for Rare Diseases) and Joint Action for Rare Cancers and working with both Joint Actions on ERN related activities. Due to the multisystem needs of many rare diseases, EURORDIS has been developing interoperability mechanisms between the different ERNs and supporting the implementation of ERNs in line with the EUCERD recommendations and the strategic development of ERNs e.g. RD Action workshops on Virtual Healthcare and Data Sharing and Registry.

EURORDIS continues to implement its policy on ERNs, CoE, expert networks & healthcare pathways on rare diseases through: Promoting the long-term EURORDIS strategic vision on ERN through the CEGRD; Developing a fit for propose ERN IT Platform based on the needs of the ERNs; Developing an ERN sustainable business model through the VALUeHEALTH project; Connecting healthcare and research under ERNs and research networks or research infrastructure across the translational research pathway; Engaging with international consortia for undiagnosed conditions to raise awareness of ERNs and stimulate discussions on models for undiagnosed patients in ERNs.

EURORDIS is also a partner on the Joint action For Rare Cancers and took part in the JARC kick off meeting and kick off meetings for each work package, ensuring the work package scope is aligned to the needs of the RC ERNs.

### Operational implementation

EURORDIS supported the development of all ERN network applications, particular the structured development of patient involvement and empowerment into the

network governance structures, advocating for patient representatives to be formal voting members of the network boards (for further information please see section 3.1.)

#### 1.2.11 Advocate for Health Technology Assessment methods and a European HTA Agency which addresses rare disease patient needs

Regarding the European Commission Inception Impact Assessment for the future of the European Cooperation on HTA, EURORDIS expressed its preferences for a dedicated scientific secretariat. The scientific secretariat could be hosted by a new European Agency, or by the EMA. It seems however preferable to create a dedicated HTA agency, with strong synergies with the EMA, for a possible merger in the future. The agency could be funded by a mix of private (fees from industry) and public funding (EC, MS) for the evaluation of all technologies (medicines, complex surgery, complex medical devices, health apps, connected devices, in vitro diagnostics) for all domains (including cost and economic aspects, organisations, social, ethical, patients aspects), even for pharmaceuticals (but not systematically).



Ideally, some HTA agencies would conduct the assessment of a new technology jointly, and will use the joint work for their national procedures. Other agencies that do not participate in the joint work will also use the joint report for their own procedures (voluntary joint work and mandatory use of the work by all HTA agencies). There is a consensus on many methods for HTA, but not all. Typically, methods for cost and economic analysis differ greatly between agencies, and this is a source of incomprehension for European citizens. Therefore, EURORDIS proposes that research for consensus methods for all aspects where EUnetHTA partners did not come to an agreement yet would continue. This would also apply to horizon scanning methods for example.

Patients and their organisations that are invited to participate in HTA procedures, that agree with the methods upfront and with the consultation modalities, should accept the outcome of the assessment, even if disappointing. Duties and responsibilities should be defined for patients' organisations that accept to take part in a HTA.



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### 1.2.12 Advocate in support of rare disease research:

*Within Horizon 2020, the overarching programme for research and innovation of the EU, the research areas prioritised by the Health, Demographic Change and Wellbeing Programme include specific topics for rare disease research, in line with IRDiRC priorities and the Regulation establishing Horizon 2020. Moreover, the E-RARE ERA-Net is currently publishing joint calls for rare diseases with a view to better align national programmes.*

Two specific rare disease specific topics were launched in 2016 within the Work Programme 2016-2017:

- + **"New therapies for rare diseases"**, that followed the extremely popular call 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). With 65 MEUR available, the new call aims to fund between 10 and 15 clinical trials of substances that obtained orphan designation.
- + **"Diagnostic characterisation of rare diseases"**, a topic that aims to develop research on molecular diagnoses for a large number of undiagnosed rare diseases with a total amount of funding 15 MEUR.

On both topics, EURORDIS disseminated the relevant information to members via its usual communication channels. EURORDIS also participated as potential partner in consortia putting together applications for funding under the research topic on diagnostics.

#### European Joint Programme Cofund (EJP)

In view of the adoption of the Work Programme 2018-2020, EURORDIS focused its advocacy activities on the adoption of a new instrument for rare disease research, the European Joint Programme **Cofund**, that would ensure longer term sustainability of rare disease research as well as better integration of research initiatives in the field, including research infrastructures. This indispensable and transformative tool would integrate E-Rare research funding with a strategic policy framework for RD research. In view of the preparatory discussions, EURORDIS developed a document with insights and patients' preferred options for the new financial instrument. EURORDIS supported the adoption of the instrument as the best available tool to respond to the address research needs in the area of rare diseases; it also encouraged the connections of research infrastructures with ERNs as research conducive-environment, and the inclusion of training activities for patients, researchers and healthcare providers to be performed under the EJP Cofund.

#### Joint Research Centre (JRC) European Platform of Rare Disease Patient Registries

EURORDIS continued its support to the establishment of the Joint Research Centre (JRC) European Platform of Rare Disease Patient Registries. With Yann Le Cam appointment as a member of the JRC Advisory Technical Group, EURORDIS could participate to the first meeting of the group and participate to the Interoperability Workshop in November, 30th. It focused on the alignment of the JRC Platform on RD Registration with the European Reference Networks (ERNs) IT Platform and other RD-related (IT) platforms being developed in the framework of European Commission supported projects.

Thanks to the RD-Connect project and the Rare Barometer programme, EURORDIS was able to define the patients' position on patient preferences on informed consent and data protection for research in light of the new EU Data Protection Regulation.

Importantly, research on rare diseases was identified by EURORDIS' National Alliances as the focus of the Rare Diseases Day 2017 campaign, with the slogan, 'With research, the possibilities are limitless'. Throughout 2016, EURORDIS, its members and 'friends of Rare Diseases Day' all over the world worked to prepare the 2017 February events, an opportunity on a global scale to call on all researchers, universities, students, companies, policymakers and clinicians to do more research and to make them aware of the importance of research for the rare disease community.

At the international level, EURORDIS pursued its active participation to the International Rare Diseases Research Consortium (IRDiRC) launched in 2011. EURORDIS continued to be represented in the governing body of the IRDiRC, its Scientific Committees and Task Forces. In 2016, Yann Le Cam completed his mandate as Chair of the Therapies Scientific Committee (SC) that provides guidance targeting pre-clinical and clinical research for developing new therapies for rare diseases. Gema Chicano, EURORDIS Board member, and Virginie Hivert, Therapeutic Development Director at EURORDIS, are members of respectively the Interdisciplinary SC and Therapies SC.

With the ambitious goals of IRDiRC being reached ahead of time (200 new therapies and means to diagnose most rare diseases by 2020), in 2016 the reflection has started on the new goals for the initiative, in view of their discussion and launch at the 3<sup>rd</sup> IRDiRC conference due in early 2017 (Paris, 8-9 February 2017).

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### 1.2.13 Advocate to improve access to and quality of rare disease diagnosis:

*Throughout 2016, EURORDIS continued to advocate for improved access to and quality of rare disease diagnosis.*

#### Patients without a diagnosis

EURORDIS, together with SWAN UK (the support group run by Genetic Alliance UK), the Wilhelm Foundation, Rare Voices Australia (RVA), the Canadian Organization for Rare Disorders (CORD), the Advocacy Service for Rare and Intractable Diseases' stakeholders in Japan (ASrid) and the National Organization for Rare Disorders (NORD) jointly developed and released in October 2016 an article listing **5 recommendations to address the specific needs of patients without a diagnosis** and urging all stakeholders to recognise undiagnosed patients as a specific population within the rare disease community.

EURORDIS is contributing to the development of a formal network of patient associations specifically supporting patients living with syndromes without a name and rare conditions within the different European countries (SWAN Europe). After a first meeting in Rome in April 2016, terms of reference for prospective members have been developed and activities for the network are being defined (upcoming workshop in March 2017 in Barcelona).

An international network of clinical centres, Undiagnosed Diseases Network International (UDNI), was initiated in



2014 to address unmet needs of undiagnosed patients at a global level. EURORDIS has attended 2 meetings held in 2016 (Vienna, February and Tokyo, November). NORD, EURORDIS and the Wilhelm Foundation have collaboratively developed and proposed a patient engagement structure for the board of the UDNI to consider during the last meeting. EURORDIS also presented the international joint recommendations for undiagnosed to this consortium which have now been endorsed by UDNI and can be downloaded from the UDNI website.

### Genome editing

EURORDIS participated in several meetings and workshops in 2016 on the topic of genome editing.

- + “Responsible use of genome editing/CRISPR/Cas9 in research” organised by the French Inserm Ethics Committee with a wide range of European stakeholders. Stakeholders (including EURORDIS) were able to present diverse positions and to suggest recommendations captured in a manuscript currently in submission to an academic journal. The resulting general principles aim at guiding research involving genome editing technology and ensuring satisfactory compliance with ethical standards.
- + “Human genome editing in the EU” hosted by the Federation of European Academies of Medicine (FEAM), the UK Academy of Medical Sciences, and the French Academy of Medicine.

Due to an intense interest from patient groups on the recent development in genome editing technology, EURORDIS together with a dedicated working group on genome editing comprising several members of the Patient Advisory Council of RD-Connect have organised and held an interactive webinar to discuss current scientific advances and related ethical issues. This webinar aimed to start informing and engaging rare disease patient

representatives on the complex topic of genome editing. Around 40 people, mostly patient representatives from rare disease organisations joined the webinar on Tuesday 20<sup>th</sup> October and were able to actively participate by asking questions directly to the experts. This working group, coordinated by EURORDIS also organised a follow up workshop on 4<sup>th</sup> November in Paris for rare disease patient representatives across Europe to participate in an open discussion with experts on several aspects of gene therapy and genome editing specifically on scientific progress and its relevance for rare diseases, associated ELSI issues as well as on the perspectives from the regulatory and biotech sectors.

### Patient involvement in Biobanks & Registries

**RD-Connect:** RD-Connect is a global infrastructure linking up data from rare disease research projects in a central resource for researchers across the world. It is developing an integrated platform in which omics data will be combined with clinical phenotype information and biomaterial availability, accessible online and query able with a suite of analysis tools.

Input of patient representatives into RD-Connect activities is managed by EURORDIS through the Patient Advisory Council and Patient and Ethics Council, which have been highly active throughout the project to date and provided valuable guidance on the project’s direction, particularly in ethically challenging areas relating to data sharing where risk and benefit must be carefully evaluated. To enable patient representatives to have a more direct and visible input into RD-Connect activities, in 2016 members of the Patient Advisory Council have been nominated to engage with each of the RD-Connect technical work packages. This not only enables the technical experts to have direct input from the PAC, but also strengthens the commitment and engagement of the PAC members, supports capacity building, and improves dissemination of the project’s



outputs to the wider rare disease patient community. Downstream communication of the project's activities will also further improve with the launch of a dedicated section for patients on the website that is developed and managed by patients themselves. The two-way exchange of information extends beyond RD-Connect, with regular participation of patient representatives in other European consortia and networks to ensure that rare disease patients' needs are integrated within the development of best practices in RD research. This includes activities such as the development of the EU Platform on Rare Diseases registration by the JRC and the BBMRI stakeholders' forum.

**Biobanks:** EURORDIS is a member of the BBMRI Stakeholder Forum and participated in several meetings in 2016 (chaired by Alistair Kent, Genetic Alliance UK). BBMRI-ERIC and representatives of patient advocacy groups representing areas of expertise on genetics, rare diseases, chronic diseases, healthy ageing/prevention, degenerative diseases, cancer, obesity, and infectious diseases met to identify the key topics for the years to come and define the detailed parameters of how a constructive dialogue can be put into place. The group discussed specifically the points to consider from a patient perspective within the upcoming code of conduct to support the future implementation of the General Data Protection Regulation. In line with RD-Connect activities of the PAC, EURORDIS raised issues related to historical samples, informed consent, return of results, carrier status and sustainability. EURORDIS also participated in the Europe Biobank Week that was held in Vienna in September 2016.

**Registries:** EURORDIS participated in the patient registry workshop organised by findacure in London on 16<sup>th</sup> September by giving a presentation to the participants

(patient groups from the UK) on information about the different types of registries, the types of data used in registries, explaining how these can be beneficial and useful for rare disease patients, why patient groups should be involved in registries and how RD-Connect supports patient registries.

EURORDIS participated in the first meeting of the technical advisory group and the first Interoperability workshop (organised by the JRC in the framework of the EU Platform on Rare Diseases Registration 7-8 April 2016 in Luxembourg). EURORDIS gave a presentation on the interoperability expectations from the perspective of patients, highlighting the need to assure coherence with tools developed within RD-Connect, to ensure patients empowerment and capacity building for adequate and full involvement of patient representatives in the governance and activities of rare disease registries and reinforce the commitment of all stakeholders involved by developing a collaborative and flexible framework.

### IRDIRC/GA4GH Joint Task Force

EURORDIS is a member of a Joint Privacy-Preserving Data Linkage (PPRL) Task Force between IRDiRC and Global Alliance for Genomic Health which aims to explore and recommend one or more approaches to linking data records within and across institutions while preserving individual privacy. Following a face to face meeting in Paris in December 2016, the team agreed to further explore an existing approach developed through the European Network for Cancer Research in Children and Adolescents (ENCCA) with some modifications for increased flexibility to accommodate a range of jurisdictional laws (including the use of a model allowing federated regional storage systems), and increased scalability.

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## 1.2.14 Voicing the social needs of people with a rare disease and advocating for the integration of rare diseases into social policy



In the first trimester of 2015, EURORDIS concluded the drafting of the Commission Expert Group Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies, following EURORDIS' previous work within the EUCERD Joint-Action Working for Rare Diseases (EJA) and as part of a drafting group coordinated by the European Commission. The recommendations, unanimously adopted in by all MS last April, advise MS and the EC on issues that should be considered when organising holistic care of RD patients within national health and social care systems.

EURORDIS has contributed to the wide dissemination of the recommendations amongst RD and social stakeholders. ECRD 2016 Edinburgh provided the opportunity for EURORDIS to invite the EC to present the recommendations to RD stakeholders at European level and to promote case studies and reflections supporting the implementation of the recommendations.

During this last year, EURORDIS also continued to support national alliances to promote the integration of rare diseases into social policies and services via the capacity building workshop at EMM 2016 Edinburgh, focused social policy and social innovation, and via the organisation of a webinar to support the organisation of national workshops focused on social services and policies.

In other to further voice the needs of people living with a rare disease and their carers, EURORDIS launched its long awaited survey on the impact of RD on daily life, within the scope of the INNOVCare project and via the Rare Barometer programme. The survey was widely disseminated via EURORDIS and its members. Directed at patients and carers, the questionnaire was composed of questions on the challenges generated by RD, on access to services and coordination of care, as well as on questions regarding employment, school and the economic burden of RD. EURORDIS will be publishing the results in 2017.

From the second semester of 2016, EURORDIS reinforced its social policy advocacy activities at European level, initiating a map of stakeholders and policy opportunities at EU level in the social field, including within issues of social services, disability and employment.

Finally, with the active engagement and support of the Social Policy Advisory Group, EURORDIS contributed to the European Commission open consultation on the European Pillar of Social Rights, ensuring that the needs of people living with a RD and their carers are represented in this consultation feeding into the future reference framework to screen the employment and social performance of MS.

Further actions done by EURORDIS to promote the integration of rare diseases into social services and policies as well as the integration of health and social services,

are described in the section "patient engagement in social care".

The Social Policy Advisory Group has provided continuous grassroots and expert input, advice and support to the different activities and actions.

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

EURORDIS has been working on the promotion of rare diseases as an international public health priority for almost a decade. The aim has been to directly promote rare diseases towards relevant institutions at international level; to provide advocacy tools for patient groups to advocate towards their national authorities thereby serving as a basis for patient empowerment locally; and to enhance international cooperation in the field of rare diseases.

To reach this goal EURORDIS has been implicated in the following initiatives

#### Rare Diseases International



Rare Diseases International (RDI) is an initiative that aims at creating an informal 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 voice

In 2016, EURORDIS played a central role in the efforts for the development of an international rare disease movement together with the major international patient organisations with which EURORDIS has signed partnership agreements. Rare Diseases International continued to grow and promote rare diseases as an international public health priority; the RDI governance structure was consolidated and the first RDI Council was elected in May 2016 hosted back to back with the EURORDIS Membership Meeting 2016 Edinburgh. In addition, Rare Diseases International supported the establishment of the NGO Committee for Rare Diseases and its official launch at the United Nations.



## NGO Committee for Rare Diseases

The NGO Committee for Rare Diseases is a substantive committee established under the umbrella of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO), the principal interface between the world's NGOs and the UN system. Its goal is to bring visibility and understanding about rare diseases to the UN and to promote rare diseases as a priority in global health, research, and social and medical care as part of the UN 2030 Agenda: the Sustainable Development Goals (SDGs), the bearing principle of which is: "Leave no one behind".

The Committee is now one of the CoNGO Committees. A Resolution for its creation was adopted at the 2014 CoNGO General Assembly after 34 members voted in favour of its creation.



Its formal existence was declared by the CoNGO President at an instigation meeting on 20 October 2015 in New York. At the beginning of 2016, a series of meetings were set-up in New York to establish contact with United Nations agencies and Permanent Missions to sensitise them to the Committee's cause and gather their support in the organisation of a first official meeting. The Committee was publicly launched at the United Nations in November 2016, with the presentation of the Founding Act, endorsed by all participants and with the decision to work towards securing a UN General Assembly resolution on rare diseases as a long-term goal. In 2017, the NGO Committee for Rare Diseases will establish its Executive Board and populate its broader membership with civil society partners.

### Official launch of the NGO Committee For Rare Diseases at the United Nations – 11 november 2016

The first official meeting of the NGO Committee for Rare Diseases took place on 11 November 2016 at the United Nations headquarters in New York under the patronage of her majesty Queen Silvia of Sweden and with the active support and encouragement of the governments of Belgium, France and Malta, the Permanent Mission of Estonia to the UN and the Swedish Ministry of Health and Social Affairs. Over 100 participants from all continents were present on the day coming from civil society, UN bodies, national governments, academia and industry.

During the meeting, the coherence between the challenges of the rare diseases community and the SDGs Agenda was confirmed by members of United Nations agencies, in particular when considering the bearing principle of the agenda: "Leave no one behind". The endorsement of the Founding Act by all those present made it clear that

great opportunities for global action and advocacy for rare diseases exist and can be facilitated through closer collaboration with UN agencies. The NGO Committee for rare diseases vowed to take-on precisely this role, acting as an advocacy platform working towards securing a UN resolution on rare diseases.

“ If we are serious about leaving no one behind, then we cannot leave behind people who have rare diseases just because they are few”

Nata Menabde, World Health Organisation,  
11 november 2016, UN HQs



## 1.3 Advocate Rare Diseases as a Priority in the next Decade 2020-2030

In 2016, EURORDIS initiated the development of the concept of a **European Parliament Interest Group on Rare Diseases** to be subsequently expanded into a **Rare Disease Network of Members of national and regional parliaments**, in collaboration with National Alliances. A first design of was proposed

During the entire year 2016 EURORDIS advocated to secure funding for a **Foresight Study on Rare Diseases in 2030** to identify long-term policy scenarios with participatory methodologies ("Rare 2030: a participatory foresight study for policy-making on rare diseases"). In November 2016, the Pilot Project for a Foresight Study was presented to the Commission Expert Group on Rare Diseases to Member States representatives to promote its relevance for the long-term RD policy-making at the EU and national level.

In particular, EURORDIS cooperated with MEPs that supported the presentation of a Pilot Project at the European Parliament as well as relevant European Commission services in order to secure financial and political support. Promoted by Françoise Grossetête MEP, Frederique Ries MEP and Cristian Busoi MEP, the Pilot Project received the support of the European Parliament throughout the entire budgetary procedure for the year 2017 at the European Parliament although it was not

endorsed by the Council (December 2016).

In 2016, EURORDIS started its advocacy towards the adoption of a new **EU Council Recommendation for a EU integrated national strategy on rare diseases** to be adopted before 2020 and the adoption of the new EU Multiannual Financial Framework 2020-2025 and related Programmes. The viability of the concept was tested with top European Commission officials and then with the Maltese government, in view of the upcoming Presidency of the EU Council (first half of 2017). In parallel, EURORDIS received the support of key Members of the European Parliament who drafted a letter of support addressed to the Presidency of the EU Council, inviting the Council to call for a new Council Recommendation. The letter was eventually sent in early 2017. The case for new soft law ten years after a successful Council Recommendation in 2009 was made and well received by all involved actors.

Last but not least, in 2016 EURORDIS started to explore feasibility of the adoption of a **Resolution of the UN General Assembly on Rare Diseases** through the UN NGO Committee for Rare Diseases and a core group of UN Member States in collaboration with UN Health Diplomacy group, RDI and all stakeholders.

## 1.4 Gathering patient experience and perspective for evidence-based advocacy

### EURORDIS Rare Barometer Programme: Generating new data from patient experience

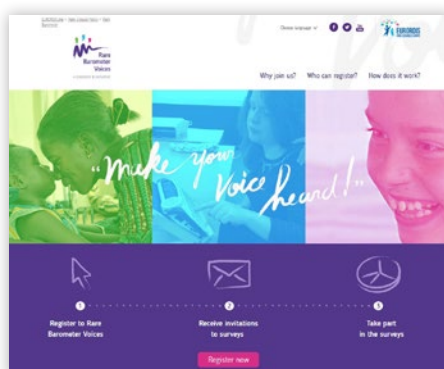
*Rare Barometer was created and launched in 2015 and consists of surveys aiming to collect qualitative & quantitative data on the experiences, needs & expectations of RD patients, their families and carers in order to facilitate and streamline the inclusion of patient perspectives in EURORDIS policy and decision-making processes. As part of the Rare Barometer Programme, Rare Barometer Voices, an online panel of people living with a rare disease who are willing to participate in EURORDIS' surveys and studies, was also launched. The webpage and the surveys are translated in 23 languages and the project covers 48 countries.*

Throughout 2016, a big effort was made for the wide dissemination of Rare Barometer and the recruitment of members. Communication material was designed (leaflets, email template that describe the project, face book posts) and translated in 23 languages to facilitate the patient organisations dissemination work. 4300 Rare Barometer Voices participants have been recruited so far.

The first quantitative survey (Impact of rare diseases on daily life stage 1, 3000 participants) and the second one (Impact of rare disease on daily life stage 2, 2000 participants) have been carried out. The two surveys were available in 23 languages and carried out in 48 countries. The first stage dealt with coordination of care, access to social care services and cost of the disease. The second

stage with the impact of rare diseases on employment, schooling and well-being. The survey was designed with the help of the Topic Expert Committee, composed of EURORDIS member organisations, academic and corporate partners.

Focus groups on genomic data sharing were carried out and analysed. 22 rare diseases patients participated. The focus groups took place during the EURORDIS Summer School and participation in the focus groups was volunteer-based among EURORDIS Summer School participants. This event offered the possibility to gather English-speaking rare disease patients, coming from different countries/ regions of the world and representing a various range of diseases. The report of the qualitative study was finalised in October.





# 2. PATIENT EMPOWERMENT: Building the network & capacities

## 2.1 Community-Building, Networking & Capacity-building of Patient Advocates

### 2.1.1 Membership

51 new members joined EURORDIS in 2016 with the addition of one new country: Lithuania.

At the end of 2016, EURORDIS had 733 members in 64 countries, 39 of which are European countries, 27 being members of the European Union.

### 2.1.2 EURORDIS Membership Meeting 2016 Edinburgh

*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 comprised the Annual General Assembly and several workshops. The majority of participants (75%) are EURORDIS member organisations and other patient organisations and about 25% of the participants represent policy makers, industry and academia. Each Membership meeting has specific focus on issues of high importance.*

The EMM 2016 Edinburgh took place May 26, back to back with the European Conference on Rare Diseases and Orphan Products, ECRD. The programme committee of EMM 2016 was made up of 10 members, including EURORDIS Directors and staff.

The event attracted 200 patient representatives from 31 countries. At the Extraordinary General Assembly, the updated statutes of EURORDIS were discussed and

approved. Following the General Assembly, four Capacity Building Workshops were held: European Reference Networks (ERN) and ePAGs; RareConnect; Social Policy and social innovation; Good clinical practice in the context of ERN.

EURORDIS offered 40 travel fellowships to Patient Advocates from 17 countries.

### 2.1.3 Council of National Alliances (CNA)

*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 39 national alliances, 36 of which constitute the CNA.

The CNA's main activities in 2016 were 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 ERNs. As always, the CNA also worked on the preparation and coordination of Rare Disease Day 2016.

By the end of 2016, 26 National Alliances within Europe had signed the Common Goals & Mutual Commitments document.



#### 2.1.4 Council of European Federations (CEF)

*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 November to discuss issues that are important across Europe and across diseases. For the third time, part of the meeting took place in conjunction with the CNA (Council of National Alliances) to discuss cross cutting issues and share experiences. This part of the meeting focused on: ERN & ePAGS; Public consultation on the future of HTA cooperation; Position paper "International joint recommendations to address specific needs of undiagnosed RD patients"; Position paper "Compassionate Use"; Rare Barometer & INNOVCare project.

The second part (CEF only) focused on What's next for the CEF? Moving towards common goals? ; how are ePAGS impacting on European Federations?; RD Action concept paper on Prevention; Peer-to-peer session. The second day of the meeting was dedicated to a training on Genome Editing. This training session was also open to National Alliance representatives and other stakeholders and gathered 61 participants.

EURORDIS continued for the 7<sup>th</sup> 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). In 2016, EURORDIS gave 13 European RD Federations financial support to help them organise their different meetings. A total of 23 650 € was granted for 18 meetings.

#### 2.1.5 European Network of Help Lines for Rare Diseases

*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.*



Help Lines  
for Rare  
Diseases

In 2016, 15 help lines from 11 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, Myastenia Gravis Romania), Spain (SIO-Feder and ENERCA), Switzerland (Info Maladies Rares), Hungary (Lifebelt, Information Centre for the Rare Disease Patients), Denmark (Rare Disorders Denmark, and Ireland (National Rare Diseases Office).

In addition to its annual face-to-face meeting, the network conducted its 9<sup>th</sup> Caller Profile Analysis in October, based on all enquiries received. The network also had a discussion on which resources exist to respond to enquiries about natural, alternative, complementary and traditional medicine.



## 2.1.6 European Patient Advocacy Groups (ePAGs) & European Reference Networks

### European Patient Advocacy Groups (ePAGs)

In early 2016, EURORDIS created the European Patient Advocacy Groups (ePAGs), aligned to the scope of the different ERN applications, to support an ERN informed rare disease community able to meet the ambitious goals set out in the EUCERD Addendum's recommendation for ERNs. The establishment of the ePAGs and election of ePAG representatives enabled a uniform and democratic structure for patient involvement in ERNs network boards, clinical committees and working groups.

Following the successful elections of 85 ePAG representatives, EURORDIS supported their involvement in the development of ERN applications through a series of individual ePAG focus groups with Network Coordinators throughout the year. Patient representatives contributed (and co-authored) the ERN vision, scope and strategic plans outlined in the network applications. Throughout 2016, EURORDIS continued to identify patient representatives to be actively involved in and formal members of the ERN disease specific clinical networks.

EURORDIS also reach out to the wider patient community through an ePAG matchmaker initiative. Nearly 1000 patient groups were successfully mapped, both members

and non-members of EURORDIS, aligning them to the scope of the thematically grouped ERNs. These patient groups form a wider patient stakeholder group for the ePAG representatives and Network Coordinators to link in with and canvas their views to focus their ERN engagement and communication activities accordingly.

Furthermore, EURORDIS developed a ePAG Leadership Programme with the aim to build the capacity of ePAG representatives to act as equal and valued partners in the ERN governance structure. In 2016, EURORDIS led two ePAG Focus Groups to capture the needs of ePAG representatives and tailor the scope and content of the ePAG Leadership Programme. The ePAG Leadership Programme's modules consist of:

- + Capacity building webinars and bit-size seminars on technical aspect of ERNs, e.g.: care pathway, registries, virtual healthcare and data sharing, etc.
- + Peer coaching support through regular virtual group meetings
- + Leadership training to enhance the impact of the patient message
- + Mentoring programme to give independent leadership support and space for reflection

### Building the capacities of the EURORDIS membership & their readiness to support European Reference Networks

EURORDIS led capacity building activities to support EURORDIS' membership, National Alliances and European Federations and Networks be ready for the first call for European Reference Networks applications, specifically at the:

- + European Conference for Rare Diseases & Orphan Products – Edinburgh, dedicated conference stream on Game Changer in Care Provision with five ERN sessions (May 2016) covering, 1. The Establishment of the ERN model for rare diseases; 2. How to structure healthcare for better health outcomes; 3. Health Outcomes; 4. Interoperability is a state of mind, and; 5. Will I benefit from an ERN in reality?

- + ERN capacity building workshop at EURORDIS Membership Meeting, Edinburgh (May 2016) developing understanding and knowledge of the emerging ERN landscape and structuring patient involvement through ePAG groups
- + National Alliance webinar advocating on anchoring ERNs into national healthcare systems
- + CEF & CNA capacity building, Paris (November 2016) focused on outcome of the technical assessment, update on ERN applications and the development of ePAG groups
- + EURORDIS continued to proactively communicate about new developments of ERNs over the year, restructured and updated EURORDIS ERN specific webpages, EURORDIS eNews and updated fact sheets on ERNs.



**RARE  
DISEASES  
INTERNATIONAL**

A EURORDIS INITIATIVE

## 2.1.7 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 voice.*

The 2<sup>nd</sup> RDI annual meeting was held on May 25th, back to back with the ECRD Edinburgh 2016. Almost 90 participants attended, mostly patient organisations (RDI members, national alliances, international federations) and research organisations, selected donor companies. The meeting was preceded by the nomination and election of the first RDI Council, the governance structure of Rare Diseases International. Members elected to the Council were:



- + John Dart, Debra International
- + Ramaya Muthyala, I-ORD
- + Durhane Wong-Rieger, CORD
- + Megan Fookes, RVA
- + Lisa Phelps, NORD
- + Yann Le Cam, EURORDIS
- + Alfredo Toledo, ALIBER

6 out of the 7 elected members of the RDI Council were present at the RDI meeting and presented the 3 year RDI Action Plan. In addition they attended a short meeting for members only where they presented themselves and spelled out their vision for the initiative. Throughout the year, the RDI Council worked on the development and adoption of the RDI by-laws and the organisation of an RDI Policy Event to be held in Geneva in 2017.

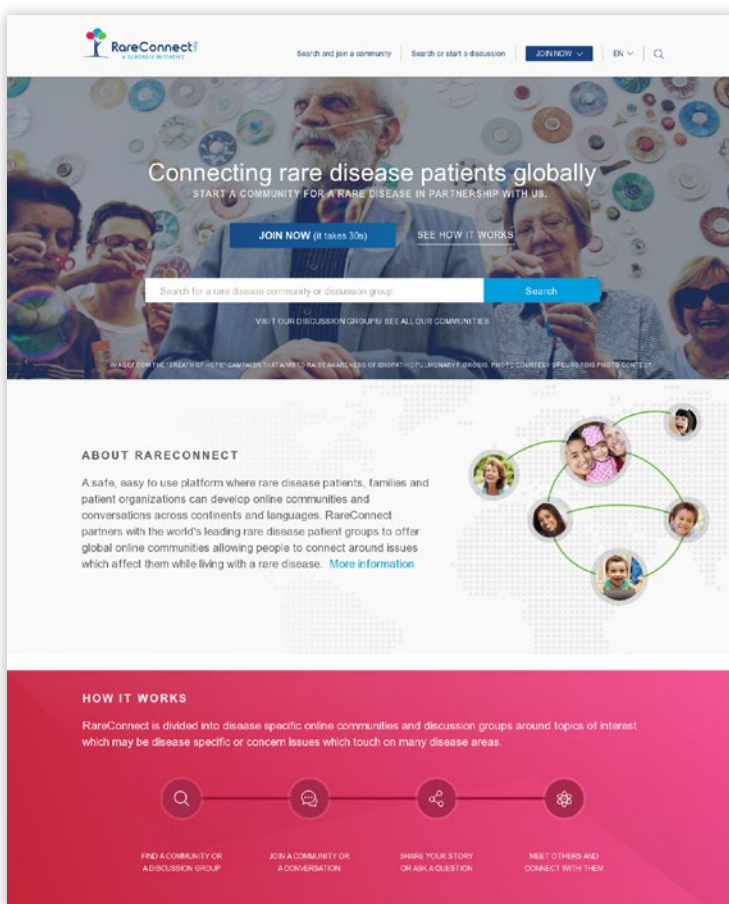
### 2.1.8 RareConnect

*RareConnect.org is a EURORDIS initiative which provides a platform for rare disease patients and patient organisations 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 90 disease-specific communities created in partnership with 750 patient groups and managed with the support of 305 volunteer moderators.*



2016 saw RareConnect plan for future growth by investing in changes which will allow it to deliver an improved user experience to mobile visitors who now account for over 50% of traffic to the site. These innovations are planned for release in 2017. The platform also added 3 new languages: Russian, Serbian and Croatian as well as a new map feature allowing visitors to easily find other affected patients or families near them.

During 2016 the RareConnect team organised several capacity-building activities including a workshop during the European Conference on Rare Diseases in Edinburgh as well as webinars on topics such as organising an awareness day, gene therapy or usage of twitter for rare disease patient advocacy. The RareConnect team also organised several web meetings to build relationships between researchers and patient organisations.



The global reach of RareConnect continues to grow with traffic during 2016 nearing 1 million visitors from 226 countries. Subscribed members surpassed 26,000 amounting to a 30% increase compared to 2015.



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### 2.1.9 Volunteers

*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.*

In 2016, EURORDIS was proud to rely on **447 volunteers** including **89 dedicated volunteer patient advocates**, **70 volunteer ePAG representatives**, **1 for fund raising**, **1 office support volunteer** and **286 volunteer moderators** of online communities of Rare Disease Patients, within the activity “RareConnect” (for more detailed information please refer to the volunteer section in cross-cutting).

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### 2.1.10 Webinars

EURORDIS webinars offer an interactive way to engage with members and the wider public. Members can participate from wherever they are at no additional costs.

Webinars focus on: providing policy updates (for example on European Reference Networks); involving patients in consultations (for example the draft EURORDIS access position paper); providing capacity-building trainings for patient advocates (for example on ERNs in context of national plans); or can be informative on a topical subject (for example genome editing).

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### 2.1.11 EURORDIS Trainings

#### 2.1.11.1 ExPRESS: Expert Patients and Researchers EURORDIS Summer School

*The EURORDIS Summer School was initiated in 2008 as part of our continued commitment to empowering people living with rare diseases. This four-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 2016 Summer School was called “ExPRESS 2016” for Expert Patients and Researchers and provided participants with a fundamental understanding of the processes involved in medicines development, the time required and the different stages of clinical research.

For the 2016 version, a much greater emphasis was placed on the pre-training aspect of the Summer School Programme. The pre-training is comprised of training blocks that the trainees can complete online before they arrive in Barcelona for the face-to-face session.

The training session in Barcelona is designed to maximise interaction between the trainees and trainers but also amongst the trainees themselves but also to present “state of the art” developments in patient advocacy. All of the trainers have many years of experience interacting with patients, some of them are indeed patients themselves. They are also members of EMA committees, ethicists, professors of pharmacy, academics, regulators, and payers many of whom have been involved in the Summer School since it was launched in 2008.

44 participants attended representing 13 countries and 28 diseases. The participants were selected based on a call for candidates.

This year in partnership with a COST Action BM1207, nine researchers were able to participate. Five participants representing Nutrition and Medical Devices were also included in the EURORDIS Summer School in partnership with European Clinical Research Infrastructure Network project (ECRIN).

#### 2.1.11.2 Training Resources Centre on the EURORDIS website

*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.*

As of 2016, a 5-month pre-training programme has been formalised and implemented for accepted participants of the Summer School. The pre-training is comprised of training blocks that the trainees can complete online before they arrive in Barcelona for the face-to-face session. Each block or “Unit” covers specific topics such as Medical Research Ethics, Statistics, Regulatory Procedures, European Medicines Agency, Benefit Risk, HTA and Market Access. The programme can be followed by any interested individuals via the EURORDIS website at time and place of their choice.

#### 2.1.11.3 Participation to EUPATI

*The European Patients’ Academy (EUPATI) is a pan-European Innovative Medicines Initiative project of 33 organisations including the European Patients’ Forum, the European Genetic Alliance, the European AIDS Treatment Group, and EURORDIS, universities, not-for-profit organisations and twenty one pharmaceutical companies members of EFPIA. The project ran from February 2012 to January 2017. Its aim was to train patient experts, advocates, patients and the public on medicines development in order to increase the capacity and capability of patients to understand and contribute to medicines research and development and to improve the availability of “objective, reliable, patient-friendly information for the public”.*

EUPATI developed training and education materials for three audiences. Audience 1 consisted of 100 patient experts who followed a 12 month blended learning programme. EURORDIS played a key role in developing the syllabus for the course and providing training materials as well as organising the two face-to-face 4.5-day programmes that were run for each of the two cohorts. Conducted in English, the course were generally well accepted. Audience 2 was 12000 patient advocates who were given access to online training in several languages via a web-based educational toolbox, aiming to reach patient advocates across Europe. Audience 3 provided information for the public with the aim of targeting 100 000 Europeans. The reach to audience 3 was evaluated based on social media interest in the project.

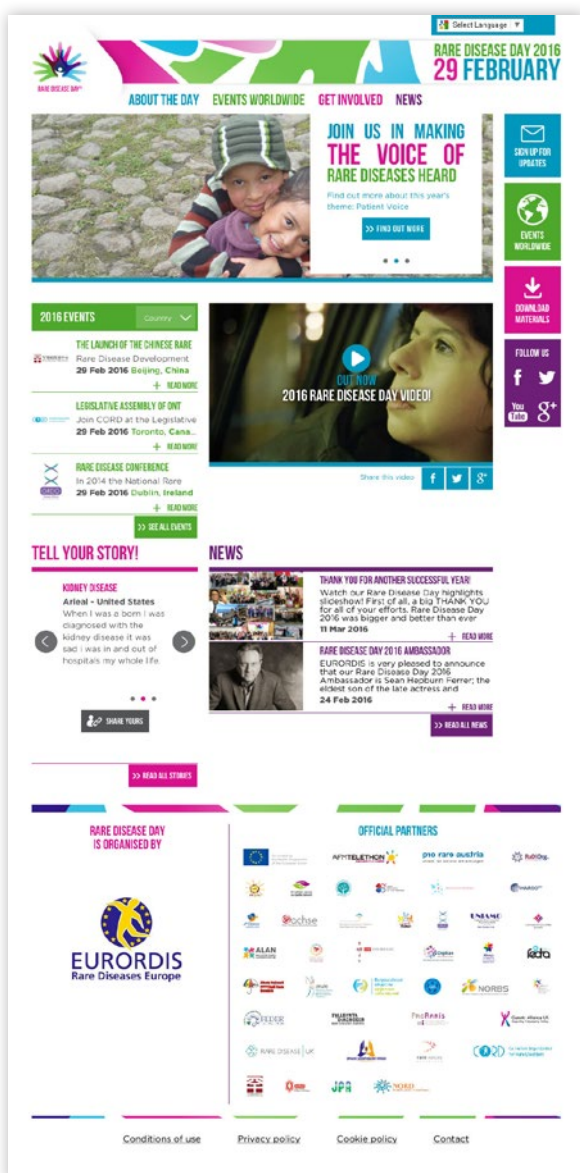
EUPATI offered a platform for patients to learn from each other and share experiences. This is probably the most valuable outcome of the project. The trainees in Audience 1 forged strong ties which they plan to maintain via an alumni network. A second phase called EUPATI 2 has been launched in 2017. EURORDIS will continue its involvement albeit at the more modest level.

## 2.2 Raising Awareness & Informing

### 2.2.1 Rare Disease Day 2016

*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.*

The 9<sup>th</sup> edition of the Rare Disease Day Campaign was held on 29<sup>th</sup> February 2016. The campaign was implemented in over 84 countries, including all 28 EU countries plus 10 new countries Aruba, Indonesia, Libya, Tanzania, Uganda, Zimbabwe, Tunisia, Andorra, Mauritius, Moldova. EURORDIS and 39 Rare Disease National Alliances, together with patient groups acting as country organisers, mobilised thousands of patient organisations throughout 5 continents. Events around the world were held by over 400 associations in Europe, North and South America, Africa, Asia and Australia.



The new look for the Rare Disease Day 2016 website was more dynamic and engaging. Average pages visited per session went up by 81%. This year there were 178 patient organisations and alliances signed up on a dedicated section of the website called 'Friends of Rare Disease Day', which includes several public institutions. The website is tablet and mobile responsive making the Rare Disease Day site the most accessible it's ever been. The number of Mobile phone users reached a peak of 60% of the total visitors on 29 February viewing from a smartphone.

For the 5<sup>th</sup> year in a row EURORDIS produced a well-received video for Rare Disease Day that exemplified this year's theme of Patient Voice and celebrated the special moments in the lives of people living with a rare disease. The video's success was in part due to the star performer Elisa, who is living with Williams syndrome and her parents Sergio and Catia. EURORDIS worked with Enfin Bref Production, Publicis Life Brands International, Renaud Cohen and the Théâtre du Châtelet in Paris, who all made the production of the video possible. The video was translated into over 34 languages, which is up from 27 languages in 2015.

As with previous years, EURORDIS hosted a **symposium** in Brussels on the occasion of Rare Disease Day (for further information please refer to the relevant item in the Advocacy section of the report).



## 2.2.2 European Conference on Rare Diseases & Orphan Products - ECRD 2016 Edinburgh

*The European Conference on Rare Diseases & Orphan Products is the unique platform/forum across all rare diseases and all European countries, bringing together all stakeholders – patient representatives, academics, health care professionals, industry, payers, regulators and policy makers. It is a biennial event, providing the state of the art of the rare disease environment, monitoring and benchmarking initiatives. It covers research, development of new treatments, health care, social care, information, public health and support at European, national and regional levels. It complements national and regional conferences, enhancing efforts of all stakeholders. There is no competition with them, but efforts fully respect initiatives of all, aiming at integrating EU and national policies and actions.*

ECRD 2016 took place at the Edinburgh International Conference Centre (EICC) on 27 - 28 May 2016 in Edinburgh, Scotland, UK. 757 people attended from 48 countries, representing all stakeholders. 115 session chairs, speakers and panellists participated. 39 fellows were present on-site representing 17 countries.

The official website ([rare-diseases.eu](http://rare-diseases.eu)) was available in six languages: English, German, French, Spanish, Italian and Russian. A mobile app available for the second time with 50% of participants having downloaded it.

The conference partners included: DIA (co-organiser), the European Medicines Agency (EMA) and in particular the Committee for Orphan Medicinal Products (COMP), FDA's Office of Orphan Products Development (OOPD), NHS National Institute for Health Research, Orphanet, the European Society of Human Genetics (ESHG), European Hospital and Healthcare Federation (HOPE), EuropaBio-EFPIA, European Confederation of Pharmaceutical Entrepreneurs (EUCOPE), the European Federation of Internal Medicine (EFIM), Genetic Alliance UK and The Scottish Government.

An 8-page Executive Summary of ECRD 2016 was prepared and disseminated by EURORDIS to over 7000 contacts and by the conference partners.

ECRD 2018 will be held in Vienna, Austria on 10 – 12 May 2018.



26-28 May 2016 Edinburgh

European Conference on Rare Diseases & Orphan Products



## 2.2.3 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 360.000 visits annually.*

The EURORDIS website provides information relating to the role of patient organisations in the development of rare disease and orphan drug policy in patient-friendly language translated into 7 languages, while also outlining the activities provided by EURORDIS.

New website sections added in 2016 include the new Get Involved section, the section on European Reference Networks

## 2.2.4 eNews & Member news

### 2.2.4.1. eNews

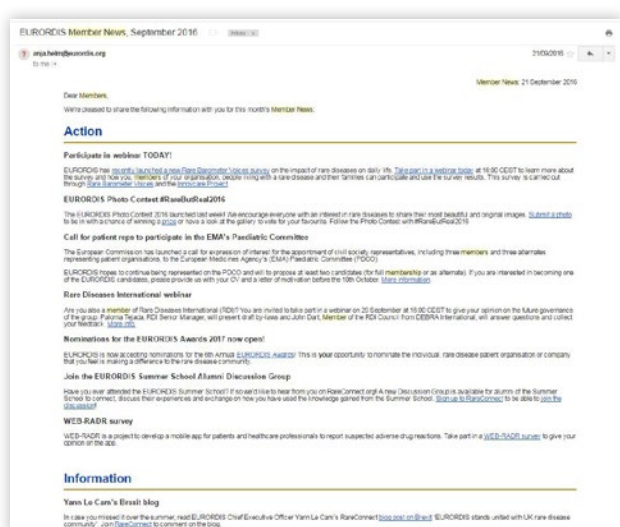
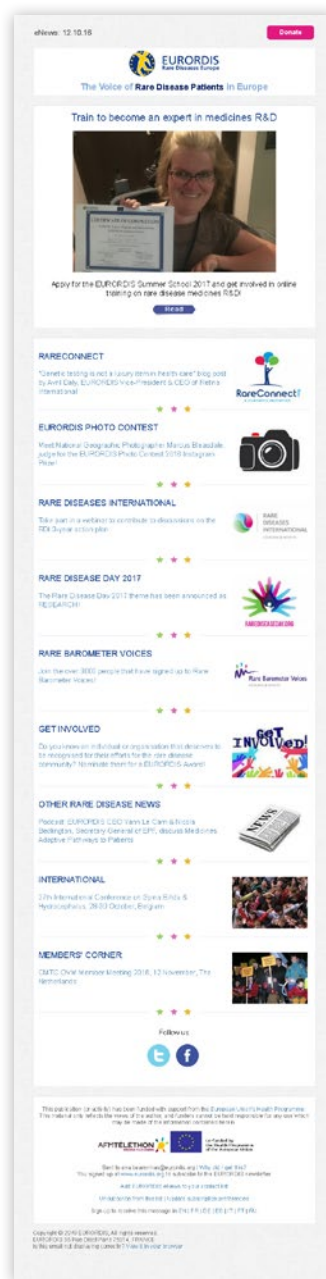
The EURORDIS eNews is bi-monthly news report in 7 languages that communicates breaking news of interest to patient advocates, people living with a rare disease and their families and policy makers. Each eNews features a lead article (devoted to important news in the rare disease community or EURORDIS 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.

Our eNews is constructed with a lead story which relates to EURORDIS activity. This e-news publication, free of charge, gives stakeholders an update on the latest EURORDIS activity, as well as other relevant news in the rare disease community. It is an opportunity for our members to disseminate information about their local events.

The eNews is entirely written, constructed and disseminated in-house by EURORDIS staff or automatic programs created by EURORDIS staff. The only element which is outsourced is the 6 translations of the English language.

Each eNews lead-story appears on the website (eurordis.org) in full and feeds the homepage on a weekly-basis in all 7 languages of the website (English, French, Spanish, Italian, German, Portuguese and Russian). The archive is also kept in the news and events section of the EURORDIS.org website. Finally, a feature was developed on the website which links previous topic related articles to the lead story. In the sidebar of the website, you can see the list of previous related eNews.

Lead stories topics in 2016 included: Register for the European Conference on Rare Diseases 2016; Register for Rare Barometer Voices to make your voice heard!; New recommendations to improve social care for rare diseases; Train to become an expert in rare disease game changers at ECRD 2016.



### 2.2.4.2 Member news

Our Member News is distributed once a month (around the middle of the month) to over 1500 contacts at our member organisations. Its simple design has ensured a high open rate. It provides links to information and activities in a simple, streamlined text template to ensure relevant information is reaching EURORDIS members as directly as possible. Each Member News is translated into the 7 languages of the EURORDIS website.

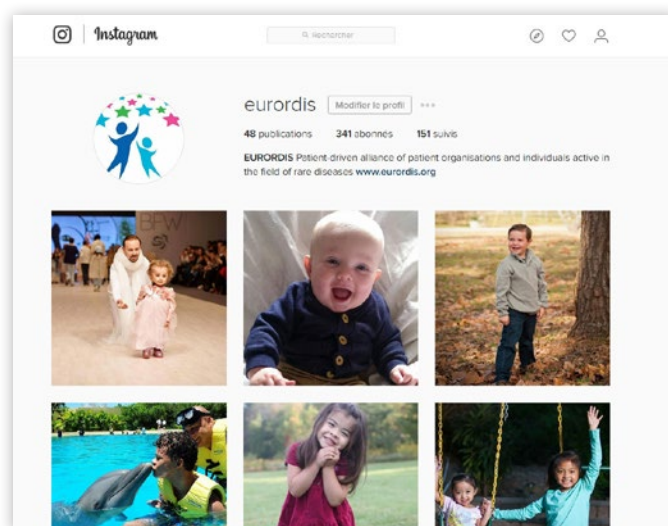
The Member News is divided into 3 sections:

- + **Action:** information that we want to provide to our members so that they take an action. Whether it be signing up for a webinar or registering for an event.
- + **Feedback:** when EURORDIS requires feedback on a document or the participation of its members in a consultative process.
- + **Information:** any information that is important for members of EURORDIS but does not require an action or feedback to be given.



## 2.2.5 Social Media

EURORDIS has its own Facebook page, Twitter account, Flickr account, YouTube channel, Google + and Instagram account.

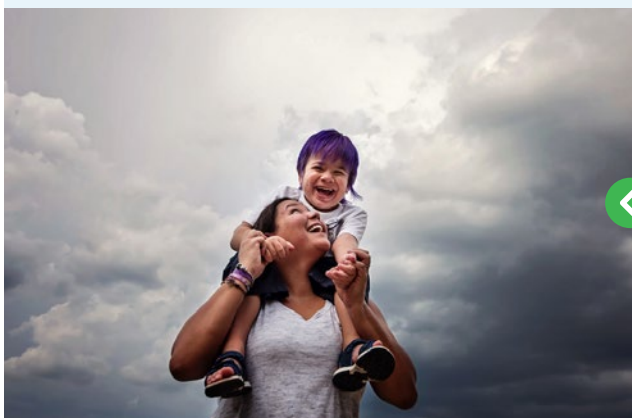


### 2.2.6 EURORDIS Photo Contest

Initiated in 2006, the EURORDIS Photo Contest is an annual event that raises awareness about rare diseases to the general public, activates and builds our network of people living with a rare disease and their families and helps us collect photos which can be used in EURORDIS' printed materials, presentations and on the web.

The photo contest was a great success with 400 photos received from 54 countries. 3 Winners were chosen by public vote, by professional photographers Rick Guidotti (Expert's Choice) and Marcus Bleasdale (Instagram Prize)

The winner of the **Public Vote** was a photo of Briana from Romania entitled "Feel free". She is enjoying a moment of happiness during her short vacation at the Black Sea. She is living with spastic tetraparesis and high myopia.



The winning **Expert's Choice** photo was 'Joy in the storm' picturing Rowen and his mum Christel from the USA. Rowen is living with Sanfilippo syndrome (MPS III).

The **Instagram Prize** went to a photo called 'The path to the dream' featuring Lomeiko. She is 5 years old and living with osteogenesis imperfecta. The photo shows her participating in the Toners Children's Fashion Day in Belarus.





## 2.2.7 EURORDIS Awards

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

2016 marked the fifth EURORDIS Awards for outstanding accomplishments in the field of rare diseases, which were presented in Brussels on the occasion of Rare Disease Day. The 2016 EURORDIS honorees were as follows:

- + Policy Maker Award**  
Cristian-Silviu Buşoi, Member of the European Parliament, Romania
- + Volunteer Award**  
Tsveta Schyns-Liharska, Belgium/Bulgaria
- + Media Award**  
France Télévisions - AFM-Téléthon, France
- + Patient Organisation Award**  
UNIQUE - The Rare Chromosome Disorder Support Group, UK
- + Scientific Award**  
Prof. Dr. Peter N. Robinson, Institute for Medical Genetics, Universitätsklinikum Charité, Germany
- + European Rare Disease Leadership Award**  
Joint winners: Antoni Montserrat Moliner, Jarek Waligóra and Michael Hübel, Directorate General of Health and Food Safety (DG-SANTE) within the European Commission, Belgium
- + Company Award**  
Actelion Pharmaceuticals Ltd., Switzerland
- + Lifetime Achievement Award**  
Renza Barbon Galuppi, Italy





# 3. PATIENT ENGAGEMENT: Roles in decision-making

## 3.1 Patient Engagement in Healthcare

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### European Reference Networks

*EURORDIS has been the central actor in the development of European Reference Networks, turning the initial idea into reality. Our advocacy work to create a framework for ERNs where patients and clinicians are equal partners started in 2005 and today spans over a decade. EURORDIS delivered a patient-centred vision for ERNs into 24 concrete therapeutic thematic networks, ensuring all rare diseases have a home in their combined structure, and providing an optimal framework to meet the multisystem needs of rare disease patients.*

EURORDIS supported the development of all ERN network applications, particular the structured development of patient involvement and empowerment into the network governance structures, advocating for patient representatives to be formal voting members of the network boards.

EURORDIS successfully advocated for the approval of all 24 ERNs to ensure the optimal framework for ERNs to meet the needs of multisystem rare disease patients.

EURORDIS created European Patient Advocacy Groups (ePAGs), aligned to the scope of the different ERN applications in early 2016 to support an ERN informed rare disease community able to meet the ambitious goals set out in the EUCERD Addendum's recommendation for ERNs. The establishment of the ePAGs and election of ePAG representatives enabled a uniform and democratic structure for patient involvement in ERNs network boards, clinical committees and working groups. EURORDIS supported their involvement in the development of ERN applications through a series of individual ePAG focus groups with Network Coordinators throughout the year

EURORDIS has completed a review of all Network Applications and developed summary reports of the applications as capacity building material for ePAG representatives and to inform the wider patient community about the ERNs. EURORDIS has completed a transversal analysis of the network themes, including Clinical Guidelines & Outcomes, Research and Registries, Training and Education, and Patient Empowerment. This work will support the development of bit sized capacity building material and the development of transversal focus groups in 2017.

EURORDIS also led capacity building activities to support EURORDIS' membership, National Alliances and European Federations and Networks be ready for the first call for European Reference Networks applications at the ECRD 2016 Edinburgh, EMM 2016 Edinburgh, CNA & CEF meetings.

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### Participate in the development of long-term projects on the methodologies for creation and evaluation of best clinical practices

EURORDIS has been involved in the project RARE-BestPractices which aims at improving clinical management of rare disease patients and narrowing the existing gap in quality of health care among EU member states as well as in other countries. This will be achieved by collecting, evaluating and disseminating best practices as well as sharing knowledge. The ultimate impact of the RARE-Bestpractices project should be an improvement of health outcomes and quality of life for rare diseases patients, through a reduction of inequalities in health care



and an increased equity of access to better diagnostics and therapeutics at EU level.

EURORDIS has been involved by building capacities of rare disease patient organisations and of people living with rare diseases on the importance, use and benefits of guidelines and HTA (Health Technology Assessment), through the dissemination of information as well as through training activities targeted at patient advocates. EURORDIS has also been involved in the development and in the implementation of the methodology for guidelines for rare diseases, ensuring that this methodology places patients' need foremost.

In 2016, EURORDIS was involved in: a training organised in Milan, in February 2016 on a course for health care

guidelines developers on treatment of rare diseases with several patient representatives attending; and a workshop during the EMM Edinburgh 2016 on Clinical guidelines in the context of ERN (speakers from Italian National Institute for Health, Healthcare Improvement Scotland, and two patient representatives from DEBRA International and VOSP Netherlands).

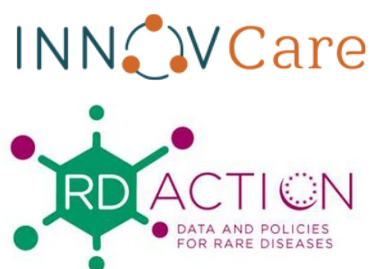
The project ended on December 31<sup>st</sup> with a final meeting and general assembly held in November. The main topic that were assessed as the top priority were the: Guidelines repository; Rare Journal; Training material to develop healthcare guidelines; Database RareGap (to retrieve research topic in gap analysing). EURORDIS pushed to have the strongest links between orphaned and the RBP main deliverable (the guidelines repository).

## 3.2 Patient Engagement in Social Care

### Support the national alliances in their action in advancing the integration of rare diseases into social services and policies at national level

During this last year, EURORDIS continued to **support national alliances to promote the integration of RD into social policies and services** via the capacity building workshop at EMM 2016 Edinburgh and the organisation of a webinar to support the organisation of national workshops focused on social services and policies.

### Promote integration of rare diseases into social services



In 2016 EURORDIS continued its focus on social policies, mainly through: its involvement in the EU-funded projects **RD-Action** (2015-2018) and **INNOVCare** (2015-2018); the launch of the **INNOVCare/Rare Barometer survey** on the impact of RD on daily life; the **mapping of stakeholders and policy issues at EU level**; and the promotion of the **representation of people living with a RD and their carers in relevant policies at European level**.

From their adoption in spring, EURORDIS promoted the wide dissemination of the **Commission Expert Group Recommendations to Support the Incorporation of Rare Diseases into Social Policies and Services** via EURORDIS media and events, as well as through ECRD 2016 Edinburgh and the workshops organised within the INNOVCare project.

Furthermore, EURORDIS has created an **Advisory Group within the INNOVCare project**, composed of 21 representatives of competent authorities from 16 European countries, with whom EURORDIS and the project partners engage into discussions on how to promote the implementation of the Recommendations and on how to move forward at national level to promote holistic care for people living with a RD and their carers.

Within the INNOVCare project, EURORDIS has initiated the activities of the secretariat of the future **European Network of Resource Centres for Rare Diseases – RareResourceNet**. The network will be composed of resource centres for rare diseases and other specialised social services for rare diseases or complex chronic conditions/disabilities and will focus on advancing holistic high quality care for people living with a RD and their carers in Europe. 2016 saw the network take its first steps with the support of EURORDIS: composition of a Steering Group (SG) and Working Group (WG); first drafts of the network' vision, mission, organisational statement and membership criteria, following face to face and online brainstorm meetings with the SG and the WG.

The **dissemination of case studies on social services** for RD and of the dedicated section on EURORDIS website continued, via EURORDIS eNews.

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 RD amongst social workers and to draw attention to the need for training social services providers by signing a **Memorandum of Understanding** and coordinating various common dissemination actions **with the International Federation of Social Workers Europe**.

Further actions done by EURORDIS to promote the integration of rare diseases into social policy are described in the section "voicing the needs of people living with a rare disease and advocating for the integration of rare diseases into social policy".

## Promoting integrated health and social care for rare diseases

Also within the INNOVCare project, and in order to further demonstrate the complexity of care coordination and care pathways for people living with a RD and their carers, EURORDIS launched a **survey on the impact of RD on daily life** (via the Rare Barometer programme) and organised **visits to a selection of European countries to collect information on the state of the art of care provision** and of care coordination.

EURORDIS continued to promote integrated care for RD in 2016 via the various activities of the **INNOVCare project** and the **involvement of the International Foundation for Integrated Care in these initiatives**. EURORDIS also joined the summer school on integrated care promoted by the Foundation.

In the autumn, EURORDIS organised the **INNOVCare workshop on Improving Integrated Care for People Living with Rare Diseases and Complex Conditions** (Sweden, September 2016) and co-organised the

**INNOVCare workshop on Workshop on Scaling Innovative Care Delivery for Rare Diseases and Complex Conditions** (Austria, October) – both focused on case management and other initiatives aiming at promoting holistic care for people with a RD in European countries. The workshops included multi-stakeholder discussions between competent authorities (INNOVCare Advisory Group), patient representatives (from the Social Policy Advisory Group), social services, healthcare providers and academics.

In parallel, EURORDIS has led and encouraged several discussions between the INNOVCare project teams, in order to **support the patient-centred design, implementation and evaluation of the pilot of case management taking place within the INNOVCare project** in 2016. As responsible for the external and internal communication of INNOVCare, EURORDIS also coordinated the elaboration of the INNOVCare logo, image and website.

An INNOVCare update 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.

## 3.3 Patient Engagement in Research

### International Consortium for Rare Disease Research



*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 has been involved in the International Consortium for Rare Disease Research (IRDiRC) since its launch. In particular in 2016, EURORDIS has been present in the **Executive Committee/Consortium Assembly**, in the **Therapies Scientific Committee** and in the **Interdisciplinary Scientific Committee**:

Béatrice de Montleau, EURORDIS patient representative in ExCo/CA and Yann Le Cam, CEO of EURORDIS, ExCo and Chair of IRDiRC Therapies Scientific Committee (TSC) until end of October 2016/TSC Member after October 2016 and Gema Chicano Saura, EURORDIS Board member, ISC member since February 2016 and Virginie Hivert, EURORDIS Therapeutic Development Director, TSC member.

In 2016, EURORDIS has also been involved in several IRDiRC Taskforces:

#### Taskforce on PCOMs (Patient-Centered Outcome Measures):

Yann Le Cam and Virginie Hivert pursued the actions of the previous year by working on the post-workshop report of the 'Workshop to discuss actions to improve clinical research in the field of rare diseases' held in November 2015, Paris, France.

An article to be submitted to a scientific journal is also currently in preparation within the Taskforce.

#### Taskforce on SPCT (Small Population Clinical Trials):

March 3, 2016, London, UK: Workshop held at EMA to discuss actions to reach agreement between the different stakeholders on appropriate small population studies – Yann Le Cam and Virginie Hivert participated to this workshop.

#### Taskforce on Data Mining / Repurposing:

Virginie Hivert has been involved in Preparatory work and in a workshop between experts in the field to discuss between the different stakeholders on data mining and repurposing held on 16<sup>th</sup> November 2016 in Barcelona, Spain. Recommendations will be issued in a post-workshop document.

#### IRDiRC/GA4GH Joint Task Force:

EURORDIS is a member of a Joint Privacy-Preserving Data Linkage (PPRL) Task Force between IRDiRC and Global Alliance for Genomic Health which aims to explore and recommend one or more approaches to linking data records within and across institutions while preserving



individual privacy. Following a face to face meeting in Paris in December 2016, the team agreed to further explore an existing approach developed through the European Network for Cancer Research in Children and Adolescents (ENCCA) with some modifications for increased flexibility to accommodate a range of jurisdictional laws (including the use of a model allowing federated regional storage systems), and increased scalability.

## Engagement in upcoming genetic developments:

EURORDIS participated in several meetings and workshops in 2016 on the topic of genome editing:

- + “Responsible use of genome editing/CRISPR/Cas9 in research” organised by the French Inserm Ethics Committee with a wide range of European stakeholders. Stakeholders (including EURORDIS) were able to present diverse positions and to suggest recommendations captured in a manuscript currently in submission to an academic journal. The resulting general principles aim at guiding research involving genome editing technology and ensuring satisfactory compliance with ethical standards.
- + “Human genome editing in the EU” hosted by the Federation of European Academies of Medicine (FEAM), the UK Academy of Medical Sciences, and the French Academy of Medicine.

Due to an intense interest from patient groups on the recent development in genome editing technology, EURORDIS together with a dedicated working group on genome editing comprising several members of the Patient Advisory Council of RD-Connect have organised and held an interactive webinar to discuss current scientific advances and related ethical issues. This webinar aimed to start informing and engaging rare disease patient representatives on the complex topic of genome editing. Around 40 people, mostly patient representatives from rare disease organisations joined the webinar on Tuesday 20<sup>th</sup> October and were able to actively participate by asking questions directly to the experts. This working group, coordinated by EURORDIS also organised a follow up workshop on 4<sup>th</sup> November in Paris for rare disease patient representatives across Europe to participate in an open discussion with experts on several aspects of gene therapy and genome editing specifically on scientific progress and its relevance for rare diseases, associated ELSI issues as well as on the perspectives from the regulatory and biotech sectors.

## Genetics Clinic of the Future (GCOF)

*The Genetics Clinic of the Future project has the following main objectives: 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 on the genetics clinic of the future; to implement key Science with and for Society (SwafS) issues; 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.*

Within this project, EURORDIS collaborates 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.

In 2016, EURORDIS organised two focus groups of 2h with 8-10 patients representatives during the EURORDIS Summer School (ExPRESS) in Barcelona during the first week of June 2016. The objective of the focus groups was to gather the perspectives of patients on considerations supporting the control and sharing of next generation sequencing data such as: location of the data (own country, Europe, US, others); domain of use (research, clinical care, any profit, not profit); perspectives on publication and disclosure; data security measures; intellectual property etc.

The report from the focus groups will be finalised in 2017.

## Involvement in E-rare-3

“E-Rare-3” is a project to pursue and expand the activities in accelerating the development of new diagnostics and therapeutics for patients suffering from 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. Until now EURORDIS has been involved in the E-Rare programme as an observer with the representative participating in the E-Rare External Advisory Board. 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’ organizations 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. EURORDIS will coordinate a network of “scientific officers” (volunteers) from funding research patient organisations to find 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).

## Participation in the Web-RADR project



*EURORDIS is member of the Web-RADR consortium, an Innovative Medicines Initiative investigating the potential for publicly available social media data for identifying drug safety issues ([www.web-radr.eu](http://www.web-radr.eu)). A second objective is to develop a mobile app for patients and healthcare professionals to report suspected adverse drug reactions to national EU regulators.*

The purpose of this social media analysis, led by Epidemico, is to provide access to classified social media data via a visualisation platform for signal identification and confirmation. Formal analysis of emerging social media data could better predict problems with marketed medicines products and inform better design of future medicines products. Data from social media could provide earlier estimates of the magnitude and nature of a safety signal. However, these data have not previously been used for safety surveillance. Careful consideration must therefore be given to how to process the information to make it suitable while meeting, keeping to data protection requirements.

A second pillar of the abovementioned Web-RADR projects consists in the design of a mobile application to engage both the public and healthcare practitioners around issues of real-time pharmacovigilance. The app will have two main functions: alerting, and reporting.

The application will alert users about new warnings from official channels (such as alerts from regulatory agencies about safety issues and warnings), informal channels (such as news media), and anonymised user submissions. Using forms tailored separately to the public and clinicians, the WP3a team will provide a user-friendly tool for reporting information about ADRs.

Reports received via the mobile app will be compared to those received via established reporting schemes for completeness, quality and value for detection of safety issues.

The app will be designed to accommodate additional languages. The steps for expanding to additional countries involves: translating and adapting the form, adding new product lists, and establishing the necessary attendant database connections.

As of end of 2016, the app was launched in the United Kingdom, Netherlands and Croatia. In 2017, it will be launched in Zambia and Burkina Faso.

### Mobile App Survey for Healthcare Professionals and Patients

Work Package 3B of the WEB-RADR project has launched two surveys, for healthcare professionals and for patients and consumers of medicines, to determine opinions about a mobile app which can be used to report adverse drug reactions (ADRs) and to receive safety information about medicines.

The results of the survey will be used to improve WEB-RADR's mobile apps, and to increase general knowledge about mobile two-way communication; the reporting of ADRs and availability of news and safety alerts about particular medicines.

EURORDIS is particularly involved in this survey, which is the second phase of a research on the facilitators and the barriers for the use of a health application. Focus groups were consulted in 2015 and the outcome of this first research were published in the Drug Safety Journal in 2016 (read here: <http://link.springer.com/article/10.1007%2F540264-016-0494-X>).

The second phase is a large online survey to healthcare professionals and patients which was conducted during Summer 2016. In total, 636 patients and 3901 healthcare professionals were included in the analyses and publication is in progress.

## 3.4 Patient Engagement in lifecycle development

### 3.4.1 Pre-marketing authorisation

#### European Medicines Agency



EUROPEAN MEDICINES AGENCY  
SCIENCE MEDICINES HEALTH

*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 686 dossiers in 2016 through the work of the scientific committees they belong to, as well as to the activities of several adhoc 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 2016, 46 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.

Applications submitted in response to Call for Expression of Interest for CAT membership were submitted in September 2015. After an internal Call for Expression of Interest within its Members, EURORDIS nominated Michele Lipucci di Paola (current member) and Christos Sotirelis (expert patient). Michele Lipucci has been nominated by the Commission as alternate and started his mandate in July 2016. Christos Sotirelis was on the reserve list and is attending the CAT meetings as an expert with the support of EURORDIS.

In October 2016, applications responding to the European Commission's Call for Expression of Interest were submitted for the PDCO membership. After an internal Call for Expression of Interest within its Members, EURORDIS proposed Dimitris Athanassiou and Kerry Leeson-Beevers

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 2016, PCWP members worked in particular on information on medicines (Session on communication and information on medicines, 8 March), on PRIME and adaptive pathways initiatives, on the PRAC strategy on measuring the impact of Pharmacovigilance activities, on Access to EudraVigilance data to Patients and Health Care professionals, on the proactive publication of clinical study reports; redaction of commercially confidential information, and on the EMA consultation on post-authorisation efficacy studies.

The Working Party established five new topic groups:

- + 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
- + Acknowledge and promote visibility of patient input in the Agency's activities
- + Training: explore synergies with existing training initiatives, explore methods to further enhance support for patients involved in EMA activities
- + 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

- + Involvement of young people / children

For topic one, recommendations to the EMA Management Board were finalised on 17 October 2016. For topic two, two sets of recommendations were finalised:

- + How to improve acknowledgement and promotion of patient input into EMA activities by EMA
- + How to improve acknowledgement and promotion of patient input into EMA activities by the organisations

For topic three, the EMA Management Board endorsed the recommendations in March 2016. For topic four, recommendations are still in progress. For topic five, "Rules of Procedure" (guidance) establishing methods for involving/consulting young people within EMA activities are prepared and should be endorsed by the EMA Management Board in early 2017.

### European Medicines Agency Management Board

In 2016 Yann Le Cam, Chief Executive Officer of EURORDIS, was appointed to the Management Board of the European Medicines Agency (EMA).

The EMA Board is made up of representatives of each of the 28 EU Member States, the European Commission, the European Parliament, two civil society organisations, and doctor and veterinarian organisations.

"I am delighted to have been appointed to the EMA Board. This is not a personal win. This is a victory for the rare disease community. After 20 years of campaigning to raise awareness of rare diseases in Europe, orphan medicines, other rare disease therapies and paediatric medicines represent a large proportion of new medicines approved each year. We are also recognised for our leadership to shape agendas and promote innovative approaches."

## 3.4.2 Post-marketing authorisation

### SCOPE Joint Action

Since 2013, EURORDIS is one of the advisors to SCOPE Joint Action (Strengthening Collaboration for Operating Pharmacovigilance in Europe), funded by the Consumers, Health and Food Executive Agency (CHAFEA). SCOPE will end in 2017.

SCOPE was created to support pharmacovigilance operations in Europe following new requirements introduced by the European pharmacovigilance legislation of June 2012.

SCOPE is gathering information and expertise on how regulators in Member States run their national pharmacovigilance systems. Using this information, a variety of tools are developed, including guidance documents, pharmacovigilance training materials and other tools to support best practice. The Joint Action supports consistent pharmacovigilance operations approaches throughout the EU network, benefiting the safety monitoring of medicines and communication outputs, thereby helping to safeguard public health.

SCOPE consists of eight work packages, three of which focused on practical aspects of the project - coordination, dissemination and evaluation. The other five work packages are associated to specific pharmacovigilance topics of Adverse Drug Reaction (ADR) collection, signal

management, risk communication, quality management systems and lifecycle pharmacovigilance.

More information can be found here: <http://www.scopejointaction.eu>

The UK's Medicines and Healthcare Products Regulatory Agency (MHRA) is the Joint Action coordinator, and is responsible for the coordination and dissemination work packages.

One of the many important results is a guideline for the Collaboration with Patient Organisations to Promote and Support Patient ADR Reporting, which can be found here: [http://www.scopejointaction.eu/\\_assets/files/WP4-DEL5-Collaboration-with-Patient-Organisations.pdf](http://www.scopejointaction.eu/_assets/files/WP4-DEL5-Collaboration-with-Patient-Organisations.pdf).

### Health Technology Assessment

As the third European Joint Action started its activities in September 2016 only, no procedures involving patients took place during the course of 2016.

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

*The Task Force represent 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.*

As the term of the task force corresponds to the one of the Patients' and Consumers' Working Party at the EMA, a call to EURORDIS volunteers was launched to renew the task force members (among Eurordis Summer School and EUPATI alumni). Because of this renewal, only one **DITA Task Force meeting was held** in Paris on 15/09 to launch the new task force.

DITA members exchanged information and elaborated contributions to: EMA consultation on post-authorisation

efficacy studies; European Commission HTA inception impact assessment; Eurordis position on compassionate use programmes; Eurordis position on access to medicines; European Commission consultation on off-label use of medicines.

DITA task force members presented Eurordis views/data in the following conferences (not exhaustive): Slovak government conference on medicines shortages, Bratislava, 17-18 November; EU Brain Council conference on off-label use, European Parliament, 27 September; EUnetHTA JA3 kick-off meeting, Amsterdam, 2 March; SCOPE joint action flagship event, London, 22-23 November; DIA-EMA workshop on medication errors, London, 20 October.

**Review of EMA documents for the public or contributions to EMA consultations:** European Public Assessment Reports for the Public (5), Package Leaflets (8). Since 2007 when the procedure to review EPAR summaries and PL was established for authorised medicines in the EU, 60 EPAR summaries and 91 PL were reviewed, for a total of 151 documents.

**On HTA,** DITA task force members responded to the consultation on the **"Inception Impact Assessment"**.

Off-label use of medicine

- + A survey on "Treatment information" was conducted and valid results obtained from 1,401 respondents.
- + This survey explored how patients are actually taking their medicines, how they understand the black symbol for additional monitoring, how they see the use of off-label products, and how they report adverse drug reactions.

Several volunteers attended the Drug Information Association EuroMeeting in Hamburg, 6-8 April, with presentations on adaptive pathways and conditional authorisation.

Several volunteers attended the European conference of the International Society of Pharmacoeconomics and Outcomes Research 29/10-2/11, Vienna, and presented on patient engagement.





# 4. CROSS-CUTTING PRIORITIES

## 4.1 Governance

### 4.1.1 EURORDIS Board of Directors

During the General Assembly held in Edinburgh on 29 May 2016, EURORDIS full members voted on five vacant positions on the Board of Directors, re-electing Terkel Andersen, Danish Haemophilia Society, Denmark; Vlasta Zmazek, Croatian Alliance for Rare Diseases, Croatia; John Dart, DEBRA International, UK; Geske Wehr, European Network for Ichthyosis e.V, Germany; Anne-Sophie Lapointe, Vaincre les Maladies Lysosomales, France. Anne-Sophie Lapointe replaces Nick Sireau, AKU UK, who resigned from the Board of Directors in November 2015 for personal reasons.

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.

### 4.1.2 EURORDIS Statutes & By-Laws

As a matter of good governance, the EURORDIS Board of Directors decided on the re-assessment and update of the EURORDIS Statutes, a process which started in 2015. The new Statutes were adopted at the General Assembly in Edinburgh on 26 May 2016. The new Statutes are lighter in content providing the overall structure and governance framework of the organisation. In addition, the new Statutes include an official name change of EURORDIS to EURORDIS – Rare Diseases Europe, in accordance with the Common Goals & Mutual Commitments between EURORDIS and its National Alliances.

Whereas the new Statutes provide the overall governance structure, the EURORDIS Board of Directors are also working on consolidating By-laws that set out the rules of the internal processes by which EURORDIS operates.

The new compiled by-laws will be presented at the General Assembly in Budapest in May 2017.



### 4.1.3 Strategic Partnerships (MoUs)

EURORDIS has developed partnerships with several European and international not-for-profit organisations to work on transversal issues relevant for patients affected with rare diseases.

Staff and EURORDIS volunteers engage in a range of different activities depending on the level and type of involvement with international NGO partners. The partners are:

#### NORD – The US Organization for Rare Disorders:



For the past 15 years, EURORDIS and NORD have been joining efforts to improve the life of rare disease patients on both sides of the Atlantic. NORD is a member of the EURORDIS Council of National Alliances of rare disease patient organisations and thus active in the Rare Disease Day Steering Committee. NORD representatives also attended the European Conference on Rare Diseases and Orphan Medicinal Products 2016 Edinburgh as well as the meeting of RDI. NORD also played an active part in Rare Diseases International as a member of the RDI Council governance board, and on the official inauguration of the NGO Committee for Rare Diseases in New York.

#### CORD – The Canadian Organization for Rare Disorders



EURORDIS and CORD have been collaborating together for many years. CORD is a member of the EURORDIS Council of National Alliances of rare disease patient organisations. In 2016, CORD and EURORDIS continued their close collaboration with CORD playing an active part in Rare Diseases International as a member of the RDI Council governance board. CORD also attended the European Conference on Rare Diseases and Orphan Medicinal Products 2016 Edinburgh.

#### JPA – The Japan Patients' Association

EURORDIS and the Japan Patients Association (JPA) signed a Memorandum of Understanding on 26 May 2013, bringing patient advocates from Europe and Japan together to promote rare diseases as an international health priority. JPA played an active part in Rare Diseases International as a member of the RDI Preformation group prior to official election for the RDI Council in May 2016. JPA also attended the ECRD 2016 Edinburgh and the meeting of Rare Diseases International



#### RVA – Rare Voices Australia

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 is currently playing an active part in Rare Diseases International as a member of the RDI Council governance board.

#### RPU – Russian Patients' Union



**RUSSIAN PATIENTS  
ASSOCIATION**  
Health for all

EURORDIS and the Russian Patients' Union signed a memorandum of understanding in May 2015. EURORDIS representatives organised a country visit to the RPU headquarters in Moscow in August 2016 and carried out meetings with local patient organisations.

#### EPF – The European Patients' Forum

EURORDIS works on transversal issues with EPF: EU Directive on cross-border healthcare; revision of the EU Directive on Clinical Trials and information to patients. Anders Olason, former EURORDIS Board member, was the current President of EPF until 2016, representing EURORDIS.



#### ICORD – The International Conference on Rare Diseases and Orphan Drugs

In 2015, ICORD and RDI signed a Memorandum of Understanding in order to mutually strengthen the international rare disease movement. EURORDIS and RDI supported the 11<sup>th</sup> ICORD conference in Cape Town, South Africa in October 2016 providing fellowships to the conference and sending representatives to participate in the event.



#### RADOIR - Rare Diseases Foundation of Iran

EURORDIS representatives visited RADOIR headquarters in Tehran, Iran, in July 2016. RADOIR foundation aims to improve the quality of life for patients living with a rare disease and to increase public awareness about the burden of rare diseases on patients, their families and the community. EURORDIS and RADOIR signed a Memorandum of Understanding as part of the continued effort to stress the international dimension of the rare disease movement and the global benefits to be gained by international collaboration in this field.



EURORDIS also has partnerships with a number of learned societies:

- + European Federation of Internal Medicine (EFIM)
- + European Hospital & Healthcare Federation (HOPE) -
- + International Federation of Social Workers Europe (IFSW-Europe)
- + European Society of Human Genetics (ESHG)
- + International Society for Pharmacoeconomics and Outcomes Research (ISPOR)



## 4.2 Human resources

### 4.2.1 EURORDIS Staff

The team comprised 41 people, as of December 2016. 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 2016:

- + **Zoe Alahouzou**, Deputy to the CEO, has temporarily left on maternity leave
- + **Erwan Berjonneau**, Research Executive Rare Barometer, has joined EURORDIS
- + **Annette Dumas**, Social Policy Senior Advisor, joined EURORDIS
- + **Susan Foster**, Administrative Manager, joined EURORDIS
- + **Clara Hervas**, European Public Affairs Junior Manager, has joined EURORDIS
- + **Marie Meunier**, Executive Assistant to the Chief Executive Officer, has joined EURORDIS
- + **Celine Parisse**, Administrative Manager and Budget Controller, has left EURORDIS
- + **Carmen Lasheras Ruiz**, RareConnect Project Manager, has left EURORDIS
- + **Jennifer Steele**, Public Affairs Junior Manager, has left EURORDIS
- + **Juliette Sénecat**, Health & Social Projects Manager, has left EURORDIS
- + **Tania Webster**, Executive Assistant to the Chief Executive Officer, has left EURORDIS
- + **Emilie Zingg**, Junior Events Manager, joined EURORDIS



## Inauguration of the Rare Disease Platform in Barcelona

In March 2016, EURORDIS launched a new Rare Disease Platform in Barcelona housed at the historic Santa Apollònia Pavilion of the UNESCO world heritage Sant Pau Art Nouveau site, Barcelona bringing together the activities of local, national, European and international rare disease patient groups. The Platform hosts the new offices for EURORDIS (including its RareConnect initiative and members of team that support EURORDIS activities at the European Medicines Agency), the Plataforma Malalties Minoritàries and other rare disease organisations in the future.

To mark the occasion, an inauguration event was organised which included speakers such as Encarna Guillén, Regional Minister of Health, Region of Murcia; Albert Salazar, Director, Hospital Sant Pau de Barcelona; Alba Ancochea, Director, FEDER.

The inauguration evening included discussions on the situation for rare disease patients locally in the region of Catalonia and nationally in Spain, on rare disease initiatives at a European and international level, as well as testimonials from several people whose family members are living with a rare disease.



### 4.2.2 EURORDIS Volunteers

In 2016, EURORDIS was proud to rely on 447 volunteers including 89 dedicated volunteer patient advocates, 70 volunteer ePAG representatives, 1 for fund raising, 1 office support volunteer and 286 volunteer moderators of online communities of Rare Disease Patients, within the activity "RareConnect".

The specific role of volunteer patient advocates consists in sharing their knowledge and experience to advocate for better national and European public health policy

measures in favour of rare diseases. They participate in committees, working groups, Task Forces (e.g. DITA and RD Connect), and speak at international conferences.

All the volunteers are governed by the EURORDIS Charter of Volunteers, adopted by the EURORDIS General Assembly on 8 May 2014 in Berlin. This Charter sets out the values of EURORDIS, the volunteers' commitments as well as the EURORDIS' commitments towards its volunteers.



### Focus on EURORDIS volunteer patient advocates:

They are considered as experts. Since the creation of EURORDIS, they have greatly contributed to shaping EU rare disease policies. Most of them are either patients or parents of patients living with rare diseases.

They are selected amongst the EURORDIS' membership base via a Call for Expression of Interest and rigorous selection criteria to join a specific group of volunteers, and/or to candidate to a EU high level committee. The volunteers must: a) be committed to the cause of rare diseases, b) master English, c) have a long-standing advocacy track record in the field of rare diseases.

As a result, our strict rules have enabled us to always propose good candidates to EMA and EC's committees, and to be nominated by the European Commission.

Most of the EURORDIS Volunteer patient advocates belong to different internal working groups and Task Forces:

#### **EPAC: European Public Affairs Committee**

This internal committee plays an active and key role in EURORDIS' advocacy activities. The EPAC members discuss all relevant advocacy issues for people living with rare diseases and their families, and they can provide their comments on EURORDIS' positions on some specific issues. The EPAC is governed by Rules of Procedure. As of end 2016, it was composed of 46 members: 18 staff members (CEO, Directors and managers) involved in advocacy as well as 28 volunteers including the Board members, 3 former Board members, members of the Therapeutic Action Group (TAG) and members of the Policy Action Group (PAG) with one member being also on the PAG-Rare Cancer. The EPAC members have a mandate to represent EURORDIS. Further information is provided in table D.2.3.

#### **TAG: Therapeutic Action Group**

The TAG includes 8 EURORDIS volunteers who are member, alternate or observer on the Scientific Committees and Working Party at the EMA (CAT, COMP, PDCO, PCWP). Further information is provided in table D.2.2.

#### **PAG: Policy Action Group**

The PAG includes 7 volunteers who are member or alternate on the Commission Expert Group on Rare Diseases (CEG-RD). The PAG also includes 1 staff full member of CEG-RD, and two staff members who are observers. Further information is provided in table M3.

#### **PAG - RC: Policy Action Group - Rare Cancers**

The PAG-RC includes the two EURORDIS volunteer representatives on the Commission Expert Group on Cancer Control (CEG-CC) representing Rare Cancers, as well as five volunteers from rare cancer organisations.

They are assisted by 1 staff member and are involved in raising awareness of the needs of adults and children with rare cancers. Further information is provided in table M3.

#### **DITA (Drug, Information, Transparency, Access) Task Force**

The DITA is composed of 20 volunteers who are trained (via the EURORDIS Summer School) and active on 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). Further information is provided in table D.2.4.

#### **PAC: Patient Advisory Council in RD-Connect**

Composed of 18 volunteers and coordinated by 1 staff member, the PAC is acting to inform partners from RD-Connect, EUrenOmics and NeurOmics\* of issues important to patients. The PAC ensures that all project activities have a patient-centric approach.

\* European projects funded under the seventh framework programme of the European Union's Research and Innovation programme 2007-2013.

#### **PAC - RBP: Patient Advisory Council for Rare-Best Practices**

Composed of 7 volunteers and coordinated by 1 staff member, the PAC-RBP has a role to advise on best clinical practices and on the activities of the RARE-Best Practices

\* project, to inform project partners of issues important to patients and guarantee a patient-centric approach throughout project activities.

\* European project funded under the seventh framework programme of the European Union's Research and Innovation programme 2007-2013

#### **SPAG: Social Policy Advisory Group**

Composed of 14 volunteers and coordinated by 1 staff member, the SPAG has been established to inform on rare disease patients' and families' social challenges and to advise on social policy, provision of social care and related issues - such as holistic care, social services, social innovation, disability, special education, psychological support - guaranteeing the formulation of patient-centric approaches to the different social challenges faced by people living with rare diseases.

### Focus on EURORDIS ePAGs (European Patient Advocacy Groups):

Between March and April 2016, 86 ePAGs' representatives were elected by patient organisations to represent their patient community in the development of 24 European Reference Networks (ERNs) per big therapeutic areas.

Amongst the 86 ePAGs, 16 of them are already EURORDIS volunteer patient advocates, 70 of them are exclusively

dedicated to the mission linked with the development of ERNs.

The ePAGs are governed by Terms of Reference adopted by the Board of EURORDIS as well as by the EURORDIS Charter of Volunteers.

## 4.3 Finance & Support Services

Finance and support services' activities in 2016 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 related to contractual matters.

### Contract Grants

#### Renewed

Specific Grant Agreement (Operating Grant) for year 2016 (SGA FY2016), 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.

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

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

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

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

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

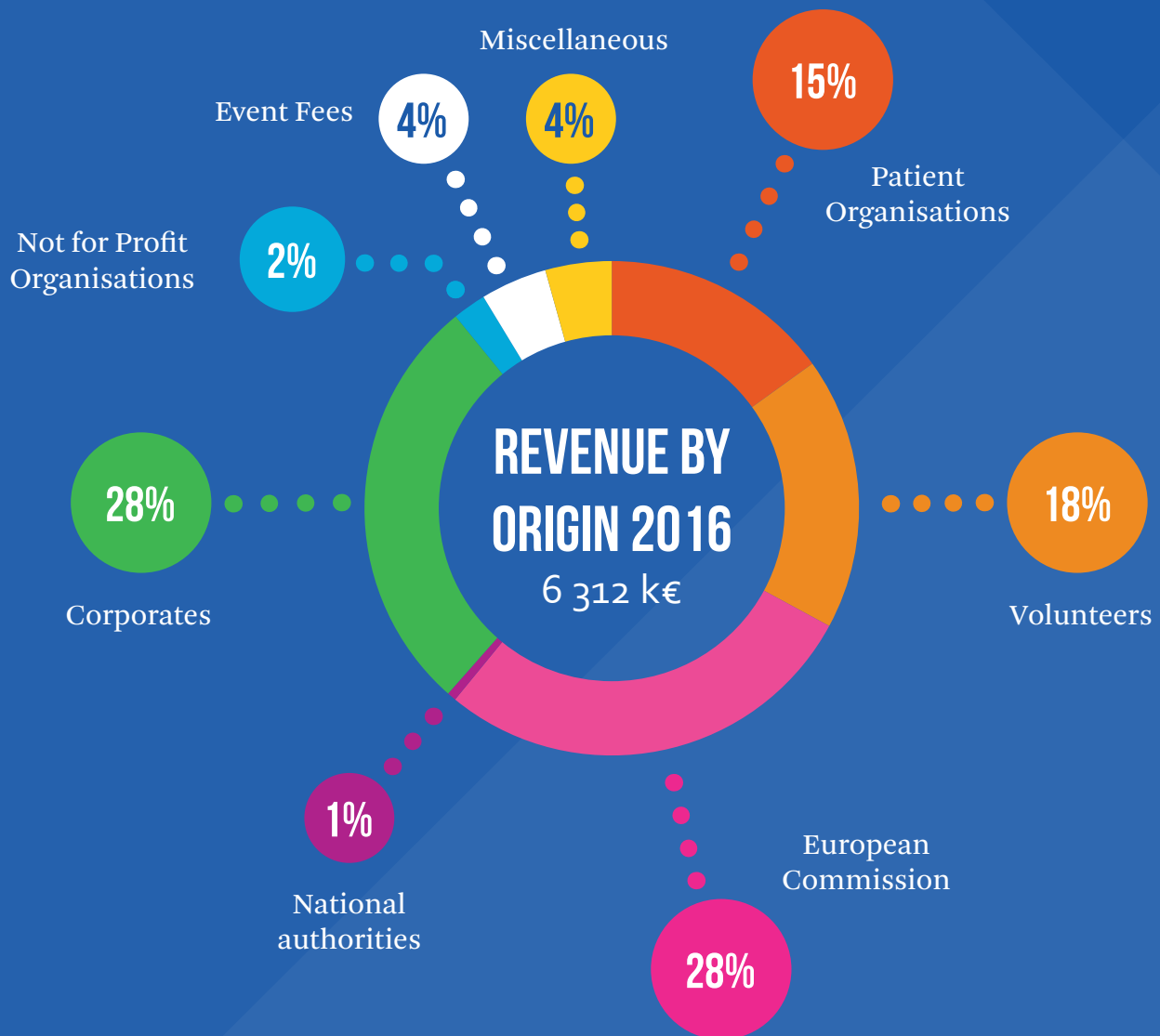
INNOVCare: Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions, DG Employment and Social Innovation (EaSI), 2015-2018

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

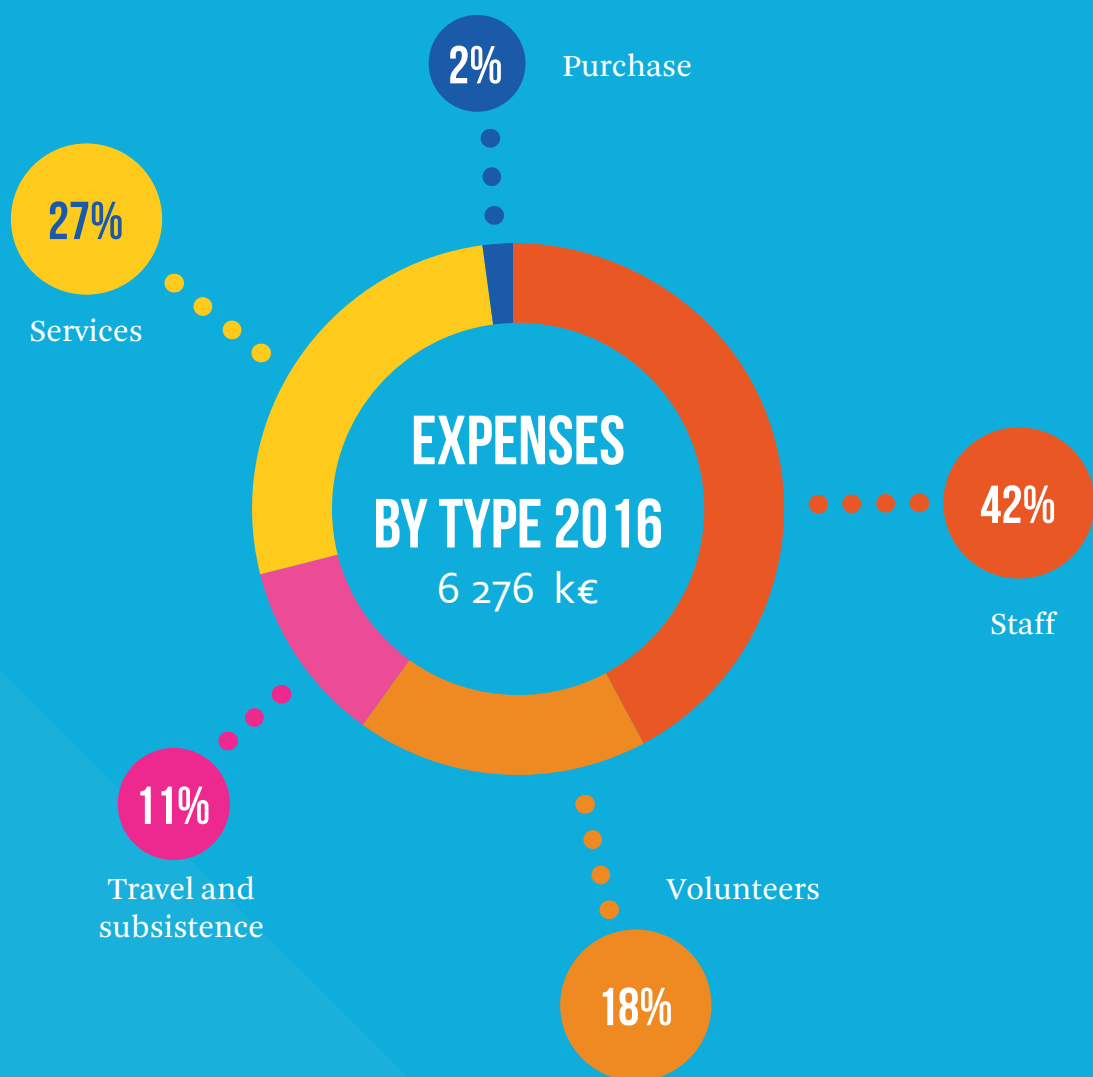




# REVENUE 2016



# EXPENSES 2016







# BOARD of directors

May 2016 - May 2017

## 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**

Vaincre les Maladies Lysosomales

 France

**Ms Françoise Salama**

AFM-Téléthon

 France

**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

**Ms Vlasta Zmazek**

Croatian Alliance for Rare Diseases

 Croatia

# MEMBERS of EURORDIS

## ALGERIA

ASSOCIATION ELAMANI POUR VENIR EN AIDE AUX MALADES SOUFFRANT DE L'ANÉMIE HÉRÉDITAIRE

## ARGENTINA

FUNDACION GEISER - GRUPO DE ENLACE, INVESTIGACION Y SOPORTE ENFERMEDADES RARAS

## ARMENIA

DOCTORS AND CHILDREN HEALTH CARE  
NEUROHEREDITARY DISEASES CHARITY ASSOCIATION

## AUSTRALIA

GENETIC ALLIANCE AUSTRALIA  
RARE VOICES AUSTRALIA

## AUSTRIA

ANGELMAN VEREIN ÖSTERREICH  
DEBRA INTERNATIONAL  
ICA ÖSTERREICH  
NF KINDER – VEREIN ZUR FÖRDERUNG DER NEUROFIBROMATOSEFORSCHUNG ÖSTERREICH  
PRO RARE AUSTRIA, ALLIANZ FÜR SELTENEN ERKRANKUNGEN  
PULMONARY HYPERTENSION ASSOCIATION EUROPE  
SELBSTHILFEGRUPPE LUNGENHOCHDRUCK - AUSTRIAN PH PATIENT GROUP  
USHER DEAFBLIND FORUM AUSTRIA

## BELARUS

BELARUSSIAN ORGANIZATION OF PATIENTS WITH MPS AND OTHER RARE GENETIC DISORDERS

## BELGIUM

22Q11 EUROPE  
ALPHA-1 PLUS ASBL  
ALS LIGA BELGIÉ  
ASSOCIATION BELGE DU SYNDROME DE MARFAN ASBL  
ASSOCIATION DE PATIENTS SOUFFRANT D'HYPERTENSION ARTÉRIELLE PULMONAIRE EN BELGIQUE  
ASSOCIATION POUR L'INFORMATION ET LA RECHERCHE SUR LES MALADIES RÉNALES GÉNÉTIQUES  
BELGISCHE ORGANISATIE VOOR KINDEREN EN VOLWASSENEN MET EEN STOFWISSELINGSZIEKTE  
BELGISCHE VERENIGING VOOR LONGFIBROSE VZW  
CONTACTGROEP MYELOOM EN WALDENSTRÖM PATIËNTEN  
DEBRA BELGIUM VZW  
EURO ATAXIA - EUROPEAN FEDERATION OF HEREDITARY ATAXIAS  
EUROPEAN CHROMOSOME 11 NETWORK  
EUROPEAN HAEMOPHILIA CONSORTIUM  
EUROPEAN HUNTINGTON ASSOCIATION  
EUROPEAN NETWORK FOR RESEARCH ON ALTERNATING HEMIPLEGIA  
EUROPEAN POLIO UNION  
FEDERATION OF EUROPEAN SCLERODERMA ASSOCIATIONS  
FEDERG - FEDERATION OF EUROPEAN ASSOCIATIONS OF PATIENTS AFFECTED BY RENAL DISEASES  
FEWS - FEDERATION OF EUROPEAN WILLIAMS SYNDROME  
GROUPE D'ENTRAÏDE BELGE DU SYNDROME GILLES DE LA TOURETTE  
HAE BELGIUM  
ICHTHYOSE BELGIQUE - ICHTHYOSIS BELGIÉ  
INTERNATIONAL FEDERATION FOR SPINA BIFIDA AND HYDROCEPHALUS (IF)  
MYELOMA PATIENTS EUROPE  
RADIOG - RARE DISEASE ORGANISATION BELGIUM  
RARE DISORDERS BELGIUM  
RELAIS 22 ASBL  
SIOP EUROPE - EUROPEAN SOCIETY FOR PAEDIATRIC ONCOLOGY  
SOBREVIVIRE VZW  
VLAAMS PATIËNTENPLATFORM VZW  
VLAAMSE VERENIGING NEUROMUSCULAIRE AANDOENINGEN VZW (NEMA)

## BENIN

ALBINOS SANS FRONTIÈRES

## BRAZIL

ASSOCIACAO BRASILEIRA DE ENFERMEDADES RARAS  
ASSOCIAÇÃO BRASILEIRA DE PARAMILOIDOSE

## BULGARIA

ASSOCIATION OF PEOPLE SUFFERING BY ACROMEGALY IN BULGARIA  
BULGARIAN ANTI-THALASSAEMIC ASSOCIATION  
BULGARIAN CYSTIC FIBROSIS ASSOCIATION  
BULGARIAN HUNTINGTON ASSOCIATION  
INFORMATION CENTRE FOR RARE DISEASES AND ORPHAN DRUGS - BULGARIAN ASSOCIATION FOR PROMOTION OF EDUCATION AND SCIENCE  
NAS - NATIONAL ASSOCIATION SARCOIDOSIS BULGARIA  
NATIONAL ALLIANCE OF PEOPLE WITH RARE DISEASES  
NATIONAL ASSOCIATION FOR CHILD SUPPORT CONGENITAL HYPOTHYROIDISM  
NATIONAL ASSOCIATION OF THE PATIENTS WITH GROWTH HORMONE DEFICIENCY  
NATIONAL GAUCHER ORGANIZATION  
NATIONAL SCLEROSIS ASSOCIATION  
NATIONAL SYRINGOMYELIA ASSOCIATION  
PHA BULGARIA  
THE BULGARIAN SOCIETY OF PATIENTS WITH PULMONARY HYPERTENSION

## BURKINA FASO

FONDATION INTERNATIONALE TIERNO ET MARIAM

## CANADA

CANADIAN ORGANIZATION FOR RARE DISORDERS  
LYMPHOMA COALITION  
PVNH SUPPORT & AWARENESS

## CHINA

CHINESE ORGANIZATION FOR RARE DISORDERS

## COLOMBIA

ASOCIACIÓN COLOMBIANA DE PACIENTES CON ENFERMEDADES DE DEPÓSITO LISOSOMAL

## CROATIA

CROATIAN ALLIANCE FOR RARE DISEASES  
DEBRA, DRUSTVO OBOLJELIH OD BULOZNE EPIDERMOLIZE

## CYPRUS

ASSOCIATION OF PATIENTS & FRIENDS OF IMD «ASPIDA ZOIS»  
CYPRUS ALLIANCE FOR RARE DISORDERS  
CYPRUS PRIMARY IMMUNODEFICIENCY ASSOCIATION AND FRIENDS  
PANCYPRIAN ASSOCIATION FOR RARE GENETIC DISEASES «UNIQUE SMILES»  
THALASSAEMIA INTERNATIONAL FEDERATION (TIF)

## CZECH REPUBLIC

CZECH ASSOCIATION OF MARFAN SYNDROME  
CZECH HUNTINGTON ASSOCIATION  
CZECH NATIONAL ASSOCIATION FOR RARE DISEASES (ČESKÁ ASOCIACE PRO VZÁCNÁ ONEMOCNĚNÍ)  
KLUB NEMOCNÝCH CYSTICKOU FIBROZOU  
META, ASSOCIATION OF PATIENTS WITH LYSOSOMAL STORAGE DISEASES  
NATIONAL ASSOCIATION OF PHENYLKETONURIA (PKU) AND SIMILAR INHERITED METABOLIC DISORDERS (DMP) NÁRODNÍ SDRUŽENÍ PKU

## DENMARK

22Q11 DENMARK  
ADDISON FORENINGEN I DANMARK  
BLÆRÆKSTROFIFORENINGEN  
CCHS DENMARK  
DANISH APERT SYNDROME ASSOCIATION /DANMARKS APERTFORENING  
DANMARKS BLODERFORENING /DANISH HAEMOPHILIA SOCIETY  
DANSK FORENING FOR NEUROFIBRMATOSIS RECKLINGHAUSEN  
EHLERS-DANLOS FORENINGEN I DANMARK  
FORENINGEN AF MÖBIUSSYNDROM I DANMARK



FORENINGEN FOR ATAKSI / HSP  
 ICHTYOSIS ASSOCIATION IN DENMARK  
 IMMUN DEFECT FORENINGEN  
 MCADD-FORENINGEN  
 MITOKONDRIE-FORENINGEN I DANMARK  
 PATIENTFORENINGEN HAE DANMARK  
 PORFYRIFORENINGEN DANMARK - PORPHYRIA ASSOCIATION DENMARK  
 RARE DISEASES DENMARK (SJAELDNE DIAGNOSER)  
 THE DANISH ASSOCIATION OF VON HIPPEL LINDAU PATIENTS AND THEIR RELATIVES  
 THE DANISH OSTEOGENESIS IMPERFECTA SOCIETY  
 WILSON PATIENTFORENINGEN

## ESTONIA

ESTONIAN PRADER WILLI SYNDROME ASSOCIATION

## FINLAND

FINNISH ASSOCIATION OF PEOPLE WITH PHYSICAL DISABILITIES  
 FINNISH CENTRAL ORGANISATION FOR SKIN PATIENTS  
 FINNISH NEURO SOCIETY  
 HARSO-RARE DISEASE ALLIANCE FINLAND  
 HARVINAISET VERKOSTO - FINNISH NETWORK FOR RARE DISEASES  
 INCLUSION FINLAND KVTL

## FRANCE

AFMKT-FRANCE  
 ALLIANCE MALADIES RARES  
 ALLIANCE SANFILIPPO  
 ALLIANCE SYNDROME DE DRAVET  
 ASS. INTERNATIONALE DE DYSTROPHIE NEURO AXONALE INFANTILE  
 ASSOCIATION «LES PETITS COURAGEUX»  
 ASSOCIATION ANÉMIES DYSERYTHROPOIÉTIQUES CONGÉNITALES  
 ASSOCIATION CONTRE LES MALADIES MITOCHONDRIALES  
 ASSOCIATION CRANIOPHARYNGIOME SOLIDARITÉ  
 ASSOCIATION DE LUTTE CONTRE L'ARACHNOÏDITE, LES DOULEURS NEUROPATHIQUES, LES PATHOLOGIES MÉNINGÉES ET SACRÉES ET AUTRES MALADIES ORPHELINES  
 ASSOCIATION DE PREFIGURATION DE LA FONDATION DENISE PICARD  
 ASSOCIATION DE SOUTIEN ET D'INFORMATION A LA LEUCÉMIE LYMPHOÏDE CHRONIQUE ET LA MALADIE DE WALDENSTRÖM  
 ASSOCIATION DES AMIS D'ANNE-LORÈNE - SYNDROME D'AICARDI  
 ASSOCIATION DES GROUPES AMITIÉ TURNER  
 ASSOCIATION DES MALADES DES VAISSEAUX DU FOIE  
 ASSOCIATION DES MALADES DU SYNDROME DE MCCUNE-ALBRIGHT  
 ASSOCIATION DES MALADES D'UN SYNDROME NÉPHROTIQUE  
 ASSOCIATION DES MALADES SOUFFRANT D'ANGIO OEDÈME PAR DÉFICIT EN C1 INHIBITEUR  
 ASSOCIATION DES PANCRÉATITES CHRONIQUES HÉRÉDITAIRES  
 ASSOCIATION DES PATIENTS DE LA MALADIE DE FABRY  
 ASSOCIATION DES PERSONNES CONCERNÉES PAR LE TREMBLEMENT ESSENTIEL  
 ASSOCIATION DES POIC  
 ASSOCIATION DU LOCKED-IN SYNDROME  
 ASSOCIATION DU NAEVUS GÉANT CONGÉNITAL  
 ASSOCIATION DU STRÜMPPELL-LORRAIN  
 ASSOCIATION FRANÇAISE CONTRE L'AMYLOSE  
 ASSOCIATION FRANÇAISE CONTRE LES MYOPATHIES  
 ASSOCIATION FRANÇAISE DE LA FIÈVRE MÉDITERRANÉENNE FAMILIALE  
 ASSOCIATION FRANÇAISE DE LA MALADIE DE BEHCET  
 ASSOCIATION FRANÇAISE DE LA MALADIE DE FANCONI  
 ASSOCIATION FRANÇAISE DE L'ATAXIE DE FRIEDREICH  
 ASSOCIATION FRANÇAISE DE L'OSTEODYSTROPHIE HÉRÉDITAIRE D'ALBRIGHT  
 ASSOCIATION FRANÇAISE DE L'OSTÉOGENÈSE IMPARFAITE  
 ASSOCIATION FRANÇAISE DE NARCOLEPSIE-CATAPLEXIE ET HYPERSONNIE  
 ASSOCIATION FRANÇAISE DES DYSPLASIES ECTODERMiques  
 ASSOCIATION FRANÇAISE DES HÉMOPHILES  
 ASSOCIATION FRANÇAISE DES MALADES ATTEINTS DE PORPHYRIES  
 ASSOCIATION FRANÇAISE DES MALADIES HÉRÉDITAIRES DU RYTHME CARDIAQUE  
 ASSOCIATION FRANÇAISE DES PATIENTS EN AUTO-TRAITEMENT  
 ASSOCIATION FRANÇAISE DES SYNDROMES D'EHLERS-DANLOS  
 ASSOCIATION FRANÇAISE DU GOUGEROT-SJÖGREN  
 ASSOCIATION FRANÇAISE DU LUPUS ET AUTRES MALADIES AUTO-IMMUNES  
 ASSOCIATION FRANÇAISE DU SYNDROME DE CORNELIA DE LANGE  
 ASSOCIATION FRANÇAISE DU SYNDROME DE RETT  
 ASSOCIATION FRANÇAISE DU SYNDROME D'ONDINE  
 ASSOCIATION FRANÇAISE LESCH-NYHAN ACTION  
 ASSOCIATION FRANÇAISE POUR LE SYNDROME DE GILLES DE LA TOURETTE

ASSOCIATION FRANÇOIS AUPETIT  
 ASSOCIATION FRANCOPHONE CONTRE LA POLYCHONDRITE CHRONIQUE ATROPHIANTE  
 ASSOCIATION FRANCOPHONE DES GLYCOGÉNOSES  
 ASSOCIATION FRANCOPHONE DU SYNDROME D'ANGELMAN  
 ASSOCIATION HEMOCHROMATOSE FRANCE  
 ASSOCIATION HISTIOCYTOSE FRANCE  
 ASSOCIATION HUNTINGTON FRANCE  
 ASSOCIATION HYPOPARATHYROIDISME FRANCE  
 ASSOCIATION ICHTYOSE FRANCE  
 ASSOCIATION ISIS  
 ASSOCIATION KERATOCÔNE  
 ASSOCIATION KOURIR  
 ASSOCIATION MALADIES FOIE ENFANTS  
 ASSOCIATION MARFANS  
 ASSOCIATION MÉDICALISTES  
 ASSOCIATION NAEVUS 2000 FRANCE EUROPE  
 ASSOCIATION NATIONALE DES CARDIAQUES CONGÉNITAUX  
 ASSOCIATION NATIONALE DU SYNDROME X FRAGILE «LE GOËLAND»  
 ASSOCIATION NEUROFIBROMATOSES & RECKLINGHAUSEN  
 ASSOCIATION PEMPHIGUS – PEMPHIGOÏDES FRANCE  
 ASSOCIATION POUR LUTTE CONTRE LES MALADIES INFLAMMATOIRES DU FOIE ET DES VOIES BILIAIRES  
 ASSOCIATION POUR AIDER ET INFORMER LES SYRINGOMYÉLIQUES EUROPÉENS RÉUNIS  
 ASSOCIATION POUR LA LUTTE CONTRE L'ALCAPTONURIE  
 ASSOCIATION POUR LA RECHERCHE SUR LA SCLÉROSE LATÉRALE AMYOTROPHIQUE  
 ASSOCIATION POUR LA RECHERCHE SUR L'ATROPHIE MULTISYSTÉMATISÉE INFORMATION-SOUTIEN EN EUROPE  
 ASSOCIATION POUR L'AIDE AUX PERSONNES CONCERNÉES PAR LES MALADIES RARES MUCKLE WELLS SYNDROME ET CINCA  
 ASSOCIATION POUR L'INFORMATION ET LA PRÉVENTION DE LA DRÉPANOCYTOSE  
 ASSOCIATION POUR L'INFORMATION ET LA RECHERCHE SUR LE SYNDROME DE SAPHO  
 ASSOCIATION POUR L'INFORMATION ET LA RECHERCHE SUR LES MALADIES RÉNALES GÉNÉTIQUES  
 ASSOCIATION SANS DIAGNOSTIC ET UNIQUE  
 ASSOCIATION SCLÉROSE TUBÉREUSE DE BOURNEVILLE  
 ASSOCIATION SOS DESMOÏDE  
 ASSOCIATION SPINA BIFIDA ET HANDICAPS ASSOCIÉS  
 ASSOCIATION SURRÉNALES  
 ASSOCIATION SYNDROME DE KLEINE-LEVIN  
 ASSOCIATION SYNDROME DE ROKITANSKY - MRKH  
 ASSOCIATION THERA WANKA  
 CHARCOT-MARIE-TOOTH FRANCE  
 CUTIS LAXA INTERNATIONALE  
 DEBRA FRANCE  
 DRAVET SYNDROME EUROPEAN FEDERATION  
 ENFANTS DE LA LUNE ASSOCIATION POUR LE XERODERMA PIGMENTOSUM  
 EUROPEAN FEDERATION OF ASSOCIATIONS OF PATIENTS WITH HAEMOCHROMATOSIS  
 FEDERATION EURO-HSP  
 FITIMA EUROPE - FONDATION INTERNATIONAL TIerno ET MARIAM  
 FOP FRANCE  
 FRANCE LYMPHANGIOLEIOMYOMATOSE (FLAM)  
 FRANCE LYMPHOME ESPoir  
 GENERATION 22  
 GENESPOIR: ASSOCIATION FRANÇAISE DES ALBINISMES  
 GENIRIS  
 HTAPFRANCE  
 HYPOPHOSPHATASIE EUROPE  
 INCONTINENTIA PIGMENTI FRANCE  
 INFLAM'CEIL  
 LES ENFANTS DU JARDIN  
 LES PETITS BOURDONS  
 LIGUE CONTRE LA CARDIOMYOPATHIE  
 LIGUE NATIONALE CONTRE LE CANCER  
 LUPUS FRANCE  
 MOSAÏQUES - ASSOCIATION DES «X FRAGILE»  
 NAEVUS GLOBAL  
 POLYPOSES FAMILIALES - APTEPF  
 PRADER WILLI FRANCE  
 RETINA FRANCE  
 SANFILIPPO SUD  
 SOS GLOBI  
 SYNDROME DE MOEBIUS FRANCE  
 UNION HUNTINGTON ESPoir  
 UNION NATIONALE DES ASSOCIATIONS PARENTS ET AMIS PERSONNES HANDICAPÉES MENTALES

VAINCRE LA MUCOVISCIDOSE

VAINCRE LES MALADIES LYSOSOMALES

VALENTIN - ASSOCIATION DES PORTEURS D'ANOMALIES CHROMOSOMIQUES

VHL FRANCE

XTRAORDINAIRE

## GEORGIA

GEORGIAN FOUNDATION FOR GENETIC AND RARE DISEASES

## GERMANY

ACHSE ALLIANZ CHRONISCHER SELTENER ERKRANKUNGEN E.V.

AHC-DEUTSCHLAND EV

AKTION BENNI & CO E.V.

ALPHA 1 NETZWERK E.V.

BSHV FÜR KINDER, JUGENDLICHE UND ERWACHSENE MIT SELTENEN, CHRONISCHEN SKELETTERKRANKUNGEN E.V.

BUNDESVERBAND ANGEBORENE GEFÄSSFEHLBILDUNGEN E.V.

BUNDESVERBAND DER CLUSTERKOPFSCHMERZEN-SELBSTHILFE-GRUPPEN

BUNDESVERBAND KLEINWÜCHSIGE MENSCHEN UND IHRE FAMILIEN E.V.

BUNDESVERBAND POLIOMYELITIS EV

CHARGE SYNDROM EV

CYSTIC FIBROSIS EUROPE

CYSTINOSE SELBSTHILFE E.V.

DEUTSCHE EHLERS-DANLOS-INITIATIVE

DEUTSCHE GBS INITIATIVE E.V.

DEUTSCHE INTERESSENGEMEINSCHAFT PKU

DEUTSCHE KLINEFELTER-SYNDROM VEREINIGUNG E.V.

DEUTSCHE NARKOLEPSIE GESELLSCHAFT E.V.

DEUTSCHE SARKOIDOSE VEREINIGUNG GEMEINNÜTZIGER E.V.

DEUTSCHE SELBSTHILFE ANGEBORENE IMMUNDEFEKTE EV

DEUTSCHE SYRINGOMYELIE UND CHIARI MALFORMATION DSCM E.V.

DEUTSCHSPRACHIGE SELBSTHILFEGRUPPE FÜR ALKAPTONURIE E.V.

EHLERS-DANLOS-SELBSTHILFE E.V.

EUROPEAN ASSOCIATION OF PATIENT ORGANISATIONS OF SARCOIDOSIS (EPOS)

EUROPEAN CONGENITAL HEART DISEASE ORGANISATION

EUROPEAN MEN ALLIANCE

EUROPEAN NETWORK FOR ICHTHYOSIS - ENI

FAUN STIFTUNG

GAUCHER GESELLSCHAFT DEUTSCHLAND E.V.

GESELLSCHAFT FÜR MUKOPOLYSACCHARIDOSEN E.V.

HAE VEREINIGUNG E.V. (HEREDITARY ANGIOEDEMA)

HOFFNUNGSBAUM E.V.

HSP-SELBSTHILFEGRUPPE DEUTSCHLAND E.V.

ICA-DEUTSCHLAND E.V. FÖRDERVEREIN INTERSTITIELLE CYSTITIS

INTERESSENGEMEINSCHAFT EPIDERMOLYSIS BULLOSA - DEBRA DEUTSCHLAND

INTERESSENGEMEINSCHAFT FRAGILES-X E.V.

KINDER MIT DELETIONSSYNDROM 22Q11 (KIDS-22Q11) EV

KINDER-AUGEN-KREBS-STIFTUNG

KINDERNETZWERK E.V.

KINDNESS FOR KIDS FOUNDATION

LEBEN MIT BEHCET - SÜDDEUTSCHLAND

LEBEN MIT BEHCET IN DEUTSCHLAND

LEONA E.V.

MARFAN EUROPE NETWORK

MASTOZYTOTOSE INITIATIVE SELBSTHILFENETZWERK

MORBUS-OSLER SELBSTHILFE EV

MPN-NETZWERK E.V.

MULTINATIONAL INTERSTITIAL CYSTITIS ASSOCIATION (MICA)

NCL-GRUPPE DEUTSCHLAND E.V.

NEPHIE

NETZWERK HYPOPHYSEN- UND NEBENNIERENERKRANKUNGEN E. V.

OIFE - OSTEOGENEIS IMPERFECTA FEDERATION EUROPE

PATIENTEN- UND SELBSTHILFEORGANISATION FÜR KINDER UND ERWACHSENE MIT KRANKER SPEISERÖHRE

PRO RETINA DEUTSCHLAND E.V.

PULMONALE HYPERTONIE E.V.

SANFILIPPO INITIATIVE E.V.

SELBSTHILFE EPP E.V.

SELBSTHILFE ICHTHYOSE E.V.

SELBSTHILFEGRUPPE EKTODERMALE DYSPLASIE E.V.

SELBSTHILFEGRUPPE FÜR PXE - ERKRANKTE DEUTSCHLANDS E.V.

SELBSTHILFEGRUPPE GLYKOGENOSE DEUTSCHLAND E.V.

SELBSTHILFEORGANISATION FÜR MENSCHEN MIT ANOREKTALFEHLBILDUNGEN

SKLERODERMIE SELBSTHILFE E.V.

SMA EUROPE

SOS-DESMOID E.V.

STIFF-PERSON-SYNDROM SELBSTHILFE DEUTSCHLAND E.V.

THE FEDERATION OF ESOPHAGEAL ATRESIA AND TRACHEO-ESOPHAGEAL FISTULA SUPPORT GROUPS E.V.

TOM WAHLIG STIFTUNG

VEREIN AHC18+

VEREIN VHL (VON HIPPEL - LINDAU) BETROFFENER FAMILIEN E.V.

ZNM - ZUSAMMEN STARK! E. V.

## GREECE

ASSOCIATION OF GREEK FRIENDS FOR PAEDIATRIC IMMUNOLOGY

ASSOCIATION OF GREEK FRIENDS FOR PAEDIATRIC IMMUNOLOGY

PRIMARY IMMUNODEFICIENCIES «HARMONY»

CHILD'S HEART

GREEK ALLIANCE FOR RARE DISEASE

HELLENIC CYSTIC FIBROSIS ASSOCIATION

HELLENIC LEAGUE AGAINST RHEUMATISM

HELLENIC MYASTHENIA GRAVIS ASSOCIATION

HELLENIC SOCIETY SUPPORTING CHILDREN WITH GENETIC DISORDERS

«TO MELLON»

PANHellenic ASSOCIATION OF PATIENTS & FRIENDS WITH

NEUROFIBROMATOSIS «LIFE WITH NF»

PANHELLINIK ASSOCIATION OF PATIENTS WITH LYSOSOMAL

DISORDERS

PARENTS AND FRIENDS OF PEOPLE WITH RETT SYNDROME

ASSOCIATION

PRADER WILLI SYNDROME ASSOCIATION HELLAS

SOCIETY OF PATIENTS AND FRIENDS OF PATIENTS WITH INHERITED

METABOLIC DISEASE

TUBEROUS SCLEROSIS ASSOCIATION OF GREECE

VHLFA ALLIANCE IN GREECE

## GUATEMALA

ASOCIACIÓN NACIONAL GUATEMALTECA PARA LAS ENFERMEDADES

DE DEPÓSITO LISOSOMAL

PROCRECE

## HONG KONG, SAR OF CHINA

JOSHUA HELLMANN FOUNDATION FOR ORPHAN DISEASE

## HUNGARY

(HUNGARIAN HAEMOPHILIA SOCIETY) MAGYAR HEMOFÍLIA EGYESÜLET

PRIMER IMMUNHIÁNYOS BETEGEK EGYESÜLETE

RARE DISEASES HUNGARY - HUNGARIAN FEDERATION OF PEOPLE WITH RARE AND CONGENITAL DISEASES

## ICELAND

AHC FEDERATION OF EUROPE

ALTERNATING HEMIPLEGIA OF CHILDHOOD ASSOCIATION OF ICELAND

EINSTÖK BÖRN - SUPPORT GROUP FOR CHILDREN WITH RARE DISORDERS

GUDRUN'S RETT SYNDROME RESEARCH TRUST

HYPOPARATHYROIDISM EUROPE (HPTH EUROPE)

THE ICELANDIC CHILDHOOD CANCER PARENT ORGANIZATION

## INDIA

INDIAN ORGANIZATION FOR RARE DISEASES

ORGANIZATION FOR RARE DISEASES INDIA

## IRAN

CHARITY FOUNDATION FOR SPECIAL DISEASES

RARE DISEASE FOUNDATION OF IRAN

## IRELAND

ALPHA ONE FOUNDATION

ATAXIA IRELAND

BARRETTOWN SERIOUS FUN

CYSTINOSIS IRELAND

FIGHTING BLINDNESS

FRIEDREICH'S ATAXIA RESEARCH ALLIANCE

GENETIC AND RARE DISORDERS ORGANISATION (GRDO)

HUNTINGTON'S DISEASE ASSOCIATION OF IRELAND

IRISH CANCER SOCIETY

MUSCULAR DYSTROPHY IRELAND

NEUROFIBROMATOSIS ASSOCIATION OF IRELAND

SICKLE CELL AND THALASSAEMIA IRELAND

SYRINGOMYELIA IRELAND

THE CAVAN TOMMY HOEY TRUST

THE IRISH FRAGILE X SOCIETY

## ITALY

ABC ASSOCIAZIONE BAMBINI CRI DU CHAT

ACONDROPLASIA - INSIEME PER CRESCERE - ONLUS

AMICI DELLA PORFIRIA - SAN PIO DA PIETRELCINA ONLUS

ANGELI NOONAN

ASSOCIAZIONE PERSONE CON MALATTIE REUMATICHE - APMAR

ONLUS



ASSOCIAZIONE DEL BAMBINO EMOPATICO  
ASSOCIAZIONE FAMIGLIE DI SOGGETTI CON DEFICIT DELL'ORMONE DELLA CRESCITA ED ALTRE PATOLOGIE  
ASSOCIAZIONE ITALIANA CISTITE INTERSTIZIALE  
ASSOCIAZIONE ITALIANA DEI CARDIOPATICI CONGENITI ADULTI - ITALIAN GUCH ASSOCIATION  
ASSOCIAZIONE ITALIANA ESTROFIA VESCICALE-EPISPADIA ONLUS  
ASSOCIAZIONE ITALIANA GAUCHER ONLUS - AIG  
ASSOCIAZIONE ITALIANA GLICOGENOSI  
ASSOCIAZIONE ITALIANA LEUCODISTROFIE UNITE (AILU)  
ASSOCIAZIONE ITALIANA LINFOISTIOCITOSI EMOFAGOCITICA MARIO RICCIARDI'S BROTHERS PROGETTO HLH ONLUS  
ASSOCIAZIONE ITALIANA MALATI DI ALCAPTONURIA  
ASSOCIAZIONE ITALIANA MALFORMAZIONE DI ARNOLD-CHIARI CHILD  
ASSOCIAZIONE ITALIANA MIASTENIA E MALATTIE IMMUNODEGENERATIVE - AMICI DEL BESTA ONLUS  
ASSOCIAZIONE ITALIANA MIASTENIA ONLUS  
ASSOCIAZIONE ITALIANA MUCOPOLISACCARIDOSI E MALATTIE AFFINI  
ASSOCIAZIONE ITALIANA NIEMANN PICK E MALATTIE AFFINI - ONLUS  
ASSOCIAZIONE ITALIANA PER LA LOTTA ALLE PHTS  
ASSOCIAZIONE ITALIANA PER LE MALFORMAZIONI ANORETTALI  
ASSOCIAZIONE ITALIANA SCLEROSI LATERALE AMIOTROFICA (SEZ. LOMBARDIA)  
ASSOCIAZIONE ITALIANA SINDROME DI POLAND  
ASSOCIAZIONE ITALIANA SINDROME E MALATTIA DI BEHÇET (SIMBA)  
ASSOCIAZIONE ITALIANA SIRINGOMIELIA E ARNOLD CHIARI  
ASSOCIAZIONE ITALIANA SOSTEGNO MALATTIE METABOLICHE EREDITARIE ONLUS  
ASSOCIAZIONE ITALIANA TELEANGECTASIA EMORRAGICA ITALIANA - HHT ONLUS  
ASSOCIAZIONE LAM ITALIA ONLUS  
ASSOCIAZIONE MALATI DI HAILEY HAILEY DISEASE  
ASSOCIAZIONE MALATI DI PORFIRIA  
ASSOCIAZIONE MALATTIE RARE DELL'ALTA MURGIA ONLUS  
ASSOCIAZIONE NAZIONALE ITALIANA MALATI SINDROME DI SJOGREN  
ASSOCIAZIONE PER L'INFORMAZIONE E LO STUDIO DELLA ACONDROPLASIA  
ASSOCIAZIONE PERSONE WILLIAMS ITALIA ONLUS  
ASSOCIAZIONE SCLEROSI TUBEROSA  
ASSOCIAZIONE SINDROME NEFROSICA ITALIA  
ASSOCIAZIONE STUDIO MALATTIE METABOLICHE EREDITARIE ONLUS  
ASSOCIAZIONE VENETA PER LA LOTTA ALLA TALASSEMIA  
CIDP ITALIA ONLUS  
COSTELLO.CFC - ASSOCIAZIONE ITALIANA SINDROME DI COSTELLO - CARDIOFACIOCUTANEA - RASOPATIE - ONLUS  
DEBRA ITALIA ONLUS – ASSOCIAZIONE PER LA RICERCA SULL'EPIDERMOLISI BOLLOSA  
DRAVET ITALIA ONLUS  
FEDERAZIONE NAZIONALE PRADER WILLI  
FOP ITALIA ONLUS  
GLI AMICI DI DANIELA  
INCONTINENTIA PIGMENTI ASSOCIAZIONE ITALIANA ONLUS  
LEGA ITALIANA SCLEROSI SISTEMICA ONLUS  
LND FAMIGLIE ITALIANE ONLUS  
P63 EEC SYNDROME INTERNATIONAL NET WORK WORD COMMUNICATION - MALATTIE RARE ONLUS  
PANDAS ITALIA  
PARENT PROJECT ONLUS  
PKS KIDS ITALIA ONLUS  
RETINA ITALIA ONLUS  
RING 14 INTERNATIONAL  
SOD ITALIA - ASSOCIAZIONE ITALIANA DISPLASIA SETTO OTTICA E IPOPLASIA DEL NERVO OTTICO  
UNIAMO - FEDERAZIONE ITALIANA MALATTIE RARE  
UNIONE ITALIANA ITTIOSI  
UNITED ONLUS  
VISUS - ASSOCIAZIONE TRA AFFETTI DA RETINITE PIGMENTOSA  
XLPDR INTERNATIONAL ASSOCIATION

## JAPAN

JAPAN PATIENT ASSOCIATION

## KAZAKHSTAN

PATIENTS WITH CANCER AND RARE DISEASES SUPPORT ASSOCIATION

## LATVIA

ASSOCIATION OF PEOPLE WITH SPECIAL NEEDS «MOTUS VITA»  
EMPTY NOSE SYNDROME INTERNATIONAL ASSOCIATION  
LATVIAN ALLIANCE FOR RARE DISEASES  
RARE DISEASE ASSOCIATION «CALADRIUS»

## LEBANON

LEBANESE ASSOCIATION FOR NEUROMUSCULAR DISEASES

## LITHUANIA

COMMUNITY OF ONCOHEMATOLOGICAL PATIENT ORGANISATIONS KRAUJAS

## LUXEMBOURG

ALAN ASBL

DENII ASBL SYNDROME DE MARFAN

EEN HÄERZ FIR KRIIBSKRANK KANNER ASBL

FONDATIOUN KRIIBSKRANK KANNER

RETT SYNDROME EUROPE

## MALAYSIA

MALAYSIA METABOLIC SOCIETY

MALAYSIAN RARE DISORDERS SOCIETY

## MEXICO

PROYECTO PIDE UN DESEO MEXICO IAP

RED SANFILIPPO

## MOROCCO

ASSOCIATION MAROCAINE DE LA FIÈVRE MÉDITERRANÉENNE FAMILIALE ET DES AUTRES FIEVRES RÉCURRENTES  
SUN'HOP

## NEPAL

GBS/CIDP FOUNDATION NEPAL

## NETHERLANDS

ALS PATIENTS CONNECTED

AUTOSOMAAL DOMINANT CEREBELLAIRE ATAXIE-VERENIGING NEDERLAND

BIJNERVERENIGING NVACP

CHILDHOOD CANCER INTERNATIONAL

CMTC-OVM

EUROPEAN SOCIETY FOR PHENYLKETONURIA

EUROPEAN VHL (VON HIPPEL-LINDAU) FEDERATION

EUROPEAN WALDENSTRÖM MACROGLOBULINEMIA NETWORK

FABRY SUPPORT & INFORMATIE GROEP NEDERLAND

FIBRODYSPLASIA OSSIFICANS PROGRESSIVA STICHTING NEDERLAND

FSHD EUROPE

INTERNATIONAL MITO PATIENTS

INTERNATIONAL PAINFUL BLADDER FOUNDATION

INTERNATIONAL PORPHYRIA PATIENT NETWORK

INTERSTITIËLE CYSTITIS PATIENTENVERENIGING

ITP PATIENTENVERENIGING

MSS (MARSHALL-SMITH SYNDROME) RESEARCH FOUNDATION

NEDERLANDSE HYPOFYSE STICHTING (DUTCH PITUITARY FOUNDATION)

NEDERLANDSE PHENYLKETONURIE VERENIGING / DUTCH PKU ASSOCIATION

NEPHCEUROPE

NETHERLANDS ASSOCIATION OF PATIENTS WITH STERNO COSTO CLAVICULAR HYPEROSTOSIS

NETHERLANDS LIVER PATIENTS ASSOCIATION

NEUROFIBROMATOSE VERENIGING NEDERLAND

OSCAR NEDERLAND

PSC PATIENTS EUROPE

STICHTING AA & PNH CONTACTGROEP

STICHTING AMYLOIDOSE NEDERLAND SAN

STICHTING HISTIOCYTOSE NEDERLAND

STICHTING IJZERSTERK

STICHTING NET-GROEP

STICHTING SHWACHMAN DIAMOND SYNDROME SUPPORT HOLLAND

STICHTING STOFWISSELKRACHT

STICHTING TERRE - RETT SYNDROME FOUNDATION

STICHTING VOOR AFWEERSTOORNISSEN

UNITED PARENT PROJECTS MUSCULAR DYSTROPHY

VASCULITIS STICHTING (FORMERLY KNOWN AS FRIEDRICH WEGENER STICHTING)

VERENIGING SAMENWERKENDE OUDER EN PATIËNTENORGANISATIES (VSOP)

VERENIGING SPIERZIEKTEN NEDERLAND - DUTCH ASSOCIATION FOR NEUROMUSCULAR DISEASE

VOLWASSENEN, KINDEREN EN STOFWISSELINGSZIEKTEN

WORLD ALLIANCE OF PITUITARY ORGANIZATIONS

## NEW ZEALAND

NEW ZEALAND ORGANISATION FOR RARE DISORDERS

## NORWAY

ANIRIDI NORGE

ANIRIDIA EUROPE

FRAMBU - RESOURCE CENTRE FOR RARE DISORDERS

MORBUS ADDISON ASSOCIATION NORWAY

MPS FORENINGEN NORGE

**NORDIC HYPOPARA ORGANISATION**

NORSK FORENING FOR ARVELIG SPASTISK PARAPARESE /ATAKSI

NORSK FORENING FOR EHLERS-DANLOS SYNDROM

NORSK FORENING FOR OSTEOGENESIS IMPERFECTA

NORSK FORENING FOR TUBEROS SKLEROSE

OSLERFORENINGEN NORGE

**POLAND**

DEBRA POLSKA

FUNDACJA SANFILIPPO

FUNDACJA SMA (SMA FOUNDATION POLAND)

FUNDACJA UMIEC POMAGAC (FOUNDATION FOR RD MPS)

MATIO-FUNDACJI POMOCY RODZINOM I CHORYM NA

MUKOWISCYDOZE

POLISH NATIONAL FORUM ON THE TREATMENT OF ORPHAN DISEASES

- ORPHAN

POLISH PKU AND RD ASSOCIATION «ARS VIVENDI»

POLISH SOCIETY OF MPS AND RELATED DISEASES

POLSKIE STOWARZYSZENIE NA RZECZ OSÓB Z AHC

THE DINA RADZIWILOWA CHILD'S HEART FOUNDATION

**PORTUGAL**

ALIANÇA PORTUGUESA DE ASSOCIAÇÕES DAS DOENÇAS RARAS

APLL - ASSOCIAÇÃO PORTUGUESA DE LEUCEMIAS E LINFOMAS

ASSOCIAÇÃO NACIONAL DE DISPLASIAS OSSEAS

ASSOCIAÇÃO NACIONAL DE FIBROSE QUÍSTICA

ASSOCIAÇÃO PORTUGUESA CDG E OUTRAS DOENÇAS METABÓLICAS

ASSOCIAÇÃO PORTUGUESA DE CHARCOT-MARIE-TOOTH

ASSOCIAÇÃO PORTUGUESA DE DOENTES NEUROMUSCULARES

ASSOCIAÇÃO SANFILIPPO PORTUGAL

FEDRA - FEDERAÇÃO PORTUGUESA DE DOENÇAS RARAS

LIGUA NACIONAL PARA O ESTUDO E APOIO DA DEFICIÊNCIA MENTAL

RARÍSSIMAS - ASSOCIAÇÃO NACIONAL DE DEFICIÊNCIAS MENTAIS E

RARAS

**ROMANIA**

ASOCIATIA COPILUL MEU-INIMA MEA

ASOCIATIA PERSOANELOR CU TALASEMIE MAJORA

ASOCIATIA ROMÂNĂ DE CANCERE RAR

ASOCIATIA ROMANA SPINA BIFIDA SI HIDROCEFALIE

ASOCIATIA WERDNIG HOFFMAN AWH

ASOCIATIA WILLIAMS SYNDROME

ROMANIAN MYASTHENIA GRAVIS ASSOCIATION

ROMANIAN NATIONAL ALLIANCE FOR RARE DISEASES

ROMANIAN PRADER WILLI ASSOCIATION

**RUSSIAN FEDERATION**

FABRY RUSSIA

HELP TO CYSTIC FIBROSIS PATIENTS

INTERREGIONAL PUBLIC ORGANISATION FOR GAUCHER DISEASE

MPS RUSSIA

NATIONAL ASSOCIATION OF PATIENTS WITH RARE DISEASES

«GENETICA»

NGO «FRAGILE CHILDREN»

RUSSIAN ASSOCIATION OF RARE DISEASES

RUSSIAN INTERREGIONAL PUBLIC ORGANIZATION «INTER-REGIONAL

SUPPORT CENTRE FOR PATIENTS WITH ANIRIDIA «IRIS»

RUSSIAN PATIENT UNION

RUSSIAN RETT SYNDROME ASSOCIATION

SPIPORZ UNION OF PATIENTS WITH RARE DISEASES AND RARE

DISEASE PATIENTS ORGANISATIONS

THE ASSOCIATION OF PRIMARY IMMUNODEFICIENCY PATIENTS

**SERBIA**

CHILD RARE DISEASE SUPPORT AND RESEARCH ASSOCIATION LIFE

CHILDHOOD CANCER PARENT ORGANISATION «ZVONCICA»

LYMPHOMA PATIENTS' ASSOCIATION

NATIONAL ORGANIZATION FOR RARE DISEASES, SERBIA

**SINGAPORE**

RARE DISORDERS SOCIETY (SINGAPORE)

**SLOVAKIA**

DEBRA SR

ORGANISATION OF MUSCULAR DYSTROPHY IN THE SLOVAK REPUBLIC

SLOVAK ALLIANCE OF RARE DISEASES

SLOVAK CYSTIC FIBROSIS ASSOCIATION

ZDRUZENIE OJEDLINELÝCH GENETICKÝCH OCHORENIE

**SLOVENIA**

ASSOCIATION OF PATIENTS WITH BLOOD DISEASES - DRUSTVO

BOLNIKOV S KRVNIMI BOLEZNIMI

DEBRA SLOVENIA - DRUSTVO DEBRA SLOVENIJA

**EAMDA EUROPEAN ALLIANCE OF NEUROMUSCULAR DISORDERS**

ASSOCIATIONS

FABRY PATIENTS ASSOCIATION SLOVENIA / DRUŠTVO BOLNIKOV S

FABRYJEVO BOLEZNIJO SLOVENIJE

FOUNDATION OF CHILD NEUROLOGY

**SOUTH AFRICA**

PRIMARY IMMUNODEFICIENCY NETWORK OF SOUTH AFRICA

RARE DISEASE SOCIETY OF SOUTH AFRICA

**SPAIN**

ALIANZA ESPAÑOLA DE FAMILIAS DE VON HIPPEL LINDAU

ASOCIACIÓ CATALANA DE LAS NEUROFIBROMATOSIS

ASOCIACIÓ D'AFECTATS DE SÍRINGOMIÈLIA

ASOCIACIÓN ANDALUZA CONTRA LA FIBROSIS QUÍSTICA

ASOCIACIÓN ANDALUZA DE PACIENTES CON SÍNDROME DE TOURETTE

Y TRASTORNOS ASOCIADOS

ASOCIACION CHIARI Y SÍRINGOMIELIA DEL PRINCIPADO DE ASTURIAS

ASOCIACIÓN DE AFECTADOS DE NEUROFIBROMATOSIS

ASOCIACIÓN DE AFECTADOS POR DISPLASIA ECTODÉRMICA

ASOCIACION DE ATROFIA DE NERVIÓ OPTICO DE LEBER

ASOCIACIÓN DE DEFICIENCIAS DE CRECIMIENTO Y DESARROLLO

ASOCIACIÓN DE EPIDERMOLISIS BULLOSA DE ESPAÑA (DEBRA

ESPAÑA)

ASOCIACIÓN DE ESCLERODERMIA CASTELLON

ASOCIACION DE FAMILIARES Y ENFERMOS DE FPI Y PFP

ASOCIACIÓN DE HEMOGLOBINURIA PAROXÍSTICA NOCTURNA

ASOCIACIÓN DE HUESOS DE CRISTAL DE ESPAÑA

ASOCIACIÓN DE LAS MUCOPOLISACARIDOSIS Y SÍNDROMES

RELACIONADOS

ASOCIACIÓN DE NEVUS GIGANTE CONGÉNITO

ASOCIACION ESPAÑOLA DEL SÍNDROME CDG, DEFECTOS

CONGÉNITOS DE LA GLICOSILACION

ASOCIACIÓN ESPAÑOLA ANIRIDIA A.E.A.

ASOCIACIÓN ESPAÑOLA DE ANGIOEDEMA FAMILIAR

ASOCIACIÓN ESPAÑOLA DE ENFERMOS DE GLUCOGENOSIS

ASOCIACIÓN ESPAÑOLA DE ENFERMOS Y FAMILIARES DE LA

ENFERMEDAD DE GAUCHER ESPAÑA

ASOCIACIÓN ESPAÑOLA DE ESCLERODERMIA

ASOCIACIÓN ESPAÑOLA DE FAMILIARES Y ENFERMOS DE WILSON

ASOCIACION ESPAÑOLA DE FIBRODISPLASIA OSIFICANTE PROGRESIVA

ASOCIACIÓN ESPAÑOLA DE FIEBRE MEDITERRANEA FAMILIAR

ASOCIACIÓN ESPAÑOLA DE ICTIOSIS

ASOCIACIÓN ESPAÑOLA DE PARAPARESIA ESPÁSTICA FAMILIAR

STRÜMPPELL-LORRAIN

ASOCIACIÓN ESPAÑOLA DE PORFIRIA

ASOCIACIÓN ESPAÑOLA DE SÍNDROME DE POLAND

ASOCIACIÓN ESPAÑOLA DEL SÍNDROME DE JOUBERT

ASOCIACIÓN ESPAÑOLA SÍNDROME DE SJÖGREN

ASOCIACIÓN HHT ESPAÑA

ASOCIACIÓN LUPUS DE MÁLAGA Y AUTOINMUNES

ASOCIACION MIRADAS QUE HABLAN DUPLICACION MECP2

ASOCIACIÓN NACIONAL AMIGOS DE ARNOLD CHIARI

ASOCIACIÓN NACIONAL DE AFECTADOS POR SÍNDROMES DE EHLERS

DANLOS E HIPERLAXITUD

ASOCIACIÓN NACIONAL DE HIPERTENSIÓN PULMONAR

ASOCIACIÓN NACIONAL SÍNDROME DE APERTY Y OTRAS

CRANEOSINOSTOSIS SINDRÓMICAS

ASOCIACIÓN SÍNDROME ANGELMAN

ASOCIACIÓN SÍNDROME DE LOWE DE ESPAÑA

ASOCIACIÓN SÍNDROME LESCH NYHAN ESPAÑA

ASOCIACION STOP SANFILIPPO

ASOCIACIÓ CATALANA DE LA DELECCIÓ 22Q

ASOCIACIÓ CATALANA DE SÍNDROME DE SJÖGREN

ASOCIACIÓ D'APRÀXIA OCULAR I MALALTIES ASSOCIADES

ASOCIACIÓN CATALANA DE ENFERMEDADES NEUROMUSCULARES

ASSOCIATION OF RELATIVES AND PEOPLE AFFECTED BY

LIPODISTROPHYES

D' GENES ASOCIACIÓN DE ENFERMEDADES RARAS DE MURCIA

DRAVET SYNDROME FOUNDATION, DELEGACION EN ESPAÑA

DUCHENNE PARENT PROJECT ESPAÑA

EUROPEAN NETWORK FOR RARE AND CONGENITAL ANAEMIAS

FEDERACIÓ CATALANA DE MALALTIES MINORITÀRIES

FEDERACIÓN DE ASOCIACIONES DE RETINOSIS PIGMENTARIA DE

ESPAÑA

FEDERACIÓN DE ATAXIAS DE ESPAÑA

FEDERACIÓN ESPAÑOLA DE ENFERMEDADES NEUROMUSCULARES

FEDERACIÓN ESPAÑOLA DE ENFERMEDADES RARAS

FEDERACIÓN ESPAÑOLA DE FIBROSIS QUÍSTICA

FEDERACIÓN ESPAÑOLA DE HEMOFILIA

FEDERACIÓN ESPAÑOLA DE MALFORMACIÓN DE CHIARI Y PATOLOGÍAS

ASOCIADAS



FEDERACION ESPANOLA DE PADRES CON NINOS CON CANCER  
FEDERACIÓN ESPAÑOLA DEL SÍNDROME X FRÁGIL  
FUNDACIÓN ANDRÉS MARCIO, NIÑOS CONTRA LA LAMINOPATIA  
FUNDACION MENUDOS CORAZONES  
FUNDACIÓN NIEMANN PICK DE ESPAÑA  
FUNDACIÓN SÍNDROME 5P MENOS  
FUNDACIÓN SÍNDROME DE MOEBIUS  
FUNDACION SÍNDROME DE WEST  
FUNDACIÓN SÍNDROME WOLF HIRSCHHORN O 4P-  
MIQUEL VALLS FOUNDATION (FUNDACION PRIVADA CATALANA DE  
ESCLEROSI LATERAL AMIOTROFICA)

## SWEDEN

AGRENSKA  
AORTA DISSEKTION FÖRENINGEN SKANDINAVIEN  
EUROPEAN DYSMELIA REFERENCE INFORMATION CENTRE  
NEUROLOGISKT HANDIKAPPADES RIKSFÖRBUND / SWEDISH  
ASSOCIATION OF PERSONS WITH NEUROLOGICAL DISABILITIES  
PRADER WILLI SYNDROME ASSOCIATION IN SWEDEN  
PRIMÄR IMMUNBRIST ORGANISATIONEN  
RARE DISEASES SWEDEN (RIKSFÖRBUNDET SÄLLSYNTA DIAGNOSER)  
SVENSKA MARFANFÖRENINGEN  
SWEDISH CYSTIC FIBROSIS ASSOCIATION  
SWEDISH EDS ASSOCIATION (EDS RIKSFÖRBUND)  
SWEDISH MPS SOCIETY  
THE SWEDISH COOPERATIVE BODY OF ORGANIZATIONS OF DISABLED  
PEOPLE  
WILHELM FOUNDATION

## SWITZERLAND

ASSOCIATION ENFANCE ET MALADIES ORPHELINES  
ASSOCIATION SUISSE ROMANDE INTERVENANT CONTRE LES  
MALADIES NEURO-MUSCULAIRES  
BLACKSWAN FOUNDATION  
FONDATION SANFILIPPO SUISSE  
FOUNDATION ORPHANHEALTHCARE  
FRAXAS - ASSOCIATION X FRAGILE SUISSE  
HAEI - HEREDITARY ANGIOEDEMA INTERNATIONAL ASSOCIATION  
HHT-SWISS  
LEUKEMIA PATIENT ADVOCATES FOUNDATION  
MARFAN FOUNDATION SWITZERLAND (MARFAN STIFTUNG SCHWEIZ)  
PRADER WILLI SYNDROM VEREINIGUNG SCHWEIZ  
PRORARIS  
RETINA INTERNATIONAL  
SCHWEIZERISCHE GESELLSCHAFT FÜR PORPHYRIE  
SELBSTHILFE ICHTHYOSE SCHWEIZ  
SMA SCHWEIZ

## TAIWAN , PROVINCE OF CHINA

TAIWAN FOUNDATION FOR RARE DISORDERS

## THE FORMER YUGOSLAV REPUBLIC OF MACEDONIA

LIFE WITH CHALLENGES  
NATIONAL ALLIANCE FOR RARE DISEASES OF R. MACEDONIA  
ZDRUZENIE ZA CISTICNA FIBROZA (MACEDONIAN CF ASSOCIATION)

## TURKEY

KIFDER  
MUKOPOLISAKKARIDOZ VE BENZERI LIZOZOMAL DEPO HASTALIKLARI  
DERNERGI

## UKRAINE

ASSOCIATION OF PATIENTS WITH PULMONATY HYPERTENSION  
CHARITABLE FOUNDATION OF SISTER DALILA  
KHARKIV'S CHARITABLE FOUNDATION - «CHILDREN WITH SPINAL  
MUSCULAR ATROPHY»  
NON-GOVERNMENTAL ORGANIZATION «RARE DISEASES OF UKRAINE»  
UKRAINIAN ASSOCIATION CRYSTAL PEOPLE  
UKRAINIAN NATIONAL CHARITABLE FUND ZAPORUKA  
UKRAINIAN UNION OF PATIENTS' ORGANISATIONS

## UNITED KINGDOM OF GREAT BRITAIN AND NORTHERN IRELAND

ADVOCACY FOR NEUROACANTHOCYTOSIS PATIENTS  
AKU SOCIETY  
ALD LIFE  
ALSTROM SYNDROME EUROPE  
ALSTROM SYNDROME UK  
ANN EDGAR CHARITABLE TRUST  
ANNABELLE'S CHALLENGE  
ASSOCIATION FOR MULTIPLE ENDOCRINE NEOPLASIA DISORDERS  
ATAXIA UK

BATTEN DISEASE FAMILY ASSOCIATION  
BEHCETS SYNDROME SOCIETY  
BRITTLE BONE SOCIETY  
CANCER 52  
CAVERNOMA ALLIANCE UK  
CHILD GROWTH FOUNDATION  
CHILD LUNG FOUNDATION  
CHILDREN LIVING WITH INHERITED METABOLIC DISEASES  
CILIOPATHY ALLIANCE  
CONTACT A FAMILY  
CYSTINOSIS FOUNDATION OF THE UK  
DANCING EYE SYNDROME SUPPORT TRUST  
DEGOS DISEASE SUPPORT NETWORK  
ECTODERMAL DYSPLASIA SOCIETY  
EUROPEAN GAUCHER ALLIANCE  
EUROPEAN MYASTHENIA GRAVIS ASSOCIATION  
EUROPEAN TUBEROUS SCLEROSIS COMPLEX ASSOCIATION  
FABRY INTERNATIONAL NETWORK  
FETAL ANTI CONVULSANT SYNDROME ASSOCIATION  
FINDACURE: THE FUNDAEMENTAL DISEASES PARTNERSHIP  
FOP FRIENDS  
GAUCHERS ASSOCIATION UK  
GENETIC ALLIANCE UK  
INTERNATIONAL BRAIN TUMOUR ALLIANCE  
INTERNATIONAL NIEMANN-PICK DISEASE ALLIANCE  
IPOPI - INTERNATIONAL PATIENT ORGANIZATION FOR PRIMARY  
IMMUNODEFICIENCIES  
JOINING JACK  
LEBER'S HEREDITARY OPTIC NEUROPATHY SOCIETY  
LYMPHANGIOMATOSIS & GORHAM DISEASE ALLIANCE EUROPE  
MAX APPEAL  
MEBO RESEARCH  
MPS SOCIETY  
MYAWARE  
MYOTUBULAR TRUST  
NEWLIFE FOUNDATION FOR DISABLED CHILDREN  
NIEMANN-PICK DISEASE GROUP UK  
NORTHERN IRELAND RARE DISEASE PARTNERSHIP  
ONE IN A MILLION - PSEUDOMYXOMA SURVIVOR  
ORGANISATION FOR ANTI-CONVULSANT SYNDROME  
POLYCYSTIC KIDNEY DISEASE CHARITY (PKDC)  
PRADER WILLI SYNDROME ASSOCIATION UK  
PSC SUPPORT  
RARE DISEASES UK  
RING 20 RESEARCH AND SUPPORT UK CIO  
SPINAL MUSCULAR ATROPHY SUPPORT UK  
STIFF MAN SYNDROME SUPPORT GROUP AND CHARITY  
STURGE-WEBER UK  
SYNCOPE TRUST AND REFLEX ANOXIC SEIZURES  
THE AADC RESEARCH TRUST CHILDREN'S CHARITY  
THE CHROMOSOME 18 REGISTRY AND RESEARCH SOCIETY (EUROPE)  
THE CURE & ACTION FOR TAY-SACHS (CATS) FOUNDATION  
THE FRAGILE X SOCIETY  
THE PBC FOUNDATION (UK) LTD  
THE ULTRA RARE DISEASES, DISORDERS & DISABILITIES FOUNDATION  
TUBEROUS SCLEROSIS ASSOCIATION  
UK MASTOCYTOSIS SUPPORT GROUP  
UNIQUE - THE RARE CHROMOSOME DISORDER SUPPORT GROUP  
UNITED KINGDOM THALASSAEMIA SOCIETY  
UVEITIS INFORMATION GROUP  
VASCULITIS UK (THE VASCULITIS TRUST)

## UNITED STATES

ALSTROM SYNDROME INTERNATIONAL  
APS FOUNDATION OF AMERICA, INC (APSA)  
CHORDOMA FOUNDATION  
CURE PSP  
CYSTINOSIS FOUNDATION  
FMD CHAT  
INTERNATIONAL PEMPHIGUS & PEMPHIGOID FOUNDATION  
INTERNATIONAL WALDENSTROM'S MACROGLOBULINEMIA  
FOUNDATION  
NORD NATIONAL ORGANIZATION FOR RARE DISORDERS  
NTM INFO & RESEARCH  
THE CUSHING SUPPORT & RESEARCH FOUNDATION  
THE OXALOSIS & HYPEROXALURIA FOUNDATION

## URUGUAY

ASOCIACION ACONDROPLASIA URUGUAY



# CONFERENCES & Workshops 2016

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**‘Pharmacovigilance challenges: what’s new in 2016?’, Paris, France, 29 November**

**Mathieu Boudes:** “Patients’ organisation involvement in EU INITIATIVES and Q&A”

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**Rare Best Practices (a platform for sharing best practices for the management of rare diseases), Final conference, Rome, Italy, 24 November**

**Mathieu Boudes:** “Better patient health outcomes require the best of research and healthcare: a need for a common infrastructure”

Juliette Sénecat also represented EURORDIS

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**Annual Symposium of the Centre for Regulatory Science (CORS) within the Faculty of Health and Medical Sciences, University of Copenhagen, Denmark, 24 November**

**Terkel Andersen:** “The Empowered Patient in the Regulatory Science Framework”

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**European Parliament: Group discussion on “Improving Patients’ Access to Medicines: What role for the European Parliament?”, Strasbourg, France, 23 November**

**Yann Le Cam and Valentina Bottarelli** represented EURORDIS in the discussion on “Access to Medicines”

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**IX International Congress on Rare Diseases – “Congreso Internacional de Enfermedades Raras”, Murcia, Spain, 17-20 November**

**Raquel Castro:** INNOVCare project: improving the coordination between health, social and local services for rare diseases (“Proyecto INNOVCare: Mejorando la coordinación entre atención sanitaria, servicios sociales y recursos locales para Enfermedades Raras”)

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**B-debate: “Drug Repurposing for Rare Diseases”, Barcelona, Spain, 17-18 November**

**Virginie Hivert,** panellist in the session “Drug Repurposing for Rare Diseases: from research to patients”

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**“Shortages of Human Medicines in the European Union”, under the auspices of the Slovak Presidency of the Council of the European Union, Bratislava, Slovakia, 17-18 November**

**François Houÿez:** “Shortages of medicines: proposals from patients and healthcare Professionals”

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**World Orphan Drug Congress Europe, Brussels, Belgium 16-17 November**

**Mathieu Boudes,** Jean-Louis Roux and Ariane Weinman represented EURORDIS

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**4<sup>th</sup> International Conference on Rare and Undiagnosed Diseases, Tokyo, Japan, 16-17 November**

**Virginie Bros-Facer:** “International Joint Recommendations to address specific needs of Undiagnosed Rare Disease Patients”

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**XXI International Conference - The Pontifical Council for Health Care Workers: “Towards a Culture of Health that is Welcoming and Supportive at the Service of People with Rare and Neglected Pathologies”, Rome, Italy, 11 November**

**Simona Bellagambi:** “The Mission of EURORDIS at the Service of People living with Rare Diseases”

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**“9<sup>th</sup> European Public Health Conference” – pre-conference event: “Enhancing evidence-informed decision-making to support resilient and sustainable health systems in Europe, Vienna, Austria, Vienna, Austria, 9 November**

**Valentina Bottarelli:** “Policies and care models for rare disease patients – EURORDIS Operating Grant”

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**Ontario Health Association (OHA) conference, Ontario, Canada, 8 November**

**Matt Johnson** together with Accreditation Canada International: “Developing the European Reference Networks’ assessment scheme”



Seminar on Respite Care: interactive and participative day on various models of respite services – “Colloque - Journée interactive et participative : Variations sur le thème du répit”, Tain l’Hermitage, France, 4 November

**Raquel Castro:** Respite services for people living with a rare disease in Europe (“Services de répit pour les personnes atteintes de maladies rares en Europe”)

ISPOR 6<sup>th</sup> Patient Representatives Roundtable (PRR) during the ISPOR 19<sup>th</sup> Annual European Congress, Vienna, Austria, 29 October -2 November

**Chris Sotirelis** represented EURORDIS at the ISPOR 6<sup>th</sup> Patient Representatives Roundtable

Cambridge Rare Disease Network, Robinson College, Cambridge, United Kingdom, 25 October

**Avril Daly** represented EURORDIS.

Colloque Recherche en Sciences Humaines et Sociales dans les maladies rares, Fondation Maladies Rares, Paris, France, 20 October

**Sandra Courbier** represented EURORDIS.

RareX / ICORD (International Conference on Rare Diseases and Orphan Drugs), Cape Town, South Africa 19-22 October

**Raquel Castro:** “Overcoming unmet social and daily life needs of people living with a rare disease”

“Improving organ donation and transplant across the EU: A cross-condition campaign”, organised by PHA Europe, under the auspices of MEP Hilde Vautmans, MEP Ivan Jacovic and MEP Alojz Peterle - European Parliament in Brussels, Belgium, 18 October

**Valentina Bottarelli** represented EURORDIS.

European Society Gene and Cell Therapy (ESGCT) / International Society for Stem Cell Research (ISSCR) Congress: “Changing the Face of Modern Medicine: Stem Cells & Gene Therapy”, Florence, Italy, 17-21 October

**Chris Sotirelis** and **Michele Lipucci** represented EURORDIS.

Makers’s Fair for new inventors (within the European Biotechnology Week), Rome, Italy, 17-18 October

**Simona Bellagambi** represented EURORDIS to address the needs of patients living with rare diseases and their families.

UK NIHR Rare Diseases Translational Research Collaboration Patient Day, Cambridge, UK, 12 October

**Eva Bearryman** represented EURORDIS.

“La Recherche Clinique et le développement du médicament : Tous acteurs !” (clinical research and drug development: all actors), Institut Imagine, Paris, France, 11 October

**Sandra Courbier:** “Rare Barometer Voices: Quantitative Surveys on Rare Diseases in Europe”

**François Houyez:** “Partenariats Associations/Promoteurs/ Investigateurs : Quels atouts pour le malade? ”

Joint DIA/EFGCP/EMA Better Medicines for Children Conference 2016, EMA, London, UK, 10- 11 October

**Virginie Hivert:** “Lessons learnt from 10 years of Paediatric Regulation – the Patients’ perspective”

Escuela de formacion de FEDER/CREER, Burgos, Spain, 8-9 October

**Sandra Courbier:** “Rare Barometer Voices – A new advocacy tool”

« Quels enjeux pour un 3<sup>ème</sup> Plan national Maladies Rares? » (What are the key issues for a 3<sup>rd</sup> National Plan for Rare Diseases), Assemblée Nationale, Paris, France, 4 October

**Anne-Sophie Lapointe:** Panellist in the session on ‘how to improve patients’ access to treatments: the challenges in research, assessment and funding.’

**Ariane Weinman** attended as EURORDIS’ representative.

7<sup>th</sup> South Eastern European Cystic Fibrosis (CF) conference, Skopje, Macedonia, 29-30 September

**Vlasta Zmazek:** Presentation of EURORDIS, Rare Diseases in Europe

Etats Généraux de l’innovation: “Parcours de santé, patient - Parcours de vie, usager - Maladie rare, maladie chronique et vieillissement : Vers un nouveau modèle tout au long de la vie ? », organisé par DESSEIN Action Santé, Paris, France, 29 September

**Yann Le Cam:** keynote speaker

IMI - Innovative Medicines Initiative - Advanced Therapies workshop of the IMI Stakeholder Forum, Brussels, Belgium, 29 September

**Chris Sotirelis** represented EURORDIS

3<sup>rd</sup> Annual Patient Advocacy Summit, organised by MassBio, Cambridge, Massachusetts, USA, 28, September

**Claudia Hirawatt** participated as EURORDIS’ representative

“Safeguarding patient safety and quality of care in Europe: Good practice for the off-label use of medicines” hosted by MEP Piernicola Pedicini, European Parliament, Brussels, Belgium, 27 September

**François Houyez** represented EURORDIS

“Meeting with patient/ consumers” organisations on HTA (health technology assessment), organised by DG SANTE – European Commission, Brussels, Belgium, 27 September

**Valentina Bottarelli** represented EURORDIS

4<sup>th</sup> International Summer School: “Rare Disease & Orphan Drug Registries” – organised by the ISS (Istituto Superiore di Sanità), Rome, Italy, 26-28 September

**Simona Bellagambi** (with **Fernanda De Angelis**): Interactive session: “Patients’ needs & experiences: Needs, strengths and weaknesses, patients driven registry experience”

MPNE Ocular conference: “Fighting For Our Lives”, 24 September, Reading UK

**Chris Sotirelis:** “Overview of EURORDIS activities and issues of access to orphan medicines”

“Introduction to patient registries and RD Connect”

EUCelLEX Final International Conference “Engaging stakeholders for responsible stem cells research”, Paris, France, 22-23 September

**Virginie Bros-Facer**, panellist in the round table discussion “Creating a European ELSI (Ethical Legal and Social Issues) Task Force”

PEALS (Policy, Ethics and Life Sciences) International Symposium 2016 - Genomic Nations, University of Newcastle, UK, 20-21 September

**Virginie Bros-Facer** represented EURORDIS

Second Orphan Drug Day: “European legislation on orphan drugs: 15 years of success, what future? “, Rome, Italy, 20 September

**Simona Bellagambi:** “Before and after the advent of orphan medicinal products: 2<sup>nd</sup> generation patients”

#### 4<sup>th</sup> Nordic Conference on Rare Diseases, Copenhagen, Denmark, 19-20 September

**Terkel Andersen**, President of EURORDIS and **Anders Olauson**, Chairman of Ågrenska: "Rare Diseases and Disabilities in European and International Perspective"

#### Findacure Patient Registry Workshop, London, UK, 16 September

**Chris Sotirelis**: "Introduction to patient registries and RD Connect"

#### Europe Biobank Week, Vienna, Austria, 13-14 September

**Virginie Bros-Facer** and **Marieke van Meel** represented EURORDIS in the framework of RD-Connect project

**Evidence from the perspective of the EMA and from an HTA perspective: What evidence is needed and what are the differences between admission and benefit assessments are really necessary?** organised by ACHSE, the German National Alliance for Rare Diseases, Berlin, Germany, 13 September

**Jean-Louis Roux**: "A European perspective: how can we optimize evidentiary demands and cooperation between regulatory and HTA agencies in order to improve access to more and safer drugs within the European Union and beyond?"

#### Italian Parliament, Rome, Italy, 26 July

**Terkel Andersen**: presentation of EURORDIS' activities in the field of rare diseases at European and international levels

#### Access to Orphan Drugs & Other Rare Disease Therapies in Romania, Bucharest, Romania, 29 June

**Yann Le Cam**: "The EURORDIS Call on Payers to Improve Patient Access to Rare Disease Therapies"

#### Workshop "Maladies Rares: Les Opportunités Européennes" (Rare Diseases: European (research) opportunities in Europe), Bordeaux University, France

**Virginie Bros-Facer**: "La Recherche sur les Maladies Rares en Europe" (Research on Rare Diseases in Europe)

#### 2<sup>nd</sup> ESO Masterclass in Cancer Patient Advocacy: 'Working Towards Stronger and more Effective Advocacy in Europe', Milan, Italy, 24 - 26 June

**Yann Le Cam**: "Meeting the challenge of leading a patient organisation"

#### 21<sup>st</sup> Congress of the European Hematology Association, Capacity Building Training with Patients, Copenhagen, Denmark, 10 June

**Matt Johnson** and **Lenja Wiehe**: "Introduction on the 'European Patient Advocacy Groups – ePAGs' and their role in European Reference Networks"

#### EULAR Annual European Congress of Rheumatology 2016, London, United Kingdom 08 - 11 June

**Chris Sotirelis**, 9 June 2016: EURORDIS representative: "Rare disease patients? Perspective on improving access to orphan drugs and opportunities for better collaboration"

#### 2016 Annual Workshop of the European Network of Paediatric Research at the EMA (Enpr-EMA), London, United Kingdom, 2 June

**Kerry Leeson-Beevers** represented EURORDIS

**Thematic session of the "Collège des Économistes de la Santé" (Health Economists College): "Integrating the patients' voice in the health decision making process", Paris, France, 2 June**

**Anne-Sophie Lapointe**, speaker in the Round Table on pricing and reimbursement decision making processes

#### Personalised Medicine Conference 2016, Brussels, Belgium, 1-2 June

**Virginie Hivert**, panellist in "Drivers and challenges in getting personalised medicine to the market"

#### Seminar on medical-social care of people living with a rare disease - "Prise en charge médico-sociale des personnes atteintes de maladies rares", Paris, France, 19 May

**Raquel Castro**: "The challenge for people living with a rare disease: European perspective" ("Le challenge Européen pour les personnes atteintes de maladies rares")

#### Cystic Fibrosis workshop on fundraising, Leuven, Belgium, 18 - 19 May

**Yann Le Cam**: "European funding opportunities"

#### Masterclass: "Patient Involvement in the Regulatory Arena", Faculty of Pharmacy – University of Lisbon, Portugal, 13 May

**Yann Le Cam**: "Patients' Involvement in the Regulatory Arena"

#### H2020 European Health/Rare Diseases Brokerage Event– Oslo 2016, Norway, 12-13 May

**Anders Olauson**: "Rare diseases – Why do we need research from the patients' perspective?"

#### CML (Chronic Myeloid Leukemia) Horizons 2016: "Learn, Share, Grow". Ljubljana, Slovenia, 6-8 May

**Patrice Régnier**: "In kind contributions in Financial Reports"

#### RARE-Bestpractices General Assembly, Tenerife, Spain, 29 April

**Juliette Sénecat**: "Activities and Work Plan regarding Patient Involvement in the RARE-Bestpractices Project"

#### IMI Workshop: "Patient Engagement Strategy for Innovative Medicines", Brussels, Belgium, 27-28 April

**Yann Le Cam**: "Way forward – The views of patients"

#### World Orphan Drug Congress, Washington D.C., USA, 20-22 April

**Yann Le Cam**:

"Europe's focus on re-engineering the process rather than new legislations to get more, better, faster, cheaper orphan drugs"

"EURORDIS –European Organisation for Rare Diseases: improving the quality of life of people living with rare through the promotion of health policies and patient empowerment and training"

#### SWAN (Syndromes Without A Name) Europe discussion meeting, Rome, Italy, 16 April

**Virginie Bros-Facer**: "Undiagnosed Disease Network – International and Joint Position Paper"

**Marta Campabadal**: "Welcome to the online community for undiagnosed conditions".

**Simona Bellagambi** and **Anne-Sophie Lapointe** also represented EURORDIS.

#### 10<sup>th</sup> International Congress FOP (Fibrodysplasia Ossificans Progressiva), Livorno, Italy, 15-16 April

**Anja Helm**: "European Rare Disease Federations – EURORDIS' perspectives"



DIA (Drug Information Association) 28<sup>th</sup> Annual EuroMeeting: “Innovation - Do You Win by Being IN?”, Hamburg, Germany, 6-8 April

**Yann Le Cam**, session Chair: “Medicinal Products in Need”

**Virginie Hivert**: “The Patient’s Perspectives on OMPs and Significant Benefit”

**François Houÿez**, Panellist in the session: “Adaptive Pathways and Conditional Approval”

Audition au Comité National d’Ethique, Paris, France, 4 April

**Yann Le Cam** represented EURORDIS

National meeting of the French patient organisation for Angelman Syndrome, Paris, France, 2 April

**Lara Chappell** represented EURORDIS

Tertiary Prevention in Oncology, Paris, France, 24 March

**Ariane Weinman** represented EURORDIS

Geniris (aniridia and iris rare disorders) General Assembly Meeting, Paris, France, 19 March

**Lara Chappell**: Presentation of EURORDIS

Brains for Brain (B4B)– European Task Force on Brain and Neurodegenerative Lysosomal Storage Disorders  
10<sup>th</sup> B4B Workshop and InNerMeD 3<sup>rd</sup> Open Conference, Madrid, Spain, 16-19 March

**Matt Johnson**: “Toward the European Reference Networks: concept and vision”

“Fostering Responsible Research with CRISPR-Cas 9”, Paris, France, 16 March

**Virginie Bros-Facer** represented EURORDIS

RE(ACT) - International Congress on Research of Rare and Orphan Diseases, Barcelona, Spain, 9 - 12 March

**Yann Le Cam**: Keynote speaker at the public opening ceremony

RD-Connect Annual Meeting, Barcelona, Spain, 9 - 11 March  
Patient Advisory Council (PAC) meeting, 9 March

**Virginie Bros-Facer**, Session Chair

PAC members, participants in the meeting: **Joseph Irwin**, **Lydie Lemmonier**, **Marita Pohlschmidt**, **Daniel Renault**, **Françoise Rouvaut**, **Chris Sotirelis**, **Oliver Timmis**.

“International Experience Exchange for Patient Organisation”, Copenhagen, Denmark, 2-3 March

**Dimitrios Synodinos** represented EURORDIS

Findacure Scientific Conference, London, UK, 29 February

**Virginie Hivert** represented EURORDIS

IX International Rare Disease Day: “La Voce Del Paziente - Unitevi a noi per far sentire la voce delle malattie rare” (the voice of patients – Join us in making the voice of rare diseases heard), jointly organised by UNIAMO and CNMR National Centre for Rare Diseases at the ISS Italian National Institute of Health, Rome Italy, 29 February

**Simona Bellagambi**: “RareConnect – Online community”

Día mundial de las enfermedades raras “La voz del paciente” (International Rare Disease Day, “the voice of patients”), under the aegis of UNESCO Chair in Bioethics and Health Law, University of Murcia, Spain, 29 February

**Gema Chicano**: “Las enfermedades raras en la actualidad: un reto ético y jurídico” (rare diseases today: an ethical and legal challenge)

“Hospital Medicine”, Athens, Greece, 23 February

**Dimitrios Synodinos** represented EURORDIS

2<sup>nd</sup> Conference on Rare Diseases of the Salento region: “The voice of patients: From Salento region to Europe”, Lecce, Italy, 22 February

**Simona Bellagambi**: “The role of patients in Europe”

Third International Conference on Rare and Undiagnosed Diseases, Vienna, Austria, 18 February

**Virginie Bros-Facer**: “EURORDIS and Undiagnosed Patients”

Information day: Dialogue between young patients affected by rare cancers, their parents and healthcare professionals and representatives of medical institutions, Paris, France, 13 February

**David Oziel**: “How to find validated information on diseases on the internet”

Croatian National Conference on Rare Diseases and Symposium on Rare Diseases, Zagreb, 12-13 February

**Raquel Castro**: “Integration of rare diseases into social services and policies: identifying the challenges and becoming part of the solution”

International course for health care guidelines developers on treatments of rare diseases, Milan, Italy, 10-12 February

**Juliette Sénécat**: “Methods for Patient Involvement”

Journée des Centres de Référence Maladies Rares / Info Day for RD Centres of Reference, Ministry of Health, Paris, France, 9 February

**Yann Le Cam**: panellist in the Round Table on European Reference Networks

**Anne-Sophie Lapointe** and **Ariane Weinman** attended as EURORDIS’ representatives.

“Better research for better health: A holistic approach to challenges & opportunities”, European Commission, Brussels, Belgium, 21 January

**Valentina Bottarelli** and **Jean-Louix Roux** attended as EURORDIS’ representatives

# ACKNOWLEDGEMENTS

EURORDIS would like to thank the following organisations and companies for their financial support in 2016:

## Patient Organisations and Public Entities

### AFM - TÉLÉTHON

The "Association Française contre les Myopathies", for the annual core activities grant, the office space they make available to the organisation free of charge and their in-kind production of the Rare Disease Day 2017 video.



### EUROPEAN COMMISSION

DG Health and Food Safety

- The Operating Grant for year 2016
- RD-ACTION – Rare Diseases Joint Action – Data and policies for Rare Diseases
- JARC – Joint Action on Rare Cancers



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
- RARE-Bestpractices – A platform for sharing best practices for the management of rare diseases
- Genetics Clinic of the Future – A stepping stone approach towards the Genetics Clinic of the Future
- COST – European Cooperation in Science and Technology
- The Innovative Medicines Initiative-Joint Undertaking (IMI-JU) project:
  - ADAPT SMART - Accelerated Development of Appropriate Patient Therapies - a Sustainable, Multi-stakeholder Approach from Research to Treatment-outcomes
  - European Patients' Academy on Therapeutic Innovation (EUPATI)
  - Web-Radr - Recognising Adverse Drug Reactions



### 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



### VISITSCOTLAND

MARKETING EDINBURGH LTD

European Conference on Rare Diseases 2016





# Health Sector Corporates

Diversification of funding is a key success factor to minimise potential conflict of interest with donors. EURORDIS has diversified its pharmaceutical and biotechnology sector companies' sponsorship from 51 to 53 different companies in 2016. 6 health sector companies also contributed to the 2016 incomes. Companies have supported EURORDIS through the EURORDIS Round Table of Companies<sup>1</sup>, the European Conference on Rare Diseases Edinburgh 2016, the Multi-Stakeholder Symposium, the EURORDIS Black Pearl Evening<sup>2</sup>, as well as international initiatives such as Rare Barometer<sup>3</sup>, RareConnect™<sup>4</sup> and Rare Diseases International<sup>5</sup>. The breakdown of each company's donations by project is detailed on the EURORDIS website on the "Corporate revenue" tab of the "Financial Information" section.

## Top five donors

1

SHIRE



2

PFIZER



3

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4

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5

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

<sup>2</sup> <http://www.blackpearl.eurordis.org>

<sup>3</sup> <http://www.eurordis.org/voices>










<sup>4</sup> <http://www.rareconnect.org>

<sup>5</sup> <http://www.rarediseasesinternational.org>

## Other Pharmaceutical Companies & Health Sector Corporates



## In-kind contributors

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## Other contributors

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EUROPEAN FEDERATION OF PHARMACEUTICAL INDUSTRIES AND ASSOCIATIONS	 European Federation of Pharmaceutical Industries and Associations	 EVERYLIFE FOUNDATION FOR RARE DISEASES	EVERYLIFE FOUNDATION FOR RARE DISEASES
KINDNESS FOR KIDS	 KINDNESS FOR KIDS	Foundation for children with rare diseases Moving mountains for kids in need.	





**EURORDIS**  
RARE DISEASES EUROPE

# WORKPLAN

## 2017

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# ACTION PLAN

## 2017

## 1. Patient Advocacy

### 1.1 OUR ADVOCACY GOALS WITHIN OUR STRATEGY PRIORITIES FOR 2015-2020

- + 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
- + Promote cross-border rare disease expertise and knowledge generation and sharing to improve quality of care diagnostic, medical care & social care at local level
- + Promote access to cross-border healthcare and making possible patient mobility
- + Promoting research and bridging patient's perspective and researcher activities
- + Addressing the new issues of genetic testing, genetic counselling & new-born screening
- + Voicing /expressing patient preferences in sharing of health and genetic data in rare diseases information systems and repositories

### 1.2 OUR ADVOCACY ACTIONS IN 2017 TO REACH OUR GOALS

Supporting the implementation of the EU strategy on RDs adopted in 2008 with the "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:

#### 1.2.1 The European Commission Expert Group on Rare Diseases (CEG-RD) (2014-2017):

- + The Expert Group provides advice and expertise to the Commission in formulating and implementing the Union's activities in the field of rare diseases and foster exchanges of relevant experience, policies and practices between the Member States and the various parties involved.
- + Participation of four EURORDIS patient representatives and their four alternates, nominated by the EC, participate in CEG-RD meetings taking place twice a year, together with two observers from the EURORDIS public affairs team.
- + The eight patient representatives in CEG-RD form the EURORDIS Policy Action Group (PAG).

#### EURORDIS Policy Action Group (PAG):

- + Established in 2010 along with the creation of the EU Committee of Experts on Rare Diseases (EUCERD), renewed with the creation of the CEG-RD in 2013;
- + Supported by the EURORDIS Public Affairs staff;
- + Teams up with other volunteers and EURORDIS staff to provide the patients' view on issues dealt with by the CEG-RD, such as: National strategies and national plans; centres of expertise, European Reference Networks (ERNs); cross-border healthcare; codification and classification of rare diseases; guidelines on diagnostic 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;
- + Contributes the RD patients' perspective to the recommendations of CEG-RD;
- + Participates in the European Joint Action on Rare

Diseases – RD-ACTION – through contributing to the Work Package “Rare Disease Policy”.

- + In 2017, the mandate of the CEG-RD will be renewed for another three-year period. EURORDIS will select candidate patient representatives to serve in the Committee through an open call addressed to all its membership and will prepare the application for the selected candidates. Following the nomination by the EC of the patient representatives for the new mandate, EURORDIS will renew the membership of its PAG and define our priorities for next three years

### 1.2.2 The European Commission Expert Group on Cancer Control (CEG-CC) (2015-2018):

- + The CEG-CC assists the Commission in the preparation of legislative proposals and policy initiatives in the field of cancer. One EURORDIS patient representative and his alternate, nominated by the EC, are members of the CEG-CC and participate to its meetings to voice the needs and expectations from patients with rare cancers. The EURORDIS patient representative is also a nominated member of CEG-RD to make the link between the two Expert Groups.
- + The two EURORDIS representatives (full member and alternate) are members of the EURORDIS Policy Action Group on Rare Cancers (PAG-RC).

### EURORDIS Policy Action Group on Rare Cancers (PAG-RC):

- + Established in July 2014 following the EURORDIS call for expression of interest addressed to its members, it is made up of six patient representatives nominated by the Board, covering rare cancers in adults, both solid tumors and hematological malignancies, as well as pediatric cancers;
- + Supported by the EURORDIS Public Affairs staff;
- + Follows up the work carried out by the CEG-CC to provide the patients' views in the field of RC and contributes to the making of relevant policies;
- + Produced the EURORDIS Table mapping out similarities and differences between rare diseases and rare cancers;
- + Contributes to other initiative and projects, such as the EU Joint Action on Rare Cancers (JARC).

### 1.2.3 2<sup>nd</sup> European Union Joint Action on Rare Diseases – RD-ACTION (2015-2018): Promoting the Implementation of Recommendations on Policy, Information and harmonised coding system for Rare Diseases

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 to disseminate EU policy outcomes and new strategies, which included the organisation of the European Conference on Rare Diseases and Orphan Medicinal products (ECRD, 27-28 May 2016, Edinburgh).

- + Continue to coordinate the organisation of up to 25 multi-stakeholder national workshops, organised locally by the RD National Alliances, with a view to foster the implementation of national plans for rare disease and the integration of EU policies at national level.

- + Develop common material for the workshops to integrate national policy and EU policy, showing how patients in Member states benefit from European action through:

- Shared resources relevant EU policy documents and EURORDIS guidance material, organised by topic area that are chosen for the national workshops;
- Specific webinars to provide information and advocacy tools, and to build capacities of national patient advocates in specific topic areas that are chosen for the national workshops (ERNs, access to therapies, research, social policy, etc.) or other emerging policy priorities that are developed at EU level and need to be integrated into national health policy systems

- + Promote exchanges of information, experience, good practices and concerns between National Alliances, the EURORDIS Advisors and the staff through means such as a national plan mailing list and the eNews.
- + Support National Alliances to advocate for the development of national healthcare networks, care pathways and increase access to highly specialised healthcare. Connecting endorsed healthcare provider members, national hub and affiliated partners and their pathways to European Reference Networks, anchoring ERNs into national healthcare systems.

### Work Package 6: Rare Disease Policy:

- + In this WP led by Newcastle University, EURORDIS is the main partner to propose policy priorities to the consultative group of Member states and to implement the activities participating to the Consultative Group of RD Action, selecting policy priority areas.
- + Actively contribute to the development of specific policies by preparing and participating to RD-ACTION workshops, and contributing to policy papers, on: creating interoperability across ERNs, virtual healthcare, data sharing and registries in ERNs; knowledge generation in ERNs, support to research and therapy development within ERNs; prevention in the field of rare diseases; public health indicators on rare diseases; information in the field of rare diseases; and other policy areas to be yet determined by the Consultative Group.
- + In addition, EURORDIS coordinates the contribution of National Alliances to the national sections of the State-of-the-Art report on rare diseases, as a way to secure patient involvement in the regular monitoring of the implementation of national RD policies, Strategies and Plans. In EU each country, a tripartite team with Member States' representatives on CEG-RD, Orphanet national coordinators and RD National Alliances contribute to the annual update of the State of the Art report.

### 1.2.4 European Union Joint Action on Rare Cancers (JARC) (2016-2019): Promoting EU Rare Cancer policy

EURORDIS is involved in five Work Packages out of 10 to contribute to the objectives of the JARC:

- + Prioritise rare cancers (RCs) in the agenda of the EU and Member States;
- + Develop a position on prevalence and incidence of rare cancers which may help refine future national strategy on cancers or on rare diseases as well as orphan product status



- + Develop innovative and shared solutions, mainly to be implemented through the future European Reference Networks in the field of Rare Cancers, in the areas of quality of care, research, education and state of the art definition on prevention, diagnosis and treatment of rare cancers.

#### 1.2.5 Advocate to improve the regulatory process for orphan medicinal products

- + Follow up on 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, by looking at the implementation and impact of the new Notice which will come into force beginning 2017.
- + Follow-up on the EURORDIS answer to the public consultation on the Concept of 'similar medicinal product' in the context of the orphan legislation
- + Develop EURORDIS position for the public consultation on the report of the 10 years of the Paediatric Regulation
- + Create links with ERNs to inform COMP decisions (e.g. use of hospital pharmacy preparations)
- + Contribute to the review of current initiatives in the regulatory field as undertaken by the Safe and Timely Access to Medicines for Patients (STAMP)

#### 1.2.6 Advocate to improve the patient access to rare disease therapies and promote a new business model sustainable for society

- + Contribute to new approaches for the engagement of patients in the benefit/risk evaluation, taking into consideration patient preferences by participating in the IMI2 project PREFER as a member of the Stakeholder Advisory Board and collaborating with EMA (responding to the IMI2 call on Patient Preferences Elicitation and collaborating with EMA)
- + Continue advocating in diverse relevant fora for the establishment of mechanisms aiming to ensure that people living with rare diseases have access to more, better, cheaper treatments that reach the patient faster, by contributing to the establishment of an economic model that is sustainable and is conducive to the development of therapies that are affordable and at a fair price.
- + Contribute to the European Parliament debate on the Own Initiative Report on EU Options on Improving Access to Medicines by bringing forward the specific issues and concerns of rare disease patients in having access to therapies and make sure that they are integrated into the EP Report.
- + Organise the **2<sup>nd</sup> EURORDIS Multi-Stakeholder Symposium** to debate with stakeholders the content of the EURORDIS "possibility paper" and start the collective negotiation
- + Contribute to the ongoing review of current and future initiatives in the regulatory field as undertaken by the **Safe and Timely Access to Medicines for Patients (STAMP)**
- + Support dialogue on **Medicine Adaptive Development Pathways (MAPPs)** with EFPIA and EMA and contribute actively through IMI ADAPT-SMART project, and help to more clearly communicate the merits of this proposed approach (as well as clear examples of where Adaptive Pathways may provide tangible benefits to patients)

- + 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.

- + Promote the organisation of multi-stakeholder meetings on specific medical areas under the auspice of EMA

- + 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 and advance the concept of a "European table for price negotiation" with National Competent Authorities for Pricing and Reimbursement, and continue to offer ad hoc support to partners of the Benelux-Austria joint collaboration platform (with a view to its gradual enlargement to other EU Member States)

- + Contribute to reflections on Joint Negotiation and Joint Purchasing, Managed Entry Agreement, Transactional Pricing, Differential Pricing, Parallel Trade, Discount for Uncertainties,

- + Promote where appropriate such innovative approaches in Member States and national plans on rare diseases and increase direct cooperation with leaders from the rare disease therapies sector.

- + **Shortages of medicines:** Advocate for the creation of a European forum to discuss in response to the European Council initiative in the context of the Presidency by Slovak Republic: Contribute to the European debate on off-label use of medicines, by contributing to the European Commission report, by developing a position on the subject based on the survey "Treatment Information on the Medicines You Take". Create a database on off-label use of medicines in rare diseases.

- + Develop a EURORDIS "possibility paper" on options to improve patients' access to orphan medicines - with proposals for a new and more virtuous access model - in collaboration with external stakeholders of reference, covering R&D new model, collaborative registries, supportive regulatory processes, formative HTA, fair pricing and affordability.

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

- + Contribute to the EMA policy on access to clinical trials data. After 5 years of actions, since October 2016, all clinical reports from clinical trials part of marketing authorisation applications are now public for the priority 2017 is to define how individual patient data could be released without compromising the anonymity of patients.

- + Adopt a European position on compassionate access programmes and advocate to improve national schemes for compassionate use
- + 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 Joint Action 3)

#### 1.2.8 Advocate to improve Access to Care for rare disease patients

- + Promote and raise awareness on the findings of the **EURORDIS Access Campaign** on difficulties in accessing treatments in the context of the economic and financial crisis.
- + Identify advocacy actions targeted specifically to Member States' governments and include in the national debate on the National Plan's and Strategies' implementation (notably, RD-ACTION National Workshops)
- + Consider developing recommendations based on the results of the Access Campaign survey to be available on EURORDIS website and widely disseminated to relevant stakeholders
- + Continue to convey our views and positions in all appropriate forums and conferences in Europe and beyond
- + In emergency situations, take action to support patients' access to therapies

#### 1.2.9 Advocate to improve access to medical devices for patients with rare diseases

- + Analyse the impact of the newly adopted European legislation on medical devices and monitor its implementation
- + Promote the importance of involving patients in the evaluation process by notifying bodies
- + Explore avenues of collaboration and dialogue with the medical device industry with a view to promote better inclusion of patients and patient preferences into medical devices development, technology assessment and accessibility

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

- + Continue the promotion of rare diseases as a research priority in Horizon 2020, Work Programme 2018-2020 and in the Innovative Medicines Initiatives
- + Continue supporting rare diseases as a public health priority in the 3<sup>rd</sup> EU Public Health Programme 'Health for Growth' in view of the last Annual Work Programmes and advocate on the rare disease community's priorities
- + Promote the concept of European Rare Disease Clinical Research Network, embedded in ERNs and in EU Research Infrastructures and develop recommendations through IRDiRC
- + Promote and take active part in the development of the European Joint Programme on Rare Diseases for integration and long-term support of rare disease research infrastructures supportive of European healthcare networks and clinical research

#### 1.2.11 Advocate for progress in Patient's Rights to Cross-border Healthcare:

- + Continue monitoring the implementation of the Cross Border Healthcare Directive, identify and take actions in support of the implementation of its multiple strands:
- + Collect patient feedback, monitoring the implementation, disseminating information and promoting access to National Contact Points.
- + Support the European Patient Forum, In coordination with its other members, in the close monitoring of the implementation of patient's rights' across the EU and at national level;
- + Develop an action plan to improve integration of National Contact Points (NCP) with European Rare Disease Helplines and other relevant source of information on rare diseases (notably Orphanet) to create better knowledge of and responsiveness to specificities of rare disease patients; in particular the recognition by NCP of patients referral by experts from HCPs in ERN, the use of European Best Practice of Diagnosis and Care as valid basket of care by NCP and the access to gene or cell therapies approved in EU in a limited number of hospital centres requiring patient mobility.

#### 1.2.12 Advocate for the development of the ERNs form and functions and the integration of ERNs with wider health, social and research infrastructures.

Actively cooperate with EURORDIS' partners under the Rare Disease Joint Action (RD-ACTION) to develop and shape emerging policy areas for ERNs:

- + Develop and promote the long-term EURORDIS strategic vision on ERN
- + Develop and disseminate the vision and the case studies that articulate the potential of ERNs in order to drive a common approach across ERNs
- + Work with EURORDIS' partners under the Rare Disease Joint Action (RD-ACTION) to develop and shape emerging policy areas for ERNs, including for eHealth, IT, data sharing, clinical guideline development, clinical outcome and research, interaction with European health assessment bodies (EMA, EUnetHTA, MOCA)
- + Support the development and monitor implementation of plans of successful ERNs and support new or re-submitted ERNs applications undergoing a technical assessment
- + Support the development of the definition and representation of Affiliated Partners in MS who are not members of an ERN, ensuring balanced network member representation
- + Promote the adoption of ERN evidence-base practice, guidelines etc. in local treating hospitals and drive quality outcomes at local level
- + Support the phased expansion of scope of rare diseases covered in successful ERNs, making steps to ensure all rare diseases have a home under one ERN
- + Improve understanding of RDs by working with ERNs to map needs of RD population through their networks
- + Develop a "knowledge generation" approach in ERNs, through sharing the vision and outcome-driven best practice and emerging innovation. Build consensus on this approach across ERNs through RD-ACTION workshops

- + Advocate for EC seed funding to all ERNs beyond the 10 to be supported from 2017 in support of minimum coordination, and advocate for access to structural fund, digital funds (CEF) and research funds, with ERN status as a restricted advantage
- + Explore sustainable funding models for ERNs
- + Support and foster inter-ERN collaboration and the integration to support interoperability of eHealth, IT Platform, quality of data and registries and standardised outcomes
- + Align core research infrastructure with ERNs governance – e.g.: ERNs research working groups linked to EU projects and their thinking/methodologies
- + Build interoperability and inter-ERN collaboration for the care and management of multi-system rare diseases that sit within multiple ERNs.
- + Connect healthcare and research under ERNs and research networks or research infrastructure across the translational research pathway, including ERCT with RC ERNs and undiagnosed disease networks with RD ERNs.
- + Improve quality of databases, registries to enable therapies and drug development for conditions without a treatment and research into undiagnosed populations
- + Achieve common codification and quality of data collected and shared for research, therapy development and faster diagnosis
- + Advocate for adequate support of the Rare Disease European Reference Networks and consistent infrastructures for data collection and research
- + Explore best avenues to connect ERNs to research infrastructures and promote these connections, in the framework of existing and planned European initiatives such as the European Joint Programme
- + Promote systematic patient registration based the JRC EU Platform for RD Patient Registries

#### **1.2.13 Advocate for Health Technology Assessment methods and a European HTA Agency which addresses rare disease patient needs**

- + **Contribute to the implementation of the “Strategy for EU cooperation on Health Technology Assessment”**
- + Contribute to the European Commission consultation on the Inception Impact Assessment and adopt a position on the future of HTA cooperation in Europe. Encourage each EURORDIS member to respond to the consultation and coordinate the process.
- + Prepare for a key legislative proposal in 2018 and in case of adoption of a position in favour of the creation of a European HTA agency, organise patient advocacy in towards this aim
- + Participate to the HTA Network Discussion paper on how to “facilitate appropriate involvement of all interested stakeholders in the European collaboration in HTA, notably patients, health professionals, healthcare industry, and payers”
- + Propose arrangements necessary to continue the EU cooperation on HTA after 2020
- + Contribute to the Reflection paper on efficient joint cooperation on HTA of Medical Technologies and their reuse of assessments at national/regional levels
- + Participate in the evaluation of the impact of EU

cooperation on HTA (including awareness on HTA methods and conclusions, improved transparency, decreased complexity and costs for developers)

- + Share experiences of approaches to situations where evidence is limited and/or are major uncertainties
- + Exchange information with the relevant EU Fora (the Network of Competent Authorities in Pricing and Reimbursement (CAPR), the Commission Expert Group on Safe and Timely Access to Medicines for Patients (STAMP) and others

#### **1.2.14 Advocate in support of rare disease research:**

- + Engage EURORDIS in European research infrastructures and research, policy debates
- + Support the establishment of the Joint Research Centre (JRC) European Platform of Rare Disease Patient Registries, through direct contribution to the JRC Advisory Technical Group and participation to the interoperability workshops
- + Promote research objectives in Executive and Scientific Committees and Task Forces of IRDiRC
- + Elaborate and promote the objective of Rare Disease Clinical Research Networks, articulated with ERNs
- + Define the patient position through the RD-Connect project and the Rare Barometer programme, including patient preferences on data protection in light of the new EU Data Protection Regulation
- + Consolidate advocacy actions to integrate rare disease priorities in the Horizon 2020 Work Programme 2018-2020.
- + Promote and support advocacy action at European and National levels towards the inclusion of the European Joint Programme on Rare Diseases into the Work Programme 2018-2020 as an indispensable and transformative tool to integrate eRare research policy and funding, IRDiRC Scientific Support and research policy, use of research infrastructures and data platform by Rare Disease European Reference Networks, research activities of Orphanet, research and healthcare patient training and engagement activities of EURORDIS and trainings for researchers and health care providers

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

- + Discuss with pharmaceutical companies, biotechs involved in advanced therapies, as well as other stakeholders: patient representatives, EMA, HTA bodies, MoCA payers and NCPs
- + Compose and launch a group of interested parties to identify specific issues and elaborate solutions on:
  - The issue of uncertainties and patient preferences at the time of B/R Assessment
  - The specific issues in value demonstration
  - The specific issues in the delivery models and patient access to few centres in Europe, diagnosis across Europe, preparation and follow up of treatments across Europe, very long term data collection
  - The specific issues in the business model and patient access, fair pricing of advanced therapies, agreement



of price in countries where treatment is not delivered, role of NCP, payment on outcome, payment at treatment time or over years of life gained, differential pricing...

### 1.2.16 Advocate to improve access to and quality of rare disease diagnosis:

- + Contribute to the work carried out within RD -ACTION on genetic testing, counselling, genetic screening, newborn screening and next generation sequencing
- + Monitor developments and participate to relevant policy events and platforms in the field of companion diagnostics and precision medicine, with a view to define if appropriate a EURORDIS position
- + Monitor the implementation of the in vitro diagnostics newly adopted legislation and analyse relevance and impact for rare disease patients.
- + Representing the Rare Disease Patient Community into the H2020 proposal application to the call SC1-PM-03-2017: Diagnostic characterisation of rare diseases
- + Disseminate the International Joint Recommendation to address specific needs of undiagnosed and rare disease patients (EURORDIS' initiative) and develop advocacy actions specifically for undiagnosed patients
- + Contribute to the development of SWAN Europe - a coalition of groups, organisations and support networks working with families and/or patients affected by syndromes without a name and/or undiagnosed conditions
- + Contribute to the development of a Patient Advisory Council within Undiagnosed Diseases Network International

### 1.2.17 Voicing the social needs of people with a rare disease and advocating for the integration of rare diseases into social policy

- + Elaborate EURORDIS' position paper on social challenges;
- + Through the RD-Action: background paper on integrating rare diseases into social services and workshop focused on the role of health services in supporting the integration of rare diseases into social services;
- + Support National Alliances to promote the integration of rare diseases into social policies and services at national level via capacity building workshop at EMM 2016 Edinburgh;
- + Identify social policy and disability emerging topics and areas of work at European level;
- + Partner with European organisations active in the social and disability fields i.e. European Disability Forum, European Association of Service Providers for Persons with Disabilities, Eurocarers;
- + Ensure the representation of people living with a rare disease and their families in relevant policy documents at European level;
- + Engage with Members of the European Parliament active in social policy, including in disability and employment.

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

- + Support to **RDI** as the global alliance of rare disease patients across all rare diseases and all nationalities to speak with one voice on behalf of 350 million patients and their families
- + Organisation of an RDI Rare Disease Day Policy Event in Geneva to engage with the World Health Organization, the United Nations Secretary-General's High-Level Panel on Access to Medicines and the Office of the High Commissioner for Human Rights
- + Setting up of a Working Group on Rare Disease Research to develop RDI's position on research, guide advocacy and awareness activities to support rare disease research, define the research priorities of the international patient community and support appointed RDI patient representatives in the International Rare Disease Research Consortium.
- + Expand the EUCERD Joint Action Report on the State of the Art of Rare Disease Activities in Europe beyond Europe in 5 pilot countries: Canada, Australia, China, Russia and Argentina.
- + Setting up of an Advocacy Standing Committee to recommend positions for adoption or endorsement by the Council, further promote the Joint Declaration 'Rare Diseases: an International Public Health Challenge'
- + Appointing RDI representatives to the NGO Committee for Rare Diseases, United Nations, New York
- + Setting up of a UN System Strategy Working Group to advise on UN -related matters and guide the work of the RDI representatives appointed.
- + Support the **UN NGO Committee for Rare Diseases** to consolidate rare diseases in the UN Sustainable Development Goals 2030 (SDGs), the inclusion of rare diseases in the UN Development, Disability, Women & Youth Programmes, the Right to Health, the Access to innovative medicines and essential medicines, the work programme of the WHO and start introducing rare diseases in UNICEF and UNESCO programmes.

## 1.3 ADVOCATE RARE DISEASES AS A PRIORITY IN THE NEXT DECADE 2020-2030

- + Advocate toward a **EU Council Recommendation for a EU integrated national strategy on rare diseases** before 2020 during mandate of CEG-RD 2017-2020, to inform policy scenarios, before the new EU Multiannual Financial Framework 2020-2025 and related Programmes
- + Advocate toward a **Resolution of the UN General Assembly on Rare Diseases** through the UN NGO Committee for Rare Diseases and a core group of UN Member States in collaboration with UN Health Diplomacy group, RDI and all stakeholders
- + Create a **European Parliament Interest Group on Rare Diseases** and initiate a **Rare Disease Network of Members of national and regional parliaments**, in collaboration with National Alliances
- + **Foresight Study 2030**: Continue the advocacy to secure funding (2018-2020) for the Foresight Study Rare Diseases 2030 to identify long-term policy scenarios with participatory methodologies:

- Continue cooperation with MEPs that supported the presentation of a Pilot Project at the European Parliament as well as relevant European Commission services in order to secure financial and political support.
- Present and promote the Foresight Study to the Commission Expert Group on Rare Diseases to present the project to Member States representatives and promote its relevance for the long-term RD policy-making at EU and national level.
- + Seek complementary funding and identify potential scientific partners.

## 1.4 EURORDIS RARE BAROMETER PROGRAMME: GENERATING NEW DATA FROM PATIENT EXPERIENCE

**Gathering patient experience and perspective for evidence-based advocacy:**

**EURORDIS Rare Barometer programme** is a patient derived knowledge survey programme launched in 2015

In 2017, the following activities are planned:

- + Disseminate results of qualitative surveys, based on focus groups and Delphi-method, carried out in 2016 on use and sharing of genomic data and health related data, through scientific publications, and communication tools with infographics for patient advocates
- + Perform in 2017 a quantitative survey on use and sharing of genomic and health-related data in order to complete qualitative work done in 2016, and disseminate results through scientific publications and patient advocate communication tools
- + Disseminate the results of the quantitative survey performed on the impact of rare diseases on daily life through scientific publications and patient advocate communication tools
- + Develop the survey contact database Rare Barometer Voices (panel of people who answer on a regular basis to EURORDIS Rare Barometer surveys) by increasing the number of participants by 8000 participants so to have representativeness per country and per thematic grouping of rare diseases based on ERN groupings.
- + Diversify Rare Barometer Voices sources of recruitment (centres of expertise, resource centres...) and increase patient organisations participation in the Programme
- + Reinforce the Programme's governance, by bringing to life the activities of the Advisory Committee, fulfilling its membership goals and establishing a process to involve members in the ongoing work
- + Develop the scientific network around the Programme
- + Watch important topics for EURORDIS members and the rare disease community at large and explore possibilities for carrying out surveys on these issues to facilitate and streamline the inclusion of patient perspectives into EURORDIS advocacy work
- + Carry out 2 additional quantitative surveys through 2017 on EURORDIS advocacy priorities
- + Develop the "Question of the month": a survey question asked to the Rare Barometer Voices participants each month and disseminated on social media
- + Implement RareConnect survey template within RareConnect communities

# 2. Patient Empowerment

## 2.1 BUILDING THE COMMUNITY & NETWORKING

### 2.1.1 Membership

- + Maintain the EURORDIS Membership over 724 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

**2.1.1.1 ERN Content - EURORDIS will support the development of the ERNs form and functions e.g.: what ERN will do in terms of governance, strategies, action plans, membership and clinical services, through RD-ACTION work package 6 and through ePAGs.**

- + Developing and promote the long-term EURORDIS strategic vision on ERN
- + Develop and disseminate the vision and the case studies that articulate the potential of ERNs in order to drive a common approach across ERNs
- + Work with EURORDIS' partners under the Rare Disease Joint Action (RD-ACTION) to develop and shape emerging policy areas for ERNs, including for eHealth,

IT, data sharing, clinical guideline development, clinical outcome and research, interaction with European health assessment bodies (EMA, EUnetHTA, MOCA)

- + Support the development and monitor implementation of plans of successful ERNs and support new or re-submitted ERNs applications undergoing a technical assessment
- + Support the development of the definition and representation of Affiliated Partners in MS who are not members of an ERN, ensuring balanced network member representation
- + Promote the adoption of ERN evidence-base practice, guidelines etc. in local treating hospitals and drive quality outcomes at local level
- + Support the phased expansion of scope of rare diseases covered in successful ERNs, making steps to ensure all rare diseases have a home under one ERN
- + Improve understanding of RDs by working with ERNs to map needs of RD population through their networks
- + Develop a "knowledge generation" approach in ERNs, through sharing the vision and case studies of outcome-driven best practice and emerging innovation. Build consensus on this approach across ERNs through RD-ACTION workshops

### 2.1.1.2 ERN Integration - EURORDIS will, through RD-ACTION, support and foster inter-ERN collaboration and the integration of ERNs with wider health, social and research infrastructures.

- + Supporting interoperability of eHealth, IT Platform, quality of data and registries and standardised outcomes
- + Aligning core research infrastructure with ERNs governances – e.g.: ERNs research working groups linked to EU projects and their thinking/methodologies
- + Build interoperability and inter-ERN collaboration for the care and management of multi-system rare diseases that sit within multiple ERNs.
- + Connecting healthcare and research under ERNs and research networks or research infrastructure across the translational research pathway, including ERCT with RC ERNs and undiagnosed disease networks with RD ERNs.
- + Improve quality of databases, registries to enable therapies and drug development for conditions without a treatment and research into undiagnosed populations
- + Achieve common codification and quality of data collected and shared for research, therapy development and faster diagnosis

### 2.1.2 Outreach

#### **Outreach to patient groups in Central and Eastern Europe, Balkans, Russia and Caucasus, 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
- + 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 2017 Budapest and to the EURORDIS Summer School 2017 Barcelona

### 2.1.3 CNA

#### **Empower 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 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

### 2.1.4 CEF

#### **Build capacities of the European network of 56 disease-specific European / International Federations & Networks through:**

- + Sharing information, experience, good practices and guidance on CEF web section and RareTogether!
- + Organise common activities 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
- + Expanding the **EURORDIS Programme to Support European Federations & Networks** with money for their governance meetings, membership meetings, first European conferences on their disease

### 2.1.5 ePAG & ERNs

#### **European Patient Advocacy Groups (ePAG)**

- + Support European Patient Advocacy Groups (ePAG) actions to enhance 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
- + Support ePAG representatives interact with clinical steering committees of ERNs and define priority actions for ePAGs in 2017
- + Further develop the roles and functions of ePAGs within the context of the ERN governance structure
- + Support ERN steering committees and ePAGs to develop and conduct patient experience surveys
- + Organise a workshop at the EMM 2017 Budapest dedicated to ERN and ePAGs
- + Further develop and pilot the ePAG Leadership Programme to build the capacity of ePAG representatives to represent the patient voice within the ERN governance structures
- + Organise face-to-face workshops for ePAG Representatives at the EMM 2017 Budapest dedicated to building their leadership skill
- + Further develop the Patient Matchmaker Initiative to match European patient organisations with ERNs according to thematic groupings and across different ERNs, create a virtual network of patient organisations for each ERN, covering a wide range of rare diseases and all Member States, and map and connect patient organisations across the EU to ensure an equitable voice of patient community within ERNs
- + Build ePAG online communities in terms of members and content using Facebook Workspace communities
- + Support ERN steering committees to develop and conduct patient experience surveys



## European Reference Networks (ERNs)

### Build the capacities of the rare disease patient groups to engage meaningfully in European Reference Networks

- + Support the development of European 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 and care, information to patients and outreach to patients
- + Support the ePAGs and National Alliances to help expand the ERN in 2017, by stimulating Health Care Providers (HCP) application as Full Members or Affiliated Members so to progressively complete geographic coverage and disease coverage
- + Organise a series of workshops at the EURORDIS Membership Meeting 2017 in Budapest dedicated to ERNs and their Network Members (HCP & Affiliated Partners)
  - Develop EURORDIS website section and produce updated fact sheet on ERN
- + Provide support and advice to Member States on the ERN Assessment Framework and, in particular, on how to best use this framework at national level, in order to ensure that ultimately all Member States establish their own national accreditation process for both HCP as Full Members and Affiliated Partners

## 2.1.6 RDI

### Rare Diseases International (RDI): the global patient voice

RDI is the Global Alliance of rare disease patients across all rare diseases and all nationalities. Activities planned for 2017 include:

- + Further recruitment of members and promotion of the initiative
- + Organisation of an **RDI Rare Disease Day Policy Event** in Geneva to engage with the World Health Organization, the United Nations Secretary-General's High-Level Panel on Access to Medicines and the Office of the High Commissioner for Human Rights
- + Setting up of a **Working Group on Rare Disease Research** to develop RDI's position on research, guide advocacy and awareness activities to support rare disease research, define the research priorities of the international patient community and support **appointed RDI patient representatives in the International Rare Disease Research Consortium**.
- + Organisation of an **RDI Annual Membership Meeting** back to back to EURORDIS Summer School in Barcelona in June
- + RDI Fellows to attend **Expert Patient and Researcher EURORDIS Summer School (ExPRESS) in English and Spanish** edition (first, organised with FEDER and Catalan RD Platform) with additional parallel session for non-European patient groups.
- + Expand the EUCERD Joint Action **Report on the State of the Art of Rare Disease Activities** in Europe beyond Europe in 5 pilot countries: Canada, Australia, China, Russia and Argentina.
- + Involvement in **ICORD conference, Beijing, China;**

participation in Program Committee and liaison with Chinese Organisation for Rare Disorders (CORD)

- + Promotion of RDI and strengthening of collaboration with ALIBER at the **5<sup>th</sup> Iberoamerican Rare Disease Patients Meeting** in Brazil, second half 2017
- + Setting up of an **Advocacy Standing Committee** to recommend positions for adoption or endorsement by the Council, further promote the Joint Declaration '**Rare Diseases: an International Public Health Challenge**'
- + Setting up of a **UN System Strategy Working Group** to guide the work of the **RDI representatives appointed to the NGO Committee for Rare Diseases**, United Nations, New York

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

- + **Rare Diseases International (RDI)**, a network of rare disease patient organisations - organisation of regular RDI meetings and touch points throughout 2017, and further expansion of the RDI membership in connection with all other international efforts to date (UN NGO Committee, ICORD, etc).
- + The promotion of the joint declaration '**Rare Diseases: an International Public Health Challenge**' by major rare disease patient organisations
- + Continue growing and developing the **UN NGO Committee for Rare Diseases**, with approximately two meetings in the course of 2017. Our actions next year will chiefly consist in consolidating the structure and membership of the Committee, but also to give it a more robust work programme and methodology for the years to come, including working towards a Resolution on Rare Diseases at the UN General Assembly
- + Signing of MoUs with international patient organisations
- + The international development of Rare Disease Day
- + Continued active participation in the International Rare Disease Research Consortium

## 2.1.7 RareConnect

### Play central role in the evolution of the RareConnect project by:

- + Implementing transfer of RareConnect to SickKids Hospital, Canada
- + Assisting in the development of new governance structure in which EURORDIS will participate. EURORDIS will represent the patient voice within the new RareConnect structure
- + Supply RareConnect with experienced online community managers
- + Implicate RareConnect in relevant core EURORDIS activities as appropriate, such Rare Barometer

## 2.1.8 Volunteers

### Support EURORDIS volunteers' involvement through:

Volunteers, participating in NGO partnerships, in European projects and representing patients in the European Commission and EMA working groups and committees

- + Encourage the participation in training programmes and meetings such as the EURORDIS Summer School, webinars, EURORDIS Membership Meetings and European Conferences on Rare Diseases and Orphan Medicinal Products (ECRDs)[AW1]

- + Brief volunteers on key topics, access to shared reference documents and public presentations
- + Create specific working groups and task forces involving volunteers by their areas of expertise such as for instance: Orphan drugs, advanced Therapies, paediatric drugs, access to treatments and information to patients, best clinical practice guidelines, registries and databases, social services for rare diseases, European and national policy on rare diseases and/or on rare cancers
- + Maintain and expand the volunteer section on the EURORDIS website

## 2.2 BUILDING THE CAPACITY OF PATIENT ADVOCATES

### 2.2.1 Website

#### Maintain content quality and information architecture of EURORDIS Website:

- + Centre content on target audiences: patient organisations and advocates, patient and families, other stakeholders
- + Maintain navigation and user-friendliness of website
- + Maintain quality, updated information in seven languages (EN, FR, DE, ES, IT, PT, RU), especially on the homepage, news and events sections and newsletter pages
- + Further develop content and facilitate access via three focal points: the core EURORDIS.org website, EURORDIS Initiative (Rare Disease Day, RareConnect, Rare Barometer, Rare Diseases International, Black Pearl Evening, ECRD, Help Lines and UN NGO Committee for Rare Diseases) and EURORDIS Social media and other media interfaces (Facebook, Twitter, Instagram, YouTube, EURORDIS TV)
- + Develop new web sections on key advocacy priorities, EURORDIS programmes and projects

### 2.2.2 Member News

Publish and continue to build content and consultation with members via monthly Member News (sent to all EURORDIS member organisations)

### 2.2.3 Webinars

- + Develop webinar tool and staff expertise, including training, informational and consultative webinars on a variety of topics
- + Webinars target one of several audiences: EURORDIS members, the general public, or a closed audience (such as those people that have registered for a EURORDIS event).
- + Webinars are held to educate participants on a new topic (for example, genome editing), to update on new policy developments (for example, European Reference Networks), to engage participants in a EURORDIS or European Commission consultation or to inform participants on the technical background of a topic that we be explored in an event.

### 2.2.4 EMM

Organise EURORDIS Membership Meeting 2017 Budapest on 19 & 20 May, including General Assembly, Conference and 4 capacity-building workshops. The capacity building workshops are dedicated to the following themes: ePAGS, Alternative therapies, Patient organisation management and fundraising, Social innovation.

### 2.2.5 CNA Workshop

Organising two workshops of the Council of National Alliances, the first one at the occasion of the Membership Meeting 2017 Budapest and the second at the end of the year in Brussels. These workshops are the opportunity for National Alliances' representatives to exchange information, learn and exchange about advocacy issues and European activities and build their capacities on Rare Disease policy issues.

### 2.2.6 CEF Workshop

Organising a two-day workshop of the Council of European Federations, including a one day training course. The theme of the training 2017 is yet to be determined.

### 2.2.7 ERN – ePAG Leadership Training

- + Organise face-to-face workshops for ePAG Representatives at the EMM 2017 Budapest dedicated to building their leadership skill
- + Further develop the Patient Matchmaker Initiative to match European patient organisations with ERNs according to thematic groupings and across different ERNs, create a virtual network of patient organisations for each ERN, covering a wide range of rare diseases and all Member States, and map and connect patient organisations across the EU to ensure an equitable voice of patient community within ERN

### 2.2.8 EURORDIS Summer Schools

Capacity building programmes on clinical research, therapeutic development, EU regulatory processes and beyond:

#### 2.2.8.1 ExPRESS: Expert Patients and Researchers EURORDIS Summer School

- + Organise **EURORDIS ExPRESS (Expert Patients and Researchers EURORDIS Summer School)** Summer School 2017 in Barcelona in June (in collaboration with LUMC/COST Action on Exon skipping, EMA and Catalan RD Platform)
- + Incorporate 30-35 new patient advocates representing a diversity of diseases and geographical locations
- + Collaborate with LUMC to incorporate 10-15 researchers in this format of the EURORDIS Summer School
- + Collaborate with RDI to incorporate 10 patient representatives from non EU-countries
- + Collaborate with ECRIN for the funding of 5 seats for patient representatives
- + Explore new partnerships for additional funding

### 2.2.8.2 Training Resources Centre on the EURORDIS website

- + 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 and interactive training modules.
- + 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.

### 2.2.8.3 Follow-up of the alumni community

- + Put emphasis on strengthening links with the Summer School/ExPRESS alumni – a RareConnect community of alumni has been launched where are posted some targeted information, including the Therapeutic reports produced monthly
- + The Therapeutic report is also now disseminated through the Member News to a broader audience.
- + Take part in the DIA EuroMeeting 2017 with speakers and session chairs, liaising with the programme committee

### 2.2.8.4 Participation to EUPATI

- + Support to the EPF Programme based on 'European Patients' Academy on Therapeutic Innovation' (EUPATI) (an IMI-funded project gathering a consortium led by European Patients' Forum (EPF) with other European umbrella patient organisations from 2012 to 2016) – 20 days of in-kind support

### 2.2.9 Plan for future Capacity building programmes:

- + Develop (in accordance to the available resources) new capacity building programmes (Advance course on Access to Medicines, Summer School for Young people, etc)

### 2.2.10 Youth Empowerment School

- + Launch the RareConnect youth discussion group to promote exchanges between youth living with a rare disease
- + Consult EURORDIS members to identify those promoting activities directed to youth: exchanging and learning from their experience
- + Develop EURORDIS Youth Empowerment School (YES) training programme, based on an outline transversal to the various issues associated with rare diseases
- + Organise the Youth Empowerment School in Barcelona, in September, with 30 young patient advocates representing a diversity of diseases and countries across Europe [alternative writing: plan the organisation of the Youth Empowerment School in Barcelona, with 30 young patient advocates representing a diversity of diseases and countries across Europe]
- + Enlarge the Training Resources section of the EURORDIS website to include online training tools for youth

## 2.3 RAISING AWARENESS & INFORMING

- + Provide a cohesive voice which represents all diseases globally.
- + Build awareness of the cause of rare diseases to a larger public and to key stakeholders.
- + Inform people living with a rare disease and their families of EURORDIS activity as well as reference points where they can find additional information about their disease or groups

### 2.3.1 Rare Disease Day 2017 & 2018

#### Organise Rare Disease Day 2017 around the theme "Research" under the new 4 year overarching theme of "Patient Involvement"

- + Develop collaboration with communications agency PUBLICIS Health (as established in 2015):
  - Work with Publicis to develop concept and script for 2017 Rare Disease Day video
  - Produce the 2017 video in cooperation with Publicis and AFM-Téléthon (AFM Productions)
  - The video will again be translated into 25+ languages to be disseminated through social media and eurordis.org
  - Work with Publicis to update visuals on RareDiseaseDay.org and to produce poster concept and creative design based on video concept
- + Encourage other stakeholder groups to also participate in Rare Disease Day, using the theme, slogan and visuals
- + Continue to develop participation in Rare Disease Day
  - via social media communication and
  - event posting from patient groups around the world on rarediseaseday.org
  - News information about the campaign via email and internet

#### Plan Rare Disease Day 2018:

- + Continue to lead worldwide patient community (40 National Alliances) in deciding 2018 Theme and creative concept
- + Build campaign materials which are free and easily accessible for all non-commercial use on rarediseaseday.org
- + Improve national media dissemination of campaign information via National Alliance exchange, training and communication (For example, TV spots for video, cinema)
- + Continue to develop rarediseaseday.org technical features to build participation in Rare Disease Day of all stakeholder groups

### 2.3.2 ECRD 2018

#### Start preparations for the 9<sup>th</sup> European Conference on Rare Diseases and Orphan Products-ECRD 2018 Vienna (10-12 May 2018):

- + Programme Committee and Programme:
  - Appoint the Programme Committee and hold two F2F meetings to develop programme with multiple parallel themes. 2. Invite speakers and session chairs.



- + Develop Partnerships: Coordinate and maintain frequent relations via the multiple partnerships:
  - Official partners including ProRare Austria, EMA (COMP), FDA (OOPD), The Austrian Government, HOPE, Orphanet, ESHG, EFPIA-EuropaBio and EUCOPE;
  - Associate partners comprising over 20 rare disease National Alliances, research institutions, projects and industry trade associations;
  - Supporters including AFM-Téléthon and RD-ACTION;
  - Outreach committee comprising between 5 and 10 rare disease National Alliances.
- + Promote ECRD & registrations:
  - Plan Call for Posters;
  - Renew EURORDIS Patient Advocates Fellowship Programme;
  - Develop communication around the event and create the official website in a minimum of 3 languages (EN, DE, RU), outreach to medical journals to encourage higher attendance from healthcare professionals;
  - Propose satellite workshops for partners e.g. Orphanet, IRDiRC ExCom, etc.

### 2.3.3 Social Media

- + Continue to strengthen the EURORDIS social media channels by
  - increasing interactive content
  - strategically using new tools (ie. livestreaming, partnership with The Mighty) which appeal to our target audiences
  - targeting selected posts in 7 languages
- + EURORDIS TV and videos
  - continue to build a library of videos related to rare diseases and publicise them via our dissemination channels
  - video tape our events and publish them to be available to the public

### 2.3.4 EURORDIS Media Relations

- + In 2017 EURORDIS will continue to develop media relations with key health/ policy journalists based primarily based in Brussels, particularly through the February symposium on access. EURORDIS is also pitching to be included in the Financial Times special report on rare diseases, to be published in February 2017.
- + Internally, we will continue from 2016 to develop a wider pool of members of staff who are prepared to respond to various incoming media requests, particularly on technical policy issues.
- + To mark the 20<sup>th</sup> anniversary of EURORDIS, we will create a template press release for national alliances to adapt to their national audience/ for dissemination to their press contacts.

### 2.3.5 EURORDIS eNews

- + Publish EURORDIS eNews in 7 languages (EN, FR, DE, SP, IT, PT, RU) every second week all year round (except August)
- + Publish and continue to build content and consultation with members via monthly Member News (sent to all EURORDIS member organisations)

### 2.3.6 EURORDIS Photo Contest

- + Present Photo Contest 2016 at EURORDIS membership meeting 2017 Budapest with continued and increased presence on Instagram

### 2.3.7 EURORDIS Awards

- + Organise and hold the 6<sup>th</sup> Black Pearl Evening Event in Brussels to be preceded by the EURORDIS Awards and live video-streamed to all public and EURORDIS members via the EURORDIS website
- + EURORDIS Awards may include the following categories: European Rare Disease Leadership, Policy Maker, EURORDIS Volunteer, Patient Organisation, Company, Media, Lifetime Achievement; and the possibility for adding new award categories will be added.
- + Lead and coordinate meetings with the BP Committee and implement proposed actions
  - Build more press coverage of the RD cause in Belgium
  - Include a stronger contingent of Belgians in the committees/speakers

### 2.3.8 Helplines

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")
  - Explore sources of biomedical information to respond to queries about non-allopathic medicine (so called NATC: Natural, Alternative, Traditional, Complementary)
  - 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) in cooperation with the SCOPE Joint Action
  - Link to national plans
  - Link to ERNs

# 3. Patient Engagement

## 3.1 PATIENT ENGAGEMENT IN HEALTHCARE

### 3.1.1 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.
- + Develop a “knowledge generation” approach in ERNs, through sharing the vision and case studies of outcome-driven best practice and emerging innovation
- + Build consensus on this approach across ERNs through RD-ACTION workshops
- + Contributing to the development of patient capacity and knowledge on ERNs services and optimise patients contribute through ePAGs.

### 3.1.2 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 (RD-ACTION) to develop and shape emerging policy areas for ERNs, including for eHealth, IT, data sharing, clinical guideline development, clinical outcome and research
- + Monitor implementation of plans of successful ERNs and support new or re-submitted ERNs applications undergoing a technical assessment
- + Support the development of the definition and representation of Affiliated Partners in MS who are not members of an ERN, ensuring balanced network member representation
- + Connecting healthcare and research under ERNs and research networks or research infrastructure across the translational research pathway, including undiagnosed disease networks
- + Develop and disseminate the vision and the case studies that articulate the potential of ERNs in order to drive a common approach across ERNs
- + Build interoperability and inter-ERN collaboration for the care and management of multi-system rare diseases that sit within multiple ERNs.
- + Promote the adoption of ERN evidence-base practice, guidelines etc. in local treating hospitals and drive quality outcomes at local level
- + Anchor ERN pathways locally to enable access to expertise and improve quality
- + Improve understanding of RDs, mapping resources, optimise spending
- + Achieve common codification and quality of data collected and shared for research, therapy development and faster diagnosis
- + Improve quality of databases, registries to enable therapies and drug development for conditions without

a treatment and research into undiagnosed populations

- + Improves access to expertise through signposting patients into ERNs’ centres (Health Care Providers) and supporting patients’ rights for prior approval

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

- + Take action to help rare disease patient groups for the implementation of successful ERNs
  - + Support the development of European 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 and care, information to patients and outreach to patients
  - + Organise a series of workshops at the EURORDIS Membership Meeting 2017 in Budapest dedicated to ERNs and their Network Members (HCP & Affiliated Partners)
  - + Develop EURORDIS website section and produce updated fact sheet on ERN
  - + Partnering in the EU project RARE-BestPractices, a platform for sharing best practices for management of rare diseases, in order to:
    - 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)
    - Establish a EURORDIS working group on best clinical practices
    - Contribute to the development and the implementation of a core methodology on best practice guidelines for rare diseases (WP3)
    - 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
- ### 3.1.4 Participate in the development of long-term projects on the methodologies for creation and evaluation of best clinical practices:
- + Follow-up on the outcomes of the RARE-BestPractices EU project (a platform for sharing best practices for management of rare diseases), completed at the end of 2016, in order to:

- Continue building 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 dissemination of information and project outcomes through EURORDIS communication channels (EURORDIS website, eNews, EURORDIS membership meetings, etc.)
- Maintain the EURORDIS working group on best clinical practices
- Contribute to the dissemination of project outcomes to relevant audiences, such as EURORDIS members (including ePAGs), European Reference Networks, RD-ACTION, etc. Explore, in particular, best avenues to promote and integrate methodologies for best practice guidelines into ERNs.

## 3.2 PATIENT ENGAGEMENT IN SOCIAL CARE

### 3.2.1 Support the national alliances in their action in advancing the integration of rare diseases into social services and policies at national level

#### 3.2.2 Promote integration of rare diseases into social services:

- + Support the dissemination and implementation of the Commission Expert Group Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies via the RD-Action, across EURORDIS' activities and communication channels;
- + Support the launch of the European Network of Resource Centres for Rare Diseases and promote the exchange of good practices via the INNOVCare project;
- + Continue to disseminate information, case studies and factsheets on specialised social services for rare diseases via the dedicated section on EURORDIS website and eNews;
- + Carry on promoting training for social services providers, in cooperation with the International Federation of Social Workers Europe, with resource centres and EURORDIS members and volunteers experienced in the field;
- + Networking and partnerships with organisations active in the social field at the EU level;
- + Follow-up of social research and social innovation projects and dissemination of relevant findings.

#### 3.2.3 Promoting integrated health and social care for rare diseases

- + Promote the development of integrated care pathways via the INNOVCare project: supporting the design, evaluation and monitoring of the pilot of case management implementation at NoRo resource centre, Romania (2017-2018);
- + Promoting the implementation of integrated and holistic care for rare diseases across Europe, via the organisation of workshops and discussions with policy makers who are part of INNOVCare's advisory group – 20 representatives from national and regional competent authorities in 16 European countries;
- + Dissemination of information on integrated care and relevant issues to support the development of holistic care models for rare diseases;

- + Establishing partnerships with international organisations focused on integrated care.

## 3.3 PATIENT ENGAGEMENT IN RESEARCH

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

- + Create a dedicated website section and disseminate information, to better promote existing relevant resources including from EURORDIS members and partners.
- + 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.
- + Develop capacity building webinars and fact sheets partly based on data collected through our involvement European projects such as RD-Connect and Genetics Clinic of the Future

### 3.3.2 Support specific actions in rare cancers:

- + Support the EURORDIS volunteers in rare cancers in adults, both solid tumours and haematologic malignancies, as well as in paediatric cancers:
  - Elected ePAGs (European Patient Advocacy Groups)
  - EURORDIS Policy Action Group on Rare Cancers (PAG-RC)
  - Elected EURORDIS' representatives on the European Commission Expert Group on Cancer-Control (CEG-CC)
- + Support coordination between the European Commission Expert Group on Cancer Control (CEG-CC) and the European Commission Expert Group on Rare Diseases (CEG-RD) to maximise synergies of these two EU policy areas
- + Actively participate in the EU Joint Action on Rare Cancers (JARC), involving the EURORDIS volunteers and RC patient community at large
- + Collaborate with the European Cancer Patients Coalition (ECPC) on common policy issues and in the EU Joint Action on Rare Cancers (JARC)
- + Participation in the Rare Cancer Europe (RCE) network (EURORDIS is one of the co-founders)
- + Collaboration with the European Society of Medical Oncology (ESMO)
- + Increase membership of rare cancer patient groups
- + Increase relevant visibility of EURORDIS' involvement in rare cancer activities on the EURORDIS website

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

- + Identify a group of interested parties to reflect on the specific issues brought by the development of advanced therapies and their uptake by the civil society
  - Identification of pharmaceutical companies, biotechs involved in this field



- Identification of other stakeholders: patient representatives, HTA bodies, MoCA payers and regulators
  - Reflect on how advanced therapies development is embedded into the concept of personalized medicines
  - + Reflect on the necessity to broaden the scope of this reflection with the potential of innovation coming from new technologies (e.g. 3D printing), combined products (medical devices + chemical products, but also medical devices + ATMPs, companion diagnostics, ...)
  - + Participate in the **Genetics Clinic of the Future (GCOF) project**, led by the University of Utrecht, Netherlands. The GCOF 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
  - + 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
  - + Scale up the involvement of EURORDIS members in the ERA-Net project E-Rare involving National Funding Agencies.
  - + Participate in the governing bodies (Call Steering Committee and Network Steering Committee)
- Increased participation in the ERA-Net
- + 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

- + 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

## 3.4 PATIENT ENGAGEMENT IN THERAPEUTIC DEVELOPMENT

### 3.4.1 European Medicines Agency

- + Participate in the **EMA Committee for Orphan Medicinal Products (COMP)** with one representative member (and vice-Chair of the Committee) and two observers – participation in the Working groups of the COMP: WG on Significant Benefit, WG on Animal Models, WG on Protocol Assistance; participation to the transversal reflections of the COMP: workshop on Medical Conditions, strategic review and learning meetings + promotion of the involvement of adhoc patient experts in the discussions on significant benefit at the time of assessment of the maintenance of the orphan status at the time of Market Authorisation
- + 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 – Participation in the Working groups of the PDCO: Joint COMP-PDCO WG on Definition of Conditions; participation to the transversal reflections of the PDCO: strategic review and learning meetings + involvement of young patients in the work of the Committee, promotion of the involvement of adhoc parents/patient experts in some specific discussions
- + Participate in the **EMA Committee for Advanced Therapies (CAT)** with one alternate member and one observer - Participation in the Working groups of the CAT: Working group on Concept of Similarity; participation to the transversal reflections of the CAT: strategic review and learning meetings
- + 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
- + Contribute to new approaches for the engagement

of patients in the benefit/risk evaluation, taking into consideration patient preferences elicitation

- + 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
- + Exploit the results of the 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) (reception of all the dossiers and collaboration with SAWP Secretariat and EMA Patient relations team), and when applicable for Scientific Advisory Groups of CHMP and EMA parallel HTA/ Scientific Advice
- + 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.
- + In practice: 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 when applicable at time of marketing authorisation (European Public Assessment Reports summaries, Package Leaflets, Significant Benefit Public Reports)
- + Follow-up on collaboration between EMA and FDA on rare therapies development beyond orphan drug designation – Participation to monthly conference calls between FDA and EMA orphan offices

### 3.4.2 HTA

- + Participation of patients in EMA multi-HTA parallel scientific advice and other HTA early dialogues as proposed by EUnetHTA Joint Action

- + Represent EURORDIS in the HTA Network Stakeholders' Forum
- + Contribute to the discussions on "Economic assessments of new technologies", for example on common core economic models which could increase transferability among EU Member States
- + Articulate and coordinate protocols for the engagement of patients in both regulatory and HTA processes, following the Reflection paper on the interaction between regulatory and HTA issues
- + Participate in the launch of EUnetHTA JA3 activities for the period 2016-2019, in particular work with the JA coordinator and relevant work package leaders to involve patients in early dialogues and in joint assessment (from scoping to reporting)

### 3.4.3 MoCA

- + Participation of patients to the pilots of the MoCA initiative (Mechanisms of Coordinated Access)
  - Involvement since 2016, of patient representatives of specific medical conditions in the discussions taking place in the context of MoCA initiative. EURORDIS identifies and supports the participation to one or two patient representatives per dossiers. The support is also financial.
  - Development of a policy of patient involvement (conflict of interest, confidentiality, competing interest)
  - The discussions are tripartite: company developing the product, payers from various countries, patient representatives (ad hoc patient experts + EURORDIS representative(s))

Promote interactions of all the stakeholders: Common dialogue between EMA-HTA bodies-companies and patients are already happening. There is a need to also include payers. One possibility could be to create links between the two initiatives, MoCA and PRIME.

### 3.4.4 Promote the engagement of patient during the lifecycle of medicine development by:

- + Co-leading the setting up of a consortium to answer the call for proposal of the IMI (Call 10) aiming to enable patients to enhance their voice (in qualitative and quantitative terms) in the medicines R&D processes of treatment development: from discovery to outcomes monitoring, and to include those who are under-represented, or may not normally participate in critical decision-making (including patients not affiliated to patient groups, vulnerable populations such as minors or the elderly). In addition, it will provide a framework and guidance for all EU stakeholders about who, when and what information is needed, as well as how to engage patients to obtain beneficial and necessary input from patients and healthcare consumers
- + Participation in the Patient Focused Medicines Development (PFMD) initiative, an independent multinational coalition. Its goal is to bring together initiatives and best practices that integrate the voice of the patient throughout the lifecycle of medicines development, thereby speeding up the creation and implementation of an effective, globally standardized framework.

- + Participate as a member in MIT's NEW Drug Development ParadigmS (NEWDIGS) program which is a unique collaborative "think and do" tank focused on enhancing the capacity of the global biomedical innovation system to reliably and sustainably deliver new, better, affordable therapeutics to the right patients faster.
- + 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
  - Adopt the Guidelines for the Operation of Community Advisory Boards developed by the DITA task force and inform the EMA and EUnetHTA on these guidelines
- + Launch a call for expression of interest to developers of new health technologies to establish new Community Advisory Boards: companies that already signed the Charter, new ones with a commitment to apply to new regulatory and HTA pathways (scientific advice, PRIME, Adaptive Pathways, MOCA...) or in unmet needs in rare diseases
- + Promote Community Advisory Boards (C.A.B) among patients' organisations and create a European Network of C.A.B. Chairs and mentors, with organisation of the meetings and the training needs being taken care by Eurordis
- + 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
  - Integrate Clinical Outcome Assessment (C.O.A) in discussions in early scientific advice (e.g. PRIME) and early dialogues (EUnetHTA)
  - Promote the discussions on C.O.A in Community Advisory Boards, and invite experts accordingly during the C.A.B trainings

### 3.4.5 Promote rare disease patient spontaneous reporting on suspected adverse drug reactions of 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 project as partner and 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
- + Participate in the European Awareness Week on pharmacovigilance
- + Reflect on SCOPE outcomes on appropriate measures that Member States can take to engage patients and

their organisations in pharmacovigilance, and promote new actions for patients' organisations to play their role, such as the appointment of an official contact person for pharmacovigilance

- + Participate in EMA Public Hearings on safety of medicines in their therapeutic context

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

- + Report on the results of the survey '**Tell us how you take your treatment**', and participate in discussions on related topics (prevention of medication errors in rare diseases, patients' participation in reporting ADRs, off-label use or medicines, information needs, access issues)
- + Continue the activities in parallel to the work plan of the PCWP, in particular:
  - Provide input into the development of a GVP module on medicines in pregnancy and breastfeeding
  - Contribute to a framework for involvement and consultation of young people in different EMA activities on paediatric medicines
  - Contribute to the identification of experts through their organisations to participate in discussions on orphan designation within the COMP.
  - Contribute to the implementation of the revised action plan regarding medicinal product supply shortages by promoting best practices on communication of shortages
  - Discuss how to increase public understanding around EMA initiatives to promote early access to medicines, including the concept of 'adaptive pathways', the PRIME initiative, and the cooperation with health technology assessment (HTA) bodies and other regulatory agencies;
  - Follow up and provide input as appropriate to the EMA initiatives on the use of real-world data and patient registries
  - Organise a workshop in March to create awareness about the areas where EMA is supporting personalised medicine.
  - Follow the outcome of the workshops organised in 2016 on social media, health/apps (IMI-WEB-RADR project) and big data and contributing with expertise as required, through the continued work of the HCPWP/PCWP topic group on digital health and media (the former topic group on social media)
- + **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)



### 3.4.7 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

- + Engage EURORDIS in the Horizon 2020 project aiming at streamlining the services offered by the biomedical European research infrastructures.
- + Participate as Partner in the IMI Consortium ADAPT SMART 'Accelerated Development of Appropriate Patient Therapies: A Sustainable Multi-Stakeholder Approach from Research to Treatment'. The works of EURORDIS is facilitated by the involvement of 6 patient experts. EURORDIS is also a member of the Navigator Group, the governing body of the project
- + Bring these innovative policy approaches and activities to the IRDiRC

### 3.4.8 EURORDIS Round table of Companies: Promote dialogue with pharmaceutical & biotech companies involved in rare disease therapy development:

- + **EURORDIS Round Table of Companies:** consolidate membership and organise two workshops in 2017.

The February ERTC will again follow an exceptional format with the second of the series of multi-stakeholder symposia on "Improving Patient Access to Rare Disease Therapies" taking place in Brussels. This event is open to all stakeholder groups. This symposium will take the debate to the next level. Following on from the issues discussed at the first symposium, participants will come together to consider sustainable, decisive and long-lasting solutions to improve patients' access to rare disease therapies. The goal of this second symposium is to build and seek convergences on the current and emerging models and initiatives and to develop a process inclusive of all stakeholders to work towards solutions to improve access.

A second ERTC workshop will be held in September in Barcelona.

In addition:

- + Strengthen the dialogue with EFPIA-EuropaBio Task Force on Orphan Drugs
- + Further develop strong relations with EUCOPE
- + Expand direct dialogue with pharmaceutical and biotech companies by increasing ERTC membership and fostering member participation.
- + Expand ERTC membership to other relevant health sector companies
- + Further clarify and refine protocol for staff interactions with health sector companies, developing common practices across the organization

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- + Strengthen the dialogue with EFPIA-EuropaBio Task Force on Orphan Drugs and further develop strong relations with EUCOPE.
- + Expand direct dialogue with pharmaceutical and biotech companies by increasing ERTC membership and fostering member participation.
- + Further clarify and refine protocol for staff interactions with health sector companies, developing common practices across the organisation.
- + Expand ERTC membership to other relevant health sector companies.

## 4. Cross-Cutting priorities

### 4.1 GOVERNANCE

#### 4.1.1 EURORDIS Board of Directors

- + Organise 4 Board meetings throughout the year in addition to relevant Board of Officers conference calls
- + Organise Board elections during the EURORDIS General Assembly 2017 Budapest, to be voted on by full members of EURORDIS

#### 4.1.2 EURORDIS Statutes & By-Laws

- + Implement new EURORDIS Statutes and use them for the revision of relevant governance documents, including the by-laws
- + Draft EURORDIS by-laws, which will serve as rules governing internal processes, to be presented at the General Assembly 2017 Budapest

#### 4.1.3 Strategy 2015-2020 Implementation

- + 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

#### 4.1.4 Strategic Partnerships (MoUs)

- + Maintain strategic partnership with NORD (USA)
- + 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

## 4.2 RESOURCE DEVELOPMENT

### 4.2.1 Public Funding

- + Operating Grant
- + EU projects (Joint Action)

### 4.2.2 Health Corporate

- + Maintain overall support level from industry donors spread between 50 + companies and diverse activities within the confines of the EURORDIS Policy of Relationship with Commercial Companies, EMA Policy on Prevention of Conflict of Interest and CHAFAE rules

### 4.2.3 Other private funding and foundations

- + Engage corporate and foundation donors beyond the pharmaceutical industry to support EURORDIS projects & actions. Priorities: EURORDIS Membership Meeting; ERTC membership for select CROs and other non-pharma companies in the health sector; EURORDIS' International Initiatives: RareConnect, Rare Barometer, Rare Diseases International, training programmes and Rare Elegance event

### 4.2.4 In-kind contributions

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

### 4.2.5 Circle of Ambassadors

- + Maintain the EURORDIS International Circle of Ambassadors, bringing together 6 - 8 community leaders from Europe and North America to establish base of foundations and individuals supporting EURORDIS, including 1 cultivation event in Geneva and 1 cultivation

### 4.2.6 EURORDIS events

- + Organise the **EURORDIS Awards & Black Pearl Evening** on 21 February 2017 in Brussels and plan event for 2018.
- + Test the concept and start planning the organisation of a headline fundraising event, a night of entertainment, celebration and opportunity to raise funds for rare disease programmes. 'Rare Elegance' is anticipated to be held in London during the 4<sup>th</sup> quarter 2018.
- + 9<sup>th</sup> European Conference on Rare Diseases & Orphan

**Products – ECRD 2018 Vienna** (10-12 May 2018): In addition to the revenues generated by registration fees, EURORDIS will expand the table-top exhibition and support packages for industry and will seek additional government support and EU funding, exploring the possibility of running workshops - in the context of RD-Action - back-to-back with ECRD. The objective is to stimulate participation of more clinicians and Member State representatives and defray some travel costs of speakers for the Conference.

### 4.2.7 Individual Donors

- + Develop new donation pages on eurordis.org website including specific pages for the Black Pearl Evening and other EURORDIS events and communications initiatives
- + Appeal for individual donations using part of the 20 years of EURORDIS communications campaign

## 4.3 HUMAN RESOURCES

### 4.3.1 EURORDIS Staff

#### 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
- + 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)
- + Create the position of HTA Stakeholder Involvement Coordinator

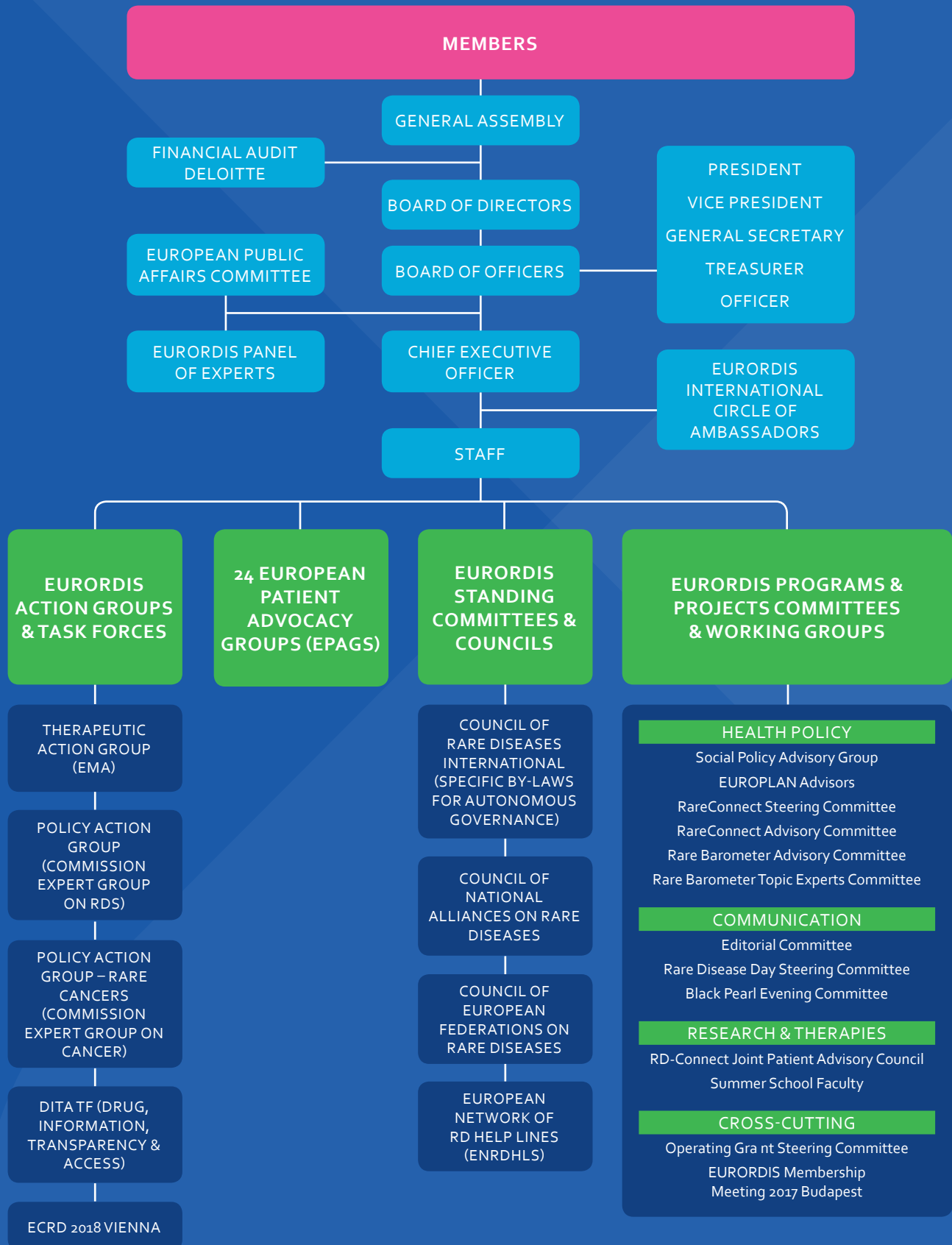
#### Seek alternative human resources:

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

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

- + Maintain decentralised structure with offices in Paris (main office), Brussels (European public affairs), Barcelona (web communications; RareConnect), Zagreb and Belgrade (RareConnect Teams in Serbia and Croatia) 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.
- + Maintain the efficiency and security of the IT infrastructure open to staff and volunteers.
- + Improve quality of video conferences and train the staff to the usage of high quality audio and video devices.
- + Ameliorate the integration of new staff, the efficiency of all HR processes such as staff reviews, trainings, coaching, etc.
- + Be the referring technician concerning the development of CRMs.

# GOVERNANCE Chart 2017





# EXTERNAL REPRESENTATION Chart 2017



## EUROPEAN NOT-FOR-PROFIT ORGANISATIONS

DIA: Drug Information Association

EFPIA Think Tank: European Federation of Pharmaceutical Industries and Associations

EUROPABIO Patients Advisory Group

EUCOPE

EPF: European Patients' Forum

EFGCP: European Forum for Good Clinical Practice

FIPRA – International Policy Advisors

Friends of Europe

Rare Cancer Europe

Social Platform

Maladies Rares Info Service (French Helpline for RDs)

Rare Disease Platform in Paris

PFMD - Patient Focused Medicines Development Initiative

## EUROPEAN NETWORK OF PARLIAMENTARIAN ADVOCATE FOR RARE DISEASES

European parliament interest group on Rare Diseases  
Advocates in national parliaments

## MEMBER OF EUROPEAN NETWORKS

E-Rare

EuroBioBank

ECRIN

BBMRI Stakeholders Forum

Treat NMD

RD-Connect

SCOPE Joint Action (Advisory Board)

OpenMedicine

IMI EUPATI

IMI ADAPT-SMART

## INTERNATIONAL INSTITUTIONS AND NOT-FOR-PROFIT ORGANISATIONS

NGO Committee for Rare Diseases  
(United Nations, New York)

NEWDIGS: New Drug Development ParadIGmS

IAPO: International Alliance of Patients' Organizations

IRDIRC: International Rare Disease Research Consortium

ICORD: International Conference on Rare Diseases and Orphan Drugs

## PARTNERSHIP LEARNED SOCIETIES

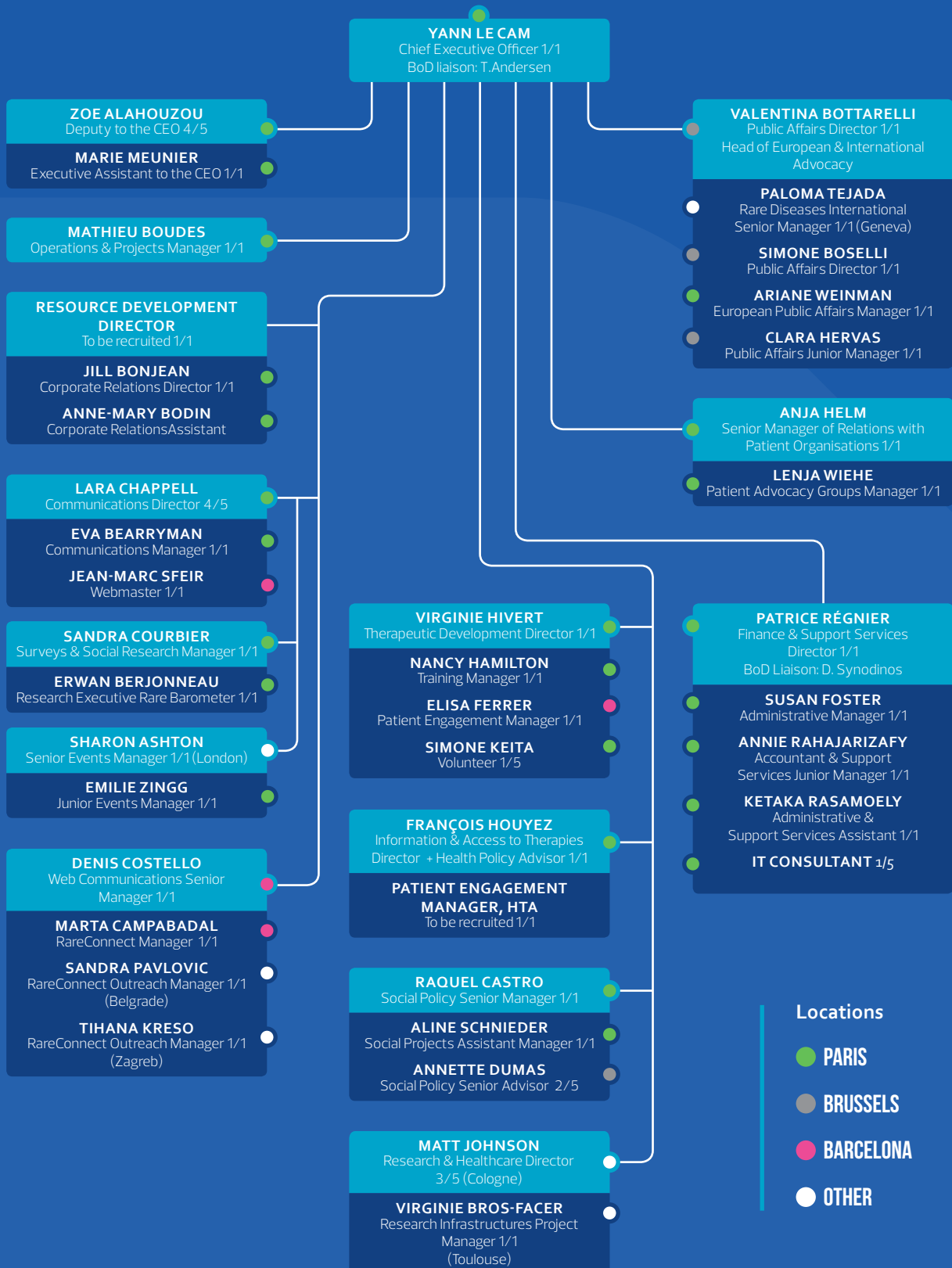
European Federation of Internal Medicine (EFIM)  
European Hospital & Healthcare Federation (HOPE) -

International Federation of Social Workers Europe (IFSW-Europe)

European Society of Human Genetics (ESHG)

International Society for Pharmaco-economics and Outcomes Research (ISPOR)

# TEAM Chart 2017

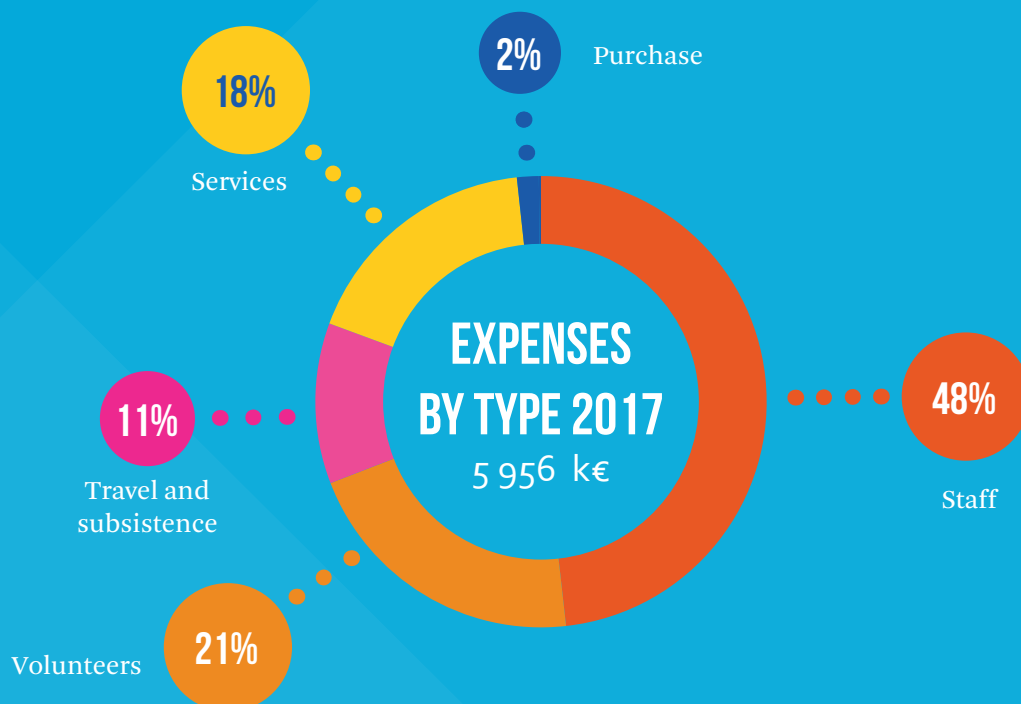
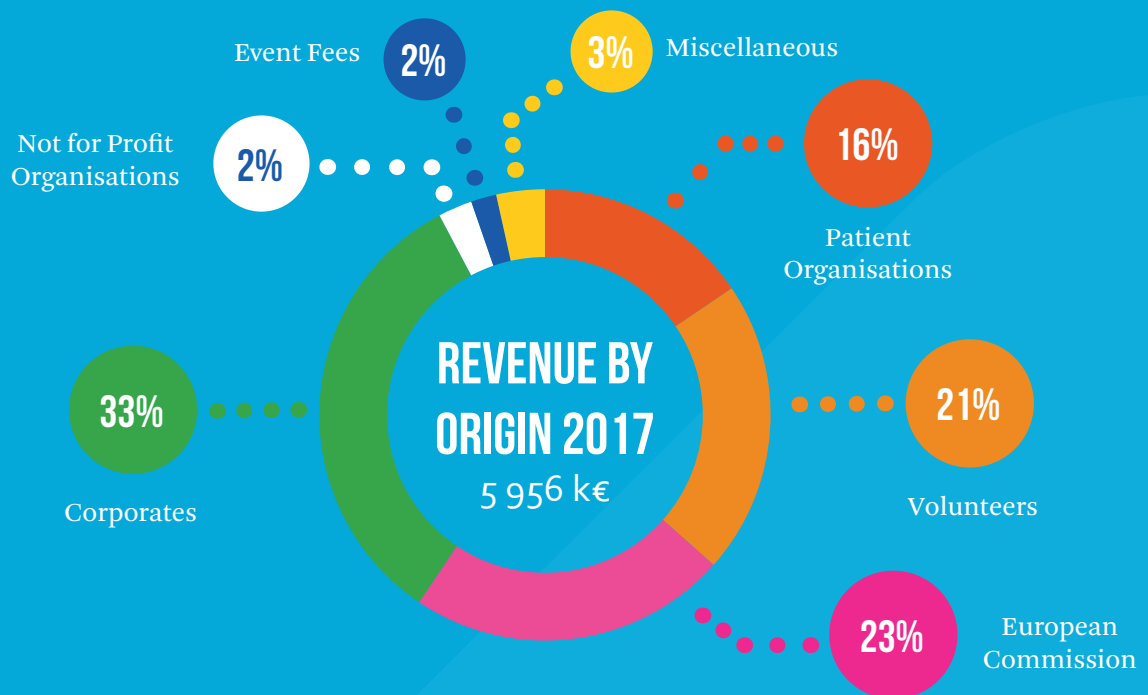


## Locations

- PARIS
- BRUSSELS
- BARCELONA
- OTHER



# REVENUE & expenses 2017



# ACRONYMS & DEFINITIONS

## EURORDIS INTERNAL COMMITTEES & TASK FORCES

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

## PROJECTS OF EURORDIS OR IN WHICH EURORDIS IS INVOLVED

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

## EURORDIS & EUROPEAN REGULATORY NETWORK

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

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

## EUROPEAN COMMISSION

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

## EURORDIS & EUROPEAN COMMISSION

<b>CEGCC</b>	Commission Expert Group on Cancer Control
<b>CEGRD</b>	Commission Experts Group on Rare Diseases - 8 patients' representatives included 2 representatives of EURORDIS and 2 Observers
<b>EU HPF</b>	EU Health Policy Forum
<b>JRC</b>	Joint Research Center EU Platform Rare Diseases Registration

## EURORDIS & NON GOVERNMENTAL PARTNERS

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

## MISCELLANEOUS

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





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