

EURODIS  
**ACTIVITY REPORT** 2017  
& WORKPLAN 2018



Living with Duchenne  
muscular dystrophy,  
Misha and his mother  
from Ukraine

# TABLE of Contents

**02**

Foreword

**06**

EURORDIS  
in brief

**07**

Strategic  
approach  
2015-2020

**09**

Activity  
Report 2017

HIGHLIGHTS 2017.....	11
PATIENT ADVOCACY .....	12
PATIENT EMPOWERMENT: BUILDING THE NETWORK & CAPACITIES .....	27
PATIENT ENGAGEMENT: ROLES IN DECISION-MAKING .....	37
CROSS-CUTTING PRIORITIES .....	46
REVENUE & EXPENSES 2017 .....	50
BOARD OF DIRECTORS MAY 2017-MAY 2018.....	52
MEMBERS OF EURORDIS.....	53
CONFERENCES & WORKSHOPS 2017.....	59
ACKNOWLEDGEMENTS .....	63

**67**

Workplan  
2018

ACTION PLAN 2018.....	68
GOVERNANCE CHART 2018 .....	75
EXTERNAL REPRESENTATION CHART 2018.....	76
TEAM CHART 2018 .....	78
REVENUE & EXPENSES 2018.....	79

**80**

Acronyms & definitions





2017 marked the 20<sup>th</sup> anniversary of EURORDIS-Rare Diseases Europe. To capture our vision for the next 10 to 20 years, EURORDIS announced 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.

This has been translated into a new identity, changing our name from the European Organisation for Rare Diseases (EURORDIS) to **EURORDIS-Rare Diseases Europe** and including a brand new logo. Officially recognising our name to include Rare Diseases Europe, was a further step to strengthen the identity of the rare disease movement, **showing unity with fellow national alliances around Europe** which are increasingly adopting this naming structure (such as Rare Diseases Denmark, Rare Diseases Italy, Rare Diseases Ireland and numerous others who are now following suite), as well as internationally with other patient groups like Rare Diseases International.

In 2017, our membership base continued to grow, reaching **779 members in 69 countries** by the end of the year and our activities, as always, focused on advocating for, empowering and engaging patients. 2017 however, also saw the sharp expansion in the complexity and scope of the activities carried out by EURORDIS and was an exciting and pivotal year that lay a lot of the groundwork for what lies ahead in 2018.

## ADVOCATING FOR PATIENTS

EURORDIS became the **first patient organisation to be invited to host an event under the Presidency of the EU Council**. The event “Integrating Research and Healthcare for Rare Diseases: A structured cooperation with high community added value” was held under the Maltese Presidency of the EU Council and offered

# FOREWORD

by the President and the Chief Executive Officer

the opportunity to explore synergies within and across the rare disease research and healthcare communities. The Maltese Presidency and the Malta Medicines Authority also hosted a **Conference on the Development and Access of Medicines for Rare Diseases** in collaboration with EURORDIS and other partners. In line with this Conference, EURORDIS **launched a declaration calling for improved mechanisms of structured voluntary cooperation across EU Member States to address the unmet needs of people living with a rare disease.**

EURORDIS continued to work towards improving access to rare disease therapies with the **second Multi-Stakeholder Symposium on Improving Patient Access to Rare Disease Therapies**, held in Brussels in February 2017. Following the reflection process of the symposium, EURORDIS launched a new position paper **‘Breaking the Access Deadlock to Leave No One Behind’** which sets out a new four-pillar approach to tackling the challenges that prevent patients’ access to care and medicines, as well as the **ambition to have 3 to 5 times more new rare disease therapies approved per year, 3 to 5 times cheaper than today by 2025.**

Another highlight of the year was the launch of the **Parliamentary Advocates for Rare Diseases**, a network of European and national

members of parliament advocating to improve the lives of people living with a rare disease. Through the network, EURORDIS aims to bring together members of parliament to ensure strong international and local action, shape political input for current and future legislation, and integrate rare diseases into all relevant policies at all levels. The launch event '**Juggling Care and Daily Life: The Balancing Act of the Rare Diseases Community**' which was organised under the auspices of MEP Francoise Grossetête, was held at the European Parliament in Brussels and included a presentation of highlights of the results of **the first European survey on the impact of rare diseases on daily life**, as carried out through Rare Barometer Voices, the EURORDIS survey initiative.

EURORDIS continued to play a central role in the efforts for the **development of an international rare disease movement through Rare Diseases International and the NGO Committee for Rare Diseases**. Rare Diseases International reached 49 members by the end of 2017. To mark the occasion of Rare Disease Day 2017, **a unique policy event on 'The Right to Health: The Rare Disease Perspective'** was held in Geneva, gathering international experts in the fields of public health,

human rights, epidemiology, scientific research and patient advocacy to discuss why and how rare diseases should be included in the global health agenda, and included representatives of the WHO, the Office of the UN High Commissioner for Human Rights, the UN Development Programme as well as patient representatives from around the world. 2017 was also marked by the **attendance and networking at a number of official United Nations events**, including the Conference of State Parties to the Convention of People with Disabilities (June), the Open-Ended Working Group on Ageing meeting (July), the High-Level Political Forum on Sustainable Development (July), the Human Rights Council (September) and the International Day of People with Disabilities (December). This preparatory work will pave the way for the future actions taken by the Committee, principally at the level of the World Health Assembly and the Human Rights Council, in 2018 and 2019.

## EMPOWERING PATIENTS

The 2017 **ExPRESS Summer School** continued for its 9th year in a row taking place in June in Barcelona and bringing together a record number of trainees from around the world. It was organised in parallel to a **first-ever Spanish version of the**



**YANN LE CAM**

Chief Executive Officer



**TERKEL ANDERSEN**

President of the Board of Officers



**Summer School**, partnering with, among others, FEDER and Plataforma Malalties Minoritaries. **Over 60 students from 25 countries**, were selected to take part in the English version of the Summer School and **43 participants from many regions of Spain as well as Central and South America**, participated in the Spanish version of the Summer School. The total number of alumni since 2008 is now 422.

In 2018, building upon our ten year experience of capacity building programmes, EURORDIS will be launching **the EURORDIS Open Academy**, which will consolidate all of our face-to-face and online training experiences and also add further trainings such as the EURORDIS Winter School on **Scientific Innovation and Translational Research**.

2017 was also a pivotal year for **RareConnect**, the online platform of rare disease patient communities. **EURORDIS transferred the platform to the Children's Hospital of Eastern Ontario Research Institute (Canada)** who in

partnership with the technology and research team at The Hospital for Sick Children, Toronto (Canada) successfully undertook developments which further improved user experience.

The Children's Hospital of Eastern Ontario Research Institute (Canada) will drive innovation in the underlying technology and interoperability with research and clinician based tools but EURORDIS will continue at the heart of the governance of the platform making the link with patient associations and members of the platform through a dedicated team of community managers.

## ENGAGING PATIENTS

**One of the dominating features of the year, was the work that materialised within the context of the European Reference Networks and the engagement of patients within them.** EURORDIS established the European Patient Advocacy Groups (ePAGs) which aligned to the scope of each ERN. ePAGs bring together patient

representatives and affiliated organisations to ensure that the patient voice is heard throughout the ERN development process. To date there are **24 ePAGs involving over 250 patient representatives and 1000 patient organisations, including non-members of EURORDIS**. In 2017, EURORDIS held regular webinars on ERNs (outcomes and indicators, eHealth and data sharing); piloted a mentoring programme for ePAG representatives and held two ePAG face to face capacity building workshops, one in Budapest in May and one Paris in October. Two key outcomes resulting from these workshops was to establish an ePAG Steering Committee in addition to topic focused transversal peer learning groups aligned to ERN priority areas.

EURORDIS also continued to reach out to the wider patient community **to raise awareness about the developments of ERNs**. We supported National Alliances through webinars focusing on actions on how to anchor ERNs into national health system and engaging with their members through a series of workshops. The development of ePAGs and ERNs were included in both the EURORDIS Membership Meeting 2018 Budapest and the meeting of the Council of European Federations in Paris. The deployment phase of the European Reference Networks will be a key area of action in 2018.

**EURORDIS continued its increased engagement in HTA activities**. Although EUnetHTA JA3 decided not to have a structured interaction with stakeholders, it did commit to involving patients as experts in its scientific assessments and other activities. Without a framework for the involvement of patients, EUnetHTA asked EURORDIS and other organisations to identify eligible patients for joint HTA procedures or Early Dialogues. **In 2017, 6 requests of patients' identification were made.**

EURORDIS also took **several actions related to the EC Proposal for a Regulation on the European HTA cooperation**. Among others, EURORDIS contributed to two documents for the European Commission's call for contribution to the HTA legislation Principles of Patients and Consumers engagement in HTA and Criteria for prioritisation of technologies for joint assessment;

and organised and presented HTA related items at the CNA/CEF meeting in Paris in October. EURORDIS also took the first steps towards the development of **a EURORDIS Task Force on HTA which will be established in 2018**.

A brief mention must also be made to other key recurrent EURORDIS activities that have become regular milestones of the rare disease community, such as **Rare Disease Day (organised in 94 countries in 2017), the EURORDIS Membership Meeting (held in Budapest in May with 250 participants from 38 countries), the Council of National Alliances and the Council of European Federations (reaching 41 National Alliances and 62 European Federations respectively)**.

None of the activities detailed in this report would be possible without the tireless dedication of the EURORDIS volunteers. In 2017, EURORDIS was privileged to rely on **428 volunteers including 68 dedicated volunteer patient advocates, 1 office support volunteer, 1 volunteer fund-raiser, 358 volunteer moderators** of online communities of rare disease patients, within the activity "RareConnect". EURORDIS volunteers have a unique insight into the complexity of different rare diseases across Europe and reinforce EURORDIS as a grassroots movement.

We know that many challenges ahead; the increase of complexity and scope of our field brings with it a growth that has a number of uncertainties, including growing financial risk. Resource diversification must be a key priority moving forward. Despite the challenges, we know that **our community is governed by common values and modes of action**. We believe in solidarity and equitability, social justice and fairness. We work through empowerment and capacity building of patient advocates, exchange and mutual support, gathering of experience, networking and partnership with all stakeholders. **Building on our collective expertise, together we will continue to tackle the challenges that lie ahead.**

”

# 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.

# 779

member patient organisations



**69** countries  
(28 EU countries)

**41** National Alliances of RD Patients Organisations

**62** European Federations of specific rare diseases

 Founded in

# 1997



# 40+

Staff members, with offices in Paris, Brussels, Barcelona

Outreach to over

# 2500

patient groups

 Over

# 420

Volunteers

**+68**  
patient advocates

**+350**  
moderators

## Vision

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

## Mission

EURORDIS-Rare Diseases Europe 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

2017

HIGHLIGHTS 2017.....	11
PATIENT ADVOCACY .....	12
PATIENT EMPOWERMENT: BUILDING THE NETWORK & CAPACITIES.....	27
PATIENT ENGAGEMENT: ROLES IN DECISION-MAKING .....	37
CROSS-CUTTING PRIORITIES .....	46
REVENUE & EXPENSES 2017 .....	50
BOARD OF DIRECTORS MAY 2017-MAY 2018.....	52
MEMBERS OF EURORDIS.....	53
CONFERENCES & WORKSHOPS 2017.....	59
ACKNOWLEDGEMENTS .....	63



# HIGHLIGHTS 2017

By the end of 2017, EURORDIS had **779 member patient organisations**, located throughout 69 different countries, and including all 28 EU countries.

The **EURORDIS Membership Meeting 2017 Budapest** was held on 19-20 May and attracted **250 participants from 38 countries**. **40 fellowships were granted from 18 countries** within the EURORDIS Patient Advocate Fellowships Programme.

**Rare Disease Day 2017** was organised in over 94 countries worldwide, including all 28 EU countries. For the 6<sup>th</sup> year in a row EURORDIS produced a well-received video for Rare Disease Day that exemplified this year's theme of Research. The video was translated into 35 languages.

The **6<sup>th</sup> EURORDIS Black Pearl Awards for outstanding accomplishments in the field of rare diseases** were presented in Brussels on the occasion of Rare Disease Day.

EURORDIS created and continued to support **24 European Patient Advocacy Groups (ePAGs)** aligned to the scope of the ERN applications, involving over **250 patient representatives and 1000 patient organisations**, including non-members of EURORDIS. In 2017, there were **2 face to face meetings of ePAG representatives** and over **77 conference calls**. In addition, EURORDIS facilitated ePAG representation and ePAG satellite meetings at 23 ERN board and kick off meetings.

EURORDIS is also **the first patient organisation to be invited to host an event under the Presidency of the EU Council**. The event "Integrating Research and Healthcare for Rare Diseases: A structured cooperation with high community added value" was held under the Maltese Presidency of the EU Council and was hosted by EURORDIS and other partners. It offered the opportunity to explore synergies within and across the rare disease research and healthcare communities.

**Rare Diseases International (RDI)** continued to expand reaching **49 members** by the end of the year. To mark the occasion of Rare Disease Day, RDI held a first of its kind **International Policy Event** in Geneva in February, which gathered over **150 international experts** in the fields of public health, human rights, epidemiology, scientific research and patient advocacy to discuss why and how rare diseases should be included in the global health agenda. RDI also organised its **3<sup>rd</sup> annual Membership Meeting** in June in Castelldefels, Barcelona, Spain. Over **50 participants from 23 countries** were able to network with patient advocates from around the

world, learn more about recent developments in international rare disease advocacy and receive information to become further involved in RDI activities

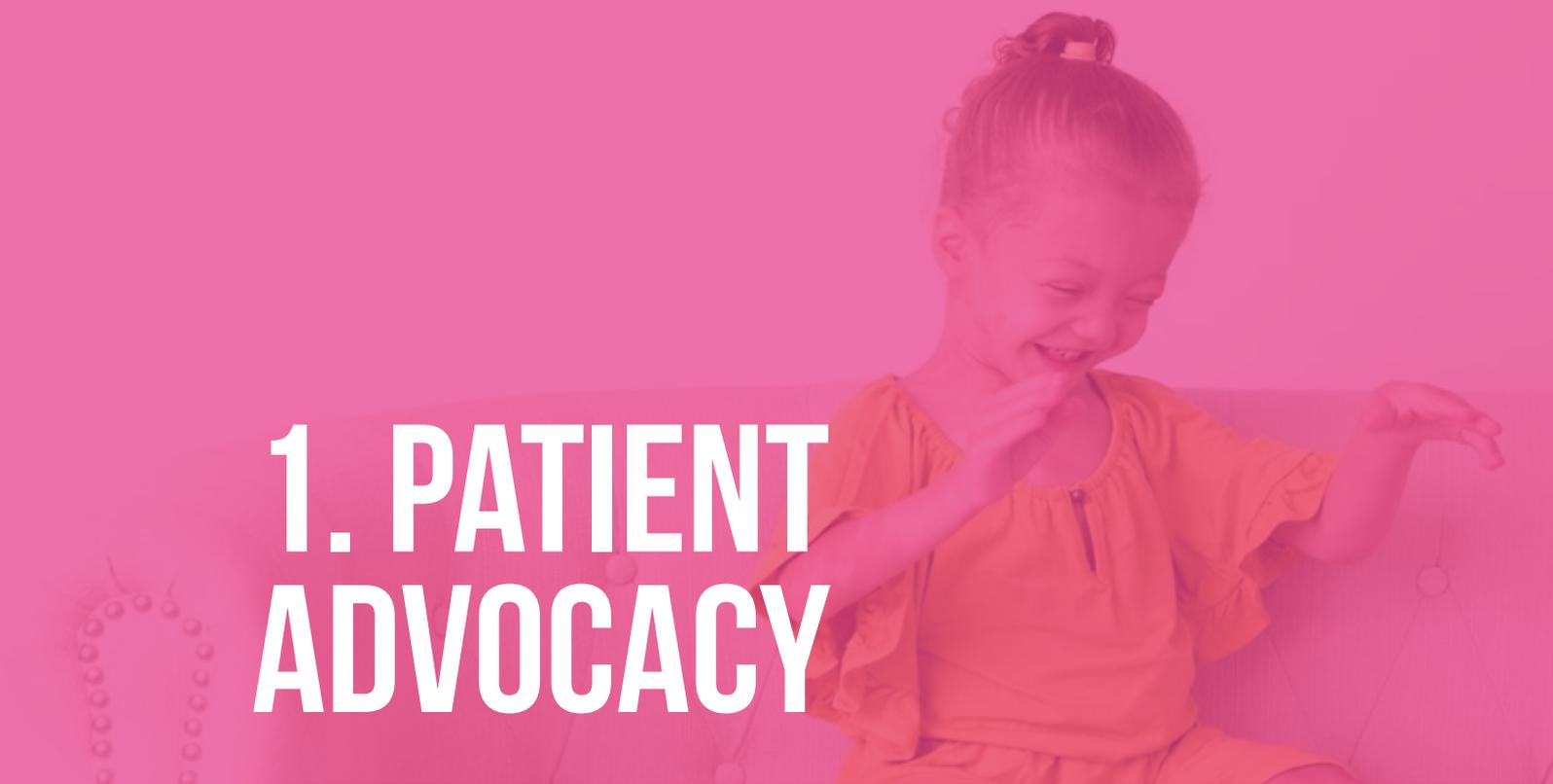
The **Rare Barometer Programme**, a permanent EURORDIS programme of quantitative and qualitative studies to collect patient experiences and expectations was further developed and promoted. Rare Barometer Voices, an online panel of people living with a rare disease who are willing to participate in EURORDIS' surveys, **recruited over 7000 participants**.

**EURORDIS continued to work towards improving access to rare disease therapies** with the **second Multi-Stakeholder Symposium on Improving Patient Access to Rare Disease Therapies**, held in Brussels in February 2017. Following the reflection process of the symposium, EURORDIS launched a new position paper '**Breaking the Access Deadlock to Leave No One Behind**' which sets out a new four-pillar approach to tackling the challenges that prevent patients' access to care and medicines, as well as the ambition to have 3 to 5 times more new rare disease therapies approved per year, 3 to 5 times cheaper than today by 2025.

The **2017 ExPRESS Summer School** took place in early June in Barcelona and brought together a record number of trainees from around the world. It was organised with new collaborations and in parallel to a **first-ever Spanish version**. **Over 60 students from 25 countries**, were selected to take part in the English version of the Summer School and **43 participants from many regions of Spain as well as Central and South America**, participated in the Spanish version of the Summer School.

In October 2017, EURORDIS launched the **Parliamentary Advocates for Rare Diseases**, a network of European and national members of parliament advocating to improve the lives of people living with a rare disease. The launch event '**Juggling Care and Daily Life: The Balancing Act of the Rare Diseases Community**' which was organised under the auspices of MEP Francoise Grossetête, was held at the European Parliament in Brussels.

EURORDIS continued to play an important role in the orphan drug development process through participation in **the European Medicine Agency's Scientific Committees** : the Committee for Orphan Medicinal Products (COMP), the Paediatric Committee (PDCO), the Committee for Advanced Therapies (CAT) and the Patients' and Consumers' Working Party (PCWP) with a total number of **172 meeting days** spent in EMA Committee's over the year.



# 1. PATIENT ADVOCACY

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

In the course of 2017, 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 2017 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 Groups on Rare Diseases and Cancer Control:

Two important platforms for policy-making in the field of rare diseases were the European Commission Expert Groups on Rare Diseases (CEG-RD) and Cancer Control (CEG-CC), where EURORDIS was represented. **In 2017, both Expert Groups ended their mandate, which was not renewed.**

The **Commission Expert Groups on Rare Diseases and Cancer Control** provided advice and expertise to the Commission in formulating and implementing the Union's activities in the field of rare diseases and cancer control respectively, fostering exchanges of relevant experience, policies and practices between the Member States and the various parties involved. Eight patient representatives were appointed member of the Expert Group on Rare

Diseases and supported by EURORDIS staff through the Policy Action Group (PAG) to provide the patients' view on issues dealt with by the CEG-RD. Likewise, two volunteer representatives of EURORDIS had been nominated to represent patients affected by rare cancers in the Commission Expert Group on Cancer Control and were supported by the PAG-RC, Policy Action Group on Rare Cancers.

With the Expert Groups having ended their mandate and not having been renewed, in the course of 2017 the Policy Action Groups stopped their activities.

The two Experts Groups and their predecessors (EUCERD, Rare Diseases Task Force RDTF) were instrumental in advancing essential areas of both EU and national policies in the field of rare diseases, such as National Plans and Strategies for Rare Diseases in EU Member States or the development of European Reference Networks (ERNs) for Rare Diseases<sup>1</sup>.

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

With the Expert Groups having ceased to exist and the imminent end of the Joint Action for Rare Diseases, RD-ACTION, in 2017 EURORDIS undertook different initiatives to prepare for the next decade of rare disease policy making. Taking stock of nearly twenty years of advances in support of rare diseases, emerging needs and challenges need to be addressed and new impulsion at European level is necessary to ensure that they are addressed with adequate actions.

### Maltese Presidency of the EU Council



In 2017, EURORDIS started to advocate for the adoption of a new policy framework that, along the lines of the 2009 Council Recommendation on an Action in the Field of Rare Diseases and the 2008 Commission Communication, could address those needs.

In early 2017, a group of key Members of the European Parliament sent a letter to the Presidency of the EU Council, inviting the Council to call for a new Council Recommendation.

The **Maltese** government, holding the **Presidency of the EU Council** in the first half of the year, gave prominence to rare diseases in the agenda of their Presidency and in the Council meeting held in La Valletta, on March, 20th. On that occasion, the Maltese Presidency also organised two side events on respectively integration of healthcare and research, and development and access to medicines for rare diseases, that EURORDIS helped organise. A delegation of European patient representatives participated in these high level meetings.

In Malta, EURORDIS launched a Declaration on rare diseases calling for improved mechanisms of structured cooperation across EU Member States to address the unmet needs of people living with a rare disease.

A first result of these efforts was the adoption in June 2017 of the **EU Council Conclusions** on voluntary cooperation between health systems, with a number of provisions relevant to rare diseases and the call on the European Commission to inform the Council about the state of implementation of the Council Recommendation and the Communication on rare diseases.

<sup>1</sup> All reports and recommendations produced by the European Commission Expert Group on Rare Diseases 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)

Following the Maltese events and with the aim to identify more accurately the long-standing and emerging needs of people living with rare diseases and the actions that would be necessary at the EU level to address them, EURORDIS initiated a broad consultation with its National Alliances during the two meetings of its Council of National Alliances held in 2017, which led to the creation of a **Working Group on Future Policy Priorities for Rare Diseases**, composed of members of National Alliances selected after having participated to a call for expression of interest. The outcomes of this process of identification of policy

priorities will be presented at the European Conference on Rare Diseases (May 2018, Vienna).

In parallel, EURORDIS followed the initial discussions within the European Commission on the next budgetary period that will lead to the adoption of the new **EU Multiannual Financial Framework 2020-2027** that will fund EU policies and programmes and prepared the reflections on priority areas for funding at the EU level. Such reflections will feed into the stakeholder consultation that European Commission plans for early 2018.

### Parliamentary Advocates for Rare Diseases

In 2017, EURORDIS continued the development of the concept of a parliamentary group on rare diseases, which culminated in the creation of the network of Parliamentary Advocates for Rare Diseases in October 2017, when a launch event was held at the European Parliament on “Juggling Care and Daily Life: the Balancing Act of the Rare Diseases Community” which included a presentation of highlights of the results of the first European survey on the impact of rare diseases on daily life, as carried out through Rare Barometer Voices, the EURORDIS survey initiative.

Through the network, EURORDIS aims to bring together members of parliament to ensure strong international and local action, shape political input for current and future legislation, and integrate rare diseases into all relevant policies at all levels.

In 2017, the core group of Parliamentary Advocates for Rare Diseases was identified among Members of the European Parliament (MEPs) who have been long standing advocates of the rare disease cause, but also –with the essential contribution of EURORDIS’ National Alliances– among MEPs who showed interest and committed to implement concrete actions in support of people living with rare diseases within the political agenda.

After the launch event, EURORDIS prepared a brainstorming meeting with parliamentarians to be held in January 2018 aimed to identify priority actions and initiatives that the network could carry out before the end of the legislative term (mid-2019). The networks of Parliament Advocates for Rare Diseases is planned to subsequently expand to include members of national and regional parliaments, in collaboration with National Alliances.



### Foresight Study on Rare Diseases in 2030

In 2017, EURORDIS continued the cooperation with the MEPs who supported the adoption of a Pilot Project at the European Parliament EURORDIS 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”). With their support and following the approval of the Council, the project was secured to the EU budget for two years of funding. In December 2017 the European Commission announced a call for proposals (expected in early 2018) to grant the funding for the Study.

### 1.3.1 2nd 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:** 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.

In addition, EURORDIS together with its member National Alliances for rare diseases, continue the EUROPLAN process, started in 2008, to foster the implementation of national plans or strategies for rare diseases, and sometimes their adoption in some EU Member States.

In 2017, National Alliances and EURORDIS organised 13 EUROPLAN conferences in Austria, Belgium, Croatia, the Czech Republic, Denmark, Hungary, Luxembourg, Macedonia, Poland, Romania, Serbia, Spain and Ukraine. The short reports highlight the key recommendations stemming from these conferences:

<http://www.rd-action.eu/europlan-rd-national-plans-conferences-reports/>

#### Work Package 6: Rare Disease Policy

In this WP led by Newcastle University, EURORDIS is a main partner to propose policy priorities to the consultative group of RD-ACTION. Policy priorities identified by the Consultative Group and then implemented within the RD-ACTION with dedicated workshops, focus on different

key aspects of the development and then deployment of European Reference Networks (see in the separate section on ERNs of this Report) EURORDIS also coordinates the contribution of National Alliances to the national sections of the State-of-the-Art report on rare diseases.



### 1.3.2 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 and 26 collaborating partners involved in the JARC including Ministries of Health, Cancer Control Programme representatives, universities, public health institutions, cancer registries, oncological institutes, research societies and 3 patients' organisations: ECPC – European Cancer Patient Coalition, CCI-E – Childhood Cancer International – Europe, and EURORDIS.

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.

The JARC is particularly involved in supporting the development of European Reference Networks for rare

cancers, namely EURACAN (solid tumours in adults), PaedCan ERN (paediatric cancers) and EuroBloodNet (including haematological malignancies in adults). It has also engaged dialogue with the ERN GENTURIS (Genetic Tumour Risk Syndrome).

EURORDIS teams up with ECPC and CCI-Europe to represent the patients' perspective in the work and future recommendations of the JARC. EURORDIS' specific mission is to: bring its expertise on the development of European Reference Networks involving elected representatives of European Patient Advocacy Groups (ePAGs) for the four above mentioned ERNs; make a link with ongoing work within RD-ACTION; identify synergies and areas of stronger collaboration between rare disease national plans and cancer national plans in EU Member States.

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

After having participated in 2016 in two public consultations launched by the European Commission addressing specific aspects of the Regulation (EC) n° 141/2000 on orphan medicinal products, in 2017, EURORDIS continued to advocate to improve the regulatory process for medicines for the patients it represents.

In particular, EURORDIS participated to a consultation on the experience acquired with the **Paediatric Regulation** (Regulation (EC) No 1901/2006 of the European Parliament and of the Council on medicinal products for paediatric use) launched by the European Commission to obtain views and feedback from stakeholders and 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. EURORDIS is in favour of a better implementation of the Regulation to develop medicinal products for paediatric use. In October 2017, the European Commission presented to the European Parliament and the Council a comprehensive report on progress made in children's medicines 10 years after the Paediatric Regulation came into force.

Following these consultations, the European Commission has launched in December 2017 the process for a **joint evaluation of the legislation on medicines for children and rare diseases**. The purpose of the evaluation is two-fold:

- 1 It will give an assessment about the strengths and weaknesses of the two pieces of legislation on orphan medicinal products and on paediatrics medicines separately and combined. It will focus on the output and results of the two regulations: in what respect have patients' needs been fulfilled, what have been the societal consequences and what has been the synergy between the two.
- 2 It will also focus on the cost-effectiveness when providing the incentives and rewards incorporated in the legislation and how they have been used in practice. And it will make a cost-benefit analysis, both from a general point of view and per group (patients, industry, payers etc). The evaluation will give a sound evidence base about the functioning of the two legal instruments from a public health and a socio-economic perspective that will be used to consider the possible need for any future changes.

This evaluation will run most likely until 2019, and it will build upon other ongoing or completed studies, such

as the above mentioned 10-year progress report on the Paediatric Regulation, and the ongoing Commission study on pharmaceutical incentives, which includes an analysis of impact of pharmaceutical incentives (such as data/market protection, market exclusivity for orphans and paediatric rewards) on innovation, availability and accessibility of medicines. This analysis relies on two studies on respectively the legal framework for Supplementary Patent Certificates (SPCs) and the impact of pharmaceutical incentives and SPCs on innovation, availability and accessibility of medicinal products (including data / market protection and market exclusivity for orphans and paediatric medicines). EURORDIS has provided contribution in 2017 to the two studies, due to be published in 2018.

In this context, EURORDIS planned and started to organise for the first ERTC workshop of the year in 2018 a full session on incentives and their role in providing support for the development of therapies for rare diseases.

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### 1.3.4 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 alia, from the large diversity of compassionate use schemes between countries (some do not have a regulatory scheme), and/or the willingness of the company to initiate one, or the lack of information on these programmes.

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

At the post-authorisation phase, obstacles come, inter alia, from the delays in deciding if the medicine should be reimbursed/covered and for whom, following the health 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 "Early Access to Medicines in Europe: Compassionate use to become a reality" was adopted by the EURORDIS Board of Directors in March 2017.

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.

Its dissemination, apart from EURORDIS working groups and members (with a webinar on 19 October with 58 participants) and an article in eNews, targeted the European Medicines Agency, the secretariat of the Heads of Medicines Agency, 86 contacts in national regulatory agencies, the European Commission. The position was presented at the Commission Expert Group on Safe and Timely Access to Medicines for Patients (STAMP) on 27 June, at Patients' and Consumers' Working Party (20 September), EMA, and at the FDA/EMA Rare Diseases Cluster (21 September). It was also sent to press and some interviews were conducted.

### At the authorisation phase

**Medicine Adaptive Pathways to Patients (MAPPS)** has continuously been promoted by EURORDIS since 2012. 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 continued to co-lead the definition of the Engagement Criteria for a product to enter this new development path. In 2017, 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. This project will be finished in April 2018 and EURORDIS will continue its activity in this field in the near future.

## 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 (when such a medicine exists and received market authorisation) 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.

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



The second Multi-Stakeholder Symposium aimed at bringing together stakeholders playing a key role in getting medicines and therapies to rare disease patients, and to continue a dialogue and cooperative process that respects the interests of all parties and that will lead to solutions for improving patients' access to rare disease therapies.

On 22-23 February 2017, nearly 400 participants including patient advocates, academics, clinicians, policymakers, regulators, investors, members of the EURORDIS Round Table of Companies and other representatives from the healthcare industry, as well as from payers and health-technology-assessment authorities, took part in the Symposium. This edition, which built on the first EURORDIS Multi-Stakeholder Symposium held in February 2016, culminated with the intention to form of a new multi-stakeholder group that will draft a Plan of Action for all stakeholders to collaborate on improving patients' access to medicines, using the 'one-text' process as a method. This process will aim to enhance and sustain trust between the various stakeholder groups, a fundamental condition to achieving this ultimate goal.

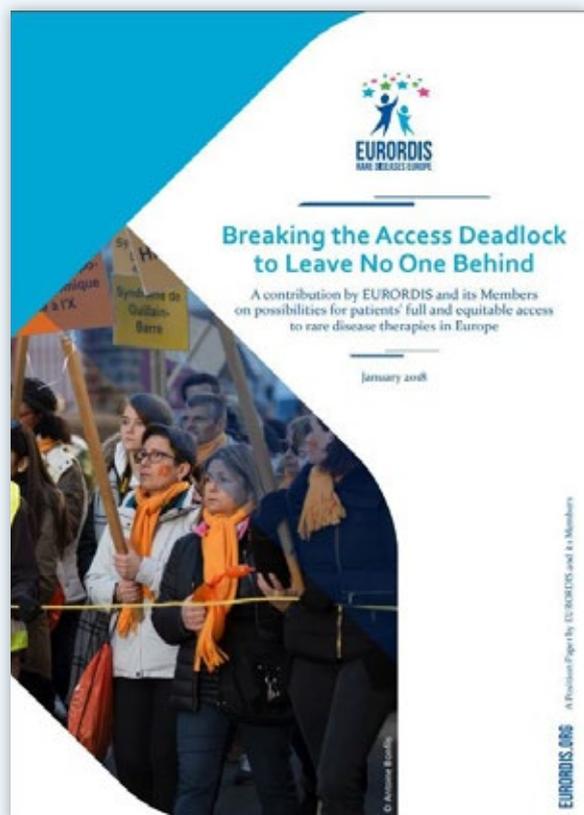
During the Symposium, it emerged that a multi-stakeholder approach is necessary to improve access to rare disease therapies. The issue calls for all the stakeholders to ally and come together if the hope of improving conditions is to be achieved. During the closing to the Symposium, the call for an approach driven from the bottom-up but supported from the top-down was issued by participants. This approach requires 3 key elements:

- + Mutual trust: both to take risks and to discuss cost and price.
- + Will: from all stakeholders (political, private and public sector).
- + Investment: in research, people and initiatives.

In addition, the current socio-political framework, in which notions of solidarity, universal health access and universal Sustainable Development Goals are bearing principles, needs to be utilised to make all stakeholders accountable and together, reach the goals of improved access to medicines for people living with a rare disease in Europe.

## Breaking the Access deadlock: from reflection to position

At this second Multi-Stakeholder Symposium, EURORDIS released a work-in-progress reflection paper ahead of the Symposium entitled 'Breaking the Access Deadlock to Leave No One Behind'. The paper offered a synthesis of EURORDIS' analysis, reflections and perspectives on the issue of access to medicines for people living with a rare disease. It expressed a set of possibilities rather than a position and is open to discussion.



Building on the proceedings of the second Symposium, and with further rounds of feedback collected from EURORDIS' members and the rare diseases community, the Board of Directors of EURORDIS in November 2017 adopted the document as a position paper "Breaking the Access Deadlock to Leave No One Behind" calling for urgent change to ensure patients' full and fast access to rare disease therapies in Europe and to tackle the challenges that prevent patients' access to care and medicines, as well as the ambition to have 3 to 5 times more new rare disease therapies approved per year, 3 to 5 times cheaper than today by 2025.

In the paper, EURORDIS calls for a new model based on a collective conversation involving all stakeholders (patients, the pharmaceutical industry, national competent authorities, national health ministries, researchers, scientists and regulators). The new position paper sets out a four-pillar approach that encompasses:

- 1 A new blueprint to cut costs and fast-track R&D;
- 2 Early dialogue and cooperation between healthcare systems on the determination of value of a medicine and on patient access;
- 3 A transparent European cooperation framework between national healthcare systems for the determination of fair prices and of sustainable healthcare budget impacts; and
- 4 A continuum approach to evidence generation linked to healthcare budget spending.

The paper also dispels misconceptions around the pricing of orphan medicines and sets out the facts on marketing authorisation and pricing of rare disease medicines.

## 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).*

Since end 2015, EURORDIS has put in place a framework for patient engagement in the MoCA procedures. EURORDIS takes care of identifying and supporting these patient experts and is also attends these meetings in person.

For each new pilot, EURORDIS' staff review the company's proposal, identify and brief the patient representatives, and provide them with administrative support and support them during the actual meeting.

EURORDIS' staff is also involved in the MoCA Steering Group, together with the Public Affairs Director, in order

to ensure the building of a sustainable framework for patient engagement in these dialogues with industry and payers. A dedicated webpage on the EURORDIS website has also been created to keep members and stakeholders up-to-date about the MoCA developments and rules of procedures.

## 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 and leading 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" that was discussed at the 2nd EURORDIS Multi-Stakeholder Symposium on Improving Patient Access to Rare Disease Therapies in February 2017.

In the same context, we have contributed to the development of a new multi-stakeholder initiative to focus on 'RealWorld Evidence to address uncertainties in complex or rare conditions that require highly specialised treatment in order to improve access to rare disease therapies', with an initial scoping meeting held in December 2017. This initial scoping meeting brought together a number of stakeholders, including patient representatives, payers, national health ministries and HTA bodies, research organisations and industry. The aim of the initiative is to collaboratively develop a paper to increase the trust of all stakeholders in evidence generation pathways.

EURORDIS also contributed to the work of the European Working Group for Value Assessment and Funding Processes in Rare Diseases (ORPH-VAL), which produced a set of Recommendations to help improve the consistency of pricing and reimbursement decisions on orphan medicines in Europe. Professor Lieven Annemans, the chair of the ORPH-VAL working group provided an overview of the 9 principles and the results from an assessment of the alignment of the principles with the P&R systems currently used in Germany, France and UK to assess OMPs at ISPOR 2017.

Building on the successful collaboration developed with the orphan therapies specialised consultancy Dolon, in 2017 we have initiated and scoped out a patient-led, collaborative, pre-competitive dialogue on patient access to gene and cell therapies for rare diseases in Europe, focusing on two issues: 1) Pricing and reimbursement (P&R), HTA, funding/financing and broader payer-related challenges to patient access to gene and cell therapies; 2) Practical requirements to ensure patients have effective access to these therapies. The ultimate goal is to identify future financing and access requirements to sustainably provide patient access to innovative therapies.

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

#### **HTA Patients' Involvement & Mentoring Programme in EUnetHTA**

The third EU Joint Action on HTA (EUnetHTA JA3) decided not to have a structured interaction with stakeholders as it was in the previous Joint Action (EUnetHTA JA2) with the EUnetHTA Stakeholder Forum, but it committed to involve patients as experts in its scientific assessments and other activities.

Without a framework for the involvement of patients, EUnetHTA asked EURORDIS and other organisations of the Stakeholder Pool or other European umbrella organisations to identify eligible patients for joint HTA procedures or Early Dialogues.

In 2017, 6 requests of patients' identification were made: 2 for rapid HTA for pharmaceuticals where patients could participate (Midaustorin for acute myeloid Leukaemia,

Alecensa for advanced non-small cell lung cancer) and one where no patient could be identified on time (Regorafenib for liver carcinoma). Consultations of patients were conducted by telephone interviews. For rapid HTA of medical devices, no patients could be identified (3 requests received: one for femtosecond laser-assisted cataract surgery, one for stool DNA testing for early detection of colorectal cancer and one for C-reactive protein point-of-care testing (POCT) to guide antimicrobial prescribing in primary care for respiratory tract infections).

#### **EURORDIS actions related to the EC Proposal for a Regulation on the European HTA cooperation**

In response to the European Commission public consultation on how to strengthen the EU cooperation on HTA, EURORDIS emphasised the need for the mandatory use of European Joint Clinical Assessments, for the mandatory participation of industry, for the consideration of patient aspects and organisational aspects when needed in the joint assessments, and for a responsible role of patients' organisations in HTA.

E-meetings with members were organised both to present the consultation and then to explain its outcomes.

EURORDIS' views were presented at the I-COM meeting at the European Parliament (8 February), the European Academy of Law conference on pharmaceutical law (23-24 February), the anniversary of the Spanish HTA coordination in Zaragoza (27-28 April), the HTAi annual conference in Roma (17-21 June), at a DIA conference in Basel (25-26 October).

As member of the HTA Stakeholder Pool created by the European Commission to interact with the HTA Network, EURORDIS contributed to the writing and adoption of two documents: Principles for the Engagement of Patients and Consumers in HTA and Criteria for the prioritisation of technologies for joint assessment.

The joint workshop of the Council of National Alliances (CNA) and the Council of European Federations (CEF) in November in Paris included a presentation on Last developments in EUnetHTA and on the future Legislation on HTA and on Patient engagement in HTA.

### **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.

The EMA created a catalogue of shortages for pharmaceuticals authorised via the centralised procedure only. The catalogue can be consulted on the EMA web site ("Shortages catalogue"). For all shortages affecting medicines to treat rare diseases, the EMA consults EURORDIS on the information for the public.

**Off-label.** Following a first survey on off-babel 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.

Based on this survey, EURORDIS responded to the Study on off-label use of medicinal products in the European Union<sup>2</sup>, conducted by Nivel, the National Institute for Public Health and Environment (Netherlands) and EPHA, on behalf of the European Commission.

The study covers the public health aspects related to the off-label use of medicinal products. In particular, it investigates the balance between the benefits and risks that off-label use has for patients, and the regulatory framework for the off-label use of medicines. Applying a wide range of methods, including a systematic review of scientific literature and grey literature, a legal analysis, interviews with stakeholders and an expert meeting, the study provides information on a variety of aspects of off-label use. These include the prevalence and incidence of off-label use and its drivers as well as a description of the national frameworks, regulatory and other, governing off-label use of medicinal products in the various EU Member States. A factual analysis is provided of how authorities have addressed the issue of off-label use and the different ways patients, healthcare professionals and industry react to this. The report does not provide any recommendations.

### 1.3.5 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:*

<https://www.eurordis.org/access-campaign-participate>

The EURORDIS Access Campaign survey is a permanent process, data will be analysed every three years as new reports come in.

### 1.3.6 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 2017, 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 and in the Innovative Medicines Initiatives.
- + Continued supporting rare diseases as a public health priority in the 3rd EU Public Health Programme 'Health for Growth' following the mid-term review of the Programme. In a dedicated event at the European

Parliament, EURORDIS advocated in favour of the added value of EU co-funded cross-border cooperation, such as in Joint Actions, and the crucial role of Operating Grants to maintain the independence of health NGOs and their active engagement in patient-centred policy making at the EU level.

- + Following the adoption of the White Paper on the Future of Europe by the President of the European Commission, Jean-Claude Juncker, which includes a scenario whereby health policy would no longer be pursued at the EU level, EURORDIS joined a broad campaign of EU health stakeholders calling for a continued and enhanced EU action in the field of health (#EU4Health) The campaigners also demanded not to dismantle a dedicated EU funding programme for health in response to the dismantling of the programme which seems to be contemplated.
- + Promoted and took an active part in the development and negotiations on 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. To gather support for the initiative and promote the added value of such a collaborative approach, in March 2017, under the auspices of the Maltese Presidency of the EU Council, EURORDIS co-organised in Valletta a meeting with key stakeholders on "Integrating Research and Healthcare for Rare Diseases: A structured cooperation with high community added value".

### 1.3.7 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 (EPF) in the monitoring of the implementation of patient's rights' across the EU and at national level. after contributing to the development and adoption of the EPF Position Statement on the Directive on patients' rights in cross-border care, EURORDIS continued to participate in follow-up work to shed light on the shortcomings of the implementation in many Member States, on the low awareness among EU citizens of their rights and

<sup>2</sup> [https://ec.europa.eu/health/sites/health/files/files/documents/2017\\_02\\_28\\_final\\_study\\_report\\_on\\_off-label\\_use\\_.pdf](https://ec.europa.eu/health/sites/health/files/files/documents/2017_02_28_final_study_report_on_off-label_use_.pdf)

on what needs doing more urgently from the patient perspective. In December, EURORDIS joined a Round Table organised by EPF on the implementation of the Directive on cross border patients' rights.

+ Throughout 2017, EURORDIS continued to receive enquiries from individual rare disease patients and families on their specific experience in the provision of care across borders and the enforcement of their patients' rights under the EU legislation, and to provide ad hoc advice.

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



On 1st March 2017, following at least a decade of legislative groundwork and preparation across the European Union, 24 European Reference Networks, a new EU infrastructure created with the sole purpose to improving access and quality of diagnosis, care and treatment for the 30 million people living with a rare, complex disease or needing highly specialised healthcare, were successfully launched. The ERN Conference in Vilnius, held on 9-10 March 2017, was a celebration of the success of patient and clinical partnership, seeing the formal launch of all 24 ERNs as a historical landmark event for the rare disease community.

The first year of ERN implementation focused on the investment of expertise, capacity and commitment of the European Commission, Member States and ERN Members (both clinicians and patients) to establish the ERN governance structure and infrastructure. To enable the interoperability and connection of over 300 hospitals across 26 Member States, making the first tangible steps to tackle the needs and inequalities in rare diseases.

#### Operational implementation

EURORDIS supported the establishment ePAGs, newly formed forums of patients aligned to the scope of each ERN, building a threefold increase of ePAG patient representatives and strengthening their capabilities. EURORDIS supports ePAG representatives through regular virtual meetings, to be active in each ERN kick-off meetings and network meetings throughout the year. EURORDIS was active in the ERN kick-off meetings to foster a culture of partnership and developed an ERN Patient Involvement Guide as a practice aid for both clinicians and patient representatives to harness their meaningful involvement in the ERNs.

#### Engagement and capacity building

EURORDIS strongly believes that a strong partnership between ERNs and the rare disease patient community, aligning needs and harnessing our collective knowledge and experiences, will optimise the opportunity ERNs hold for our community. EURORDIS remains committed to demonstrating 'proof of concept' of patient involvement in ERNs and has established a leadership programme consisting of regular webinars on ERNs (outcomes and indicators, eHealth and data sharing); piloted a mentoring programme for ePAG representatives; and held two ePAG face to face capacity building workshops, one in Budapest in May and one Paris in October. Two key outcomes resulting from these workshops was to establish an ePAG Steering

Committee in addition to topic focused transversal peer learning groups aligned to ERN priority areas.

EURORDIS continued to reach out to the wider patient community, to raise awareness and inform of the developments of ERNs. It also started to support National Alliances through webinars focusing action on anchoring ERNs into national health system and engaging with their members through a series of an engagement workshops, which is continuing in 2018.

#### Technical and strategic preparation

EURORDIS continues to engage with all actors involved in ERNs, advocating on important issues and topics for the rare disease community and facilitating the preparation for the full deployment of ERNs. More specifically, this included: the managed expansion of ERN membership in a stepwise approach, firstly addressing gaps in expertise and geography; the initiation of the dialogue and collaboration between ERNs & the industry community through our 25th ERTC (EURORDIS Round Table of Companies) workshop in Barcelona; the centralisation of patient experience survey for all ERNs to build consistency and robust approach; the support to HCP leads to have capacity to act and be active in ERNs through raising awareness within their hospital, through supporting EC Hospital Directors workshop and ERN symposiums; and, the engagement with MEPs in funding prioritises for ERNs in the preparation of setting MFF 2020-2025.

EURORDIS is an RD-ACTION partner and participated in RD-ACTION workshops, which were co-organised with DG SANTE and the ERNs, securing funding for ePAG fellowships. These included the following workshops: Using Standards and Embedding Good Practices to Enable Interoperable Data-Sharing in ERNs' Workshop (April 2017), Indicators and Outcomes for ERNs (June 2017) and Clinical Practice Guidelines' Workshop (Dec 2017). EURORDIS was also an active member in the ERN Network Coordinator' Monitoring Working Group and Clinical Guidelines Working Group, contributing to the development of the ERN Monitoring Framework and starting the development of the ERN methodologies for the development of clinical practice guidelines.

### 1.3.9 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 Horizon 2020 Work Programme 2016-2017 on health research:

- + "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 for Rare Diseases (EJP for RD)

In view of the publication of the EJP for RD within the Work Programme 2018-2020 (Oct 2017) supported by EURORDIS' advocacy activities, the first and second concept papers describing the proposed approach, consortium and draft content of the planned activities were presented to the European Commission and Member States representatives in April and October respectively. EURORDIS is co-leader of the training activities within the consortium and therefore a key member of the Operating Group leading on the development of the proposal to be submitted in April 2018. 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

The JRC released the first interoperability tool of the EU RD Platform: the "Set of Common Data Elements for RD Registration" in April 2017. According to the needs expressed by stakeholders, the Set of Common Data Elements constitutes the first important building block for the EU RD Platform. It is the result of a dedicated Working Group composed of experts from projects related to common data sets: EUCERD Joint Action, EPIRARE and RD-Connect, as well as the JRC's EU RD Platform team. The Set is recommended as a constitutive element for all RD registries in Europe. This first step towards interoperability of registries is being offered to the European Reference Network's (ERNs) existing registries and registries under development and to all other RD registries at national, regional, local level in the Member States, to patient organisations, researchers and healthcare providers, thus covering the whole range of the EU RD Platform's stakeholders.

The JRC has organised in 2017 the first training sessions on the use and implementation of the "Set of Common Data Elements" and the interoperability tools under development for all interested RD Registries. EURORDIS supports and encourages implementation of the JRC Set of Common Data Elements through our activities and interactions with the ERNs, advocating for its future implementation within the new EU IT Platform and for the addition of a mandatory requirement in all EC funded registries through Health and Research programmes.

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

Throughout 2017, EURORDIS continued to advocate for improved access to and quality of rare disease diagnosis.

### New collaborative H2020-funded projects on diagnostic characterisation of rare diseases

A large consortium led by the University of Tübingen, the Radboud university medical center Nijmegen and the University of Leicester has successfully acquired a € 15 million grant for the Solve-RD research program. The consortium in which EURORDIS is a partner will use the funding to improve the diagnosis of rare diseases. Solve-RD echoes the ambitious goals set out by IRDiRC to deliver diagnostic tests for most rare diseases (RD) by 2020 and fully integrates with the formation of ERNs. The main ambitions of the project are:

- + to solve large numbers of RD, for which a molecular cause is not yet known, by sophisticated combined Omics approaches, and
- + to improve diagnostics of RD patients through contribution to, participation in and implementation of a "genetic knowledge web" which is based on shared knowledge about genes, genomic variants and phenotypes.

The entire Solve-RD project will be placed into the newly developing ERN environment which opens a unique window of opportunity for substantial progress for diagnostic research on unsolved RD cases. The 5-year project will start in January 2018.

## Patients without a diagnosis

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). The last network meeting was held at the EURORDIS office in Barcelona in March 2017 confirming current membership and sharing best practices and relevant activities. The official press release announcing the establishment of the network was published on 28th April 2017 on "Undiagnosed Children's Day" (<https://www.undiagnosed.org.uk/news-events/news/introducing-swan-europe/>).

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 attended the annual meeting held in September 2017 in Stockholm. NORD, EURORDIS and the Wilhelm Foundation have collaboratively developed a patient engagement working group which was adopted by for the board of the UDNI. As UDNI matures and expands further, a Board Committee for Patient Engagement will be created within the next 2 years. This suggestion was accepted by the consortium. In addition, EURORDIS presented the perspectives of rare disease patients on large-scale genomic data sharing for research collected within the framework of the RD-Connect project. EURORDIS is a member of the UDNI data sharing working group also created during this last meeting.

In 2017, EURORDIS created a dedicated webpage on its website to promote its activities related to undiagnosed rare diseases and provides relevant information and resources for the undiagnosed and rare patient community: <https://www.eurordis.org/content/undiagnosed-rare-diseases>

## 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. Downstream communication of the project's activities has further improved 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 perspectives and concerns of rare disease patients on the General Data Protection Regulation (GDPR) which were presented by EURORDIS at several workshops throughout 2017 organised by Microsoft, the EU Cloud in Health Advisory Council, EPF, E-Rare, BBMRI-ERIC as well as DG CONNECT, DG JUST, DG SANTE and DG RTD.

## Patient involvement in Biobanks & Registries

**Biobanks:** EURORDIS is a member of the **BBMRI Stakeholder Forum** and participated in several meetings in 2017 (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.

### Registries:

- + EURORDIS participated in the rare disease registry multi-stakeholders workshop organised by CHAFAE and CIBERER in Madrid on 21-22 March by giving a presentation on EURORDIS' projects and advocacy activities related to rare disease patient registries, explaining why patient groups should be involved in registries.
- + EURORDIS participated on 18-22 September 2017 in the 5th International Summer School for rare diseases and orphan drug registries organized by ISS (the Italian Institute for Health) in Rome. EURORDIS presented the planned activities of the ERNs on research and registries highlighting the specific roles of the European Patient Advocacy Groups (ePAGs) involved in the 24 ERNs.
- + Through RD-Connect and the activities of the Patient Advisory Council chaired by EURORDIS within the project, EURORDIS participated in the development of a manuscript for academic publication on the quality aspects and criteria specific for rare disease patient registries.

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### 1.3.11 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 two main initiatives: 1) Rare Disease International (RDI), and 2) the NGO Committee for Rare Diseases.

## 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.

Rare Diseases International continued to grow and promote rare diseases as an international public health priority reaching 49 members by the end of 2017: 27 national alliances of rare disease patient groups and 11 international disease-specific federations of patient groups, 4 pan-regional networks, as well as 7 associate members.

EURORDIS is an elected member of the RDI Council and continued to host the initiative. To mark the occasion of Rare Disease Day 2017, Rare Diseases International, in partnership with EURORDIS-Rare Diseases Europe, held a unique policy event in Geneva, Switzerland, co-organised with the BlackSwan Foundation, gathering international experts in the fields of public health, human rights, epidemiology, scientific research and patient advocacy to discuss why and how rare diseases should be included in the global health agenda. Over 150 people attended and speakers included representatives of the World Health Organization (WHO), the UN Development Programme (UNDP), the UN Secretary General's High-Level Panel on Access to Medicines, the Organisation for Economic Co-operation and Development (OCDE) and the International Rare Diseases Research Consortium, as well as the Special Rapporteur to the UN on Health and patient representatives from around the world.

RDI also organized its third annual meeting in Barcelona, Spain, in June 2017. Over 50 participants from 23 countries were able to network with patient advocates from around the world, learn more about recent developments in international rare disease advocacy and receive information to become further involved in RDI activities.

15 RDI fellows from India, Malaysia, South Africa and six Latin American countries, stayed on to attend the EXPRESS 2017 Expert Patient and Researcher EURORDIS Summer School (English and Spanish versions) that took place in Barcelona the week following the RDI annual meeting (June 5-9, 2017).



**RARE  
DISEASES  
INTERNATIONAL**



## NGO Committee for Rare Diseases



**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 in 40 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. 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 order to achieve its goals, the Committee aims to act as a forum of interested parties such as NGOs from the field of rare diseases and beyond, United Nations bodies and agencies, as well as individual experts. 2017 represented a period of reflection and mapping out of key stakeholders and advocacy platforms within the United Nations. November 2017 marked the official creation of the Inception Executive Board of the Committee, with officers from key organisations: Ågrenska, EURORDIS-Rare Diseases Europe, International Alliance of Patients' Organizations, International Alliance of Women, International Federation for Spina Bifida and Hydrocephalus, Word Federation

of Hemophilia, and with the global patient voice being represented through Rare Diseases International (RDI), the global alliance of people living with a rare disease of all nationalities across all rare diseases.

The year 2017 was also marked by the attendance and networking at a number of official United Nations events, including the Conference of State Parties to the Convention of People with Disabilities (June), the Open-Ended Working Group on Ageing meeting (July), the High-Level Political Forum on Sustainable Development (July), the Human Rights Council (September) and the International Day of

People with Disabilities (December). The Committee also collaborated in the first policy event of RDI 'The Right to Health: The Rare Disease Perspective' in February 2017 in Geneva. In addition, a number of working meetings in collaboration with RDI were also organised with a number of Permanent Missions to the UN and officials of the World Health Organisation in Geneva in November 2017. This preparatory work will pave the way for the future actions taken by the Committee, principally at the level of the World Health Assembly and the Human Rights Council, in 2018 and 2019. The Committee also plans to hold a high-level meeting on rare diseases in New York in early 2019.

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

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

*Rare Barometer consists of surveys aiming to collect qualitative & quantitative data on the experiences, needs & expectations of RD patients and their families 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, is the tool used to carry out quantitative surveys. Rare disease patients can register from all over the world. The webpage and the surveys are translated in 23 languages.*

Throughout 2017, Rare Barometer Voices reached more than 7000 patients and represented 83 countries in the world. Four quantitative and one qualitative surveys were carried out. Each survey was designed in collaboration with a Topic Expert Committee composed of experts on the topic, including other patient organisations. Three survey reports and an infographic were drafted and disseminated

throughout EURORDIS' communication channels and networks. The possibility to sort each survey carried out by ERN grouping was developed (results can also be sorted by country and disease when possible).

Surveys results were presented in several events and conferences, including EURORDIS's workshops at the Membership Meeting in Budapest, EUROPLAN workshops, and the Launch of European Network of Parliamentary Advocates for RDs at the European Parliament.



# 2. PATIENT EMPOWERMENT:

## Building the network & building capacities

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

#### 2.1.1 Membership

57 new members joined EURORDIS in 2017 with the addition of the following new countries: Albania, Israel, Moldova and Montenegro.

At the end of 2017, EURORDIS had 779 members in 69 countries, 42 of which are European countries, 28 being members of the European Union.

EURORDIS Membership Meeting 2017 Budapest



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### 2.1.2 EURORDIS Membership Meeting 2017 Budapest

*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 2017 Budapest took place May 19-20. The programme committee of EMM 2017 was made up of 8 members, including EURORDIS Directors and staff. The event attracted 250 patient representatives from 37 countries. Following the General Assembly, a plenary sessions focused on "Building on Success of 20 years of advocacy for rare diseases". Saturday 20 May was dedicated to 4 workshops: European Reference Networks (ERN) and ePAGs, Natural, alternative, traditional and complementary therapies, Survival kit for small patient organisations, Social Revolution.

EURORDIS offered 40 travel fellowships to Patient Advocates from 18 countries.



Council of National Alliances (CNA)

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### 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 41 national alliances, 34 of which constitute the CNA. The CNA's main activities in 2017 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 ERN
- + the preparation and coordination of the Rare Disease Day 2018
- + the launch of the Parliamentary Advocates for Rare Diseases, a network of European and national members of parliament advocating to improve the lives of people living with a rare disease

### 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 October to discuss issues that are important across Europe and across diseases. For the fourth time, part of the meeting took place in conjunction with the CNA (Council of National Alliances) to discuss cross cutting issues and share experiences. This part of the meeting focused on: RareConnect; Rare Barometer; Orphacode: implementation of RD coding; Development of ERN & ePAGs; European Pillar of social rights; EURORDIS Position paper on social rights.

The second part (CEF only) focused on: E U N e t H T A ; Patient engagement in scientific advice; HTA done properly: the Scottish example; Patient engagement in EMA scientific advice.

The second day of the meeting was dedicated to a training on Community Advisory Boards (CABs).

EURORDIS continued for the 8<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 2017, EURORDIS gave 13 European RD Federations financial support to help them organise their different meetings. A total of 22750 € was granted for 15 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. There are 17 help lines which are members of the European Network.*

In 2017, 13 help lines from 11 countries participated in the activities: Bulgaria (ICRDOD), Croatia (Croatian help Line for rare Diseases), France (Maladies Rares Info Services), Italy (Coordinating Centre for Rare Diseases Veneto Region, and Telefono Verde Malattie Rare), Portugal (Linha Rara), Romania (NORO, Myastenia Gravis Romania), Spain (SIO-Feder), 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 participating in a face-to-face training on pharmacovigilance and how to take complex calls, organised in London, 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.



Help Lines  
for Rare  
Diseases



## 2.1.6 European Patient Advocacy Groups (ePAGs)

*European Patient Advocacy Groups (ePAGs) were launched in 2016 while the ERN network applications were being developed in answer to the EC Call for ERN Applications. 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.*

In 2017, EURORDIS organised two ePAG face to face meetings. The first meeting was held on 18 May and marked the first time ePAG representations from all 24 European Reference Networks came together to share their expectations and experiences at the first ePAG Satellite Meeting, held prior to the EURORDIS Membership Meeting 2017 Budapest. The meeting was attended by 37 ePAG representatives and was an exciting and informative day for all, coming together as a community and making our first steps towards cross-ERN collaboration.

The second meeting took place on Monday 18<sup>th</sup> December 2017 at EURORDIS headquarters in Paris. Close to 40 ePAG representatives from the 38 European Reference Networks were present in Paris and many more participated online. The face to face meeting was organized at the request of ePAG representatives specifically to work together, across all ERNs, to take stock of progress to date and discuss critical topics together, agreeing on the most effective way to progress the ePAGs in 2018.

During the meeting three task and finish groups made up of ePAG representatives were created: Defining the roles and responsibilities of ePAG representatives as well as discussing a code of conduct; Communicating to the wider patient community; Expanding the ePAG membership.

The outputs of the three groups will be used to further develop the Terms of Reference in 2018 and adapt them to the current situation of the ERNs and ePAGs. In addition to the meetings organized by EURORDIS, EURORDIS facilitated ePAG representation and ePAG satellite meetings at 23 ERN board and kick off meetings throughout 2017.

EURORDIS held around 77 conference calls with the 24 ePAGs and ERNs throughout the year to discuss topics of relevance with the ERN leads and ePAG representatives. Much of this focused on strengthening and formalising patient engagement in ERNs such as the development of patient boards and the recruitment of ePAG representatives.

Another priority activity area in terms of the ePAGs in 2017, was to further develop the ePAG online communities. Facebook Workplace in 2017 was developed and ePAG representatives were invited to join.

In 2017, EURORDIS continued the ePAG Matchmaker Initiative, to register the interest of patient organisations in Europe to become ePAG Members, and to collaborate, consult and be kept informed on the development of ERNs. Approximately, 2000 patients are now members of the 24 ePAGs.



### 2.1.7 RareConnect

*RareConnect.org is an online 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, by EURORDIS, RareConnect is now home to 148 disease-specific communities created in partnership with 889 patient groups and managed with the support of 407 volunteer moderators.*

2017 was a year of significant development for the platform. EURORDIS took steps to transfer the platform to the Children's Hospital of Eastern Ontario Research Institute (Canada) who in partnership with the technology and research team at The Hospital for Sick Children, Toronto (Canada) successfully undertook developments which further improved user experience through the addition of technology which adapts the platform to mobile devices. The platform has also seen the addition of Japanese language capability.



Preparatory work was also initiated to develop RareConnect as a research platform thus allowing researchers across the world to interact with patients and families through specific questionnaires and data-capture of phenotypic information. Once Ethics Committee approval for this project has been approved it will see RareConnect become interoperable with existing research and clinician-based tools in 2018. This work is largely being funded through Canadian public funding as part of a Genome Canada grant to harness multi-omics to deliver innovative diagnostic care for rare genetic disease

Now that CHEO RI will drive innovation in the underlying technology and interoperability with research and clinician based tools, EURORDIS and Rare Diseases International will continue at the heart of the governance of the platform. EURORDIS will continue to make the link with patient associations and members of the platform through a dedicated team of community managers.

### 2.1.8 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. We have developed an effective protocol for registration and to ensure efficiency/ the smooth running of webinars.*

Webinars focus on: providing policy updates (for example on European Reference Networks); involving patients in consultations (for example to develop a EURORDIS position paper in consultation with members); providing capacity-building trainings for patient advocates (eg. training on topics such as genome editing or practical skills for running a patient organisation).

Topics for 2017 webinars included: Consultation on EURORDIS positions on both access to medicines and on social policy; informational/ capacity-building webinar on genome editing; a pre-event webinar for the September workshop of the EURORDIS Round Table of Companies on ERNs to increase event participants' knowledge of the subject; legal and ethical impacts of the data protection regulation (GDPR) on data sharing.

Towards the end of 2017, the team took the steps needed to set up a new 'how to' webinar series to provide patient organisations with the necessary skills to run their organisation. The webinar series will start in 2018.

### 2.1.9 EURORDIS Trainings

#### 2.1.9.1 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.*

For the 2017 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 content for the pre-training relies on the webcasts from previous sessions of the Summer School, webinars and interactive training tools found on the EURORDIS website.

The 2017 Summer School provided participants with a fundamental understanding of the processes involved in medicines development, the time required and the different stages of clinical research. Coupled with formal presentations of the topics, the participants were divided into small groups and encouraged to share their experiences and knowledge in the context of documents provided to complement the lectures. In addition, practical 'hands-on' examples (made-up examples created for the purpose of the course) of the work performed by the representatives at the EMA Scientific Committees and working parties was presented.

62 participants attended representing 27 countries and 43 diseases. The Summer School 2017 participants have been selected based on a call for candidates. Exceptionally in 2017 participants attended under the umbrella of Rare Disease International.



### First Spanish edition of EURORDIS Summer School

Strong interest from Spanish-speaking patient representatives led to the organisation of a Spanish version of the Summer School. It welcomed **43 participants from many regions of Spain as well as Central and South America.**

The Spanish version of the Summer School was held at the same venue and time as the EURORDIS Summer School. It was organised in cooperation with EURORDIS, FEDER (Spanish Rare Disease Organisation), Plataforma Malalties Minoritàries, CIBERER (Spanish Rare Disease

Research Centre), Ministerio de Sanidad y Política Social, Rare Disease International, and Hospital de la Santa Creu I Sant Pau.

The Spanish version covered the same topics as the EURORDIS Summer School version; only the sessions on regulatory affairs focused more on the Spanish regulation. The Spanish version was video recorded so that the recordings can be used to form the pre-training for future versions of the Spanish Summer School.

### 2.1.9.2 EURORDIS participation in EUPATI

*The European Patients' Academy (EUPATI), a pan-European Innovative Medicines Initiative project of 33 organisations and twenty one pharmaceutical companies members of EPIIA. 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".*

EURORDIS is involved in EUPATI's transition phase (2017 to 2020) as a member of the Programme Committee for Cohort Three (the third group of 50 patient advocates taking part in the expert patient 10-month blended learning programme on Medicine Research and Development) and also as member of the EUPATI Steering Group which is mandated to develop and implement sustainability strategies for the project.

## 2.2 Raising Awareness & Informing



### 2.2.1 Rare Disease Day 2017

**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 10<sup>th</sup> edition of Rare Disease Day took place in 94 countries and regions on every corner of the globe. 4 new African countries had events this year: Sudan, Botswana, Nigeria and Senegal. Media from all over the world covered the day, in which politicians, researchers, medical professionals and policymakers in Europe, the US and many more countries participated.

The overarching theme was Patient Involvement with the specific theme of Research. The slogan was: With research, possibilities are limitless. The Rare Disease Day website ([rarediseasesday.org](http://rarediseasesday.org)) continued to be a central point for people living with a rare disease around the world to download the materials to hold events. They could also come to the site to see the worldwide movement and events happening on and around the day. This year there were 226 patient organisations and alliances signed up on a dedicated section of the website called 'Friends of Rare Disease Day' which includes several public institutions. They described their contribution to the campaign and committed to displaying the RDD logo on their website and linking to [rarediseasesday.org](http://rarediseasesday.org).

For the 6<sup>th</sup> year in a row EURORDIS produced a well-received video for Rare Disease Day that exemplified this year's theme of Research. On Rare Disease Day and EURORDIS social media, the video was viewed over 200 000 times. It had almost 3000 shares on Facebook and was translated into 35 languages. EURORDIS also provided 39 National Alliances with an original copy of the video in their own language with subtitles. The National Alliances disseminated locally via their own social media.

EURORDIS hosted 3 events in 2017 for Rare Disease Day:

- + Rare Diseases International 1 Policy Event The Right to Health, A Rare Disease Perspective which was held on 10 February in Geneva, Switzerland. Over 100 people attended with key speakers representing WHO and the UN;
- + 2<sup>nd</sup> Multistakeholder Symposium on Improving Access to Rare Disease Therapies held on 22-23 February in Brussels, Belgium. Close to 400 people attended the event and 1300 tweets were exchanged during the event using the hashtag: #RareEU2017. The livestream recorded a total of 687 viewers;
- + EURORDIS Black Pearl Awards held on 21 February in Brussels, Belgium, which was livestreamed and displayed on [Eurordis.org](http://Eurordis.org). Over 200 people attended the evening.

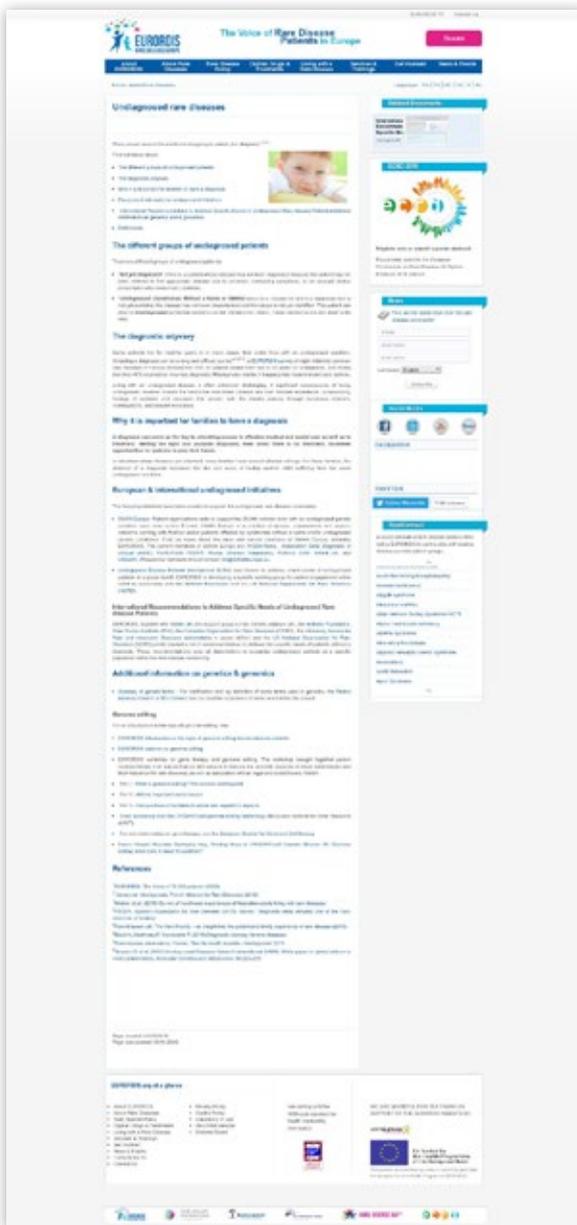


## 2.2.2 EURORDIS Website

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 430.000 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. A User Centered Design (UCD) approach was used to improve navigation and to ensure that structure, navigation architecture, content and features adapt to the expectations of all users' profiles and that an adequate user experience is achieved.

New website sections added in 2017 include: Undiagnosed; Parliamentary Advocates for Rare Diseases.



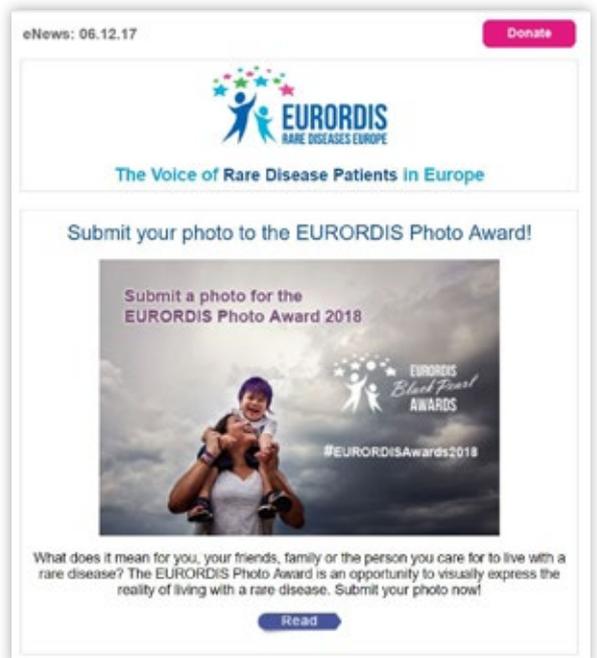
## 2.2.3 eNews & Member News

### 2.2.3.1 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.

Throughout 2017, 21 eNews issues were written, translated in 7 languages, produced and distributed via email. This publication is year-round at a frequency of 2 times per month (except in August). 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.

Lead stories topics in 2017 included: Ritje & JiePie's story: multiple system atrophy; 3,000 rare disease patients & carers voice difficulties in balancing care & life; European Patient Advocacy Groups in action!; Rare Diseases 360° at ECRD 2018 Vienna.



### 2.2.3.2 Member News

The EURORDIS Member News gives updates relevant to the rare disease patient community as well as offering a space for us to remind members of EURORDIS activities that they can participate in (such as events & webinars) and consultations to EURORDIS positions. It is translated into 6 languages and disseminated to over 1500 contacts.

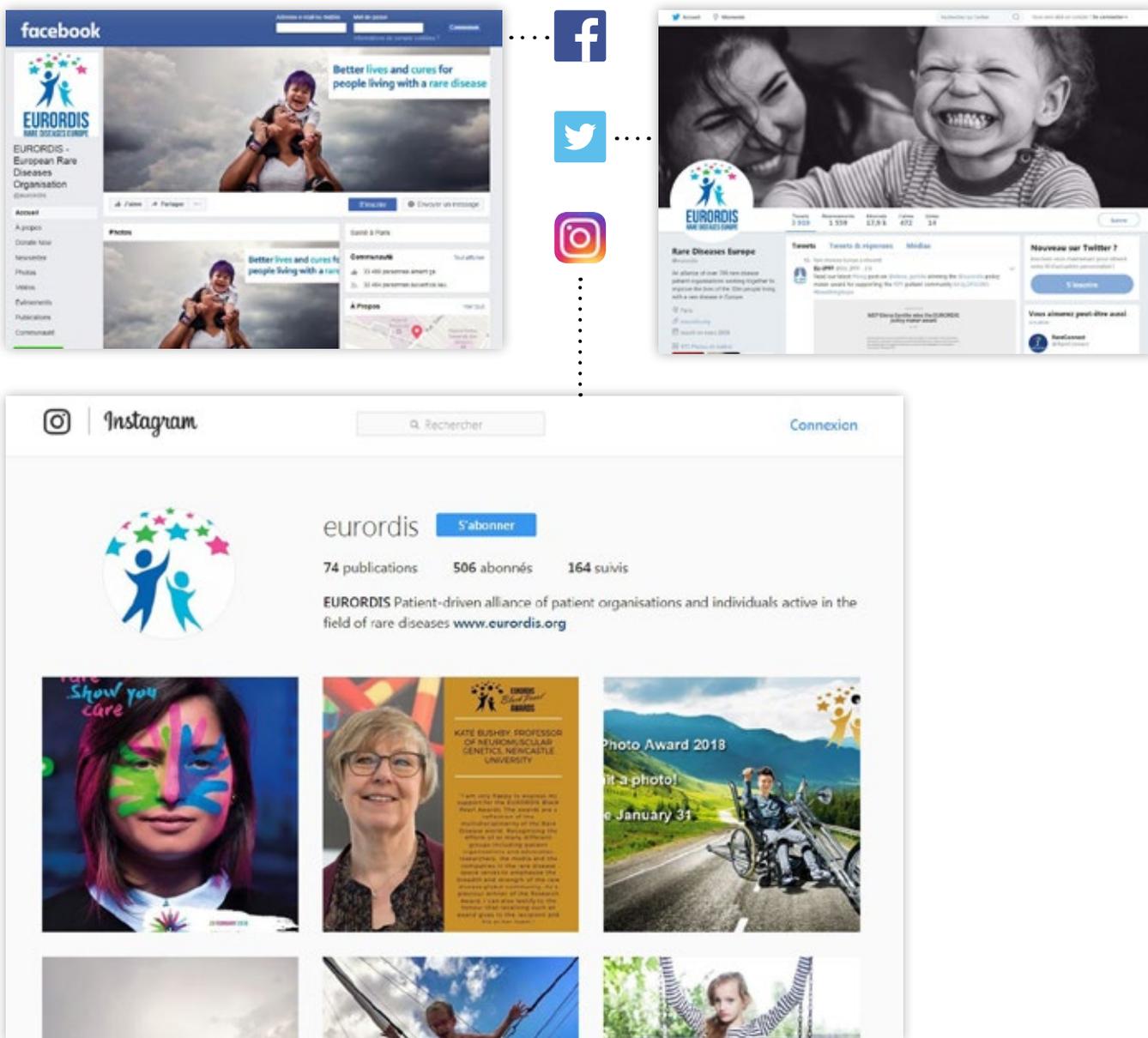
In 2017, we completed our third year of the very successful EURORDIS Member News. The member news was distributed once a month (around the middle of the month) to over 1500 contacts at our member organisations. Its simple design has ensured a high open rate. It provides links to information and activities in a streamlined text template to ensure relevant information is reaching EURORDIS members as directly as possible.

Each 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.

### 2.2.4 Social Media

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



## 2.2.5 The EURORDIS Black Pearl Awards

*The EURORDIS Black Pearl Awards recognise the outstanding commitment and achievements of patient organisations, volunteers, companies, scientists, media and policymakers who have contributed 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.*

The EURORDIS Black Pearl Awards was held in Brussels to mark the occasion of Rare Disease Day 2017. Vytenis Andriukaitis, European Commissioner for Health and Food Safety presented the awards.

Also in attendance of the event was Rare Disease Day Ambassador Sean Hepburn Ferrer.

The winner of the EURORDIS Black Pearl Awards 2017 were:

- + Policy Maker Award**  
Frédérique Ries, Member of the European Parliament, Belgium
- + Volunteer Award**  
Elizabeth Vroom, Duchenne Parent Project, the Netherlands
- + Media Award**  
Aldo Soligno, Photographer, Rare Lives Project, Italy
- + Company Award**  
GSK (GlaxoSmithKline) – Rare Diseases
- + Scientific Award**  
Dr Lucia Monaco, Chief Scientific Officer, Fondazione Telethon, Italy
- + Patient Organisation Award**  
The Dravet Syndrome European Federation, Spain
- + Lifetime Achievement Award**  
Anders Olauson, Founder of the Ågrenska Centre, Sweden





# 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 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 ePAG representatives

enabled a uniform 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.

Throughout the year EURORDIS promoted the capacity-building of ePAG members with regular calls, webinars and face to face meetings and by developing and piloting an ePAG mentoring programme. ePAG representatives grew from the initial 81 at the early stages of development to over 300, active in the 150+ Core Networks under the 24 ERNs.

EURORDIS also supported National Alliances in raising awareness regarding ERNs with their members. Workshops were held in Malta and Germany and will continue in 2018. Overall, an emphasis was put on creating a culture of engagement and collaboration within ERNs through developing an ERN Patient Involvement Guide to promote active and meaningful collaboration and partnership working, in addition to supporting the development of patient representatives role in ERNs, from being co-authors of application in 2016, to securing patient representatives as voting members of all ERN Boards and co-chairs and members of some of the Core Networks and working groups at end of 2017.

# 3.2 Patient Engagement in Social Care

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



### JUGGLING CARE AND DAILY LIFE: THE BALANCING ACT OF THE RARE DISEASE COMMUNITY

Through its survey initiative Rare Barometer Voices, EURORDIS-Rare Diseases Europe carried out the first European-wide survey on the impact of rare diseases on everyday life. The survey covered issues including coordination of care, mental health, employment and economic impact. See the full survey report at [eurordis.org/voices#studies](http://eurordis.org/voices#studies)



**30 million** people are living with a rare disease in Europe and 300 million worldwide



No cure for the vast majority of diseases and few treatments available

#### Rare diseases seriously impact everyday life

**7 in 10** patients & carers

reduced or stopped professional activity due to their or their family member's rare disease.



**8 in 10** patients & carers

have difficulties completing daily tasks (household chores, preparing meals, shopping etc.)



**2/3** of carers

spend more than 2 hours a day on disease-related tasks.



**3 times** more people

living with a rare disease and carers report being unhappy and depressed than the general population\*

\* Rare Barometer Voices sample compared to International Social Survey Programme, 2011



Rare Barometer Voices is a EURORDIS-Rare Diseases Europe online survey initiative. It brings together over 6,000 patients, carers and family members to make the voice of the rare disease community stronger. Results are shared with policy decision makers to bring about change for people living with a rare disease.

**3,071** people responded to the survey.

The survey was conducted in **23** languages across **42** countries



Thank you to all Rare Barometer Voices participants and partners!

[www.eurordis.org/content/contribute-rare-barometer-programme](http://www.eurordis.org/content/contribute-rare-barometer-programme)

For more information visit [eurordis.org/voices](http://eurordis.org/voices) or email [rare.barometer@eurordis.org](mailto:rare.barometer@eurordis.org)

This survey was conducted in the scope of the EU-funded INNOVCare project:



This project is co-funded by the European Union

### Juggling Care and Daily Life: The balancing act of the rare disease community

In 2017, EURORDIS concluded the **first Europe-wide survey on the social and daily life impact of RD, Juggling Care and Daily Life: The balancing act of the rare disease community**.

Over 3,000 people living with a RD and carers responded to this survey, conducted in 23 languages and 42 countries, via Rare Barometer Voices, the EURORDIS survey initiative. The survey and was carried out in the scope of the INNOVCare project.

The results from the survey show that **RD have a severe impact on everyday life**, including that:

- + The **time and care burden** is substantial for a majority of patients and carers;

- + RD have a strong impact on **work-life balance**, leading to absence from work, hampered professional activity, and economic burden;
- + Patients and carers are faced with **complex and hard to manage care pathways**, feel badly informed about their rights, and feel that social services are badly prepared to support them;
- + RD have a significant impact on the **mental health** of patients and carers.

The results of the survey were shared with EURORDIS members at a dedicated **workshop at EMM 2017 Budapest** and in several formats (e.g. report, presentation, infographics). EURORDIS widely disseminated the results to external stakeholders, via targeted communication actions and events, including at the launch of the network of Parliamentary Advocates for Rare Diseases (October).

In the autumn, EURORDIS started the process to elaborate **EURORDIS position paper in social policy**, aiming at advocating for person-centred and holistic care for people living with a RD and their carers (publication expected at the end of 2018). The position paper will build on robust steps taken over the last 6 years by EURORDIS and its members, via advocacy actions, social surveys and projects. Additionally, EURORDIS will work closely with its volunteers and members to write the position paper. The first consultations involving the Social Policy Advisory Group, the Council of National Alliances, the Council of European Federations and members at large took place in the last trimester of 2017.

**EURORDIS continued to widely disseminate its contribution to the European Commission consultation on the European Pillar of Social Rights (December 2016) and followed closely the launch of this European framework** and linked initiatives (i.e. Work Life Balance Directive), advocating for the needs of people living with a RD and their carers to be taken into account.

EURORDIS also continued to disseminate the **Commission Expert Group Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies**, adopted in by all EU MS in 2016.

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



### 3.2.2 Support the national alliances in their action to advance the integration of rare diseases into social services and policies

During this last year, EURORDIS continued to **support national alliances to promote the integration of rare diseases into social policies and services** via the dissemination of a tool kit to support national workshops focused on social and daily life aspects. EURORDIS board members, volunteers and staff also presented the social challenges of people with a RD at various national workshops.

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### 3.2.3 Promote integration of rare diseases into social services

In 2017 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 **first Europe-wide survey** on the social and everyday impact of RD; and the promotion of the **representation of people living with a RD and their carers in relevant policies at European level**.

EURORDIS continued to engage with the **Advisory Group of the INNOVCare project**, composed of competent authorities from European countries, to discuss how to promote the implementation of the **Commission Expert Group Recommendations to Support the Incorporation of Rare Diseases into Social Policies and Services** and on how to implement holistic care for RD.

Within the INNOVCare project, EURORDIS has continued the activities of the secretariat of the **European Network of**



**Resource Centres for Rare Diseases – RareResourceNet.** The network, composed of resource centres for RD, focuses on advancing holistic high quality care for people living with a RD and their carers in Europe. In 2017, the network adopted its vision, mission and objectives, as well as its membership criteria, membership application procedures and its draft by-laws.

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**: the Social Platform and the International Federation of Social Workers Europe, with whom EURORDIS signed a Memorandum of Understanding in 2016.

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### 3.2.4 Promoting integrated health and social care for rare diseases

EURORDIS continued to promote integrated care for RD in 2017 via the various activities of the **INNOVCare project**.

In June, EURORDIS organised the **INNOVCare Workshop on Advancing Holistic & Innovative Care for Rare Diseases & Complex Conditions** (Romania) – focused on presenting the progress of INNOVCare pilot of case management and on discussing key issues to support the implementation of holistic care for people with a RD in European countries. The workshop, attended by 46

participants from 18 countries, included multi-stakeholder discussions on key issues to implement holistic care for RD.

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 INNOVCare pilot of case management**. As responsible for the external communication of INNOVCare, EURORDIS also ensured the dissemination of the project's outcomes.



## 3.3 Patient Engagement in Research

### 3.3.1 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 and in particular in 2017, with its presence in the **Consortium Assembly**, in the **Therapies Scientific Committee** and in the **Interdisciplinary Scientific Committee**: Béatrice de Montleau, EURORDIS patient representative in Consortium Assembly; Yann Le Cam, CEO of EURORDIS, Consortium Assembly and Chair of IRDiRC Therapies Scientific Committee (TSC) until end of October 2016/TSC Member after October 2016; Gema Chicano, EURORDIS patient representative, Interdisciplinary Scientific Committee member since February 2016; Virginie Hivert, EURORDIS Therapeutic Development Director, TSC member since 2014 and Vice-Chair since March 2017.



In 2017, EURORDIS was involved in the preparation of the new roadmap of IRDiRC in order to achieve the new goals elaborated in February 2017 during the IRDiRC conference in Paris. The new IRDiRC vision for 2017-2027 is to **enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention.**

The new goals and vision have been disseminated to the entire EURORDIS network.

### 3.3.2 Engagement in upcoming genetic developments

#### Genetic Clinic of the Future (GCOF)

*The Genetic Clinic of the Future project had 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 collaborated with experts from other project partners to carry out a survey on patient perspectives. 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.

The project came to an end in June 2017 and worked on: a website hosted by the university of Leicester to explain in lay terms the different models for data sharing and to which EURORDIS contributed [https://datasharing-101.le.ac.uk/DataSharing\\_101](https://datasharing-101.le.ac.uk/DataSharing_101); as well as the production of a Policy Brief on Genetics Clinic of the future, a document that provides a backbone on which a Legal Expert Report and Ethic Expert Report will be produced.



### 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' organisations in research.

The participation of EURORDIS in E-Rare-3 opens the possibility for patients' organisations to foster their engagement in funding of research on rare disease at the transnational level. EURORDIS will coordinate a network of "scientific officers" (volunteers) from funding research patient organisations to find an innovative funding schema with patient organisations

### 3.3.3 Participation in the Web-RADR project

Web-RADR: Web Recognition of Adverse Drug Reactions (<https://web-radr.eu>)



Now used by over one billion people globally, social media websites and mobile phone apps enable and encourage users to create, share, and exchange information and experiences instantly. It was WEB-RADR's mission to develop new ways of using these technologies and channels of communication for pharmacovigilance purposes. WEB-RADR was an IMI project that ended in 2017. EURORDIS was one of the consortium partners.



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).

Throughout 2017, EURORDIS worked on the European Joint Programme on RD, as an ambitious follow up of E-Rare, to position EURORDIS in the Governance of the future programme. This effort will continue in 2018.

WEB-RADR delivered two key outcomes: a mobile phone app for the reporting of suspected ADRs to EU regulators (now available in the United Kingdom, Croatia, Netherlands, Zambia, Burkina Faso); new technical tools for data mining publicly available data shared on social media websites.

Through its work, WEB-RADR also established the regulatory framework for social media mining for ADRs; engaged patients in the awareness and reporting of safety concerns; improved methods of mining social media for pharmacovigilance; delivered a platform for monitoring effects of risk minimisation

EURORDIS took part in the valuation of enablers and barriers to the use of an app to report side effects and this work has been published:

- + Interest in a Mobile App for Two-Way Risk Communication: A Survey Study Among European Healthcare Professionals and Patients. Sieta T. de Vries, Petra Denig, Carmen Lasheras Ruiz, François Houÿez, Lisa Wong, Alastair Sutcliffe, Peter G. M. Mo. Drug Saf (2018). <https://doi.org/10.1007/s40264-018-0648-0>
- + Factors Influencing the Use of a Mobile App for Reporting Adverse Drug Reactions and Receiving Safety Information: A Qualitative Study. Sieta T. de Vries, Lisa Wong, Alastair Sutcliffe, François Houÿez, Carmen Lasheras Ruiz, Peter G. M. Mol. Drug Saf. 2017 May;40(5):443-455. <https://doi:10.1007/s40264-016-0494-x>

Next steps are the creation of a new initiative to further develop the app, adapt it to new uses and other countries. This initiative will gather WHO, FDA, EMA, MHRA, EFPIA, IMI and EURORDIS.

## 3.4 Patient Engagement in lifecycle development

### 3.4.1 Patients creating their Community Advisory Boards to engage with Industry

#### What is a CAB? A corner stone of patients' engagement with industry

*Patient Community Advisory Boards (CABs) are consulting groups established, operated and maintained by patient advocates and expert patients to discuss, in a neutral, continual and critical setting, the latest developments, challenges and issues related to medical treatments and procedures under development in your disease area. CABs, with anywhere from seven to twelve advocates, are involved in scientific as well as policy related issues (i.e., access), and they provide expert advice to all stakeholders involved in the research, development and service provision of biomedical treatment.*

Throughout 2017, EURORDIS worked on the development of the EURO-CAB programme consisting mainly in: Guidelines on how to be organised and operate a CAB; All templates needed to start discussions with industry or other sponsors; A mentoring programme with training

activities; Ad hoc guidance to help preparing meeting; an evaluation of the impact of the CABs.

The advantages to join the EURO-CAB programme for EURORDIS members are:

- + Experience: CABs benefit from experienced advice by EURORDIS staff on how to operate a CAB
- + Training: CABs benefit from EURORDIS training programmes (Summer School, Winter School)
- + Credibility: CABs benefit from EURORDIS credibility and strong governance
- + Up-to-date: CABs will be aware of initiatives along the products life-cycle (e.g. Clinical trial legislation, ethics committee, HTA Early Dialogues, Regulatory Scientific Advice, Protocol Assistance, Horizon Scanning, PRIME, MOCA, CHMP/Scientific Advisory Groups, scoping/HTA, Late dialogues, Pharmacovigilance, Variations...)
- + Visibility: CABs become more largely visible with the creation of the EURO-CAB register

To conduct the programme, a Community Advisory Board Patient Engagement Manager was recruited (Rob Camp) at end of 2017 and the programme will be launched in early 2018.

### 3.4.2 Pre-marketing authorisation

#### European Medicines Agency

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

Dedicated expert patient representatives contributed to the examination and scientific evaluation of dossiers in 2017 through the work of the scientific committees they belong to, as well as to the activities of several ad hoc working groups all along the year.

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

In addition to these permanent activities at the EMA, patient representatives are also invited on a sporadic basis to attend the scientific committees and the Scientific Advice Working Party (SAWP) as experts for their disease.

In 2017, 47 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.

In 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. Dimitrios Athanassiou has subsequently been nominated as a Member of PDCO by the European Commission for the new mandate starting in August 2017. An exchange of expertise was organised between Tsveta Schyns, former Member since the creation of PDCO and Dimitrios.

In December 2017, applications responding to the European Commission's Call for Expression of Interest were submitted for the COMP membership. After an



internal Call for Expression of Interest within its Members, EURORDIS has proposed Virginie Hivert and has issued three letters of endorsement for Julian Isla, Pauline Evers and Cathalijne van Doorne.

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 2017, PCWP members worked in particular personalised/precision medicines (workshop 14 March), on antimicrobial resistances (workshop 19 September), on pharmacovigilance (Forum on 21 September), on data-anonymisation (workshop 30 November – 1 December) and on Site and Histology – Independent Indications in Oncology (workshop 14-15 December).

Following the evaluation of the involvement of patients in oral explanations with the company, the CHMP decided to end the pilot phase and include the participation of patients as external experts as a common practice.

For the pilot, patients were invited to participate where their involvement anticipated bringing added value to the discussion, and this was decided on a case-by case basis. Patients (or carers) were selected depending on the relevance of their experience/knowledge of particular disease/condition under evaluation, and after assessment of any conflict of interest.

Two patients were usually invited to the oral explanation, accompanied by a 'mentor' (PCWP member); in addition EMA provided personal support (guidance on the work of the CHMP, issues for discussion and clear definition of their role).

Patients give their views and participate in the discussions; including asking questions to the company. They do not take part in decision-making process (leave the room prior to voting).

The evaluation concluded that:

- + The feedback from CHMP/EMA received during pilot was generally positive
- + Patients reported a very positive experience. Their participation increase transparency and trust in the work of the CHMP
- + Each case was variable depending on the topic and on patients involved
- + Involvement has been a learning curve and has improved with experience
  - More relevant questions for the patients
  - Everyone involved knows better what to expect

As the PCWP is consulted regarding the framework of interaction between patients, consumers and the EMA, a session invited representatives of the European Network of HTA agencies (EUnetHTA) to exchange on how to best involve patients in European HTA activities.

### 3.4.3 Post-marketing authorisation

#### Developing the roles of patients in monitoring the medicines they take: Vigil

*In pharmacovigilance, an important role in industry is played by the Qualified Person for Pharmacovigilance<sup>1</sup> (QPPV). This person is appointed by the company to ensure the company applies all measures provided for in the legislation to monitor their medicines. He/she ensures communication with regulatory authorities.*

On the same line, EURORDIS now proposes to create a new role for patients' representatives who would be interested to liaise with regulatory authorities about the monitoring on authorised medicines. This person would be a "Vigil", officially appointed by the national regulatory authorities to be the contact person in his/her organisation for all pharmacovigilance aspects (exchange of information, safety alerts, direct-to-patient pharmacovigilance, training, reporting suspected adverse drug reactions etc.).

<sup>1</sup> Individual named by a pharmaceutical company as the main person responsible for ensuring that the company (the product's Marketing Authorisation Holder or MAH) meets its legal obligations for the monitoring of the safety of a medicinal product on the market. See Article 8(3)(ia) of Directive 2001/83/EC [https://ec.europa.eu/health/sites/health/files/files/eudralex/vol-1/dir\\_2001\\_83\\_cons2009/2001\\_83\\_cons2009\\_en.pdf](https://ec.europa.eu/health/sites/health/files/files/eudralex/vol-1/dir_2001_83_cons2009/2001_83_cons2009_en.pdf)

This role is not foreseen in the legislation, but it is very much needed. The Vigil, would be trained on how pharmacovigilance is organised in Europe and in Member State of interest, and would be the liaison between members of an organisation or users of social networks, and national or European regulatory authorities, possibly also for industry (second step).

As an outcome of the SCOPE Joint Action in pharmacovigilance, EURORDIS proposed this idea in many conferences and has now formed the embryo of a larger coalition to develop the concept further, and EATG, NDA Reg (a consultancy firm), European Medicines Agency (EMA) and some members of its Pharmacovigilance Committee (PRAC), International Society for Pharmacovigilance (ISOP), WHO Drug Monitoring Centre (UMC), Spanish Regulatory Agency (AEMPS), United Kingdom Regulatory Agency (MHRA), and the Irish Regulatory Agency (HPRA) expressed their interest.

### Creation of a EURORDIS Task Force on Health Technology Assessment

In 2017, EURORDIS laid the groundwork for the launch of a new HTA Task Force. In December 2017, the Board of Directors approved the mandate and general objectives, the composition, the requirements for applicants and the timeframe.

The mandate of that new Task Force is to advise EURORDIS on all aspects regarding Health Technology Assessment policies and procedures.

The Task Force general objective is to raise awareness of all EURORDIS members and gather technical and scientific expertise on HTA. In so doing, the Task Force aims at strengthening EURORDIS positions, establishing and adequate knowledge of HTA practices throughout Europe in their own Organisations, facilitating the participation of patients, and share patients' views on the future of HTA in EU.

Specific requirement to be fulfilled have been established for applicants to take part in the Task Force, such as

- + Having participated in an HTA procedure, such as:
  - Early Dialogue or Parallel EMA/HTA Scientific Advice;
  - Scoping phase of the assessment;
  - Assessment of a technology at national or EU level;
  - appraisal procedure or decision committee for pricing and reimbursement
- + Having participated in an HTA training programme, such as:
  - EURORDIS Summer School
  - EUPATI and/or EUnetHTA Training
- + Being an English speaker

The Task Force will be **composed** of 12 (twelve) members for a **mandate** of 3 (three) years (2018-2021), confirmed on a yearly basis.

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

*The Task Force represent a group of 18 volunteers who are trained (via the EURORDIS Summer School) and active in issues concerning therapeutic development of medicines for rare diseases. The Task Force supports and/or advises the EURORDIS representatives who participate in EMA Scientific Committees and Working Parties, or in the European Network*

*of Health Technology Assessment (EUnetHTA) and the HTA Network (DG Sante). It is consulted on papers prepared by EURORDIS.*

Two DITA Task Force face-to-face meetings were held in Paris in 2017. One in June where the main topic was fair pricing, and a health economist explained his approach for an objective evaluation of a fair price for medicines, and one in November where the main topic was Patient Relevant Outcomes (PROs) where an expert in outcome research explained his work and views on PROs.

DITA members exchanged information and elaborated contributions to: an EMA template for herbal medicinal product monographs and the Shortage of Unituxin®.

In addition, DITA task force members presented EURORDIS views/data in EMA conferences and other conferences on regulatory affairs, including the following (list not exhaustive):

- + Workshop on antibiotic microbial resistances, EMA London, 19 September
- + Pharmacovigilance Forum, 21 September
- + Data-anonymisation workshop – a key enabler for clinical data sharing, 30 November- 1 December, EMA London, presenting on Defining sensitive data – Influence of the context of the disease on the tolerability of risk
- + Real world data contribution to regulatory and coverage decisions, 17 March, EMSP conference at the European Parliament, Brussels, presenting on Registries and Data Protection
- + EMA-FDA Rare Diseases Cluster, 27 June, EMA, presenting EURORDIS Position on Compassionate Use
- + IAPO African Regional Meeting, 4-5 July, Entebbe, Uganda, presenting by video on Public Engagement How Patients and Consumers Work with the EMA
- + DIA Value, Access & Regulatory Strategy Workshop, 25-26 October, Basel, presenting on 1) Meeting the needs of patients, 2) What does fast patient access mean? Risks, early access and trade-offs
- + United Parent Project Muscular Dystrophy meeting, 9 November, Birmingham, presenting on Drug Development, Evaluation and Reimbursement (and other technologies): Times are Changing
- + ACHSE Round Table of Companies, 14 November, Berlin, presenting on Early Access to Medicines in Europe: How to make Compassionate Use Become a Reality for All in Need

**Review of EMA documents for the public or contributions to EMA consultations:** European Public Assessment Reports for the Public (6), Package Leaflets (19). Since 2007 when the procedure to review EPAR summaries and PL was established for authorised medicines in the EU, 66 EPAR summaries and 111 PL were reviewed, for a total of 177 documents.





# 4. CROSS-CUTTING PRIORITIES

## 4.1 Governance

### 4.1.1 EURORDIS Board of Directors

During the General Assembly held in Budapest on 19 May 2017, EURORDIS full members voted on five vacant positions on the Board of Directors, electing Dimitrios Synodinos, Tuberous Sclerosis Association, Greece; Alba Ancochea, Spanish Federation of Rare Diseases (FEDER), Spain; Birthe Holm, Rare Diseases Denmark, Denmark; Dorica Dan, Romanian Prader Willi Association, Romania and Lieven Bauwens, International Federation for Spina Bifida and Hydrocephalus, Belgium. Lieven Bauwens

replaces John Dart, DEBRA International, UK, who stepped down from the Board of Directors for personal reasons.

The Board of Officers, which is 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: Geske Wehr, Germany; Treasurer: Dimitrios Synodinos, Greece; and Officer: Dorica Dan, Romania.



### 4.1.2 Partnerships with international organisations (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 also plays an active part in Rare Diseases International as a member of the RDI Council governance board.



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

EURORDIS and CORD have been collaborating together for many years and signed a Memorandum of Understanding in 2012. CORD is a member of the EURORDIS Council of National Alliances and also plays an active part in Rare Diseases International as a member of the RDI Council. CORD is also part of the governance structure of the online community platform, RareConnect.



#### 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.



#### 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. In 2017, RVA played an active part in Rare Diseases International as a member of the RDI Council.



#### RUSSIAN PATIENTS ASSOCIATION Health for all

#### RPU – Russian Patients' Union

EURORDIS and the Russian Patients' Union signed a memorandum of understanding in May 2015. RPU is a member of the Council of National Alliances.



#### RADOIR - Rare Diseases Foundation of Iran

EURORDIS signed a Memorandum of Understanding with RADOIR foundation, Iran, in 2016. Throughout 2017, RADOIR attended several EURORDIS events and worked with the RareConnect team to develop the online community platform in Farsi, for release in 2018.

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 Pharmaco-economics and Outcomes Research (ISPOR)



## 4.2 Human resources

### 4.2.1 EURORDIS Staff

The team comprised 41 people as of December 2017. The team is composed of paid staff, 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 ERN and Healthcare Advisor are based in the Brussels office. The EURORDIS RareConnect team managing the online patient communities network is based in Barcelona alongside the Social Policy & Projects team, Patient Engagement Manager and Web Technology Manager. The Rare Diseases International Director is based in Geneva and the Events Director in the UK. The Chief Executive Officer shares his time between the Paris and Brussels offices.

The following are the main changes in human resources in 2017:

- + **Zoe Alahouzu**, Deputy to the CEO, has returned from maternity leave
- + **Laura Amorini**, Web Technology Manager, left EURORDIS
- + **Simone Boselli**, Public Affairs Director, joined EURORDIS
- + **Maria Mavris**, National Expert on secondment to the EMA, left EURORDIS
- + **Robert Pleticha**, Rare Connect Manager, left EURORDIS
- + **Ketakaniaina Rasamoely**, Administrative & Support Services Assistant, left EURORDIS
- + **Jean-Louis Roux**, Public Affairs Director, left EURORDIS
- + **Matteo Scarabelli**, Patient Engagement Manager in HTA, joined EURORDIS
- + **Aline Schnieder**, Social Projects Assistant Manager, joined EURORDIS

### 4.2.2 EURORDIS Volunteers

In 2017, EURORDIS was privileged to rely on **428 volunteers** including **68 dedicated volunteer patient advocates**, **1 office support volunteer**, **1 volunteer fundraiser**, **358 volunteer moderators** of online communities of rare disease patients, within the activity "RareConnect".

Most EURORDIS volunteers are rare disease patients or family members. Due to the lack of available information for many rare diseases, patients find themselves becoming experts of their own disease and of their national health care system.

EURORDIS volunteers are involved in many different aspects of our work including the following: representing EURORDIS in EU high-level committees and in scientific committees of the European Medicines Agency (EMA); internal Task Forces and committees; representing EURORDIS in European NGOs, networks and working groups; voicing our organisation's positions in international conferences; moderating Patients' online communities. The volunteers of EURORDIS can share their respective expertise in various fields, from research to access to medical care and adapted social services as well as in sharing information on specific rare diseases.

All the volunteers are governed by the EURORDIS Charter of Volunteers, which was 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.

EURORDIS is extremely grateful to this group of dedicated individuals who offer their time and expertise to improve the lives of people living with a rare disease and their families.

## 4.3 Finance & Support Services

Finance and support services' activities in 2017 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

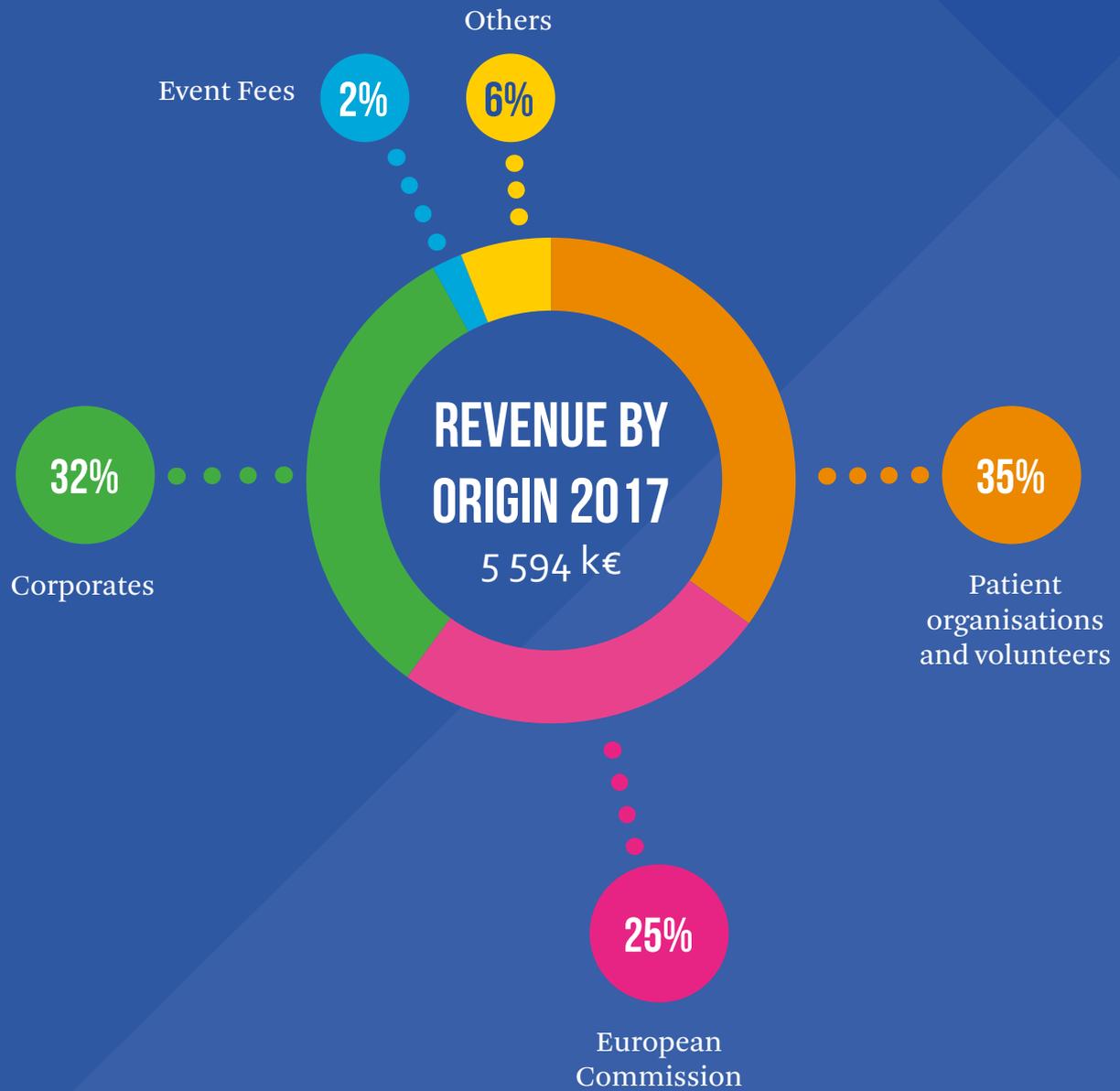
- + Specific Grant Agreement (Operating Grant) for year 2017 (SGA FY2017), single beneficiary, DG Sante, 12 months
- + eNews and Website in Russian, Association of International Pharmaceuticals Manufacturers (AIPM), 12 months
- + Framework Partnership Agreement 2015-2017 (Operating Grant), single beneficiary, DG Sante, 2015-2017
- + Genetic Clinic of the Future, a stepping stone approach towards the Genetics Clinic of the Future, DG Research, 2014-2017.

#### Ongoing

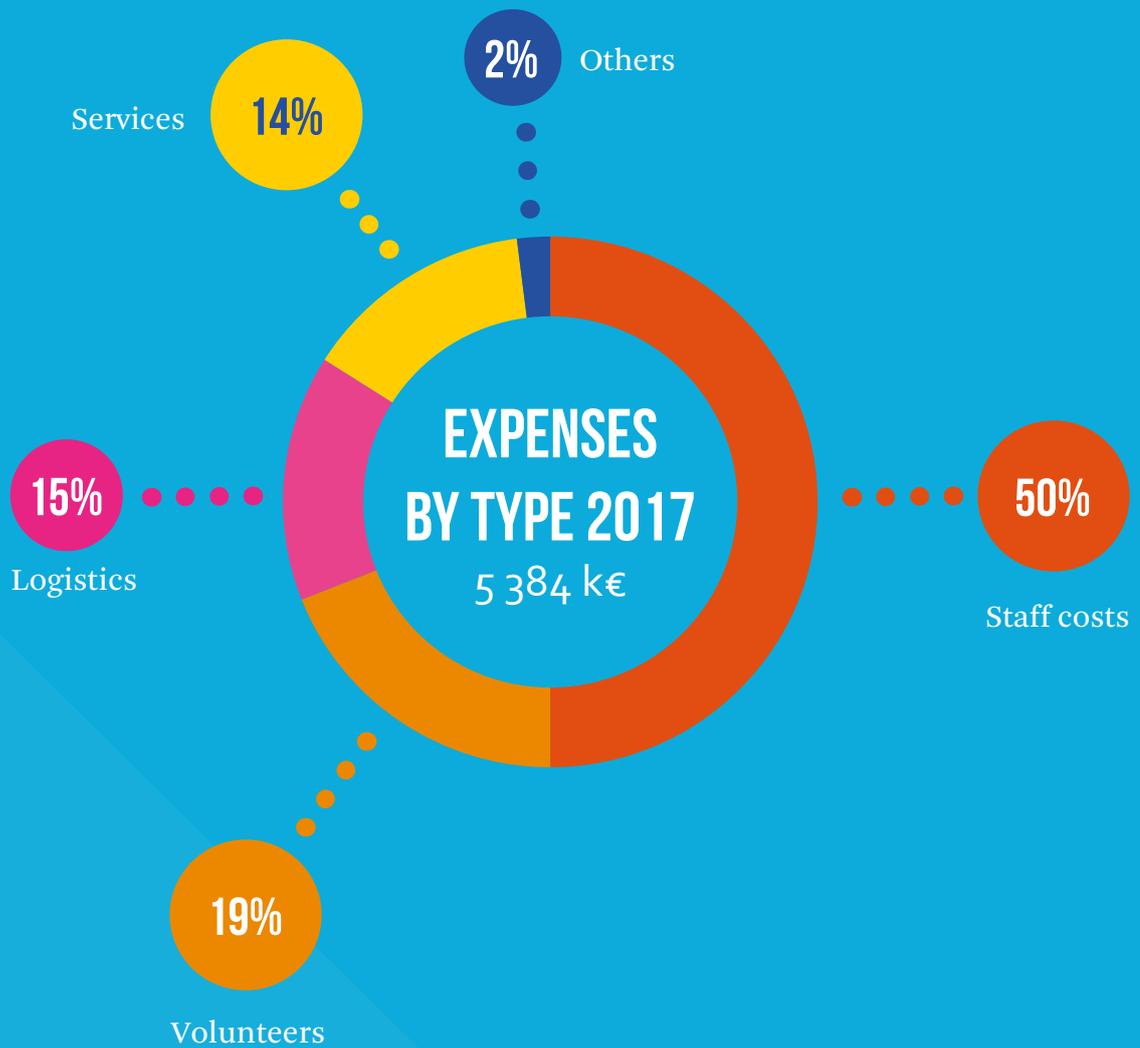
- + Advocacy and core activities, AFM-Téléthon, 2014-2017
- + Adapt-SMART: An enabling platform for the coordination of Medicines Adaptive Pathways to Patients (MAPPs) activities, Innovative Medicines Initiative (IMI), 2015-2017
- + EUPATI European Patients' Academy on Therapeutic Innovation, Innovative Medicines Initiative (IMI), 2012-17
- + 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
- + JARC: Joint Action on Rare Cancers, DG SANTE, 2016-2019
- + RD-Action: Joint Action to expand and consolidate the achievements of the former EUCERD JA, DG Sanco, 2015-2018
- + 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



# REVENUE 2017



# EXPENSES 2017





# BOARD of Directors

May 2017 - May 2018

## PRESIDENT

**Mr Terkel Andersen** Danish Haemophilia Society

 Denmark

## DIRECTORS

**Ms Alba Ancochea** Spanish Federation of Rare Diseases (FEDER)

 Spain

**Mr Lieven Bauwens** International Federation for Spina Bifida and Hydrocephalus

 Belgium

**Ms Simona Bellagambi** UNIAMO - Rare Diseases Italy

 Italy

**Ms Avril Daly**  
Vice-President Rare Diseases Ireland

 Ireland

**Ms Dorica Dan**  
Officer Romanian Prader Willi Association

 Romania

**Ms Birthe Byskov Holm** Rare Diseases Denmark

 Denmark

**Ms Anne-Sophie Lapointe** Vaincre les Maladies Lyosomales

 France

**Ms Françoise Salama** AFM-Téléthon

 France

**Mr Dimitrios Synodinos**  
Treasurer Tuberos Sclerosis Association

 Greece

**Ms Geske Wehr**  
General Secretary European Network for Ichthyosis e.V

 Germany

**Ms Vlasta Zmazek** Rare Diseases Croatia

 Croatia

# MEMBERS of EURORDIS

FULL MEMBER ..... ●  
ASSOCIATE MEMBER ..... ●

## ALBANIA

SHOQATA E SEMUNDJEVETE RRALLA - RARE DISEASE ASSOCIATION

## 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

CYSTIC FIBROSIS AUSTRALIA

GENETIC ALLIANCE AUSTRALIA

RARE VOICES AUSTRALIA

## AUSTRIA

ANGELMAN VEREIN ÖSTERREICH

DEBRA INTERNATIONAL

HAND IN HAND GEGEN TAY-SACHS UND SANDHOFF

ICA 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 BELGIË

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 NETWORK FOR RESEARCH ON ALTERNATING HEMIPLEGIA

EUROPEAN POLIO UNION

EYE HOPE FOUNDATION

FÉDÉRATION DE EUROPÉAN SCLERODERMA ASSOCIATIONS

FEDERG - FEDERATION OF EUROPEAN ASSOCIATIONS OF PATIENTS AFFECTED BY RENAL DISEASES

FEWS - FEDERATION OF EUROPEAN WILLIAMS SYNDROME

HAE BELGIUM

ICHTHYOSE BELGIQUE - ICHTHYOSIS BELGIË

INTERNATIONAL FEDERATION FOR SPINA BIFIDA AND HYDROCEPHALUS

INTERNATIONAL HUNTINGTON ASSOCIATION

MYELOMA PATIENTS EUROPE

RADIORG - RARE DISEASE ORGANISATION BELGIUM

RARE DISORDERS BELGIUM

RELAIS 22 ASBL

SIOP EUROPE - EUROPEAN SOCIETY FOR PAEDIATRIC ONCOLOGY

SOBREVIVIRE VZW

VASCULAR ANOMALY PATIENT ASSOCIATION

VLAAMS PATIËNTENPLATFORM VZW

VLAAMSE VERENIGING NEUROMUSCULAIRE AANDOENINGEN VZW (NEMA)

VLAAMSE VERENIGING VOOR ERFELIJKE

BINDWEEFSELAANDOENINGEN

## 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

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

DEBRA CROATIA

RARE DISEASES CROATIA

## 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 (CESKA

ASOCIACE PRO VZACNA ONEMOCNENI)

KLUB NEMOCNYCH CYSTICKOU FIBROZOU

META, ASSOCIATION OF PATIENTS WITH LYSOSOMAL STORAGE DISEASES

NARODNI SDRUZENI PKU A JINYCH DMP (CZECH PKU ASSOCIATION)

## DENMARK

22Q11 DENMARK

ADDISON FORENINGEN I DANMARK

BLÆREEKSTROFIFORENINGENS

CCHS DANMARK

DANISH APERT SYNDROME ASSOCIATION /DANMARKS

APERTFORENING

DANMARKS BLODERFORENING / DANISH HAEMOPHILIA SOCIETY

DANSK FORENING FOR NEUROFIBRATOSIS RECKLINGHAUSEN

EHLERS-DANLOS FORENINGEN I DANMARK

FORENINGEN AF MÖBIUSSYNDROM I DANMARK

FORENINGEN FOR ATAKSI / HSP

ICHTYOSIS ASSOCIATION IN DENMARK

IMMUN DEFJEKT FORENINGEN

MCADD-FORENINGEN

MITOKONDRIE-FORENINGEN I DANMARK  
PATIENTFORENINGEN HÆ 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

ASSOCIATION OF CANCER PATIENTS IN FINLAND (SUOMEN  
SYÖPÄPOTILAAT RY.)  
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  
RINNEKOTI FOUNDATION, NORIO CENTRE

## FRANCE

AFMKT-FRANCE  
ALLIANCE MALADIES RARES  
ALLIANCE SYNDROME DE DRAVET  
ASSOCIATION ISIS  
ASS. INTERNATIONALE DE DYSTROPHIE NEURO AXONALE  
INFANTILE  
ASSOCIATION «LES PETITS COURAGEUX»  
ASSOCIATION AMS-ARAMISE  
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 À 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 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 FRANÇAISE CONTRE L'AMYLOSE  
ASSOCIATION FRANÇAISE CONTRE LES MYOPATHIES  
ASSOCIATION FRANÇAISE DE LA FIÈVRE MÉDITERRANÉENNE  
FAMILIALE  
ASSOCIATION FRANÇAISE DE LA MALADIE DE FANCONI  
ASSOCIATION FRANÇAISE DE L'ATAXIE DE FRIEDREICH  
ASSOCIATION FRANÇAISE DE L'OSTÉODYSTROPHIE 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 DU SYNDROME PHELAN-MCDERMID  
ASSOCIATION FRANÇAISE LESCH-NYHAN ACTION  
ASSOCIATION FRANÇAISE POUR LE SYNDROME DE GILLES DE LA  
TOURETTE  
ASSOCIATION FRANCE VASCULARITES  
ASSOCIATION FRANÇOIS AUPÉTIT  
ASSOCIATION FRANCOPHONE CONTRE LA POLYCHONDRIE  
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 INTERNATIONALE MALADIES KYSTES TARLOV  
ASSOCIATION KOURIR  
ASSOCIATION MALADIES FOIE ENFANTS  
ASSOCIATION MARFANS  
ASSOCIATION MÉDICALISTES  
ASSOCIATION MICROPHALMIE FRANCE  
ASSOCIATION NAEVUS 2000 FRANCE EUROPE  
ASSOCIATION NATIONALE DES CARDIAQUES CONGÉNITAUX  
ASSOCIATION NEUROFIBROMATOSES & RECKLINGHAUSEN  
ASSOCIATION OLLIER MAFFUCCI EUROPE  
ASSOCIATION PEMPHIGUS - PEMPHIGOÏDES FRANCE  
ASSOCIATION POUR AIDER ET INFORMER LES SYRINGOMYÉLIQUES  
EUROPÉENS RÉUNIS  
ASSOCIATION POUR LA LUTTE CONTRE L'ALCAPTONURIE  
ASSOCIATION POUR LA LUTTE CONTRE LES MALADIES  
INFLAMMATOIRES DU FOIE ET DES VOIES BILIAIRES  
ASSOCIATION POUR LA RECHERCHE SUR LA SCLÉROSE LATÉRALE  
AMYOTROPHIQUE  
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 STRÜMPPELL-LORRAIN  
ASSOCIATION SURRENALES  
ASSOCIATION SYNDROME DE KLEINE-LEVIN  
ASSOCIATION SYNDROME DE MOEBIUS FRANCE  
ASSOCIATION SYNDROME DE ROKITANSKY - MRKH  
ASSOCIATION TANGUY MOYA MOYA  
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 LESCH-NYHAN DISEASE  
EUROPEAN FEDERATION OF ASSOCIATIONS OF PATIENTS WITH  
HAEMOCHROMATOSIS  
FÉDÉRATION DES MALADES DRÉPANOCYTAIRES ET  
THALASSÉMIQUES SOS GLOBI  
FEDERATION EURO-HSP  
FITIMA EUROPE - FONDATION INTERNATIONAL TIERNO ET MARIAM  
FOP FRANCE  
FRANCE LYMPHANGIOLEIOMYOMATOSE (FLAM)  
FRANCE LYMPHOME ESPOIR  
GÉNÉRATION 22  
GENESPOIR: ASSOCIATION FRANÇAISE DES ALBINISMES  
GENIRIS  
HTAP FRANCE  
HYPOPHOSPHATASIE EUROPE  
INCONTINENTIA PIGMENTI FRANCE  
INFLAM'ŒIL  
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  
OUVRIR LES YEUX - OLY  
POLYPOSES FAMILIALES - APTEPF  
PRADER WILLI FRANCE  
RETINA FRANCE  
SANFILIPPO SUD  
SOLIDARITÉ HANDICAP AUTOUR DES MALADIES RARES  
UNION HUNTINGTON ESPOIR  
UNION NATIONALE DES ASSOCIATIONS PARENTS ET AMIS PERSONNES  
HANDICAPÉES MENTALES  
UNION NATIONALE DES SYNDROMES D'EHLERS-DANLOS  
VAINCRE LA MUCOVISCIDOSE  
VAINCRE LA PAPILLOMATOSE RESPIRATOIRE RÉCURRENTE  
VAINCRE LES MALADIES LYSOSOMALES  
VALENTIN - ASSOCIATION DES PORTEURS D'ANOMALIES  
CHROMOSOMIQUES

**VHL FRANCE**

X FRAGILE 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 EV  
BSHV FÜR KINDER, JUGENDLICHE UND ERWACHSENE MIT SELTENEN, CHRONISCHEN SKELETTERKRANKUNGEN E.V.  
BUNDESVERBAND DER CLUSTERKOPFSCHMERZEN-SELBSTHILFEGRUPPEN  
BUNDESVERBAND FÜR ANGEBORENE GEFÄSSFEHLBILDUNGEN E.V.  
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 E. V  
DEUTSCHE GBS CIDP 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 E.V.  
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.  
HAEVEREINIGUNG 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 DELETIONS-SYNDROM 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  
MASTOZYTÖSE 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  
PATIENTENVERBAND FAMILIÄRE AMYLOID POLYNEUROPATHIE 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  
SKLERO DERMIE SELBSTHILFE E.V.  
SMA EUROPE  
SOS-DESMOID E.V.  
SSADH-DEFIZIT E.V  
STIFF-PERSON VEREINIGUNG 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.

**XP - FREU(N)DE MONDSCHNEINKINDER**

ZNM - ZUSAMMEN STARK! E. V.

**GREECE**

«TO MELLON» ASSOCIATION OF PEOPLE WITH GENETIC DISORDERS  
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  
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**

ASOCIACION NACIONAL GUATEMALTECA PARA LAS ENFERMEDADES DE DEPOSITO LISOSOMAL  
PRÓCRECE

**HONG KONG, SAR OF CHINA**

JOSHUA HELLMANN FOUNDATION FOR ORPHAN DISEASE

**HUNGARY**

(HUNGARIAN HAEMOPHILIA SOCIETY) MAGYAR HEMOFÍLIA EGYESÜLET  
MAGYAÓRSZÁGI MITOCHONDRÍÁLIS BETEGEK ALAPÍTVÁNYA  
PRIMER IMMUNHIANYOS BETEGEK EGYESÜLETE  
RARE DISEASES HUNGARY - RIROSZ

**ICELAND**

AHC FEDERATION OF EUROPE  
ALTERNATING HEMIPLEGIA OF CHILDHOOD ASSOCIATION OF ICELAND  
EINSTÖK BÖRN - SUPPORT GROUP FOR CHILDREN WITH RARE DISORDERS  
GÜDRUN'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  
BARRETTSTOWN SERIOUS FUN  
CYSTINOSIS IRELAND  
FIGHTING BLINDNESS  
FRIEDREICH'S ATAXIA RESEARCH ALLIANCE  
HUNTINGTON'S DISEASE ASSOCIATION OF IRELAND  
IRISH CANCER SOCIETY  
MUSCULAR DYSTROPHY IRELAND  
NEUROFIBROMATOSIS ASSOCIATION OF IRELAND  
POPSYCLE FOUNDATION  
RARE DISEASES IRELAND  
SICKLE CELL AND THALASSAEMIE IRELAND  
SYRINGOMYELIA IRELAND  
THE CAVAN TOMMY HOEY TRUST  
THE IRISH FRAGILE X SOCIETY

**ISRAEL**

ISRAEL ADULT POLYGLUCOSAN BODY DISEASE

**ITALY**

ABC ASSOCIAZIONE BAMBINI CRI DU CHAT  
ACONDROPLASIA - INSIEME PER CRESCERE - ONLUS  
AIBWS ONLUS  
AMICI DELLA PORFIRIA - SAN PIO DA PIETRELcina ONLUS  
ANGELI NOONAN  
ASSOCIAZIONE PERSONE CON MALATTIE REUMATICHE - APMAR ONLUS  
ASSOCIAZIONE CONTO ALLA ROVESCIA  
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  
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 LIGURE THALASSEMICI ONLUS  
ASSOCIAZIONE MALATI DI HAILEY HAILEY DISEASE  
ASSOCIAZIONE MALATTIE RARE DELL'ALTA MURGIA ONLUS  
ASSOCIAZIONE NAZIONALE ITALIANA MALATI SINDROME DI SJOGREN  
ASSOCIAZIONE NAZIONALE PEMFIGO/PEMFIGOIDE  
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  
GRUPPO ITALIANO PER LA LOTTA ALLA SCLERODERMIA ONLUS  
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 - RARE DISEASES ITALY  
UNIONE ITALIANA ITTIOSI  
UNITED ONLUS  
VISUS - ASSOCIAZIONE TRA AFFETTI AA 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 - MALADIES RARES LUXEMBOURG

## DEN I ASBL SYNDROME DE MARFAN

EEN HÄERZ FIR KRIIBSKRANK KANNER ASBL  
FONDATION KRIIBSKRANK KANNER

## RETT SYNDROME EUROPE

## MALAYSIA

MALAYSIA METABOLIC SOCIETY  
MALAYSIAN RARE DISORDERS SOCIETY

## MALTA

NATIONAL ALLIANCE FOR RARE DISEASES SUPPORT - MALTA

## MEXICO

PROYECTO PIDE UN DESEO MEXICO IAP  
RED SANFILIPPO

## MOLDOVA

COPIIII PLOII

## MONTENEGRO

NATIONAL ORGANISATION FOR RARE DISEASES

## MOROCCO

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

## NEPAL

GBS/CIDP FOUNDATION NEPAL

## NETHERLANDS

ALS PATIENTS CONNECTED

AUTOSOMAAL DOMINANT CEREBELLAIRE ATAXIE-VERENIGING NEDERLAND

BIJNIEVERENIGING NVACP

CHILDHOOD CANCER INTERNATIONAL

CMT-COVM

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

KAISZ CHILDREN WITH A AUTOINMUUM OR AUTOINFLAMMATORY DISEASE

MSS (MARSHALL-SMITH SYNDROME) RESEARCH FOUNDATION

NEDERLANDS NETWERK VOOR LYMFODEEM EN LIPOEDEEM

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

SARCOIDOSE BELANGENVERENIGING NEDERLAND

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 SPIERZIEKTEN NEDERLAND - DUTCH ASSOCIATION FOR NEUROMUSCULAR DISEASE

VOLWASSENEN, KINDEREN EN STOFWISSELINGSZIEKTEN

VSOP - VERENIGING SAMENWERKENDE OUDER EN PATIËNTENORGANISATIES

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 RADZIWIŁŁOWA CHILD'S HEART FOUNDATION

**PORTUGAL**

ALIANÇA PORTUGUESA DE ASSOCIAÇÕES DAS DOENÇAS RARAS

APLL - ASSOCIAÇÃO PORTUGUESA DE LEUCEMIAS E LINFOMAS

ASSOCIAÇÃO NACIONAL DE DISPLASIAS ÓSSEAS

ASSOCIAÇÃO PORTUGUESA CDG E OUTRAS DOENÇAS METABÓLICAS

ASSOCIAÇÃO PORTUGUESA DE CHARCOT-MARIE-TOOTH

ASSOCIAÇÃO PORTUGUESA DE DOENTES NEUROMUSCULARES

ASSOCIAÇÃO PORTUGUESA DE OSTEOGÊNESE IMPERFEITA

ASSOCIAÇÃO SANFILIPPO PORTUGAL

FEDRA - FEDERAÇÃO PORTUGUESA DE DOENÇAS RARAS (SUSPENDED)

RARISSIMAS - ASSOCIAÇÃO NACIONAL DE DEFICIÊNCIAS MENTAIS E

RARAS (SUSPENDED)

**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

AUTOIMMUNE DISEASES PATIENTS ASSOCIATION

MASTOCYTOSIS SUPPORT ASSOCIATION ROMANIA

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

INTERREGIONAL PUBLIC ORGANISATION OF PATIENTS WITH FABRY DISEASE «THE ROAD TO LIFE»

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 «ZVONČICA»

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 OJEDLINELYCH GENETICKYCH 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**

ACCIÓN Y CURA PARA TAY-SACHS - ACTAYS

ALIANZA ESPAÑOLA DE FAMILIAS DE VON HIPPEL LINDAU

ASOCIACIÓ CATALANA DE LAS NEUROFIBROMATOSIS

ASOCIACIÓ D'AFECTATS DE SIRINGOMIÈLIA

ASOCIACIÓN ANDALUZA CONTRA LA FIBROSIS QUÍSTICA

ASOCIACIÓN ANDALUZA DE PACIENTES CON SÍNDROME DE

TOURETTEY TRASTORNOS ASOCIADOS

ASOCIACIÓN ARTROGRIPOSIS MÚLTIPLE CONGÉNITA

ASOCIACIÓN CHIARIY SIRINGOMIÈLIA DEL PRINCIPADO DE

ASTURIAS

ASOCIACIÓN DE AFECTADOS DE NEUROFIBROMATOSIS

ASOCIACIÓN DE AFECTADOS POR DISPLASIA ECTODÉRMICA

ASOCIACIÓN DE AFECTADOS POR HIPERINSULINISMO CONGÉNITO

ASOCIACIÓN DE ATROFIA DE NERVIÓ OPTICO DE LEBER

ASOCIACIÓN DE EPIDERMOLISIS BULLOSA DE ESPAÑA (DEBRA

ESPAÑA)

ASOCIACIÓN DE ESCLERODERMIA CASTELLÓN

ASOCIACION DE FAMILIARES Y AFECTADOS POR LIPODISTROFIAS

ASOCIACION DE FAMILIARES Y ENFERMOS DE FPIY FPF

ASOCIACIÓN DE HEMOGLOBINURIA PAROXÍSTICA NOCTURNA

ASOCIACIÓN DE HUESOS DE CRISTAL DE ESPAÑA

ASOCIACIÓN DE LAS MUCOPOLISACARIDOSIS Y SÍNDROMES

RELACIONADOS

ASOCIACIÓN DE NEVUS GIGANTE CONGÉNITO

ASOCIACIÓN ESPAÑOLA DEL SÍNDROME CDG, DEFECTOS

CONGENITOS DE LA GLICOSILACION

ASOCIACIÓN ESPAÑOLA ANIRIDIA

ASOCIACIÓN ESPAÑOLA DE ANGIOEDEMA FAMILIAR

ASOCIACIÓN ESPAÑOLA DE ENFERMOS DE GLUCOGENOSIS

ASOCIACION ESPAÑOLA DE ENFERMOS DE POMPE

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 MASTOCITOSIS Y ENFERMEDADES

RELACIONADAS

ASOCIACIÓN ESPAÑOLA DE PARAPRESIA 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

ASOCIACION NACIONAL DE HIPERTENSIÓN PULMONAR

ASOCIACIÓN NACIONAL SÍNDROME DE APERTY OTRAS

CRANEOSINOSTOSIS SINDRÓMICAS

ASOCIACIÓN SÍNDROME DE ANGELMAN

ASOCIACIÓN SÍNDROME DE LOWE DE ESPAÑA

ASOCIACIÓN SÍNDROME LESCH NYHAN ESPAÑA

ASOCIACIÓN 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

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

FEDER - FEDERACIÓN ESPAÑOLA DE ENFERMEDADES RARAS

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 FIBROSIS QUÍSTICA

FEDERACIÓN ESPAÑOLA DE HEMOFILIA

FEDERACION ESPAÑOLA DE MALFORMACIÓN DE CHIARIY PATOLOGÍAS

ASOCIADAS

FEDERACION ESPANOLA DE PADRES CON NIÑOS CON CANCER

FEDERACIÓN ESPAÑOLA DEL SÍNDROME X FRÁGIL

FUNDACIÓN ANDRÉS MARCIO, NIÑOS CONTRA LA LAMINOPATÍA

FUNDACION MENUDOS CORAZONES  
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HPE-ORG  
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)  
SVENSK FÖRENING FÖR HYPOPARATYREODISM  
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  
FMF & AID GLOBAL ASSOCIATION  
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  
SAVE LIVER ASSOCIATION OF PATIENTS WITH LIVER DISEASES  
ZDRUZENIE ZA CISTICNA FIBROZA (MACEDONIAN CF ASSOCIATION)

## TURKEY

KIFDER  
MUKOPOLISAKKARIDOZ VE BENZERI LIZOZOMAL DEPO HASTALIKLARI  
DERNERGI

## UKRAINE

ASSOCIATION OF PATIENTS WITH PULMONARY 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  
UPO-UKRAINIAN ASSOCIATION OF HELP FOR PATIENTS WITH CF

## 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 - NEUROENDOCRINE TUMOUR  
SUPPORT  
ANNABELLE'S CHALLENGE  
ASSOCIATION FOR GLYCOGEN STORAGE DISEASE  
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 LUNG FOUNDATION  
EUROPEAN MYASTHENIA GRAVIS ASSOCIATION  
EUROPEAN TUBEROUS SCLEROSIS COMPLEX ASSOCIATION  
FABRY INTERNATIONAL NETWORK  
FETAL ANTI CONVULSANT SYNDROME ASSOCIATION  
FINDACURE: THE FUNDAMENTAL 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  
PITT HOPKINS UK  
POLYCYSTIC KIDNEY DISEASE CHARITY (PKDC)  
PRADER WILLI SYNDROME ASSOCIATION UK  
PSC SUPPORT  
RARE AUTOINFLAMMATORY CONDITIONS' COMMUNITY UK  
RARE DISEASES UK  
RING 20 RESEARCH AND SUPPORT UK CIO  
SPINAL MUSCULAR ATROPHY SUPPORT UK  
STIFF PERSON SUPPORT GROUP  
STURGE-WEBER UK  
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 (APSFA)  
CHORDOMA FOUNDATION  
CURE PSP  
CYSTINOSIS FOUNDATION  
FMD CHAT  
INTERNATIONAL FOXG1 FOUNDATION  
INTERNATIONAL PEMPHIGUS & PEMPHIGOID FOUNDATION  
INTERNATIONAL WALDENSTROM'S MACROGLOBULINEMIA  
FOUNDATION  
NORD NATIONAL ORGANIZATION FOR RARE DISORDERS  
NTM INFO & RESEARCH  
PTEN HAMARTOMA TUMOR SYNDROME FOUNDATION  
PURA SYNDROME FOUNDATION  
THE CUSHING SUPPORT & RESEARCH FOUNDATION  
THE OXALOSIS & HYPEROXALURIA FOUNDATION

## URUGUAY

ASOCIACION ACONDROPLASIA URUGUAY

# PARTICIPATION OF EURORDIS' REPRESENTATIVES IN PUBLIC EUROPEAN / INTERNATIONAL CONFERENCES & Workshops 2017

**2017 Next Wave Forum, Massachusetts Institute of Technology (MIT), USA, 29 December**

**Mathieu Boudes:** Panelist in the sessions "ABI: A Vision for Sustainable, Patient-Centered Drug - Development, Access, & Value" & "Stakeholder Perspectives on Platform Trials"

**EMA Workshop: "Site and Histology – Independent Indications in Oncology", London, UK, 14-15 December**

**François Houÿez:** "Views of the Patients"

**'Genetic Counselling in Genomic Medicine Symposium', University Hospital Ghent, Belgium, 7 December**

**Lieven Bauwens:** "The role of genomics in rare diseases"

**The International Symposium on "Accessibility to high-value medicines, the new frontier?", London School of Hygiene and Tropical Medicines, London, UK, 6 December**

**Yann Le Cam:** "Breaking the Access Deadlock: A "new deal" to improve access to rare diseases therapies"

**Rare Cancers Europe (RCE) – ESMO – ESO Training Course for Rare Cancer Patient Advocates (solid tumours in adults), Milan, Italy, 1-4 December**

**Ariane Weinman:** "The Power of Rare Disease Patient Advocacy"

**2nd Regional Education for Patient Representatives on Rare Diseases from Ex-Yugoslavian Countries, Vrdnik, Serbia, 1-4 December**

**Sandra Pavlovic:** "EURORDIS: RareConnect"

**EMA workshop: "Data-anonymisation– a key enabler for clinical data sharing", London, UK, 30 November- 1 December**

**François Houÿez:** "Defining sensitive data – Influence of the context of the disease on the tolerability of risk"

**TREAT-NMD conference, Frieberg, Germany, 28 November**

**Simone Boselli:** "Breaking the Access Deadlock: Patients' recommendations to leave no one behind"

**European Commission Conference «Opening up to an ERA of Social Innovation», Lisbon, Portugal, 27-28 November**

**Raquel Castro:** Panellist in the session: "Social Innovation and the Care Society"

**French Pharmacovigilance day - Pharmacovigilance by 2020, Paris, France, 27 November**

**François Houÿez:** "The Pharmaco-vigilant patient"

**RARE 2017, Paris, France, 20-21 November**

**Yann Le Cam:** « Bilan de l'action de la France en Europe et de ce que l'Europe pourrait inspirer à la France, du point de vue des malades » (Patient's perspective on the state of the art of France's activities at the European level and what Europe could inspire to French rare disease policy)

**Mathieu Boudes:** Panelist in the session: Engagement of the expert patient in all stages of the drug development process.

**Anne-Sophie Lapointe** and **Ariane Weinman** also represented EURORDIS.

**Workshop: "Role of Hospital Managers in Shaping the Future of the European Reference Networks", Rotterdam, the Netherlands, 16-17 November**

**Matt Johnson:** "The view of the patients: How can Hospitals benefit and contribute to the success of the ERNs?"

**Orphan Drug Congress Europe 2017, Barcelona, Spain, 14-15 November**

**Yann Le Cam:** Panelist in the session "Executive industry discussion: Intentions of pharmaceutical companies developing treatments for rare disease patients – What do they hope to achieve?"

**ACHSE Round Table of Companies, Berlin, Germany, 14 November**

**François Houÿez:** "Early Access to Medicines in Europe: How to make Compassionate Use Become a Reality for All in Need"

**Meeting of the "Société des Internes en Génétique de France", Paris, France, 14 November**

**Virginie Bros-Facer:** Presentation of EURORDIS

“Health Series 2017: Multi-stakeholder high level debate on the role of pharmaceutical incentives in securing Europe’s future as a leader in pharmaceutical innovation, which can deliver growth and sustainable health”, The Permanent Representation of Denmark to the EU, Brussels, Belgium, 14 November

**Simone Boselli** represented EURORDIS

Fostering Responsible Research with CRISPR-Cas9, INSERM External European Experts Meeting, Paris, France, 13 November

**Virginie Bros-Facer:** “Rare Disease Patients & Genome editing: Perspectives and Engagement”

Italian Chamber of Deputies, Rome, Italy, 9 November

**Simona Bellagambi:** Presentation of the European Parliamentary Network

“More Trust, More Data, Better Health, How does Europe grasp the innovation opportunity?”, Brussels, Belgium, 9 November

**Simone Boselli** represented EURORDIS

EuropaBio Patients Bio-Forum: “European Reference Networks: how to make the most of them?”, Brussels, Belgium, 9 November

**Matt Johnson:** “The patients’ perspective – First-hand experience with ERNs and the future role for patients”

United Parent Project Muscular Dystrophy meeting, Birmingham, UK, 9 November

**François Houÿez:** “Drug Development, Evaluation and Reimbursement (and other technologies): Times are Changing”

DIA (Drug Information Association) Workshop: “Value, Access & Regulatory Strategy” Basel, Switzerland, 25-26 October

**François Houÿez:** 1) Meeting the needs of patients  
2) What does fast patient access mean? Risks, early access and trade-offs

European Commission’s Workshop on GDPR Implementation and Health Data (co-organised by DG CONNECT, DG JUST, DG SANTE and DG RTD), Brussels, Belgium, 23 October

**Virginie Bros-Facer:** “Rare Disease Patients: Perspectives on the GDPR in health and research”

«XIII Congreso Estatal y I Iberoamericano del Trabajo Social» (XIII Spanish and Iberoamerican Social Work Congress), Mérida, Spain, 19-21 October

**Raquel Castro:** “Trabajo Social y Enfermedades Raras” (Social Work and Rare Diseases)

European Parliament: launch of the White Paper on Informal Carers of People with Cancer - Hosted by MEP Heinz Becker (EPP, Austria), Brussels, Belgium, 19 October

**Ariane Weinman** represented EURORDIS

NORD Annual Summit, Arlington, USA, 16-17 October

**Mathieu Boudes** represented EURORDIS

STOA (Science and Technology Options Assessment) European Parliament, Brussels, Belgium, 11 October

**Yann Le Cam:** “Now is the time to guarantee access to the therapies of the future”

«Thérapie Génique : des maladies rares à l’oncologie, quelles avancées, quels marchés ? » (Gene Therapy: From Rare Diseases to Oncology, which steps forwards, what markets?), Imagine Institute, Paris France, 2 October

**Ariane Weinman** represented EURORDIS.

Journée des anomalies du développement pour les personnes Sans Diagnostic et Unique, Paris, France, 22 September

**Virginie Bros-Facer:** “Initiatives Internationales SANS DIAGNOSTIC”

EMA-FDA Rare Diseases Cluster Meeting on Compassionate Use Programmes, London, UK, 21 September

**François Houÿez** represented EURORDIS

EMA Pharmacovigilance Forum, 21 September

**François Houÿez** represented EURORDIS

EMA Workshop on antibiotic microbial resistances, London, UK, 19 September

**François Houÿez** represented EURORDIS

5th International Summer School of the Centre for Rare Diseases of the Istituto Superiore di Sanità (ISS): “Rare Disease & Orphan Drug Registries and Bring Your Own Data”, Rome, Italy, 18-22 September

**Virginie Bros-Facer:** “Patients Registry in ERNs: the patients’ perspective and role of ePAGs”

European EUnetHTA JA3 Stakeholder Forum, Amsterdam, the Netherlands, 14 September

**François Houÿez:** “Views of Patients about Horizon Scanning and Topic Prioritisation”

ESMO 2017 Congress (European School of Medical Oncology): “Integrating science into oncology for a better patient outcome”, Madrid, Spain, 8-11 September

**Ariane Weinman** represented EURORDIS

WEB-RADR Stakeholder Event, London, UK, 7 September

**François Houÿez:** “The social media impact for patients”

CDDF (Cancer Drug Development Forum) Multi-Stakeholder Workshop: “Access to Innovative Oncology Drugs in Europe”, Madrid, Spain, 7 September

**Matteo Scarabelli:** “How to involve patients in HTA”

“Fit for Rare, Fit for All!” conference, Cluj-Napoca, Romania, 3 September

**François Houÿez:** “How to improve early and equal access to medicines in the EU”

5th International Conference on Rare and Undiagnosed Diseases (UDNI), Stockholm, Sweden, 30-31 August

**Virginie Bros-Facer:** “Rare Disease Patients: Perspectives and Engagement in Data Sharing and Data Protection”

Presentation of the 3rd report MonitoRARE, in collaboration with the Parliamentary Intergroup on Rare Diseases, Rome, Italy, 27 July

**Simona Bellagambi:** Presentation of the Italian situation within the European framework and based on the EUCERD Indicators

3rd World Conference on CDG, Leuven, Belgium, 15-16 July

**Raquel Castro:** “INNOVCare: bridging the gaps between health, social and local services to improve care of people living with rare and complex conditions”

IAPO African Regional Meeting, Entebbe, Uganda, 4-5 July

**François Houÿez:** “Public Engagement: How Patients and Consumers Work with the EMA” (presented by video)

How can Big Data contribute to healthcare from the point of view of: patients, doctors and mutuals/health insurance funds? Brussels, Belgium, 29 June

**Simone Boselli** presented the point of view of patients

**How to Ensure Treatments and the Respect of Patients' time across Europe? Focus on timely access to innovative medicines after the European Commission approval, European Parliament, Brussels, Belgium, 28 June**

**Simone Boselli** presented the point of view of patients on "How to ensure timely access to innovative medicines in Europe"

**EMA-FDA Rare Diseases Cluster, EMA, London, UK, 27 June**

**François Houÿez:** "EURORDIS Position on Compassionate Use"

**« La santé: un levier d'attractivité ? » (Health: a mechanism for growth?), Representation of the European Parliament in France, Paris, France, 27 June**

**Eva Bearryman** represented EURORDIS

**First ICPeMed Workshop "Innovative Concepts on Data Generation and Use of Personalised Medicine Research", Milan, Italy, 26-27 June**

**Virginie Hivert** represented EURORDIS

**Genetics Clinic of the Future (GCOF), Rotterdam, The Netherlands, 26 June**

**Mathieu Boudes** represented EURORDIS

**"Patient engagement and building value partnerships between patient associations and the healthcare industry", Milan, Italy, 20 June**

**Simona Bellagambi** represented EURORDIS

**HTAi conference, Roma, Italy, 19-21 June**

**François Houÿez:**

- 1) Advanced therapies and ethics, do we have all the answers?
- 2) How to improve early and equal access to medicines in the EU
- 3) Joint HTA

**DIA Annual Meeting, Chicago, USA, 20 June**

**Mathieu Boudes** represented EURORDIS

**EFPIA annual conference: "Unlocking tomorrow's cures", Brussels, Belgium, 14 June**

**Yann Le Cam** and **Simone Boselli** represented EURORDIS

**Second annual conference of the EC Scientific Panel for Health: "Health research in a connected and participative society", Brussels, Belgium, 9 June**

**Mathieu Boudes** represented EURORDIS

**WHY EUROPE? Experience sharing, connection of national patient organisations into the EU structures, European Parliament, Brussels, Belgium, 7 June**

**Valentina Bottarelli** and **Simone Boselli** represented EURORDIS

**Forum meeting: discussion on the development of a Code of Conduct in the framework of the General Data Protection [EU] Regulation; organised by BBMRI-ERIC, Brussels, Belgium, 7 June**

**Virginie Bros-Facer**, Panelist in the session on patients' needs

**OECD Forum 2017: "Bridging Divides", Paris, France, 6-7 June**

**Anne-Sophie Lapointe:** Panelist in the session on people-centred health

**Workshop «Advancing Holistic & Innovative Care for Rare Diseases & Complex Conditions», INNOVCare project, Cluj-Napoca, Romania, 1-2 June**

**Raquel Castro:** "Juggling care and daily life: The balancing act of the rare disease community - First Europe-wide survey on social impact of rare diseases"

**European Food Safety Authority Stakeholder Forum 31 May, Parma, presenting on the EMA Framework of Interaction with Patients and Consumers**

**François Houÿez** represented EURORDIS.

**1st Stakeholder Forum of the European Food Safety Authority (EFSA), Parma, Italy, 30-31 May**

**François Houÿez:** "EMA framework of Interaction with Patients and Consumers"

**8th Childhood Cancer International (CCI) Europe Regional Conference: "Connecting EUROPE", Rome, Italy, 12-14 May**

**Matt Bolz-Johnson:** "Patient Involvement: European Reference Networks"

**Ariane Weinman** represented EURORDIS as well.

**XI European Patients' Rights Day - European Economic and Social Committee, Brussels, Belgium, 10 May**

**François Houÿez:** "presenting on the Role the EU can Play to Make Access to Innovative Medicines More Sustainable"

**"The voice of Patients on Health Data: Saving lives and protecting patients' rights", workshop co-hosted by Microsoft, the EU Cloud in Health Advisory Council and EPF, Brussels, Belgium, 8 May**

**Virginie Bros-Facer:** "Rare Disease Patients: Perspectives and Engagement in Data Sharing and Data Protection"

**E-Rare Data Sharing and Harmonization Workshop, Berlin, Germany, 3-4 May**

**Virginie Bros-Facer:** "Patients participation in data sharing: Perspectives and engagement"

**RD-CONNECT Annual Meeting, Berlin, Germany, 1-3 May**

**Virginie Bros-Facer:** "Patient Engagement Throughout RD-CONNECT via the Patient Advisory Council"

**10th anniversary of the Spanish Network of Health Technology Assessment (HTA) Agencies, Zaragoza, Spain, 27-28 April**

**François Houÿez:** "presenting on Patients' and consumers' engaged in EMA: source of ideas for HTA"

**World Orphan Drugs Congress USA, Washington D.C., USA, 20-21 April**

**Yann Le Cam:** "Patient Engagement in Research & Product life cycle"

**"Building the Foundation for Genomic Medicine for Patients with Rare Diseases", 6th international seminar FHU – TRANSLAD (organised by the "filière" AnDDI-Rares), IMAGINE Institute, Paris, France, 20 April**

**Yann Le Cam:** panelist in the Round Table: Building the foundation for genomic medicine for patients with rare diseases

**Members Information Meeting (RIME) of the French Alliance for Rare Diseases, Paris, France, 12 April**

**Virginie Bros-Facer:** "Introduction to European Reference Networks and ePAGs – European Patient Advocacy Groups"

**DIA 29th Annual EuroMeeting: "Translational Health Care – From Bench to Bedside – and Back", Glasgow, UK, 29-31 March**

**Mathieu Boudes:** "ADAPTIVE PATHWAYS: The rare disease patients' perspectives"

**FP7 Small-population research methods projects (ASTERIX, InSpiRe, IDEAL) and regulatory application workshop, EMA, London, UK, 29-30 March**

**Elisa Ferrer** and **Kerry Leeson-Beevers** represented EURORDIS

- SCOPE joint action final conference, London, UK, 20-22 March**  
**François Houÿez:** "Collaboration to Promote and Support ADR Reporting and Feedback to Patients"
- Rare Diseases Registries Workshop – Organised by CHAFAE and CIBERER, Madrid, Spain, 21-22 March**  
**Virginie Bros-Facer:** "Patients Registry: the Patients' Perspective"
- Maltese EU Presidency event: "Integrating Research and Healthcare for Rare Diseases: a structured collaboration with high community added value", La Valetta, Malta, 20 March**  
**Yann Le Cam:** "Breaking the Access Deadlock to Leave No One Behind"  
**Simona Bellagambi** and **Avril Daly:** Panellists in the session: "Rare Diseases Research in the future: Perspectives, needs and opportunities"
- Terkel Andersen, Dorica Dan** and **Anne-Sophie Lapointe** also represented EURORDIS.
- SWAN (Syndromes Without A Name) Europe Network Meeting, Barcelona, Spain, 17-18 March**  
**Virginie Bros-Facer:** "International Initiatives - Undiagnosed Rare Diseases"
- Real world data contribution to regulatory and coverage decisions, EMSP conference at the European Parliament, Brussels, Belgium, 17 March**  
**François Houÿez:** "Registries and Data Protection"
- BioCapital Europe 2017, Amsterdam, the Netherlands, 15 March**  
**Avril Daly:** "Trends in Patient Advocacy: EURORDIS' structured approach on patient engagement in research, therapy development and health care; and focus on RETINA International"
- EMA Workshop: Personalised/precision medicines workshop, London, UK 14 March**  
**François Houÿez** represented EURORDIS.
- Fondazione Telethon: XIX Scientific Convention, Riva del Garda, Italy, 13-15 March**  
**Yann Le Cam:** OPENING LECTURE: "Leave no one behind"
- Meeting of the Commission Expert Group on Safe and Timely Access to Medicines for Patients ("STAMP"), Brussels, Belgium, 13-14 March**  
**Virginie Hivert** invited as patient representative
- 3rd European Commission Conference for European Reference Networks (ERNs) & ERN Kick-off meeting, Vilnius, Lithuania, 9-10 March 2017**  
**Yann Le Cam:** Chair of the Round Table on "ERNs and National Healthcare Systems"  
**Matt Bolz-Johnson, Valentina Bottarelli, Lenja Wiehe** represented EURORDIS as well.  
 ePAGs' representatives of each of the 24 approved ERNs attended as well.
- 7ème journée internationale des maladies rares, Lausanne, Suisse, 4 march**  
**Paloma Tejada** represented EURORDIS
- MALTA EU 2017: Structured Cooperation between Health Systems (Enhancing access to novel or highly specialized services, medicines & technologies), La Valetta, Malta, 1 - 2 March**  
**Yann Le Cam:** "Rare Diseases: How do we ensure small populations can access affordable innovations?"  
**Matt Bolz-Johnson** represented EURORDIS as well.
- Official Press Conference on launch of ERN on Rare Diseases Day, Leuven University Hospital, Belgium, 28 February**  
**Yann Le Cam:** "A patients' perspective on rare diseases in Europe"
- Italian event at the Chamber of Deputies on European Reference Networks and ePAGs on the occasion of the Rare Disease Day, Rome, Italy, 25 February,**  
**Simona Bellagambi:** Presentation of the European Patient Advocacy Groups – ePAGs - established by EURORDIS, to enhance the involvement of patient organisations in European Reference Networks for Rare Diseases
- Annual Conference on EU Law in the Pharmaceutical Sector, Brussels, Belgium, 23-24 February**  
**François Houÿez:** "How to improve early and equal access to medicines in the EU?"
- Rare Diseases International, 1st Policy Event, Geneva, Switzerland, 10 February**  
**Yann Le Cam:** "Trends in Patient Advocacy"
- IRDIRC Conference, Paris, France, 8-9 February**  
**Yann Le Cam:** "Trends in Patient Advocacy"  
**Mathieu Boudes, Virginie Hivert, Michele Lipucci di Paola** and **Chris Sotirelis** represented EURORDIS as well.
- I-Com conference, 8 February, European Parliament presenting on HTA in EU: from National best practices to EU cooperation**  
**François Houÿez** represented EURORDIS
- HTA Network meeting, Brussels, Belgium, 8 February,**  
**François Houÿez** represented EURORDIS
- The future of HTA in the EU: From national best practices to EU cooperation, European Parliament, Brussels, Belgium, 8 February**  
**François Houÿez:** Panelist in the Round Table: "EU Harmonization"
- BBMRI-ERIC operates and is developing a pan-European distributed research infrastructure of biobanks and biomolecular resources —Kick-start discussions on a code of conduct for data sharing, Brussels, Belgium, 1 February**  
**Virginie Bros-Facer** represented EURORDIS in the BBMRI Stakeholders forum
- European Cancer Congress, Amsterdam, the Netherlands, 28-30 January**  
**François Houÿez:** "Why would patients report side effects?"
- DIA (Drug Information Association), Washington DC, USA, 22-23 January**  
**François Houÿez:** "Pharmacovigilance Legislation Impact on Patient Safety Outcomes"
- National Institute for Clinical Excellence, London, UK, 17 January**  
**François Houÿez:** Bringing Patient Perspectives to NICE: Training and Accompanying Patients
- Grande conférence maladies rares 2017, organisée par l'Alliance française des maladies rares, Palais du Sénat, Paris, France, 13 January**  
**Yann Le Cam,** Panelist: « Pourquoi et comment faire des maladies rares une priorité nationale? »  
**Mathieu Boudes** and **Ariane Weinman** represented EURORDIS as well.
- Agence Nationale de Sécurité des Médicaments, ANSM, Paris, France 9 January, (about the Form to Collect Spontaneous Reports of Suspected Adverse Drug Reactions)**  
**François Houÿez** represented EURORDIS

# ACKNOWLEDGEMENTS

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## Patient Organisations and Public Entities

### AFM - TÉLÉTHON

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



### EUROPEAN COMMISSION

DG Health and Food Safety

- The Operating Grant for year 2017
- RD-ACTION – Rare Diseases Joint Action – Data and policies for Rare Diseases
- JARC – Joint Action on Rare Cancers



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### 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
- Genetics Clinic of the Future – A stepping stone approach towards the Genetics Clinic of the Future
- The Innovative Medicines Initiative-Joint Undertaking (IMIJU) project:
  - ADAPT SMART - Accelerated Development of Appropriate Patient Therapies - a Sustainable, Multistakeholder Approach from Research to Treatment outcomes
  - European Patients' Academy on Therapeutic Innovation (EUPATI)
  - Web-Radr - Recognising Adverse Drug Reactions



Co-funded by the 7th Framework Programme of the European Union



Co-funded by the Horizon 2020 Framework Programme of the European Union



### EUROPEAN COMMISSION

DG Employment, Social Affairs & Inclusion

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



Co-funded by the Employment and Social Innovation (EaSI) Programme of the European Union

# Health Sector Corporates

Diversification of funding is key to minimising potential conflict of interest with donors. EURORDIS has diversified its pharmaceutical and biotechnology sector contributors from 53 to 64 different companies in 2017. Eight other health sector companies also contributed to the 2017 incomes. Companies have supported EURORDIS through the EURORDIS Round Table of Companies<sup>1</sup>, the EURORDIS Membership Meeting Budapest 2017, the Multi-Stakeholder Symposium, the EURORDIS Black Pearl Awards<sup>2</sup>, as well as international initiatives such as Rare Disease Day, Rare Barometer<sup>3</sup>, RareConnect™<sup>4</sup>, Rare Diseases International<sup>5</sup> and the co-funding of DG SANTE Joint Action of Rare Cancers. 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	
	PFIZER	
3	CELGENE	
4	VERTEX	
5	NOVARTIS	

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

<sup>2</sup> <https://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 & Biotechnology Companies & Health Sector Corporates



In-kind contributors

BURSON-MARSTELLER			DLA PIPER UK LLP
GOOGLE			MEDIAPLANET
PUBLICIS HEALTH			REFLEX MEDIA
TROMMONS, MAINTAINED BY THE ROSETTA FOUNDATION			TRANSLATORS WITHOUT BORDERS

Other contributors

ASSOCIATION OF INTERNATIONAL PHARMACEUTICAL MANUFACTURERS



KINDNESS FOR KIDS



Foundation for children with rare diseases.  
Moving mountains for kids in need.



**EURORDIS**  
RARE DISEASES EUROPE

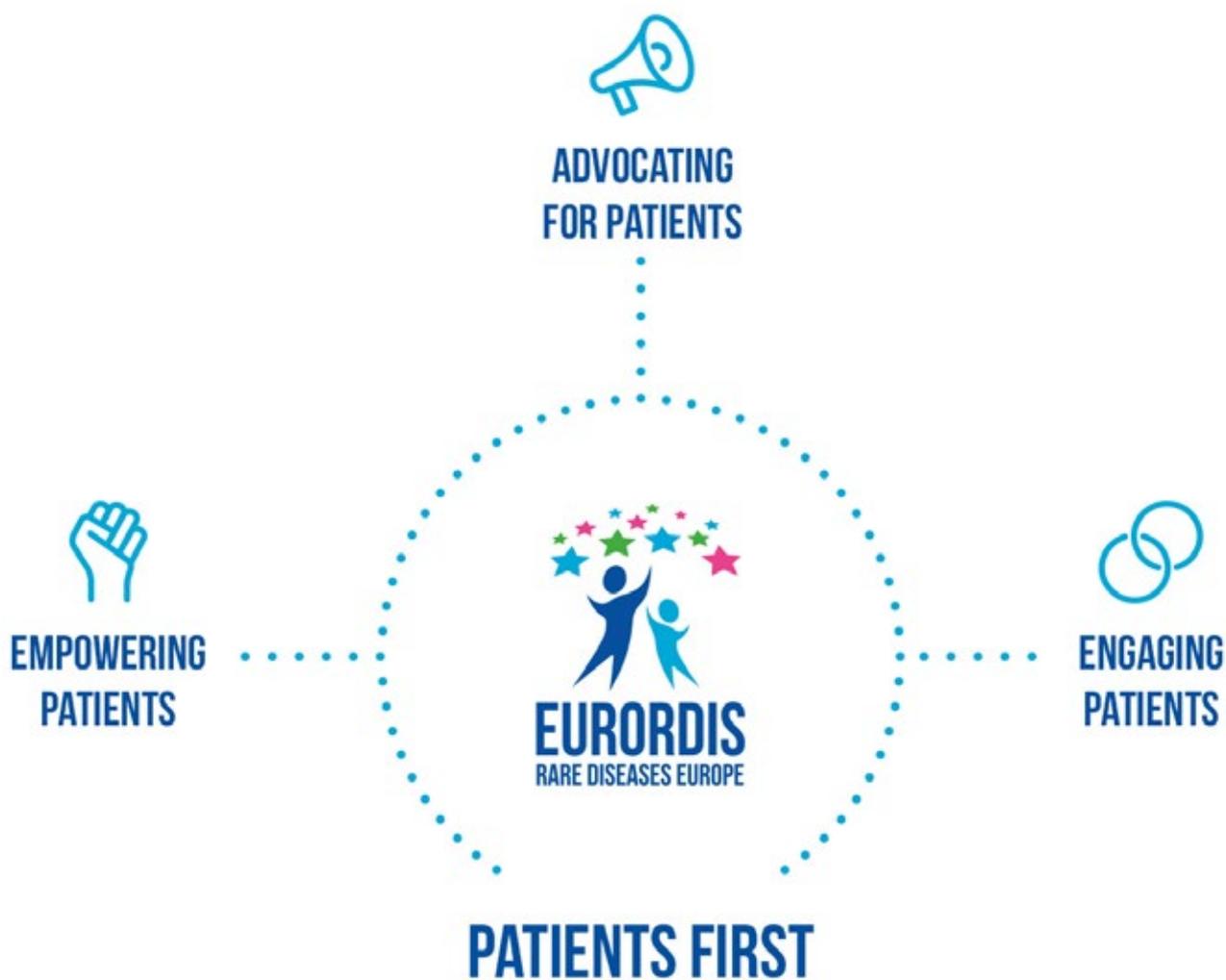
# WORKPLAN

2018

- ACTION PLAN 2018 ..... 68
- GOVERNANCE CHART 2018 ..... 75
- EXTERNAL REPRESENTATION CHART 2018..... 76
- TEAM CHART 2018 ..... 78
- REVENUE & EXPENSES 2018 ..... 79

# ACTION PLAN

## 2018



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

- + Prepare for the next decade of rare disease policy making to take the necessary steps to requalify rare diseases as a public health issue.
- + Focus on the next **EU Multiannual Financial Framework 2020-2027** that will fund EU policies and programmes. Follow the discussions and participate in the stakeholder consultation to be launched by the European Commission in early 2018.
- + Continue the development of the **Parliamentary Advocates for Rare Diseases**, launched in late 2017. Organise a brainstorming meeting with parliamentarians for the identification of priority actions and initiatives that the network can carry out before the end of the legislative term. Expand the network to include members of national and regional parliaments, in collaboration with National Alliances.
- + Continue the work with the **Working Group on Future Policy Priorities for Rare Diseases**, set up in late 2017 and composed of members of the CNA, for the purpose of identifying upcoming policy priorities. Present the outcomes at the ECRD 2018 Vienna.
- + Submit a proposal for a **Foresight Study on Rare Diseases in 2030**, within the call for proposals to be announced in early 2018, to identify long term policy scenarios with participatory methodologies.

## 1.3 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 and in the Innovative Medicines Initiatives**
- + Continue supporting **rare diseases as a public health priority in the 3rd EU Public Health Programme 'Health for Growth'** and advocate on the rare disease community's priorities
- + Engage with other EU health stakeholders in a campaign calling for **enhanced EU action in the field of health #EUHealth**
- + 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, for submission in 2018

## 1.4 ADVOCATE FOR THE DEVELOPMENT OF THE ERNS FORM AND FUNCTIONS AND THE INTEGRATION OF ERNS WITH WIDER HEALTH, SOCIAL AND RESEARCH INFRASTRUCTURES

- + Promote the deployment of ERNs & support the implementation of their key functions by: promoting further geographical and rare disease coverage; participation & contribution to the Board of Member States; participation & contribution to the ERN coordinators group and the ERN WGs; preparation of tender to support ERN implementation
- + Promote integration of ERNs into national healthcare systems, in collaboration with National Alliances (NAs) by developing a capacity-building programme to support 10 NAs through 10 local face-to-face meetings; gap analysis of potential HCP – full or affiliated - to fill expertise and geographical gaps; promoting the create a working group of National Alliances in EU MS with small populations to identify specific needs, challenges and ad hoc strategy; engaging with Board of Member States WG on integration into national healthcare systems

## 1.5 ADVOCATE FOR HEALTH TECHNOLOGY ASSESSMENT METHODS AND A EUROPEAN HTA AGENCY WHICH ADDRESSES RARE DISEASE PATIENT NEEDS

- + Prepare, adopt and disseminate a **EURORDIS Statement on the European Commission Proposal for a Regulation on the European HTA Cooperation**
- + Organise relevant meetings and contact EU policy makers

- + Take action to empower EURORDIS members to advocate nationally through a series of webinars and organise a dedicated session at the CNA meeting 15 March, Brussels
- + Organise meetings with National Alliances
- + Prepare a common position with other patient organisations which highlights the patient position and needs, for dissemination

## 1.6 ADVOCATE IN SUPPORT OF RARE DISEASE RESEARCH

- + Define the **EURORDIS position on data sharing/data protection** through the RD-Connect project and the Rare Barometer programme, in light of the new EU Data Protection Regulation

## 1.7 VOICING THE SOCIAL NEEDS OF PEOPLE WITH A RARE DISEASE AND ADVOCATING FOR THE INTEGRATION OF RARE DISEASES INTO SOCIAL POLICY

- + Continue to disseminate the results of the first European survey on the social impact of rare diseases - "Juggling care and daily life: the balancing act of the rare disease community" (conducted via Rare Barometer Voices and within the INNOVCare project) - amongst members, rare disease stakeholders and wider audiences
- + **Conclude EURORDIS' position paper on social policy** and disseminate it widely, gathering support amongst and strengthening partnerships with rare disease and social stakeholders
- + Continue to identify and act on social policy and disability emerging topics and areas of work at European level, notably within the Social Pillar initiative, to ensure the representation of people living with a rare disease and their carers in relevant EU policies
- + Engage with Members of the European Parliament active in social policy, including in disability and employment, notably via the Network of Parliamentary Advocates

## 1.8 PROMOTE RARE DISEASES AS AN INTERNATIONAL PUBLIC HEALTH PRIORITY THROUGH:

### Rare Diseases International:



RARE  
DISEASES  
INTERNATIONAL

- + Support RDI in its establishment as an **independent legal entity**: EURORDIS will assist with the development of separate RDI Statutes and the legal incorporation of the organisation in France. EURORDIS & RDI will sign a Memorandum of understanding to ensure support for the RDI initiative for the period 2019-2023

- + Continue to expand the RDI membership base and to engage members: Organise the annual RDI membership meeting in Vienna in May; hold regular webinars with members; establish a Facebook Workplace forum
- + Pursue strategic partnerships with: Orphanet; International Rare Diseases Research Consortium - Patient Advocates Committee; International Federation of Pharmaceutical Manufacturers & Associations (IFPMA) Working Group on RD

### NGO Committee for Rare Diseases:



- + Continue actions within the **UN system** jointly with RDI by joint outreach with RDI to Permanent Missions in New York and Geneva
- + Continue actions with the **WHO** jointly with RDI by developing relations with staff at the Division of Universal Health Coverage and Health Systems, staff at the Division on Access to Medicines, Medical devices and Assistive technology, Health attaches at the Permanent Missions to the UN in Geneva
- + **Work towards organising a High Level Meeting in UN Headquarters**, New York, to be held in 2019
- + Contribute to the Report of the Office of the High Commissioner for Human Rights (OHCHR): The Right to Health framework to implement health-related SDGs **to be presented at Human Rights Council 38th session (June 2018) Geneva**
- + Contribute to the Report of the UN Special Rapporteur on the rights of people with disabilities: The Right of persons with disabilities to the highest attainable standard of health **to be presented at 73rd session UN General Assembly Third Committee (Oct 2018) New York**

## 1.9 EURORDIS RARE BAROMETER PROGRAMME: GENERATING DATA FROM PATIENT EXPERIENCE



- + Continue the Rare Barometer growth internationally by developing the Rare Barometer Voices sample in countries outside Europe. Topics explored through the programme will be aligned with policy priorities at EURORDIS, including data protection and data sharing, access to treatment and diagnosis and will consider international policy priorities (Sustainable Development Goals of the UN).
- + Explore the possibility to develop a common mechanism to collect patients' experience through the ERNs through collaboration of Rare Barometer and ERNs

## 2. Patient Empowerment

### 2.1 BUILDING THE COMMUNITY & NETWORKING

#### 2.1.1 Membership

- + Maintain EURORDIS' Membership at over 800 members and ensure regular interaction
- + Maintain process of regular membership reassessment, as established in 2013

#### 2.1.2 RareConnect



- + Consolidate EURORDIS' new role at the heart of the new RareConnect governance structure
- + Examine the possibility of developing a research platform to allow researchers to interact with patients and families through specific questionnaires and data-capture of phenotypic information
- + Explore the possibility of adding further languages such as Farsi and Greek.

### 2.2 BUILDING THE CAPACITY OF PATIENT ADVOCATES

#### 2.2.1 Communications tools

- + Maintain and regularly evaluate and update EURORDIS' communications tools such as the EURORDIS website, eNews, dedicated Member News, EURORDIS' social media, webinars programme
- + Expand the EURORDIS webinar programme to include a new 'how to' webinar series for new or small patient organisations which will provide training on practical skills for how to run a patient organisation (for example, governance or communications), also including speakers from outside EURORDIS

#### 2.2.2 EURORDIS Open Academy

- + Building upon ten years of experience of capacity building programmes, EURORDIS will launch the **EURORDIS Open Academy**, which will consolidate all of our face-to-face and online training experiences and also add further trainings:



- Organise the 2018 **EURORDIS Summer School in Barcelona** including the **Spanish version** in conjunction with, among others, FEDER and the Plataforma Malalties Minoritaries
- Organise the 2018 **EURORDIS Winter School on Translational Research**

- **Develop a Leadership Programme for ePAGs**
- Look into the development and funding for the organisation of a Digital Academy
- Continue to search for relevant funding opportunities for EURORDIS Youth Empowerment School (YES) training programme and look into launching a youth volunteer group to promote exchanges and collect youth perspectives
- 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.

### 2.3 RAISING AWARENESS & INFORMING

#### 2.3.1 Rare Disease Day 2018

- + Organise Rare Disease Day 2018 in over 90 countries, launch new #ShowYourRare campaign



- + Organise two events at the European Parliament on the occasion of Rare Disease Day 2018:
  - **'Rare Lives' Exhibition Opening Event** co-organised with UNIAMO -Rare Diseases Italy. A photographic exhibition held at the European Parliament's most visible exhibition space during the entire Rare Disease Day week (Monday 26th February – Friday 2nd March). The opening event of the exhibition will present an opportunity to celebrate advances in rare disease policy and will include MEPs who have joined the newly created network of Parliamentary Advocates for Rare Diseases
  - **Event on European Reference Networks** – EURORDIS will participate in the organisation of the event by the European Reference Network on rare bone diseases (ERN-BOND) at the European Parliament entitled European Reference Networks (ERNs) accelerating and improving diagnosis for rare diseases patients

### 2.3.2 ECRD 2018 Vienna



- + Organise the European Conference on Rare Diseases and Orphan Products in Vienna on 10-12 May 2018 under the overarching theme of **"Rare Diseases 360° – collaborative strategies to leave no-one behind"**

## 3. Patient Engagement

### 3.1 PATIENT ENGAGEMENT IN EUROPEAN REFERENCE NETWORKS (ERNS) AND EUROPEAN PATIENT ADVOCACY GROUPS (EPAGS)

- + Organise a structured dialogue between ERNs and POs, promote a meaningful patient engagement in ERN activities & provide regular support to ePAGs:
  - establish common rules and policies (ePAG Constitution)
  - support the recruitment of ePAG reps to ensure optimal patient representation in all ERNs
  - facilitate the creation of and support the participation in transversal ePAG working groups and an ePAG Steering Committee
  - develop a communication framework for ePAGs to support their internal communication and help them to reach-out to the wider patient community
  - foster patient-clinical partnership through the development of patient engagement guide(s) for clinical leads, research leads and ePAG representatives
  - promote common methods to capture feedback on patient satisfaction & patient experience across the 24 ERNs in 28 Member States over the years (with Rare Barometer)
  - organise regular conference calls throughout the year for each of the 24 ePAGs
  - prepare ePAG representatives participation in 24 ERN Board Meetings and organise parallel, individual ePAG representative meetings during ERN board meetings
- + Provide support to ePAGs based on a capacity-building programme tailored to their needs:
  - Organise one meeting of the ePAG representatives back to back with ECRD 2018 Vienna & support the participation of 150 ePAG reps and a second one in Q3
  - Develop implementation plan to deliver the training strategy: hold regular webinars (at least 5 through the year) open to all ePAG representative (in partnership

with the University of Montreal- Centre of Excellence on Partnership with Patients & the Public (CEPPP) and other organisations)

- Further develop peer-to-peer capacity building ePAG mentoring programme

### 3.2 PATIENT ENGAGEMENT IN LIFECYCLE DEVELOPMENT

#### 3.2.1 Patients creating their Community Advisory Boards (CABs) to engage with Industry

- + Launch the EURO-CAB programme consisting mainly in: guidelines on how to be organised and operate a CAB; all templates needed to start discussions with industry or other sponsors; a mentoring programme with training activities; ad hoc guidance to help preparing meeting; an evaluation of the impact of the CABs
- + Organise e-meetings with industry and meetings with EURORDIS members to explain the programme to all interested parties
- + Organise the 27th ERTC workshop in Barcelona in October on CABs

#### 3.2.2 Patient engagement in medicines development (project PARADIGM)

- + Take part in project PARADIGM (Patients Active in Research and Dialogues for an Improved Generation of Medicines), funded by the Innovative Medicines Initiative, as one of the 34 public and private partners engaged in the project.

#### 3.2.3 Creation of a EURORDIS HTA Task Force

- + Launch a EURORDIS HTA Task Force with a mandate to advise EURORDIS on all aspects regarding Health Technology Assessment policies and procedures. The Task Force will be composed of 12 (twelve) members for a mandate of 3 (three) years (2018-2021), confirmed on a yearly basis.

### 3.3 PATIENT ENGAGEMENT IN THERAPEUTIC DEVELOPMENT- EMA

- + Participate in the **EMA Committee for Orphan Medicinal Products (COMP)**
- + Contribute to the **EMA Pharmacovigilance and Risk Assessment Committee (PRAC)** as external experts for rare disease therapies
- + Participate in the **EMA Paediatric Committee (PDCO)**
- + Participate in the **EMA Committee for Advanced Therapies (CAT)**
- + Explore methods for patients to contribute to or witness the benefit/risk evaluation in the **EMA Committee for Human Medicinal Products (CHMP)**
- + 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
- + 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.

### 3.4 PATIENT ENGAGEMENT IN RESEARCH

- + Continue participation in 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. Take part in all meetings and activities
- + Take an active part in the development of the **European Joint Programme on Rare Diseases** to be submitted in 2018. The objectives of the EJP are:
  - **Create a research and innovation pipeline "from bench to bedside"** ensuring rapid translation of research results into clinical applications and uptake in healthcare for the benefit of patients
  - Follow the policies and **contribute to the objectives of the International Rare Diseases Research Consortium (IRDiRC)**
  - **Improve integration, efficacy, production and social impact of research on rare diseases**



# 4. Cross-Cutting priorities

## 4.1 GOVERNANCE

### 4.1.1 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

### 4.1.2 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 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 CHAFEA rules

### 4.2.2 Other private funding and foundations

- + Engage corporate and foundation donors beyond the pharmaceutical industry to support EURORDIS projects & actions.

### 4.2.3 In-kind contributions

- + Consolidate and increase in-kind contributions from companies outside the health sector to help meet objectives for EURORDIS projects and actions.

### 4.2.4 Circle of Ambassadors

- + Maintain the EURORDIS International Circle of Ambassadors

## 4.3 EURORDIS EVENTS

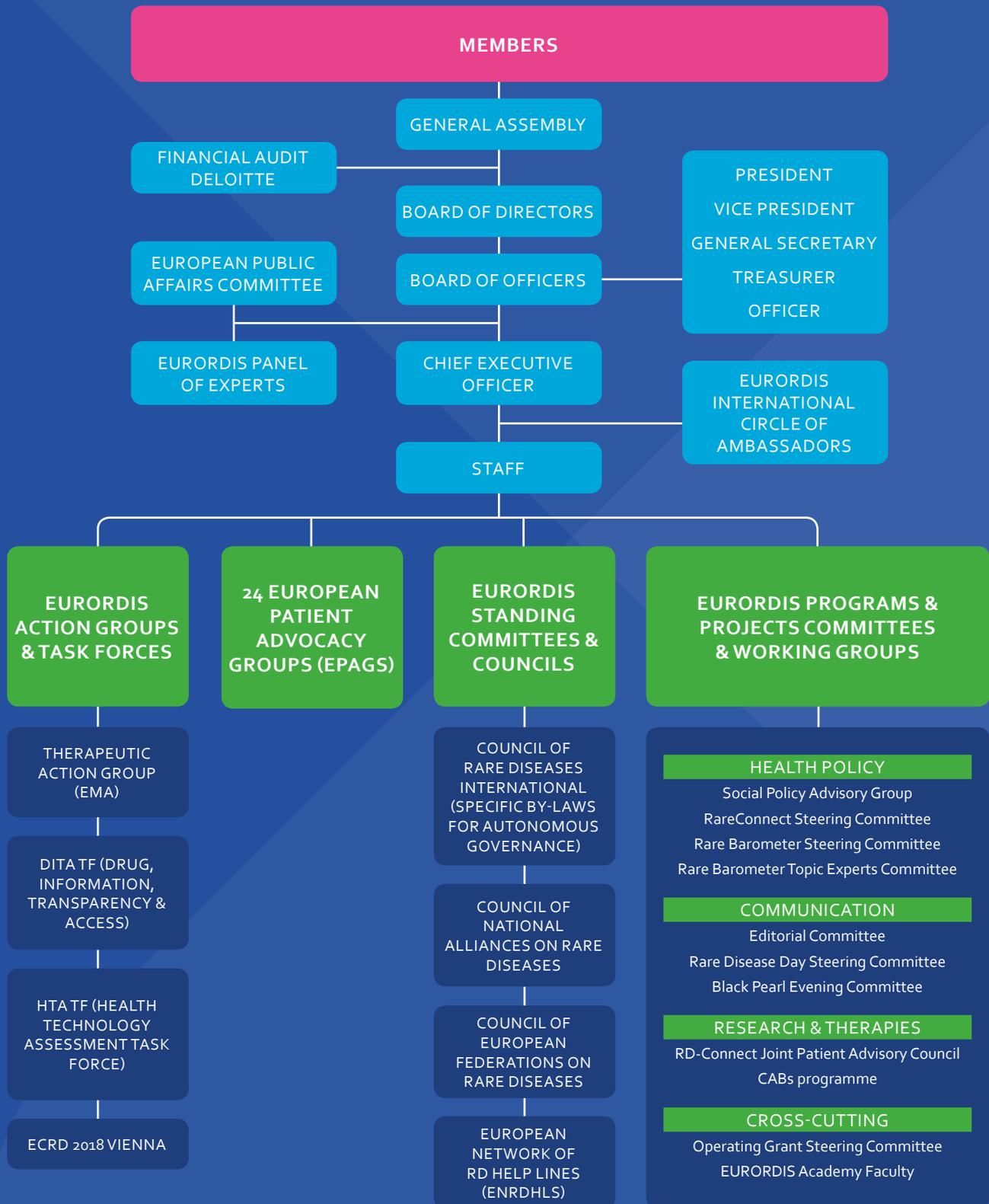
- + Organise the **EURORDIS Black Pearl Awards** in February 2018 in Brussels
- + Start planning the organisation of a fundraising event, 'Rare Elegance'
- + **9<sup>th</sup> European Conference on Rare Diseases & Orphan Products – ECRD 2018 Vienna** (10-12 May 2018) State representatives and defray some travel costs of speakers for the Conference.

## 4.4 HUMAN RESOURCES

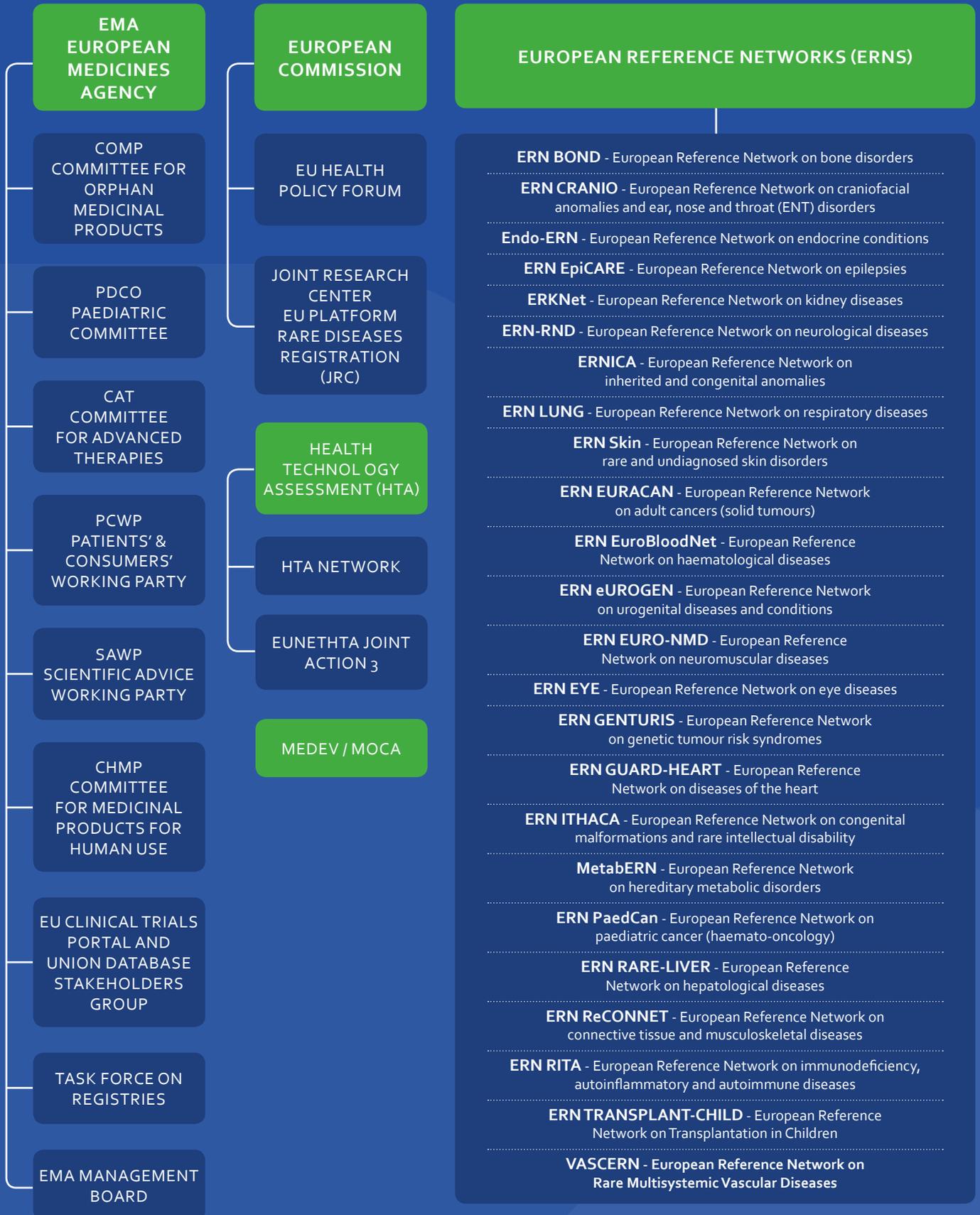
### 4.4.1 EURORDIS Staff

- + Creation of position: Resource Development Senior Manager
- + Creation of position: Communications Junior Manager
- + Creation of position: Research Policy and Project Manager
- + Creation of position: Patient Engagement Manager CABs
- + Creation of position: Patient Engagement Manager EMA, MoCA
- + Creation of position: Chief Operating Officer

# GOVERNANCE Chart 2018



# EXTERNAL REPRESENTATION Chart 2018



## EUROPEAN NOT-FOR-PROFIT ORGANISATIONS

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

Orphanet

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

Parliamentarian advocates in national assemblies

## MEMBER OF EUROPEAN NETWORKS

E-Rare

BBMRI Stakeholders Forum

RD-Connect

SCOPE Joint Action (Advisory Board)

OpenMedicine

EUPATI

PARADIGM

C4C (Connect 4 Children)

CORBEL – MIUF

Solve - RD

## INTERNATIONAL INSTITUTIONS, NOT-FOR-PROFIT ORGANISATIONS & INITIATIVES

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 Global Commission....

## 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)

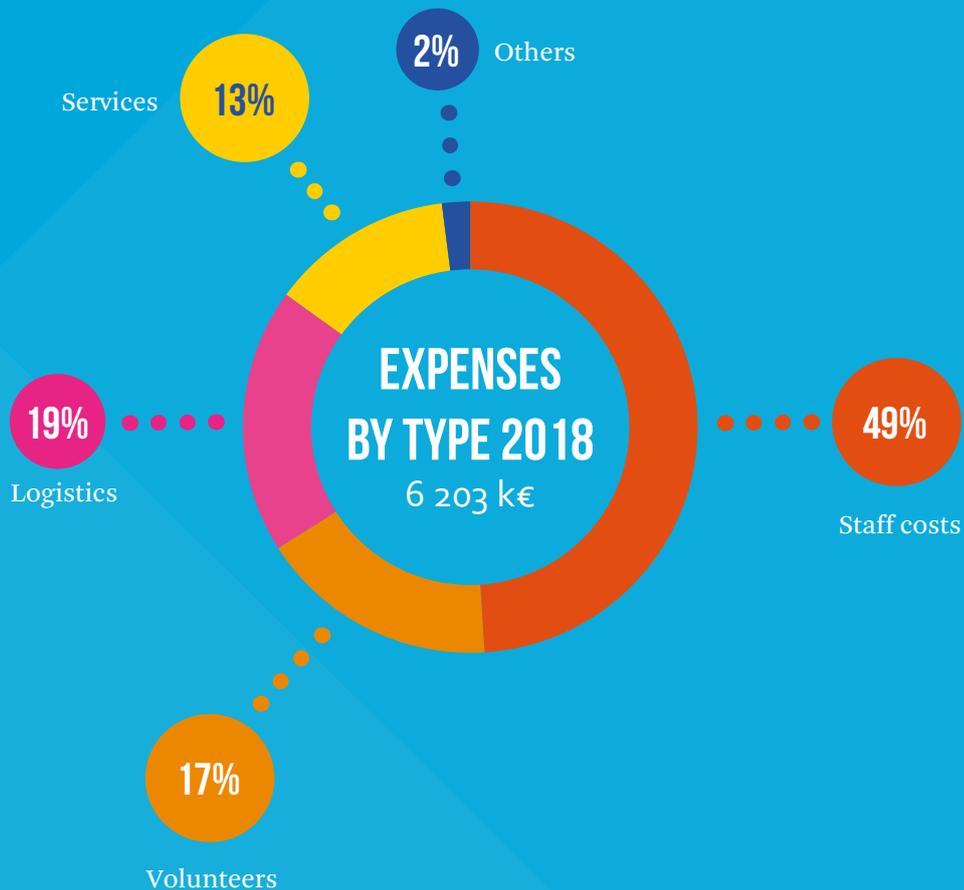
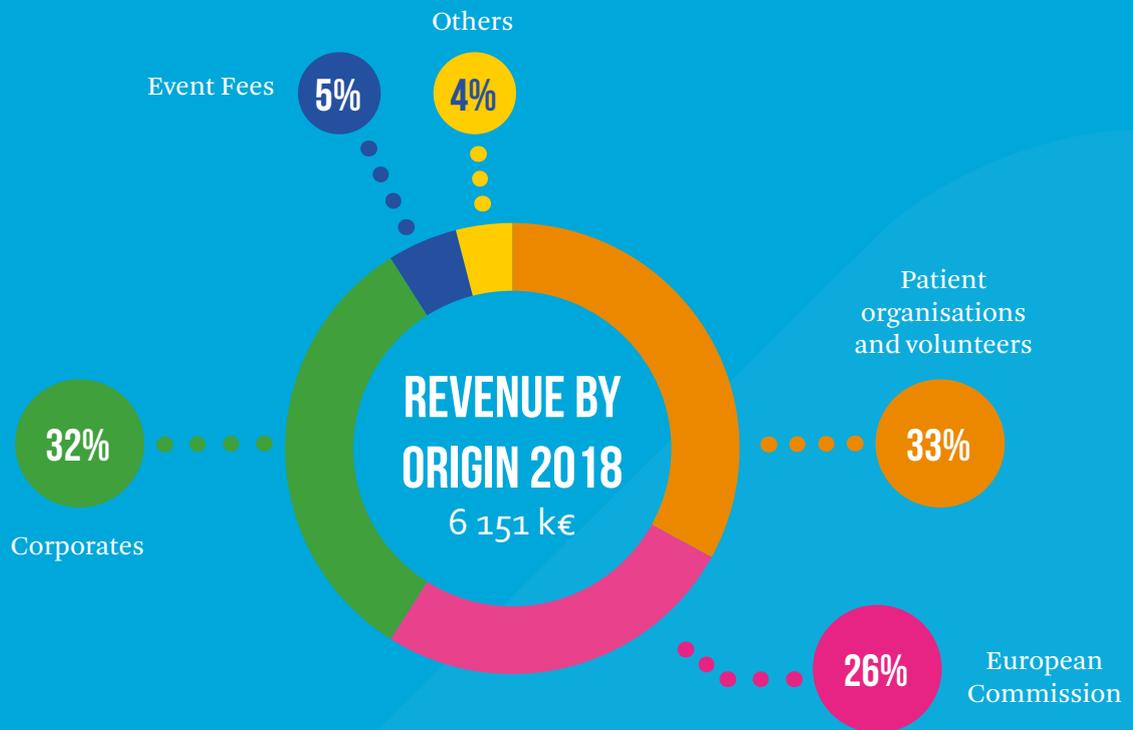
International Society for Pharmaco-economics and Outcomes Research (ISPOR)

European Hematology Association (EHA) (ongoing)

Genetic Institute for Patients, Therapies, Innovation (GIPTIS) (ongoing)



# REVENUE & expenses 2018



# 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>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>EunethTA 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

PCWP	Patients and Consumers Working Party - Richard Webst and François Houjéz represent EURORDIS
PDCO	Paediatric Drugs Committee - Tsveta Schyns represents Eurordis
PRAC	Pharmacovigilance and Risk Assessment Committee
SAWP	Scientific Advice Working Party

## EUROPEAN COMMISSION

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

## EURORDIS & EUROPEAN COMMISSION

CEGCC	Comission Expert Group on Cancer Control
CEGRD	Commission Experts Group on Rare Diseases - 8 patients' representatives included 2 representatives of EURORDIS and 2 Observers
EU HPF	EU Health Policy Forum
JRC	Joint Research Center EU Platform Rare Diseases Registration

## EURORDIS & NON GOVERNMENTAL PARTNERS

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

## MISCELLANEOUS

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



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