

EURORDIS  
**ACTIVITY REPORT** 2018  
& WORKPLAN 2019



Kacper from Poland is living  
with Kawasaki disease.

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**YANN LE CAM**

Chief Executive Officer



**TERKEL ANDERSEN**

President of the Board of Officers

# FOREWORD

by the President and the Chief Executive Officer



2018 saw a number of exciting developments within EURORDIS. As always EURORDIS' activities focused on advocating for, empowering and engaging patients.

EURORDIS' membership base continued to grow in 2018 reaching over **825 members** by the end of the year, including **49 new members in EU Member States**. EURORDIS has members in 70 countries and all 28 EU member states.

The **Council of National Alliances** reached **35 members** in 2018. 2 CNA Workshops took place in Vienna in May and in Paris in December, which was held partly in common with the Council of European Federations (CEF) (68 members reached in 2018), in order to allow for cross cutting discussions on common issues. The CNA's main activities in 2018 were the preparation and coordination of the Rare Disease Day 2019 and Rare Disease Day Strategic Review; the EURORDIS Working Group on Key policy priorities for RDs; and the creation of two new Working Groups: on Small EU countries and the Western Balkans.

The **European Conference on Rare Diseases & Orphan Products (ECRD) 2018 Vienna** was held in May 2018, and attracted **857 participants from 58 countries**. The conference remains a unique platform/forum across all rare diseases, across all European countries, bringing together all stakeholders - academics, health care professionals, industry, payers, regulators, policy makers and patient representatives.

**2018 saw the end of RD-ACTION** ([www.rd-action.eu](http://www.rd-action.eu)), the three-year Joint Action (JA) co-funded by the EU Health Programme. The JA aimed at ensuring an integrated European approach to the challenges faced by the rare disease community. Within the scope of RD-Action, EURORDIS co-organised a series of EUROPLAN conferences with national alliances (a total of 59 conferences took place between 2010-2018); supported the involvement of national alliances in the development of the State of the Art of Rare Diseases; and was particularly active in the support to the development of ERNs and discussions on transversal topics that are relevant to ERNs. In 2018, EURORDIS released a policy brief on

patient empowerment co-authored by EURORDIS and the National Centre for Rare Diseases, Istituto Superiore di Sanità in Italy. The policy brief tries to define the way in which patient empowerment is implemented and to identify the link between patient empowerment and equity, resilience and sustainability of healthcare systems, including a series of specific policy recommendations.

2018 was also an important year with regards to **voicing the social needs of people with a rare disease and advocating for the integration of rare diseases into social policy**. EURORDIS organised a '**Conference on Advancing person-centred & integrated care for rare diseases & complex conditions across Europe**', in Brussels in September during which the results of INNOVCare's pilot of case management were released in addition to a set of recommendations on integrated care for RDs. EURORDIS also strengthened its actions to raise awareness of the disabilities faced by people living with a RD and to **advocate for the recognition of the needs of the RD community within the disability agenda**. EURORDIS supported the contribution of the NGO Committee for Rare Diseases to the study of the United Nations Special Rapporteur on the Rights of People with Disabilities and also presented at the '**Work Forum on the Implementation of the UN Convention on the Rights of Persons with Disabilities**'. The **EURORDIS position paper on "Achieving Holistic Person-Centred Care to Leave no One Behind"**, aiming at advocating for measures to improve the everyday life of people living with a rare disease and their families was elaborated in 2018 was a launch in 2019.

EURORDIS continued to play a central role in the efforts for the **development of an international rare disease movement** through its involvement in **Rare Diseases International and the UN NGO Committee on Rare Diseases**.

2018 was a pivotal year for Rare Diseases International. At the 4<sup>th</sup> RDI Annual Meeting that took place on May 10<sup>th</sup> in Vienna, Austria, **RDI members unanimously adopted the statutes of the new organisation and approved the Council's decision to proceed with RDI's incorporation as a legal entity**. EURORDIS' commitment to RDI and to rare disease patients around the world remains unchanged and the two organisations signed a memorandum of

understanding that sets out how they will work together over the next five years (2018-2022).

2018 was marked by a **series of advocacy actions within the health, human rights and sustainable development agendas of the UN** that positioned the NGO Committee as an increasingly recognised actor, and catalysed support from Member States towards the cause of rare diseases. The advocacy & networking actions of the NGO Committee and RDI comprised preparatory work towards a number of actions that will take place in 2019 & 2020 principally at the **UN General Assembly and the Human Rights Council at the World Health Assembly**.

The organisation of **Rare Disease Day** remains a key EURORDIS activity which in 2018 involved **90 participating countries**. For the 7<sup>th</sup> time in a row EURORDIS proceeded with the production of a RDD video which was translated into **35 languages**. A **new interactive face-paint social media campaign, "Show your Rare"**, was also launched with great success. On the occasion of the 10 year anniversary of Rare Disease Day, a **strategic review** was commissioned to examine and prepare for the future of the initiative. The review will end in 2019.

2018 continued to be dominated by the work materialised within the context of the **European Reference Networks** and the engagement of patients within them. EURORDIS has promoted meaningful patient involvement in ERNs through the creation of the European Patient Advocacy Groups (ePAGs). ePAGs bring together patient representatives and affiliated organisations to ensure that the patient voice is heard throughout the ERN development process. There are **24 ePAGs** involving over **250 patient representatives, including non-members of EURORDIS**. In 2018, there were **2 face to face meetings of ePAG advocates** organised by EURORDIS and over **100 conference calls**. In addition to the meetings organised by EURORDIS, EURORDIS supported the **organisation of 7 individual ePAG face to face meetings** and also facilitated **ePAG representation at 12 ERN annual and Board meetings** throughout the year. Another priority area in 2018 was to **empower and train ePAG patient advocates** to help them develop soft skills and technical knowledge in areas relevant to ERNs. EURORDIS organised **one ePAG Steering Committee face-to-face training workshop, and 5 webinars** throughout the year on topics such as healthcare pathways and clinical guidelines, CPMS, influencing skills and ERNs. EURORDIS has also developed a comprehensive **Leadership training school for ePAGs** on leadership, network management, healthcare and research, which will be launched in 2019 as part of the EURORDIS Open Academy.

Building upon its experience of ten years of capacity-building programmes, EURORDIS developed and launched the **EURORDIS Open Academy consolidating all of EURORDIS' training activities**. The primary goal of the Open Academy is to empower patient advocates in the various fields where patient engagement is needed. The EURORDIS Open Academy encompasses the EURORDIS Summer School, EURORDIS Winter School, EURORDIS Digital School (to be launched in 2019) and the EURORDIS Leadership School (to be launched in 2019).

The **EURORDIS Summer School** 2018 was held for the 11<sup>th</sup> year running and had an **attendance of 52 participants representing 20 countries**.

The **EURORDIS Winter School on Scientific Innovation and Translational Research** was held for the first time in March 2018, at the Imagine Institute in Paris. **29 patient**

**representatives, from 12 countries**, participated in the training course aiming to support patient engagement in research.

EURORDIS continued to be extensively involved in the EMA's activities related to the **provision of information to patients and the public about medicines authorised** via the centralised procedure. EURORDIS spent a total number of **193 meeting days** in EMA Committee's over the year. A total of **171 EMA dossiers for public information**, were reviewed by EURORDIS staff members in order to ensure the quality of the information disseminated by the Agency to the general public.

EURORDIS continued to support the **EURORDIS Therapeutic Action Group (TAG)**, composed of the 7 patient representatives on the EMA Scientific Committees (CAT, COMP, PDCO) and PCWP (Patients and Consumers Working Party). The TAG is a forum for discussion amongst patient representatives across Committees/Working Parties of the EMA.

**Patient engagement in HTA activities.** Focus was put on informing the patient community about the European Joint Action on HTA and the European Commission Proposal for a Regulation on European Cooperation, via meetings/e-meetings with National Alliances' representatives, participation to their Boards meetings, and public conferences. On EURORDIS initiative, **14 EU umbrella patient organisations** signed a common statement on the rationale for involving patients in HTA, which was translated and made available in 6 languages.

The **EURORDIS Task Force on HTA** was launched at end 2018 and held its first face to face meeting.

The **EURORDIS EURO CAB** programme started in 2018 to support patient organisations in setting up and structuring a **Community Advisory Board (CAB)**, a group of patients who offer their expertise to sponsors of clinical research for their disease area through a transparent and effective process. A number of training webinars were organised throughout the year focusing on ethics, medical research, regulatory/EMA and HTA; and the EURO CAB programme was presented to the Council of European Federations (CEF) during the annual CEF meeting held in Paris in December.

None of the activities detailed in this report would be possible without the tireless dedication of the EURORDIS volunteers. In 2018, EURORDIS was privileged to rely on **444 volunteers**. EURORDIS volunteers have a unique insight into the complexity of different rare diseases across Europe and reinforce EURORDIS as a grassroots movement.

In 2019, we aim to **revise and improve the processes for effective volunteer management and acknowledgement**.

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. Resource diversification is 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 equity, social justice and fairness. We work through empowerment and capacity building of patient advocates, exchange and mutual support, gathering of experience, networking and partnership with all stakeholders. **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.

**826**  
member patient organisations 

**70** countries  
(28 EU countries)

**41** National Alliances of RD Patients Organisations

**68** European Federations of specific rare diseases

 Founded in  
**1997**

**6** Million € Budget

Outreach to over  
**2500**  
patient groups

 Over  
**440** Volunteers  
**100** patient advocates  
**+340** moderators

**40+**   
Staff members,  
with offices in Paris,  
Brussels, Barcelona

## 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

2018

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# HIGHLIGHTS 2018



By the end of 2018, EURORDIS had **826 member patient organisations**, located throughout 70 different countries, and including all 28 EU countries.



The **European Conference on Rare Diseases & Orphan Products (ECRD) 2018 Vienna** which was held back to back with the EMM, attracted **857 participants from 58 countries** and is the unique platform/forum across all rare diseases, across all European countries, bringing together all stakeholders - academics, health care professionals, industry, payers, regulators, policy makers and patient representatives. **ECRD 2018 Vienna met the expectations of 84% of the evaluation survey respondents**



The **Council of National Alliances had 35 members** in 2018. 2 CNA Workshops took place in Vienna in May and in Paris in December, which was again held partly in common with the **Council of European Federations (CEF) (68 members reached in 2018)**, in order to allow cross cutting discussions on common issues. The CNA's main activities in 2018 were the preparation and coordination of the Rare Disease Day 2019 and Rare Disease Day Strategic Review; the EURORDIS Working Group on Key policy priorities for RDs; and the **creation of two new CNA Working Groups: on Small EU countries and the Western Balkans.**



**Rare Disease Day 2018** involved **90 participating countries worldwide**, including all 28 EU countries. For the 7<sup>th</sup> year in a row EURORDIS produced a RDD video which was translated into **35 languages**. A new **interactive face-paint social media campaign, "Show your Rare"**, was launched with great success. A **strategic review in preparation of the future of Rare Disease Day**, was initiated and discussed at the December CAN meeting. The review will end in March 2019.



EURORDIS continued its support to the **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 2018, there were **2 face to face meetings of ePAG representatives** and over **100 conference calls**. In addition, EURORDIS supported the **organisation of 7 individual ePAG face to face meetings** and also facilitated **ePAG representation at 12 ERN annual and Board meetings** throughout the year. **Another priority area** was to empower and train ePAG patient advocates to **help them develop soft skills and technical knowledge in areas relevant to ERNs**. EURORDIS has also developed a **comprehensive Leadership training school for ePAGs** on leadership, network management, healthcare and research, which will be launched in 2019 as part of the EURORDIS Open Academy.



Building upon its experience of ten years of capacity-building programmes, EURORDIS developed and launched the **EURORDIS Open Academy consolidating all of EURORDIS' training activities**. The primary goal of the Open Academy is to empower patient advocates in the various fields where patient engagement is needed. The EURORDIS Open Academy encompasses the EURORDIS Summer School, EURORDIS Winter School, EURORDIS Digital School and the EURORDIS Leadership School. The **EURORDIS Winter School on Scientific Innovation and Translational Research** was held for the first time in March 2018, at the Imagine Institute in Paris. **29 patient representatives, from 12 countries**, participated in the training course aiming to support patient engagement in research.

2018 saw the end of the INNOVCare project. The **results of INNOVCare's pilot of case management and a set of recommendations on integrated care for RD** were released during the '**Conference on Advancing person-centred & integrated care for rare diseases & complex conditions across Europe**', organised by EURORDIS in Brussels in September. The conference was followed by high-level discussions with all stakeholders. The event brought together **80 participants from 18 European countries** and was followed online by over **280 people**.



The proposal of the **European Joint Programme for Rare Diseases** within the Work Programme 2018-2020 received a positive evaluation in August 2018 by the European Commission and will officially start in January 2019. The main goal of the EJP on RD is to **develop a sustainable ecosystem allowing a virtuous circle between RD care, research and medical innovation**. The EJP on RD will be a major driving force for collaborative RD research in Europe.



EURORDIS has a prominent role in **project PARADIGM**, a 30-month public-private partnership launched on 1 March 2018 and led by the EPF and EFPIA. Its mission is to **advance a structured, meaningful and ethical patient engagement in medicines development** and aims to develop tools and resources to allow the effective and systematic inclusion of patients and to design an innovative roadmap to ensure long-term sustainability of patient engagement.



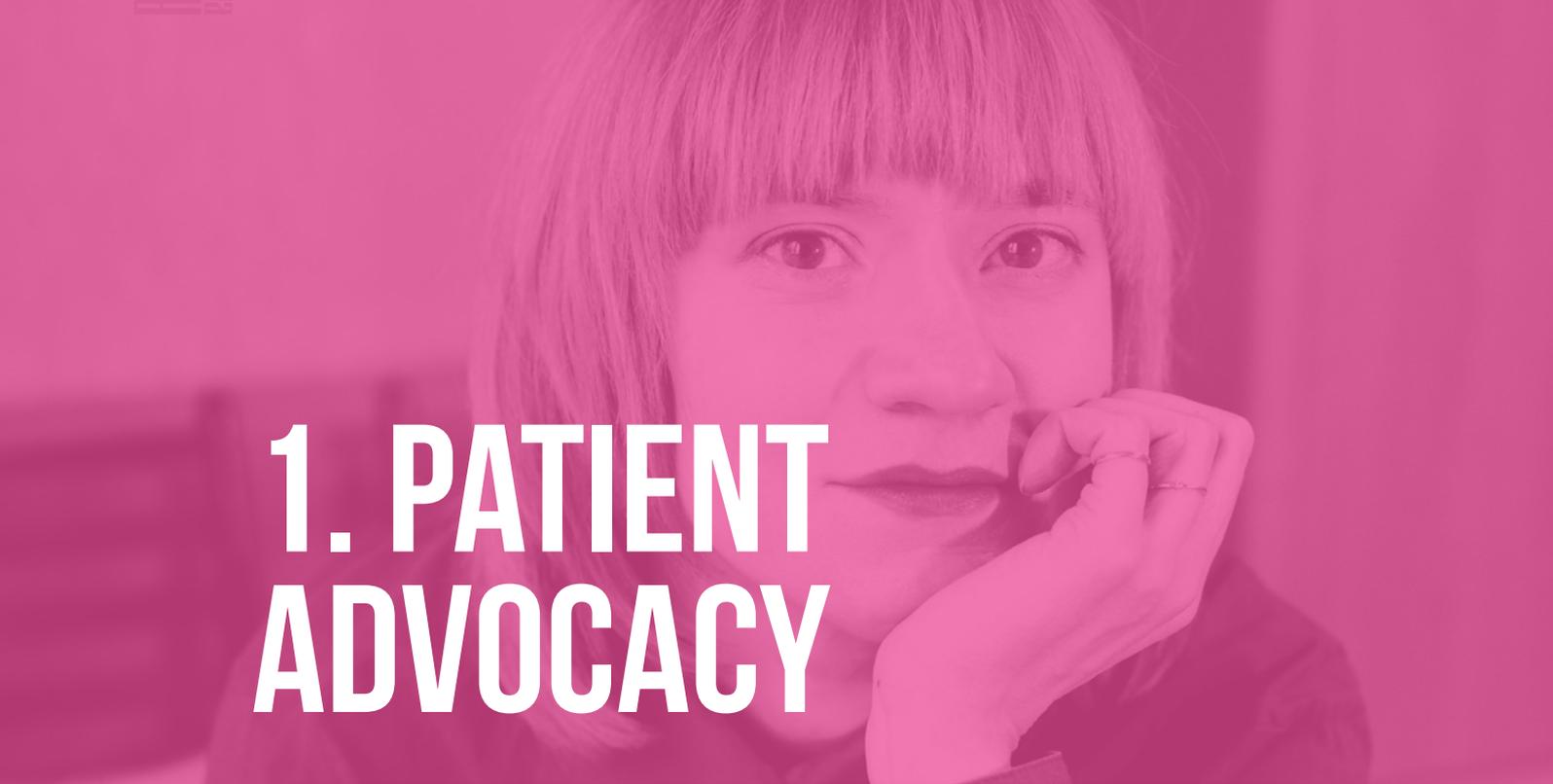
The **EURORDIS EUROCAB project** started in 2018 to **support patient organisations in setting up and structuring a Community Advisory Board (CAB)**, a group of patients who offer their expertise to sponsors of clinical research for their disease area through a transparent and effective process. A number of training webinars were organised throughout the year focusing on ethics, medical research, regulatory/EMA and HTA; and the EUROCAB programme was presented to the Council of European Federations (CEF) during the annual CEF meeting held in Paris in December.



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). EURORDIS spent a **total number of 193 meeting days in EMA Committee's over the year**. A total of **171 EMA dossiers for public information**, were reviewed by EURORDIS staff members in order to ensure the quality of the information disseminated by the Agency to the general public.



2018 was a pivotal year for **Rare Diseases International (RDI)**. At the 4<sup>th</sup> RDI Annual Meeting that took place on May 10<sup>th</sup> in Vienna, Austria, **RDI members unanimously adopted the statutes of the new organisation and approved the Council's decision to proceed with RDI's incorporation** as a legal entity. EURORDIS' commitment to RDI and to rare disease patients around the world remains unchanged and the two organisations signed a memorandum of understanding that sets out how they will work together over the next five years (2018-2022).



# 1. PATIENT ADVOCACY

## 1.1 Our advocacy goals within our strategy priorities for 2015-2020

In the course of 2018, 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 2018 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.

## 1.3 Advocate rare diseases as a priority in the next decade 2020-2030

With the Expert Groups having ceased to exist and the end of the Joint Action for Rare Diseases, RD-ACTION, in 2018 EURORDIS undertook different initiatives to prepare for the next decade of rare disease policy making. Taking stock of over twenty years of advances in support of rare diseases, unmet needs are still to be addressed while new challenges emerge. New impulsion at European level is necessary to ensure that they are addressed with adequate actions.

Therefore in 2018, EURORDIS continued to pursue opportunities to prepare for a new policy framework that could address those needs, along the lines of the 2009 Council Recommendation on an Action in the Field of Rare Diseases and the 2008 Commission Communication on Rare Diseases.

Building on the outcomes of the **Maltese Presidency of the EU Council (2017)** that gave prominence to rare diseases (including the **EU Council Conclusions on voluntary cooperation between health systems** and the launch of a EURORDIS' **Declaration on rare diseases**), EURORDIS initiated a broad consultation with its National Alliances which led to the creation of a **Working Group on Future Policy Priorities for Rare Diseases**, composed of members of National Alliances.

The Group worked 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. The outcomes of this process of identification of policy priorities were presented at the plenary session of the European Conference on Rare Diseases (May 2018, Vienna).

In parallel, EURORDIS pursued opportunities to continue the multi-stakeholder dialogue and the policy work that the dismantled Expert Groups for Rare Diseases secured for many years. Following advice from the European Commission, EURORDIS and other key partners of the former Joint Action for Rare Diseases enquired for opportunities under the **Health Policy Platform**, a virtual space coordinated by the European Commission (DG SANTE) to promote dialogue amongst stakeholder in the field of health policy. This included joint letters to high level management of DG SANTE, participation to meetings of the Health Policy Platform and regular exchanges with key Joint Action partners to set up a **"Stakeholder Network" for Rare Diseases** hosted by the Platform.

Finally, EURORDIS continued to follow the discussions on the next budgetary period that led 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 fed into the stakeholder consultation that European Commission organised in early 2018.

### Parliamentary Advocates for Rare Diseases

After the creation of the network of Parliamentary Advocates for Rare Diseases in October 2017, EURORDIS

consolidated—with the essential contribution of EURORDIS' National Alliances—this core group of Members of the European Parliament (MEPs) who have been long standing advocates of the rare disease cause and committed to implement concrete actions in support of people living with rare diseases within the political agenda.

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 January 2018, a brainstorming meeting with parliamentarians was organised with the support of Frédérique Ries MEP, aimed to identify priority actions and initiatives that the network could carry out before the end of the legislative term (mid-2019).

Further to that meeting, a group of 15 MEPs led by Parliamentary Advocates for Rare Diseases tabled an amendment promoted by EURORDIS to the EP Resolution on the **2021-2027 Multiannual Financial Framework**, asking to secure financial support to European Reference Networks within the forthcoming EU financial framework.



Also, events were organised at the European Parliament to mark the occasion of Rare Disease Day 2018, both under the auspices of the Parliamentary Advocates for Rare Diseases, hosted by Elena Gentile MEP:

- + the RareLives Photo Exhibition, a UNIAMO initiative with the support of EURORDIS-Rare Diseases Europe. Its opening was attended by 13 MEPs;
- + the event "European Reference Networks (ERNs) accelerating and improving diagnosis for rare diseases patients", a ERN-BOND initiative with the support of EURORDIS.

### Foresight Study on Rare Diseases in 2030

In 2018, EURORDIS responded as project coordinator to the call for proposals "Pilot project - Rare 2030 - a participatory foresight study for policy-making rare diseases" bringing together a consortium of eight key thought leaders in the field of rare diseases as partners: EURORDIS, Orphanet, the University of Newcastle, ISINNOVA, the Imperial College of London, the European Reference Network for Rare Metabolic Diseases and the European Reference Network for Rare Bone Diseases.

EURORDIS and project partners proposed a workplan harnessing the expertise and experience of associated and collaborating partners and a panel of experts made up of representatives of all those benefiting from the project's outcomes. This Panel of Experts including up to 250

stakeholders across the rare disease community will serve as the projects consultative body to achieve the following proposed project objectives:

- 1 Establish the **baseline knowledge** required to identify, agree on and rank scientific, technological, social, political, financial and institutional **trends and drivers** for the future governance and care of RDs in Europe.
- 2 Explore and combine trends and existing instruments to develop **consistent and plausible qualitative scenarios** revealing the needs, perceptions and preferences of the RD Community and society at large as well as the **policy options** leading to these scenarios.
- 3 Draft clear recommendations for a **road map of European RD policy 2020-2030**,
- 4 Identify solutions to sustain them beyond 2030.
- 5 Evaluate the efficacy and appropriateness of foresight and other future studies methods for RD policy planning and create long-lasting communication tools to engage the RD Community, including policy makers and society at large, to follow through with implementing these recommended policies.

The project was accepted in July 16 for a start in January 2019.

### 1.3.1 2<sup>nd</sup> European Union Joint Action on Rare Diseases – RD-ACTION (2015-2018): Promoting the Implementation of Recommendations on Policy, Information and harmonised coding system for Rare Diseases



RD-ACTION ([www.rd-action.eu](http://www.rd-action.eu)) was a three-year Joint Action (JA) co-funded by the EU Health Programme, starting in 2015 and ending in June 2018. The JA was coordinated by Orphanet at the INSERM in France. The JA aimed at ensuring an integrated European approach to the challenges faced by the Rare Disease (RD) community and at promoting, catalysing and triggering multi-stakeholder debates which are necessary both at European level, to build shared strategies, and at national level, to support the integration of EU policies on rare diseases across Member States.

EURORDIS led the WP2 on Dissemination and was particularly involved in the WP 6 Rare Disease Policy, which included the support to the development of ERNs and discussions on transversal topics that are relevant to ERNs.

The key achievements of RD-Action over the three years:

- 1 **Providing information and data on Rare Diseases (Orphanet):** the Orphanet nomenclature, (or ORPHAcodes) has been improved by making it more easily exploitable in information systems
- 2 **Improving identification of RDs in health information systems: RD-ACTION developed a tool set to assist European countries in implementing the ORPHA codes,** including standard procedures, and **practical guidance for integration, use and routine maintenance of the ORPHA nomenclature** in health information systems
- 3 **ERNs support:** Meaningful patient involvement in ERNs was promoted with the establishment of the European Patient Advocacy Groups (ePAGs) by EURORDIS. Throughout the duration of RD-ACTION, six major workshops were organised to address a particular policy area or topic in which the ERNs could add value and capitalise on past investments and lessons learned. Many of these workshops were co-organised by DG Santé and involved over 360 multi-stakeholder experts. All these six workshops led to reports, concept papers and Recommendations that are accessible online.
- 4 **State of the Art of rare disease policies in Europe and beyond:** The State of the Art of Rare Diseases provides a broad outline of the history and status quo of rare disease activities across Europe, with an emphasis on topics such as the RD legislative and policy framework, European Commission investments, achievements of the most relevant funded initiatives and research activity. It also provides an overview of the national situation in each EU Member State as well as some other countries. EURORDIS supported the involvement of NAs in the development of the State of the Art on Rare Diseases.
- 5 **Support to sustainable and resilient health systems:** In line with the Communication of the European Commission and Recommendation from the EU Council on RDs, work was carried out within RD-ACTION to support national authorities to quantify the burden of RDs and identify available resources for sustainable and resilient health systems, taking into account the principles of equity, quality and efficiency, and involving stakeholders, policy makers and civil servants in charge of national RD plans or strategies. Within this work, six Policy Briefs have been released including a policy brief on patient empowerment released in 2018 and co-authored by EURORDIS and the National Centre for Rare Diseases, Istituto Superiore di Sanità in Italy. The policy brief tries to define the way in which patient empowerment is implemented and to identify the link between patient empowerment and equity, resilience and sustainability of healthcare systems, including a series of specific policy recommendations.
- 6 **Fostering national RD plans/strategies:** The EUROPLAN National Conferences are the tool designed to promote the adoption and implementation of national plans or strategies for rare diseases in European countries. EUROPLAN National Conferences are jointly organised in each country by a National Alliance (NA) of rare disease patient organisations and EURORDIS. Between 2010 and 2018, altogether 59 EUROPLAN National Conferences took place in 25 EU Member States, as well as in Georgia, Macedonia, Russia, Serbia and Ukraine. Some National Alliances organised several EUROPLAN conferences in their country. As a result, by the end of 2018, **25 EU Member States** had put in place a national plan or strategy for rare diseases. In 2018, EURORDIS co-organised the following EUROPLAN National conferences: ITALY 9-10 February; SLOVENIA 28 February; CYPRUS 9-10 March; LITHUANIA 16 March; SLOVAKIA 25 May; BELGIUM 8 June
- 7 **ECRD 2016 Edinburgh:** Within the framework of RD-ACTION, the 8<sup>th</sup> edition of ECRD took place in Edinburgh, on 26-28 May 2016. The conference focused on "game changers in rare diseases". It attracted over 800 participants from 48 countries. The conclusions of ECRD 2016 helped inform EU policies impacting 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 partnering patients' organisations are involved across all work packages.

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).

In addition, the JARC, through the leadership of the Catalan Institute of Oncology (ICO), undertook in 2018 "a comparative analysis of the priorities and recommendations on rare and paediatric cancers based on National Cancer Control Programmes (NCCPs), Rare Diseases Plans, EU funded initiatives and priorities for patients' organisations". EURORDIS actively participated in this study together with SIOPe (the European Society for Paediatric Oncology).

## European Reference Networks for Rare Cancers

### What is an ERN?

European Reference Networks (ERNs) are networks of national centres of expertise involving nearly 1,000 healthcare providers throughout the European Union (EU) specialising in the treatment of rare and complex diseases. There are 24 ERNs and each corresponds to a broad disease grouping.

EURACAN and EuroBloodNet cover respectively solid and haematological cancers in adults, PaedCan covers paediatric cancers and GENTURIS encompasses genetic tumour risk syndromes.

### How will ERNs help patients?

ERNs facilitate virtual medical consultations on difficult cases for diagnosis and treatments. Expertise travels rather than the patient. Specialised healthcare professionals discuss patients' cases with their counterparts, and securely share images and/or biological samples within and/or outside their country. **Ultimately, this will result in providing timely, adequate and equal access to diagnosis and care for all rare disease/rare cancer patients in the EU.**

The EU Joint Action for Rare Cancers (JARC) supports the development of ERNs for rare cancers and provides recommendations on rare cancers' policy: [www.jointactionrarecancers.eu](http://www.jointactionrarecancers.eu)

### ERNs are patient-centred:

Patient representatives/advocates associated with ERNs are called European Patient Advocacy Groups ("ePAGs"). They are involved at the highest level on the ERN Boards/Steering Committees and work closely with the ERN medical experts. EURORDIS – Rare Diseases Europe – provides them with coordinated support.



## Participation in events related to Rare Cancers and publication of a leaflet on the 4 ERNs for Rare Cancers

EURORDIS participated in the following events:

- + ESMO (European Society of Medical Oncology) Congress, 19-23 October, Munich, Germany
- + Training Course of ESMO – European School of Oncology (ESO) - Rare Cancers Europe (RCE), 1-3 December, Milan, Italy: Presenting to the rare cancers patient community the experience of the rare disease patient community in promoting the adoption of national plans for rare diseases in EU Member States.

EURORDIS led the creation of a joint leaflet for the four ERNs relevant to rare cancers, namely EURACAN, PaedCan, EuroBloodNet and GENTURIS, with the support of EURACAN, the participation of all ePAGs across these four ERNs and validation from the ERN Coordinators and project managers.

### 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 2018, EURORDIS continued to advocate to improve the regulatory process for medicines for the patients it represents.

Following the launch of the process for a **joint evaluation of the legislation on medicines for children and rare diseases in 2017**, the European Commission commissioned an additional study to understand the strength and weaknesses of the Regulation on Orphan Medicinal Products. The purpose of the evaluation is two-fold:

- 1 To 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 regulations
- 2 To focus on the cost-effectiveness when providing the incentives and rewards incorporated in the legislation and how they have been used in practice.

The evaluation aims to 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 be completed most likely by Autumn 2019, and it will build upon other ongoing or completed studies, such as the 10-year progress report on the Paediatric Regulation, and the study on pharmaceutical incentives.

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, published in 2018.

EURORDIS participated in the targeted consultation organised to provide input to the evaluation, and also coordinated input from its membership, including European Federations, National Alliances and individual associations. Furthermore, EURORDIS participated in the



open public consultation prepared also by the European Commission to gather input also from the general public.

In this context, EURORDIS organised the 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. The 26<sup>th</sup> workshop, entitled "Rare diseases therapies: Do we get what we incentivise?", which aimed at taking a balanced look at the role of incentives in therapies development, address shortcomings of the current system and consider what the rare disease community can do to fulfil important unmet medical needs;

The workshop collected feedback, and explored interests and positions on the forthcoming joint evaluation of the legislative framework for orphan and paediatric medicines, as well as the current evaluation of the incentives system. The workshop represented the opportunity to discuss how to support therapies development for rare diseases and ensure access of the patients to these treatments. The workshop, attended by around 200 people, allowed us to collectively reflect on better and innovative approaches, building on what is already working.

### 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.

#### **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 continue 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 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.

### Breaking the Access Deadlock to Leave No-one Behind: continuing the efforts

Following the adoption by the Board of Directors of EURORDIS in November 2017, the position paper on "Breaking the Access Deadlock to Leave No One Behind" took centre stage in 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. EURORDIS continues to have 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 new model based on a collective conversation involving all stakeholders (patients, the pharmaceutical industry, national competent authorities, national health ministries, researchers, scientists and regulators) was heavily featured in the discussion at the 26<sup>th</sup> ERTC workshop (see above), with discussions taking place around the key role of collaboration between stakeholders and decision makers to provide accessible and affordable medicines to as many patient as possible.

The four pillar approach presented in the paper constituted the cornerstone.

### MoCA (Mechanisms of Coordinated Access)

Since end 2015, EURORDIS is putting in place a framework for patient engagement in the MoCA procedures. Elisa Ferrer, EURORDIS Patient Engagement Senior Manager and, from September 2018 Maria Cavaller, EURORDIS Patient Engagement Junior Manager are taking care of identifying and supporting these patient experts.

Regarding MoCA, for each new pilot, EURORDIS' staff is spending an average of 9 hours, which include the time to review the company's proposal, the time to identify the patients' representatives, one or more calls to brief them, the administrative support to them and the actual time in the meeting. In addition, one-hour meeting + 5 hours of travel can be also added, as the staff is most of the time physically attending the meetings.

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 has been created to keep members and stakeholders up-to-date about the MoCA

developments and rules of procedures: <https://www.eurordis.org/content/moca>

### **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 the same context, we have contributed to the development of a new multi-stakeholder initiative to focus on 'Real World Evidence to address uncertainties in complex or rare conditions that require highly specialised treatment' in order to improve access to rare disease therapies' – TRUST<sub>4</sub>RD. Through a series of stakeholder meetings during in 2018, which included patient representatives, payers, national health ministries and HTA bodies, research organisations and industry, led by Professor Lieven Annemans (University of Leuven), steps to the development of a paper to increase the trust of all stakeholders in evidence generation pathways. A discussion on the role of managing uncertainties in post authorisation phase with representatives of ERNs, national health decision makers and industry took place at the European Health Forum Gastein in October. A publication (submitted to the Orphanet Journal) and further dissemination meetings will take place throughout 2019.



At the ECRD conference 2018, EURORDIS kicked off RARE-IMPACT, a new patient-led, collaborative, pre-competitive dialogue on patient access to gene and cell therapies for rare diseases in Europe by addressing practical, clinical, economic and political challenges, in full recognition of the transformative and potentially curative nature of the new therapies, with the support of specialist consultancy Dolon, which is acting as a secretariat of the initiative, EURORDIS built a consortium of sixteen manufacturers (both for and not-for profit) and industry associations that is looking at:

- + Identifying challenges that are preventing rare disease patients accessing gene and cell therapies at European and country level and proposing actionable solutions to address these challenges;

- + Preparing external stakeholders and companies for the access challenges that are likely to be faced with gene and cell therapies;
- + Educating external stakeholders on gene and cell therapy technology and terminology;
- + Provide a pre-competitive forum in which manufacturers can share experiences and ideas.

The RARE IMPACT initiative will continue throughout 2019, when we expect to have greater understanding of challenges and solutions for better and faster access to such promising therapies.

## To prepare the reimbursement decision: the HTA momentum

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

The third EU Joint Action on HTA (EUnetHTA JA<sub>3</sub>) decided not to have a structured interaction with stakeholders as it was in the previous Joint Action (EUnetHTA JA<sub>2</sub>) with the EUnetHTA Stakeholder Forum, but it committed to involve patients as experts in its scientific assessments and other activities.

In 2018, EURORDIS received 2 requests for the identification of patients or questionnaire dissemination for Joint assessments, of which one for a rare disease.

The role of European umbrella organisations consists in advising EUnetHTA on the principles and rules for patient involvement and mentoring of patients invited to participate in their procedures.

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

EURORDIS continued informing the patients' community about the European Joint Action on HTA and the European Commission Proposal for a Regulation on European Cooperation in 2018, via meetings/e-meetings with RD National Alliances representatives, participation to their Boards meetings, and public conferences.

18 meetings/e-meetings with National Alliances representatives and their boards were held throughout the year and dedicated sessions on HTA were carried out in both CNA meetings (in March and December 2018) and the European Conference on Rare Diseases (ECRD 2018 Vienna) held in May. In addition, a series of meetings were held with EU policy makers and national contacts in conjunction with the national alliances.

On EURORDIS initiative, 14 EU umbrella patient organisations signed a common statement on the rationale for involving patients in HTA, which was translated and made available in 6 languages.

## 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-label use of medicines in rare diseases, launched in 2012, in 2016, the DITA Task Force launched an even larger survey on treatment information on rare diseases ('Tell us how you take your treatment'), with a questionnaire translated in 13 languages. 1,965 responses were collected.

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

<sup>1</sup> [https://ec.europa.eu/health/sites/health/files/documents/2017\\_02\\_28\\_final\\_study\\_report\\_on\\_off-label\\_use\\_.pdf](https://ec.europa.eu/health/sites/health/files/documents/2017_02_28_final_study_report_on_off-label_use_.pdf)

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### 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.

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### 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 2018, EURORDIS has continued to support and promote 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, including the Innovative Medicines Initiatives.
- + 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, allowing a virtuous circle between research, care and medical innovation in rare diseases.
- + Continued supporting rare diseases as a public health priority in the 3<sup>rd</sup> EU Public Health Programme 'Health for Growth', following the mid-term review of the Programme.
- + In parallel, EURORDIS followed the discussions on the next budgetary period that led to the adoption in 2018 of the proposed new **EU Multiannual Financial Framework 2020-2027 and the legislative proposals for new funding programmes**.
- + After the adoption of the European Commission proposals, EURORDIS followed the legislative work of the other EU Institutions, notably the European Parliament and the Council of the EU. Namely, EURORDIS contributed to the stakeholder consultation that European Commission organised in early 2018 and proposed an amendment, co-signed by 15 MEPs, aimed to secure financial support to European Reference Networks under the new financial framework.
- + Under the new proposed EU financial framework, "Horizon Europe", the future research and innovation programme of the EU, features rare diseases as a priority area in health research. EURORDIS worked to ensure that this priority area be maintained.
- + Importantly, under the forthcoming Multiannual Financial Framework, as from 2021 the Health Programme will be embedded into the 'European Social Fund Plus' (ESF+). EURORDIS held meetings

with responsible services of the European Commission to have better understanding of the impact this would have on health policies and funding.

- + In this renewed context, and in order to contain the potential scaling down of EU action in the field of health, EURORDIS continued to be an active member of the broad campaign of EU health stakeholders calling for a continued and enhanced EU action in the field of health (**#EU4Health**). The campaigners demand not to dismantle a dedicated EU funding programme for health in the context of the new financial framework and specifically of ESF+.
- + EURORDIS held informative meetings with European Commission' services and prepared an information session at the Council of National Alliances on the new funding schemes and opportunities for EURORDIS members and Alliances.

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### 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:

- + 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 on what needs doing more urgently from the patient perspective.
- + 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.
- + Moreover, in 2018 EURORDIS was identified as key stakeholder for contributing to the scoping exercise of the European Court of Auditors on the implementation of the Cross-border Healthcare Directive. EURORDIS staff were interviewed by the Court's auditors and provided them with relevant documentation.

### 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



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.

In 2016, EURORDIS in collaboration with the European rare disease community established 24 European Patient Advocacy Groups (ePAGs) as forums to optimise the involvement of patient representatives of the rare disease community in the 24 ERNs. Each ePAG corresponds to the scope of one of the 24 ERNs, aligning patient organisations and clinicians, experts and researchers working on the same rare or complex disease or highly specialised intervention. Today, the ePAGs consist of 255 ePAG patient advocates across the 24 ERNs. Likewise, 1760 patient organisations have registered to be part and engage in the ERN that covers the diseases that the patient organisation is representing.

A priority activity in 2018 was to establish a common governance framework for all ePAGs that can then be adapted to the specificities of each group. This governance framework describes, among other things, the role of ePAG patient advocates, outlines the selection process, eligibility criteria and sets the values and ethical principles that ePAG patient advocates must observe.

In 2018, EURORDIS organised (i) an ePAG face-to-face meeting, (ii) an ePAG f2f Steering Committee meeting (iii) an ePAG Steering Committee training session. In addition to the meetings organised by EURORDIS, EURORDIS has **supported the organization of 7 individual ePAG f2f meetings** that were attended by 49 ePAG patient advocates, with a special focus on those groups that were still forming and where a face-to-face meeting would help to identify strategic priorities and form their identity as a group. Finally, we have also facilitated ePAG representation at 12 **ERN annual and Board meetings** throughout 2018.

EURORDIS held more than 100 conference calls with ePAG patient advocates in 2018. The majority of these calls (**99 conference calls**) were organised with individual ePAG groups. We also held 7 ePAG Steering Committee calls to discuss topics of relevance to the 24 groups (cross-ERNs). In addition, EURORDIS has launched three transversal topic groups as learning forums to exchange and share information on the following areas: research and registries topics (**2 calls**), training and education (**3 calls**) and outcomes and guidelines (**2 calls**).

Another priority area this year has been to **empower and train ePAG patient advocates** to help them develop soft skills and technical knowledge in areas relevant to ERNs. EURORDIS has developed a comprehensive Leadership

training programme for ePAGs on leadership, network management, healthcare and research. 2019 will be a pilot year, where we will be testing the programme structure, delivery methods, contents and trainers.

In 2018, EURORDIS continued to support Rare Disease National Alliances on European Reference Networks. We organized workshops in 10 different countries to help raise awareness and knowledge on ERNs and engage with national stakeholders (patient organisations, clinicians and health authorities) to help them understand how they can participate and benefit from this new structure. In addition we have a group of National Alliances from smaller Member States to which we have provided tailored advice in 2 dedicated conference calls.

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 deployment of ERNs.

EURORDIS co-organised the RD-ACTION Workshop DG Sante on How ERNs can provide added-value in the area of clinical research, which was held 29-30 May 2018, EMA, London, UK

EURORDIS was also present at the 4<sup>th</sup> ERN Conference was held in Brussels on 21-22 November and opened a new stage in the networks' lifecycle, namely the deployment phase. After an intensive period of preparatory actions and awareness raising, the first 24 ERNs are operational. While outcomes and success stories have already been delivered since their launch in March 2017, numerous aspects still needed to be considered to consolidate the work and results achieved so far. This conference intended to present the main clinical and organisational outcomes of the networks, as well as to identify the challenges and margins for future improvements

On the occasion of the conference 2018, EURORDIS Recommendations on the Integration of ERNs into national health systems, a result of a consultative process that included feedback from Rare Disease National Alliances and ePAG patient advocates.

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### 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.*

#### European Joint Programme Cofund on Rare Diseases (EJP on RD)



The proposal of the EJP for RD within the Work Programme 2018-2020 has been successfully submitted in April, received a positive evaluation in August 2018 by the European Commission and will officially start in January 2019. The main goal of the EJP on RD is to develop a sustainable ecosystem allowing a virtuous circle between

RD care, research and medical innovation. The EJP on RD will be a major driving force for collaborative RD research in Europe. More specifically, the EJP on RD is a 5-year 55M € programme coordinated by Inserm in France and has secured the official participation of 88 partners across Europe including academic scientists, clinicians, European Research Infrastructures, ERNs and patient organisations. The proposed joint programme of activities ranging from research to coordination and networking activities, including training, demonstration and dissemination activities, will be structured along five main components ensuring the implementation of a comprehensive and cohesive research and innovation bench to bedside pipeline, fully in line with the Work Programme call SC1-BHC-04-2018 "Rare Disease European Joint Programme Cofund":

Pillar 0: Strategic coordination and management of the EJP RD will constitute the foundation linking all the Pillars together, enabling cross-communication, prioritisation and alignment between different activities;

Pillar 1: "funding collaborative research on RD" will foster joint transnational calls for collaborative research projects (continuation of E-RARE) resulting in financial support to third parties encompassing various aspects of rare diseases;

Pillar 2: "innovative coordinated access to data and services for transformative RD research" aiming at rationalized, optimized and increased potential of existing resources and services;

Pillar 3: "capacity building and empowerment" will be raising the level of knowledge and know-how within the RD research and care community, including through ERNs and RD patient representatives and advocates;

Pillar 4: "accelerating the translation of high potential projects and improving outcomes of clinical studies in small populations".

EURORDIS is co-leader of Pillar 3 and therefore a key member of the Operating Group. EURORDIS is also actively involved in all transversal activities (Pillar 0) and in funding collaborative research on RD (Pillar 1) mostly regarding support and development of involvement of patient organisations in research projects.

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

Throughout 2018, 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 project. 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.

To make substantial progress in diagnosis of unsolved rare diseases and to cope with the main challenges of diagnostic discovery and diagnosis-adapted patient management, Solve-RD brings together i) the most advanced and most useful diagnostic RD research infrastructure, ii) a critical mass of RD diagnostic discovery expertise stemming mainly from involved ERNs and iii) unique research cohorts.

The 5-year project started in January 2018 with the kick-off meeting held in Tübingen. Solve-RD fully integrates with the newly formed European Reference Networks (ERNs) for rare diseases which began to operate in 2017. Four ERNs (ERN-RND, -EURO-NMD, -ITHACA, and -GENTURIS) build the core of Solve-RD but the project is reaching out to patient cohorts across all 24 ERNs as well as the undiagnosed disease programmes from Spain and Italy in order to achieve its aims. EURORDIS' main activities within Solve-RD includes the development of the first edition of the capacity building programme for patient representatives on scientific innovation and translation research which took place in March 2018 at the Imagine Institute for Genetic Diseases in Paris as well as the development of a Community Engagement Task Force which was launched in November 2018 (see below for more information).

### Undiagnosed Community

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. The UDNI brings clinicians, researchers, genetic counselors, and other medical professionals from around the world together to collaborate on diagnosing the most difficult and intractable cases. Patient and patient representative participation are instrumental in ensuring the long-term success of this initiative as they can offer their expertise to the UDNI institutional, clinician, non-clinician members on how to ensure the effort is patient-focused, patient-friendly, and patient-driven. NORD, EURORDIS and the Wilhelm Foundation have collaboratively developed a patient engagement membership which was adopted by for the board of the UDNI. From December 2018, patient organisations around the world can officially apply to join the UDNI as members as long as they can demonstrate that their organization is a certified not-for-profit organization, have a Board of Directors composed of a majority rare and undiagnosed patient advocates as well as a mission statement that includes advancing access to diagnoses and show proven activities of advocating for diagnoses for the undiagnosed community.

EURORDIS is leading the development of the Community Engagement Task Force (within Solve-RD) that aims to create a united and engaged multi-stakeholder community of patients, scientists and clinicians committed to improving diagnosis and care of ultra-rare diseases and supporting the needs of the undiagnosed community.

The main objectives of the CETF are to:

- + Ensure that the patient voice is heard and represented in all stages of the project, by i) acting as a point of reference for patient voice across the Solve-RD project and ii) providing a ‘critical friend’ function to those engaged in delivering the project;
- + Demonstrate the added value of patient involvement by bringing useful and impactful input in specific areas of Solve-RD;
- + Support and facilitate engagement of stakeholders within, and across, initiatives and networks in the field of diagnosis at European and international levels (including Undiagnosed Diseases Network International, the Global Commission to end the diagnostic Odyssey for children with a rare disease, SWAN Europe).

### The Global Commission



The Global Commission to end the diagnostic odyssey for children with a rare disease is a multidisciplinary group of experts with the creativity, technological expertise and commitment required to make a major difference in the lives of millions of children and their families. In 2018, the Global Commission met via 2 TCs and 2 F2F meetings in 2018 (April in Boston and September in Seattle) to prepare the roadmaps and identify solutions and pilots.

### RD-Connect

RD-Connect is a 6-year FP7-funded European project whose funding period ended in October 2018. It aimed to develop a global infrastructure linking up data from rare disease research projects in a central resource for researchers across the world through an integrated platform in which genetic data are combined with clinical phenotype information and biomaterial availability, accessible online and query able with a suite of analysis tools. This platform is called the Genome-Phenome analysis platform (GPAP) and is used by the Solve-RD project to deposit and analysed exomes for unsolved patients. The GPAP will also be further developed within the EJP on RD. The GPAP uses a Data Access Committee (DAC) which includes several stakeholders including a representative from EURORDIS. The DAC remains active and regularly reviews all requests from clinicians and scientists wishing to access the platform to deposit and analysed exome sequences from undiagnosed patients in a research context.

Input of patient representatives into RD-Connect activities was managed by EURORDIS through the Patient Advisory Council, which have been highly active throughout the project 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. The RD-Connect consortium decided to continue its work beyond the end of the FP7 funding period (October 2018) as the RD-Connect Community.

## Patient involvement in Biobanks & Registries

### Biobanks:

EURORDIS is a member of the BBMRI Stakeholder Forum and participated in several meetings in 2018 (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.

The topics included contribution of the Stakeholder Forum to the code of conduct for health research, involvement of patients and citizens in European Biobank Week 2018 and providing ideas for the BBMRI ERIC Work Programme; such as preparation of the knowledge base on biobanking for patients/consumers. The general Rules of Procedure for the whole of the Stakeholder Forum were also established and approved.

### Registries:

EURORDIS participated on 10-12 September 2018 in the 6<sup>th</sup> 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 and the upcoming launch of a new ePAG Cross-Working Group on Research & Registries and the infographics on how to improve qualities of RD patient registries developed with the Patient Advisory Council within the RD-Connect project.

Through RD-Connect and the activities of the Patient Advisory Council chaired by EURORDIS within the project, EURORDIS participated in the development of an academic article "Recommendations for Improving the Quality of Rare Disease Registries" which was published in August 2018 in the International Journal of Environmental Research & Public Health.

### 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 the global alliance of people living with a rare disease of all nationalities across all rare diseases. RDI's mission is to be a strong common voice on behalf of rare disease patients around the world, to advocate for rare diseases as an international public health priority and to represent its members and enhance their capacities. RDI brings together national and regional rare disease patient alliances from around the world as well as international rare disease-specific federations to create the global alliance of rare disease patients and families. RDI has more than 50 member organisations from over 30 countries, that in turn represent rare disease patient groups in more than 100 countries worldwide.

2018 was a pivotal year for Rare Diseases International.

The 4<sup>th</sup> RDI Annual Meeting took place on May 10<sup>th</sup> in Vienna, Austria, back to back to the European Conference on Rare Diseases and Orphan Medicinal Products (ECRD 2018). Delegates from 33 member organisations attended the first part of the meeting, which was reserved to members only. One third of the member delegates came from outside Europe: from Argentina, Brazil, Colombia, USA, Canada and all the way from India, China, Japan, Singapore, Hong Kong and South Africa.

The Membership Meeting included an Extraordinary General Assembly called by the Council to approve the legal incorporation of RDI, as well as the 2017 Activity and Financial Report and the Action Plan and Budget for 2018. At the meeting, members present unanimously adopted



the statutes of the new organisation and approved the **Council's decision to proceed with RDI's legal incorporation**, which will be completed before the end of the year.

In order to support RDI as an independent entity, EURORDIS and RDI signed a memorandum of understanding for the duration of 5 years, during which EURORDIS will provide RDI with financial and operational support. EURORDIS will assist RDI in its legal incorporation as a separate entity.

A number of advocacy actions were carried out in 2018.

In May 2018, Rare Diseases International, represented by Paloma Tejada, delivered an official statement at the World Health Assembly of the World Health Organisation (WHO) in Geneva. The statement was the product of a joint collaboration between a number of organisations holding the status of 'special relations with the WHO' (Thalassaemia International Federation, World Federation of Hemophilia, International Alliance of Patients' Organizations and March of Dimes) and the umbrella organisations that are members of the NGO Committee for Rare Diseases, including Ågrenska, EURORDIS-Rare Diseases Europe, International Alliance of Women, International Federation for Spina Bifida and Hydrocephalus and Rare Diseases

International. The statement called on Member States to “not leave behind significant but often neglected rare diseases, each of which affect relatively small numbers of patients but collectively affect at least 300 million people globally”.

In addition, throughout 2018 several meetings took place with WHO officials and preparatory work was done towards the drafting of **an MoU with the WHO via the submission**

**of a Draft Collaborative Programme of Work.**

RDI also worked on a position paper on **Universal Health Coverage** (for publication in 2019), and carried out **Consultations on the call towards a resolution on rare diseases to be issued at the 2<sup>nd</sup> High Level Event of the NGO Committee for RDs in February 2019.**



### 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), an important 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 one in 40 CoNGO Committees, approved by 34 members at the CoNGO General Assembly in 2014, and instigated by the CoNGO President in 2015. The Committee was then publicly launched in 2016 at its First High Level Event at the United Nations. Here, the Founding Act was presented and approved, and the decision to work towards securing a UN General Assembly resolution on rare diseases as a long-term goal was agreed upon.



**NGO COMMITTEE FOR RARE DISEASES**

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. Its Executive Board counts 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, World Federation of Hemophilia,** and with the global patient voice being represented through **Rare Diseases International (RDI).**

The year 2018 was marked by a series of advocacy actions within the health, human rights and sustainable development agendas of the UN that have positioned the NGO Committee as an increasingly recognised actor, particularly within New York UN stakeholders, and has catalysed support from Member States towards the cause of rare diseases. To advocate

towards the right to health for persons affected by rare diseases, the NGO Committee submitted a contribution to the Office of the High Commissioner for Human Rights (OHCHR) for its report on health and SDGs from a human rights perspective (February); and submitted an official Written Statement to the Human Rights Council 38<sup>th</sup> session in Geneva (June). In 2018, the NGO Committee also advocated towards the recognition of the diversity within the disability community by submitting a contribution to the Special Rapporteur on the Rights of Persons with Disabilities for a report on the right to health (March); as well as by submitting an oral statement at the Conference of State Parties to the Convention on the Rights of Persons with Disabilities in New York (June). The outcome of this was the inclusion of rare diseases in the official report of the Rapporteur which was presented at the UN General Assembly (September). Finally, in 2018, in collaboration with RDI, the work towards the inclusion of rare diseases

within the Universal Health Coverage (UHC) agenda of the UN was initiated, with a high number of working meetings with Permanent Missions to the UN in New York and Geneva, as well as the participation in a panel organised at the UN HQs in NYC by the WHO and Friends of UHC on December 12 (UHC Day).

These advocacy actions and networking opportunities formed part of the preparatory work for the Second High-Level Event on Rare Diseases to be organised on February 21, 2019 at the United Nations HQs in NYC, marking Rare Disease Day. The Committee will continue working towards the engagement of Member States and UN actors by requesting co-sponsorship to the event and providing speaking opportunities. This event is expected to catalyse support towards future actions of the Committee, principally at the UN General Assembly and the Human Rights Council, and in collaboration with RDI, at the World Health Assembly, both in 2019 and 2020.

## 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.*

In 2018, Rare Barometer Voices, the panel of rare disease patients who answer on a regular basis to EURORDIS' surveys has reached more than 8000 patients (7550 in the European continent including the UK and 450 outside Europe).

This year, the programme focused on communicating about past survey results. Results from the two latest surveys were presented via infographics and each of them were translated and adapted in 23 languages. Results were sorted by different target populations (ERN therapeutic areas, countries, rare diseases, pediatric population, etc.) to help the rare disease community advocacy work. Results were presented in several conferences by the EURORDIS

staff and by the rare disease community at large.

One quantitative was carried out in 2018 on health-related data sharing and protection in order to draw recommendations on this topic and foster the inclusion of rare diseases patients' perspectives in data sharing initiatives. Results gathering 2013 respondents were analysed in collaboration with the University of Cardiff School of Social Science in order to publish them in an academic journal.

Another quantitative survey on rare disease patients' experience of treatments which will be launched in 2019 has been prepared in collaboration with a Topic Expert Committee representing patient organisations and various experts on the topic. This survey will help us to build and strengthen our positions on a number of different themes, including prioritisation of unmet treatment needs, reinforcing good clinical practices for paediatric and adult rare disease patients and the evaluation of EU orphan drug regulation and the wider EU regulatory framework.

The programme also implemented new features in the Rare Barometer Voices database to be able to launch a common patient experience survey across ERNS, enabling in particular to connect rare disease patients to HCPs affiliated to ERNS. 4 ERN networks (ERN Lung, eRKNET, GENTURIS and eUROGEN) have volunteered to participate in a pilot ERN common patient experience survey.

The multistakeholder Rare Barometer Advisory Committee, that includes patients organisations, policy advisers, academics, EURORDIS relevant staff and corporate partners, took place in December. The objective of the meeting was to discuss the strategy of the programme and future topics to be explored through Rare Barometer. Diagnosis was confirmed as a high priority subject for the rare disease community at large and thus for the programme.

# 2. PATIENT EMPOWERMENT:

## Building the network & building capacities

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

#### 2.1.1 Membership

60 new members joined EURORDIS in 2018. At the end of 2018, EURORDIS had 826 members in 70 countries, 42 of which are European countries, 28 being members of the European Union.

#### 2.1.2 EURORDIS Membership Meeting 2018 Vienna

*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 majority of participants (75%) are EURORDIS member organisations and other patient organisations and about 25% of the participants represent policy makers, industry and academia.*

The EMM 2018 Vienna took place on 10 May 2018, back to back with ECRD Vienna 2018. It comprised the General Assembly, ePAG and RDI meetings.

The ePAG meeting focused on the ePAG Constitution and Rules of Procedure; Taking Stock of what has been achieved and what are the main areas of future development;



Patient-clinician partnerships; Patients' involvement in the development of clinical guidelines.

The RDI meeting comprised the Extraordinary General Assembly of RDI and its annual meeting.

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

### 2.1.3 Council of National Alliances (CNA)

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

EURORDIS supports a network of 41 national alliances, 35 of which constitute the CNA.

In 2018, 2 CNA Workshops took place, in Brussels on 15 March and in Paris on 10-11 December. The latter was again held partly in common with the Council of European Federations (CEF), in order to allow cross-cutting discussions on common topics.

The CNA's main activities in 2018 were the work on:

- + a) the preparation and coordination of the Rare Disease Day 2019 and Rare Disease Day Strategic Review
- + b) EURORDIS Working Group on Key policy priorities for RDs

#### Creation of two CNA Working Groups on: Small EU countries and Western Balkans

The CNA endorsed the launch of two CNA working groups in 2018.

The CNA Working Group of the National Alliances from the smaller EU countries is based on an initiative launched by the Cyprus Alliance for Rare Disorders (CARD). Rare Disease patients and their organisations in small EU countries face the same challenges, due to the size of the population, the limited number of Health Care Professionals and access to treatment. The focus of this Working Group is to collaborate on European Reference Networks (ERN) and Health Care Providers (HCP), and access to Orphan Medicinal Products (OMP) and other rare disease therapies. The Internal Working Group "Small EU Countries" is led by the Cyprus Alliance for Rare Disorders, embedded into the CNA and supported by EURORDIS staff. The following countries

form part of this group: Cyprus, Estonia, Latvia, Malta, Croatia, Lithuania, Luxembourg, and Slovenia.

At the CNA meeting of December, 2018, it was decided to officially create a CNA Working Group of Western Balkan countries, based on the initiative launched by the National Organization for Rare Diseases of Serbia (NORBS). The following countries will be included in the WG: Albania, Bosnia and Herzegovina, Croatia, FYR of Macedonia, Montenegro, Serbia. The focus of this WG will be to address the needs of people living with rare diseases in the region of the Western Balkans. The main objectives will be collaboration, networking and capacity building on National Plans and Strategies for Rare Diseases

The two working groups will meet in Bucharest May 16 2019, in conjunction with the CNA meeting and the EMM 2019.



### 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: Integration of ERNs into national HC systems; ePAG Training; Rare Barometer Voices surveys; Patient Engagement in research; EURORDIS Position paper on social rights & integrated care.

The second part (CEF only) focused on: Community Advisory Boards (CABs); the Paradigm Project; and a Peer to Peer session.

The second day of the meeting was dedicated to a training on Horizon scanning, HTA and Drug Repurposing.

EURORDIS continued for the 9<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 2018, EURORDIS gave 21 European RD Federations financial support to help them organise their different meetings. A total of 39 250 € 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 16 help lines which are members of the European Network.*

In 2018, 16 help lines from 11 countries participated in the activities: Bulgaria (ICRDOD), Croatia (Croatian Help Line for Rare Diseases), France (Maladies Rares Info Services, AFM-Téléthon), Italy (Coordinating Centre for Rare Diseases Veneto Region, and Telefono Verde Malattie Rare), Portugal (Linha Rara), Romania (NORO, Myastenia Gravis Romania), Spain (SIO-Feder), Switzerland (Info Maladies Rares, Help Line Seltene Krankheiten), Hungary (Lifebelt, Information Centre for the Rare Disease Patients), Denmark (Rare Disorders Denmark), Ireland (National Rare Diseases Office) and Serbia (National Organisation for Rare Diseases of Serbia NORBS).

One help line stopped its activities (ENERCA, transformed into a European Reference Network on Rare Anaemias).

The network conducted its 11<sup>th</sup> Annual Caller Profile Analysis in October 2018 and participated in discussions on communication campaigns, on exchange of information with National Contact Points for Cross-Border care. EURORDIS organised a face to face meeting of the Help Lines in Paris in October 2018.



### 2.1.6 European Patient Advocacy Groups (ePAGs)

*In 2016, EURORDIS in collaboration with the European rare disease community established 24 European Patient Advocacy Groups (ePAGs) as forums to optimise the involvement of patient representatives of the rare disease community in the 24 ERNs. Each ePAG corresponds to the scope of one of the 24 ERNs, aligning patient organisations and clinicians, experts and researchers working on the same rare or complex disease or highly specialised intervention. Today, the ePAGs consist of 255 ePAG patient advocates across the 24 ERNs. Likewise, 1760 patient organisations have registered to be part and engage in the ERN that covers the diseases that the patient organisation is representing.*

A priority area this year has been to empower and train ePAG patient advocates to help them develop soft skills and technical knowledge in areas relevant to ERNs.

In 2018, EURORDIS has organised (i) an ePAG face-to-face meeting, (ii) an ePAG f2f Steering Committee meeting (iii) an ePAG Steering Committee training session.

The first meeting was held on 10 May and it allowed ePAG patient advocates to exchange views on key topics such as the common governance framework, patients' involvement in clinical practice guidelines and patient-clinician partnership. It was also the opportunity to receive updates on ERNs and ePAGs; patient advocates were also able to present their work and reflect on what had been achieved, what were the priorities and challenges for the future. The meeting was attended by more than 60 ePAG patient advocates and was an exciting and informative day for all, coming together as a community and fostering cross-ERN collaboration.

The second meeting took place on 13<sup>th</sup> September 2018 at EURORDIS offices in Brussels. 22 ePAG patient advocates from 15 ERNs were present. The main topic discussed was the integration of ERNs into National Health Systems. Participants were able to discuss at length and provide specific feedback to EURORDIS Recommendations on the Integration of ERNs into National Health Systems. This meeting also provided also the opportunity to reflect on the role and function of ePAG patient advocates and how to communicate that role to different audiences.

The next day, a training session on effective communication and presentation skills was organized for the ePAG



Steering Committee members. A total of 18 ePAG patient advocates attended this training workshop.

In addition to the meetings organized by EURORDIS, EURORDIS has supported the organization of 7 individual ePAG f2f meetings that were attended by 49 ePAG patient advocates, with a special focus on those groups that were still forming and where a face-to-face meeting would help to identify strategic priorities and form their identity as a group. Finally, we have also facilitated ePAG representation at 12 ERN annual and Board meetings throughout 2018.

EURORDIS held more than 100 conference calls with ePAG patient advocates in 2018. The majority of these calls were organized with individual ePAG groups. We also held 7 ePAG Steering Committee calls to discuss topics of relevance to the 24 groups (cross-ERNs). In addition, EURORDIS has launched three transversal topic groups as learning forums to exchange and share information on the following areas: research and registries topics, training and education and outcomes and guidelines.

Besides the ePAG Steering Committee face-to-face training workshop organized in September, EURORDIS has organized 5 webinars on topics such as healthcare pathways and clinical guidelines, CPMS, influencing skills and ERNs. More than 130 people followed these webinars.

EURORDIS has also developed a comprehensive Leadership training programme for ePAGs on leadership, network management, healthcare and research. Training topics include, among others, self-awareness on leadership skills, communicating with impact, conflict resolution, European Reference Networks, healthcare pathways and clinical practice guidelines, integrated and networked care, digital health and research and therapeutic development. The programme will be launched in 2019 and will be delivered with a blended format, including a series of e-learning courses (webinars) as well as a face-to-face training session. 2019 will be a pilot year, where we will be testing the programme structure, delivery methods, contents and trainers.

### 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 translated into 12 languages and is home to 180 disease-specific communities created in partnership with 940 patient groups and managed with the support of 400 volunteer moderators.*



Following the transfer of RareConnect to the Children's Hospital of Eastern Ontario Research Institute (Canada) in 2017 last year saw a significant amount of work put into the development of a research portal which will allow opt-in interaction between the research community and members of RareConnect. Following the initiation of an Ethics approval process the platform developed its preparedness to host research studies on a separate and secure environment which complies with all international norms and best practices for the treatment of such sensitive data. The platform also underwent a full review for compliance with the EU GDPR Data Directive.

### 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).

### 2.1.9 EURORDIS Trainings

#### EURORDIS Open Academy

In 2018, building upon its experience of ten years of capacity-building programmes, EURORDIS developed and launched the Open Academy consolidating all of EURORDIS' training activities. The primary goal of the Open Academy is to empower patient advocates in the various fields where patient engagement is needed. The programmes are based on a blended approach with online trainings and face-to-face components and the Academy is characterised as open as all online training modules are also made freely available via the EURORDIS Website to anyone interested in building their capacities independently. The EURORDIS Open Academy encompasses the EURORDIS Summer School, EURORDIS Winter School, EURORDIS Digital School (coming soon) and the EURORDIS Leadership School (ePAG Capacity Building Programme). The EURORDIS Winter School on Scientific Innovation and Translational Research was held for the first time in March 2018. Another key element of the EURORDIS Open Academy in 2018 was the 11<sup>th</sup> EURORDIS ExPRESS Summer School that traces the medicines development process pathway from medicines discovery,



clinical trials, aspects of the EU regulatory process right up to access for patients and was held in Barcelona in June 2018.

#### 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 2018 version, a much greater emphasis was placed on the pre-training aspect of the Summer School Programme. The pre-training is comprised of training modules that the trainees can complete online before they arrive in Barcelona for the face-to-face session. Each module covers specific topics such as Medical Research Ethics, Statistics, Regulatory Procedures, European Medicines Agency, Benefit Risk, HTA and Market Access. The trainees had approximately 2.5 weeks to complete each module.

The 2018 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.

52 participants attended representing 17 countries and 43 diseases. The Summer School 2018 participants have been selected based on a call for candidates. EURORDIS received 133 applications of which 20 were from research institutions of which 52 candidates were selected

Training material from previous Summer Schools including webcasts and slide presentations as well as interactive training tools and webinars have now be developed into a full online training programme on the EURORDIS website The training is made up of 6 separate units.

### Spanish edition of EURORDIS Summer School

A 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 participants also took part in a pre-training programme based on sessions that were video recorded during the 2017 version.

### First EURORDIS Winter School on Scientific Innovation and Translational Research

*The EURORDIS Winter School on Scientific Innovation and Translational Research consists of one week face-to-face training and of online training modules, available for free. EURORDIS launched the training with the aim of deepening patient representatives' understanding of how pre-clinical research translates into real benefits for rare disease patients. Expert speakers and researchers from across Europe deliver the face-to-face training sessions.*

*The training equips participants with knowledge and skills so they are empowered to effectively participate in discussions with the researchers, policy makers and companies responsible for research or research infrastructures.*

The first edition of the EURORDIS Winter School on scientific innovation & translational research took place on the 19-23 of March 2018, at the Imagine Institute, in Paris. Imagine Institute is a research and innovative healthcare institute, bringing together researchers, doctors and patients, with a common goal to cure genetic diseases. 29 patient representatives, from 12 countries, participated in the training course aiming to support patient engagement in research. The course covered topics such as the history of genetics, diagnostics, new technologies in gene therapy, and drug repurposing and included laboratory visits. The patient advocates attending included European

Patient Advocacy Groups (ePAGs) who represented various European Reference Networks as well as representatives of patient organisations for undiagnosed genetic conditions.



## 2.2 Raising Awareness & Informing

### 2.2.1 Rare Disease Day 2018

*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 11<sup>th</sup> edition of the day, Rare Disease Day events took place in over 90 countries and regions on every corner of the globe. 5 new countries had events this year – Togo, Ghana, Trinidad and Tobago, Cape Verde and The Syrian Arab Republic. 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 and regions participated.

The overarching theme was Patient Involvement with the specific theme of Research. The slogan was: Show Your Rare, Show You Care. 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 around the world happening on and around the day.

The interactive face-paint social media campaign "Show your Rare" was launched in 2018. The rare disease community and wider public were encouraged to show their support for raising awareness of rare diseases by sharing a selfie or photo of them wearing colourful face-paint to 'show their rare' on their social media channels. The campaign's simple and interactive call to action was popular around the world.

For the 7<sup>th</sup> year in a row EURORDIS produced a well-received video for Rare Disease Day that launched the Show Your Rare interactive campaign. On Rare Disease Day and EURORDIS social media, the video was viewed by over 350 000 people. It had over 7000 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.

On the occasion of the 10 years since the launch of the first Rare Disease Day, EURORDIS decided to carry out a strategic review for the RDD initiative in preparation for the next 10 years and to maintain a campaign that creates positive change for people living with a rare disease. The strategic review comprises three phases and includes internal and external audits, presentations, and meetings based on research and behavioral science methods. A presentation of the Strategic review was made to the Council of National Alliance on 10 December 2018. A follow up survey was sent to EURORDIS members to obtain their thoughts on the future of Rare Disease Day. The review will end in March 2019.





EURORDIS was implicated in the hosting of 3 events in 2018 for Rare Disease Day:

- + EURORDIS Black Pearl Awards Ceremony on 20 February took place in Brussels and was livestreamed online (see more information in the separate section below).
- + Rare Lives exhibition on 28 February. The exhibition of Rare Lives photos at the European Parliament was organised by UNIAMO FIMR Onlus, together with MEP Elena Gentile and with the support of EURORDIS-Rare Diseases Europe to mark the occasion of Rare Disease Day 2018.
- + The European Reference Network for rare bone diseases (ERN BOND) presented its white paper on diagnosis at an event at the European Parliament.

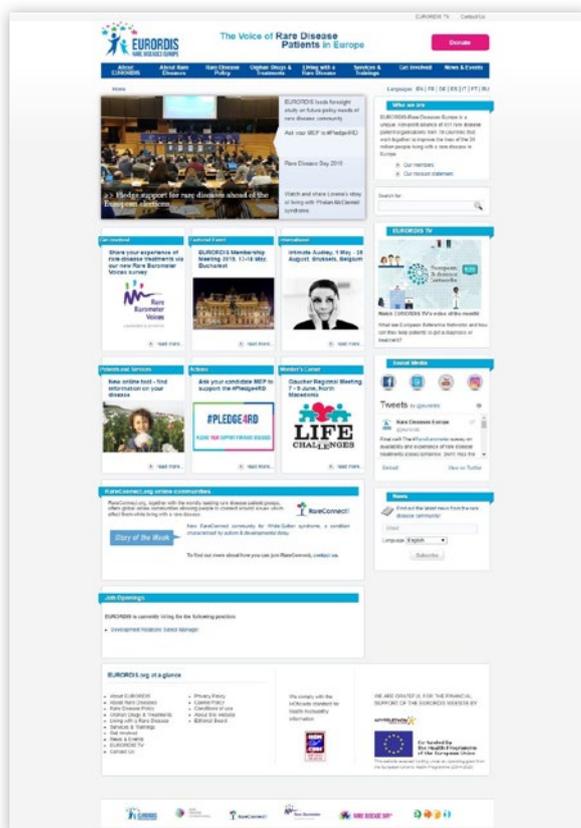


## 2.2.2 EURORDIS Website

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.

In 2018, we went through a call for tender process to select an agency to create a new eurordis.org. The agency kicked off the user research phase of the project in 2018, which will be followed up by building a new website in 2019.



### 2.2.3 eNews & Member News

#### 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 2018, 11 eNews issues were written, translated in 7 languages, produced and distributed via email. This publication is year-round at a frequency of once a 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.

Lead stories topics in 2018 included: INNOVCare Project results demonstrate need for integrated care for rare disease patients; Community Advisory Boards - connecting patients with clinical research; Show Your Rare for Rare Disease Day 2018!

### 2.2.4 Social Media

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

Social media content in 2018 included: Scheduled content taken from the eNews; Live content from events including our Rare Disease Day events in the month of February and also at our training programmes throughout the year; Spontaneous content to disseminate information of interest from and to the rare disease community, including information received from members and projects; Posts to encourage the public to register for our events, take part in our capacity-building trainings, respond to our surveys and submit photos to our Photo Award; Improved social media visuals, using templates designed by a graphic designer but easily adaptable by the internal team; Increased use of Instagram to reach a new audience; New use of Facebook live for broadcasting EURORDIS 'how to' webinars to make them more openly available to a wider public.

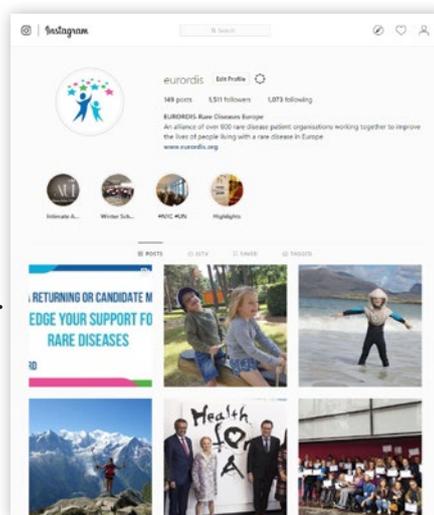
#### Member News

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 2000 contacts.

In 2018, our Member News was distributed twice a month to 2000 contacts at our member organisations. Its simple design has ensured a high open rate. It provides links to information and activities in a simple, streamlined text template to ensure relevant information is reaching EURORDIS members as directly as possible. Each Member News is translated into the 6 languages of the EURORDIS website.

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.



## 2.2.5 The EURORDIS Black Pearl Awards



The EURORDIS Black Pearl Awards recognise the outstanding achievements and ground-breaking work of those committed to improving the lives of people living with a rare disease. The Awards are presented to patient advocates, patient organisations, policy makers, scientists, companies and media at a unique annual event held every year in February to mark the occasion of Rare Disease Day. 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 2018. There were over **350 nominations**, representing **37 different countries worldwide**.

The winners of the EURORDIS Awards 2018 were:

- + **EURORDIS Young Patient Advocate:**  
Sammy Basso, Associazione Italiana Progeria Sammy Basso, Italy
- + **EURORDIS European Rare Disease Leadership Award:**  
Bruno Sepodes, Chair, Committee of Orphan Medicinal Products, European Medicines Agency, Portugal
- + **EURORDIS Policy Maker Award:**  
Elena Gentile, Member of the European Parliament, Italy

- + **EURORDIS Scientific Award:**  
Professor Michele de Luca, University of Modena and Reggio Emilia, Italy & Dr Tobias Hirsch, BG University Hospital Bergmannsheil, Germany
- + **EURORDIS Volunteer Awards:**  
Chris Sotirelis, UK Thalassaemia Society, United Kingdom, Helene and Mikk Cederroth, Wilhelm Foundation, Sweden
- + **EURORDIS Patient Organisation Award:**  
European Pulmonary Hypertension Association
- + **EURORDIS Company Award for Innovation:**  
Novartis
- + **EURORDIS Company Award for Patient Engagement:**  
The PARADIGM Project (the European Federation of Pharmaceutical Industries and Associations-EFPIA, Bayer, UCB and MSD)
- + **EURORDIS Visual and Audio Media Award:**  
Special Books by Special Kids, Christopher Ulmer, USA
- + **EURORDIS Written Media Award:**  
'On peut changer le monde en vendant des crêpes et des ballons', Serge Braun, AFM-Téléthon, France
- + **EURORDIS Lifetime Achievement Award:**  
Alastair Kent OBE, United Kingdom

# 3. PATIENT ENGAGEMENT: Roles in decision-making

## 3.1 Patient Engagement in Healthcare

### 3.1.1 European Reference Networks

*EURORDIS has been a 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 well 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 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 in 2016. The establishment of the ePAGs and ePAG representatives enables a uniform structure for patient involvement in ERNs network boards, clinical committees and working groups.

In 2018, EURORDIS supported the organization of 7 individual ePAG f2f meetings that were attended by 49 ePAG patient advocates, with a special focus on those groups that were still forming and where a face-to-face

meeting would help to identify strategic priorities and form their identity as a group. EURORDIS also facilitated ePAG representation at 12 ERN annual and Board meetings throughout 2018

More than 100 conference calls with ePAG patient advocates were held in 2018. The majority of these calls were organised with individual ePAG groups. We also held 7 ePAG Steering Committee calls to discuss topics of relevance to the 24 groups (cross-ERNs). In addition, EURORDIS launched three transversal topic groups as learning forums to exchange and share information on the following areas: research and registries topics, training and education and outcomes and guidelines.

In 2018, EURORDIS continued to support Rare Disease National Alliances on European Reference Networks. We organised workshops in 10 different countries to help raise awareness and knowledge on ERNs and engage with national stakeholders (patient organisations, clinicians and health authorities) to help them understand how they can participate and benefit from this new structure. In addition, a dedicated CNA working group of National Alliances from smaller Member States was created to which tailored advice was provided in 2 dedicated conference calls.

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### 3.1.1 Patients Active in Research and Dialogues for an Improved Generation of Medicines (PARADIGM)



PARADIGM is a 30-month public-private partnership launched on 1 March 2018 and led by the European Patients' Forum (EPF) and EFPIA. The consortium includes patient organisations, regulatory bodies, universities, non-profit organisations, Small & Medium Enterprises (SMEs), pharmaceutical companies and pharmaceutical trade associations. Its mission is to advance a structured, meaningful and ethical patient engagement in medicines development and aims to develop tools and resources to allow the effective and systematic inclusion of patients and to design an innovative roadmap to ensure long-term sustainability of patient engagement in three key decision-making points in medicines R&D: 1) the setting of research priorities, 2) the design of clinical trials, and 3) the early dialogue between regulators and HTA bodies.

EURORDIS has a prominent role in PARADIGM being part of the general assembly and leading both the sustainability strategy and the co-development of recommendations on the capabilities required by all stakeholders to implement effective patient engagement.

EURORDIS first started by assessing the needs and expectations from all stakeholders regarding patient engagement, including aspects on sustainability (e.g. financial compensation of patient participants). In addition,

a benchmarking exercise was conducted to explore existing sustainability models in similar socially impactful ecosystems (e.g. education, refugees, environment) including patient engagement initiatives (such as EUPATI or PCORI) and others related to health. A series of interviews were performed to identify what make those initiatives/organisations sustainable based on 3 pillars: 1) culture (cultural changes to make it the norm); 2) processes (how to drive all stakeholders together) and 3) resources (how to mobilize the resources to achieve the objective). Currently we are developing potential sustainability scenarios suitable for the patient engagement framework. We started by generating 4 sustainability models and the work will continue to identify the feasibility and viability of the options proposed.

The final sustainability roadmap will include:

- + Catalogue of services
- + Definition of a multi-modular framework to provide one common entry point for all stakeholders
- + Business model
- + The associated partners and the governance strategy
- + Operational plan for a sustainable long-term approach
- + Financial plan to underpin sustainability

## 3.2 Patient Engagement in Social Care

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

In 2018, EURORDIS continued to **widely disseminate 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'** (2017) - to raise awareness of all stakeholders on the serious impact of RD on everyday life.

The results of this survey, conducted via Rare Barometer Voices, were shared widely with EURORDIS members and all stakeholders, at key national and European platforms and events. The survey infographics disseminated in various media, including the European Parliament Magazine (February).

Throughout the year, EURORDIS continued the process to elaborate **EURORDIS position paper on "Achieving Holistic Person-Centred Care to Leave no One Behind"**, aiming at advocating for measures to improve the everyday life of people living with a rare disease and their families. The final draft of the paper was presented to the Council of National Alliances and to the Council of European Federations in December. The paper will be launched in May 2019, at the EURORDIS Membership Meeting in Bucharest.

This position paper built on robust steps taken over the last 7 years by EURORDIS and its members, via advocacy actions, social surveys and projects. Additionally, EURORDIS worked closely with its volunteers and members to write the position paper. Several consultations took place in 2018, involving the Social Policy Advisory Group, the Council of National Alliances, the Council of European Federations and members at large.

In parallel, EURORDIS **continued to follow the discussions on the implementation of the European Pillar of Social Rights and contributed to the European Parliament discussions on the 'Work-Life Balance' Directive proposal**. In April, EURORDIS shared a concept paper and a set of amendments proposals with Members of the European Parliament, to present the challenges of the RD community and to propose specific improvements to the proposal, to meet the work-life balance needs of people living with a RD and their families.

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. In December 2018, the Social Policy Advisory Group concluded its mandate. A new Social Policy Action Group will follow to continue supporting EURORDIS' activities in social policy and holistic care.

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

EURORDIS continued its focus on social services, mainly through its involvement in the EU-funded projects **RD-Action** (2015-2018) and **INNOVCare** (2015-2018) as well as via the promotion of the representation of people living with a RD and their carers in relevant policies at European level.

Within the INNOVCare project, EURORDIS continued to manage the secretariat of the European Network of Resource Centres for Rare Diseases-RareResourceNet and supported its transition to the Frambu resource

centre, after the end of the project (September 2018). RareResourceNet focuses on advancing holistic high quality care for people living with a RD and their carers in Europe. **Raquel Castro, EURORDIS Social Policy Director, was nominated by the EURORDIS Board of Directors, in November, to join the Board of Directors of RareResourceNet as patient representative.**

During this last year, EURORDIS also continued to strengthen the cooperation with key organisations working on social policy: the Social Platform, of which EURORDIS is a member, and the International Federation of Social Workers Europe, with whom EURORDIS has a Memorandum of Understanding.

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

## INTEGRATED CARE FOR RARE DISEASES: BRIDGING THE GAP TO IMPROVE THE LIVES OF 30 MILLION PEOPLE IN EUROPE

### RARE DISEASES

-   
 Rare, complex, chronic, disabling
-   
**30 million**  
 people affected in Europe
-   
 Fewer than **1 in 2000**  
 people affected by each rare disease
-   
 No cure for the vast majority and few treatments available
-   
 Expertise and knowledge on their consequences are scarce and difficult to access

### PEOPLE LIVING WITH A RARE DISEASE AND THEIR CARERS FACE SERIOUS CARE BURDEN<sup>1</sup>

  
**65%**  
 have to visit different health, social and local services in a short period of time

  
**7 in 10**  
 do not feel well informed about their rights



  
**67%**  
 feel that these services communicate badly between each other

  
**7 in 10**  
 find that organising care is time-consuming; 6 in 10 find it hard to manage

### INTEGRATED HEALTH AND SOCIAL CARE IS ESSENTIAL TO ENABLE PEOPLE LIVING WITH A RARE DISEASE TO

- ✓ **Overcome their care burden and to secure the services and support that they require**
- ✓ **Achieve a quality of life on an equal footing with other citizens**
- ✓ **Participate in society and in the job market to their highest potential**
- ✓ **Fully realise their fundamental human rights**

*The appointments should be multidisciplinary allowing for the various specialists to see the patient on the same day and place. Besides facilitating communication, it would avoid that the patient has to travel to different appointments and places, being absent from work: losing income, facing discrimination at the workplace and spending a lot in travel". Female, Portugal*

The European Commission Expert Group on Rare Diseases therefore recommends that "European Member States should promote measures that facilitate multidisciplinary, holistic, continuous, person-centred and participative care provision to people living with rare diseases, supporting them in the full realisation of their fundamental human rights"<sup>2</sup>.

**SHARE THIS FACTSHEET TO HELP RAISE AWARENESS OF THE NEED FOR INTEGRATED CARE FOR RARE DISEASES**

#RareDisease | #INNOVCare  
[innovcare.eu](http://innovcare.eu)

**What is Integrated Care?** "Integration is a coherent set of methods and models on the funding, administrative, organizational, service delivery and clinical levels designed to create connectivity, alignment and collaboration within and between the care and care sectors. The goal of these methods and models is to enhance quality of care and quality of life, consumer satisfaction and system efficiency"<sup>3</sup>.

Sources:  
<sup>1</sup> EURORDIS-Rare Diseases Europe (2017). Juggling care and daily life: The balancing act of the rare disease community. <http://bit.ly/SurveyRD>. Survey of over 3000 people, conducted through the EURORDIS survey initiative Rare Barometer Voices: [eurordis.org/voices](http://eurordis.org/voices)  
<sup>2</sup> Commission Expert Group on Rare Diseases (2016). Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies. <http://bit.ly/CEGRDreco>  
<sup>3</sup> International Journal of Integrated Care (IJIC) (no date). IJIC guidelines for research and theory papers. Adapted from Kodner, D., & Spreeuwenberg, C. (2002).

  
This factsheet was developed in the scope of the EU-funded INNOVCare project

  
This project is co-funded by the European Union

  
The information contained in this publication does not necessarily reflect the official position of the European Commission.

EURORDIS continued to promote integrated care for RD in 2018 via the various activities of the **INNOVCare project** and by ensuring the wide dissemination of the results of the project.

In April, EURORDIS co-organised the joint **INNOVCare and RD-Action Workshop on 'Creating a Sustainable Environment for Holistic & Innovative Care for Rare Diseases & Complex Conditions'** (65 participants from 22 countries). The workshop focused on key issues to advance

integrated care for RD, including the **role of Centres of Expertise and European Reference Networks** and the possible **impact of case management services**.

The workshop was also the occasion for EURORDIS to release a **factsheet on integrated care for rare diseases**, developed within the INNOVCare project, using results of 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' (2017).

In September, EURORDIS organised the **'Conference on Advancing person-centred & integrated care for rare diseases & complex conditions across Europe'**, in Brussels. The results of INNOVCare's pilot of case management and a set of recommendations on integrated care for RD were released at the conference, followed by high-level discussions with all stakeholders. The event brought together 80 participants from 18 European countries and was followed online by over 280 people.

The pilot of case management conducted and evaluated within the INNOVCare project resulted in important outcomes to advance integrated care for RD:

- + An increase in beneficiaries' level of information about their disease, their rights and available services as well as in their capacity to manage their own care;
- + A reduction in the burden faced by caregivers (Zarit Caregiver Burden Interview);
- + An improvement in coordination between care providers.

INNOVCare also supported the **identification of key opportunities and challenges to up-scale integrated care**

of RD and other complex conditions in European countries.

EURORDIS coordinated the **elaboration of recommendations to the EU and MS, to support the implementation of integrated care for RD**. The recommendations built on the outcomes of the project and on the inputs of all stakeholders, including: EURORDIS volunteers and members as well as the project's **Advisory Group**, managed by EURORDIS, composed of competent authorities from European countries. These recommendations directly supported the draft of the **EURORDIS Position Paper on 'Achieving Holistic Person-Centred Care to Leave no One Behind'** (to be launched in May 2019).

In the last trimester of 2018, EURORDIS ensured the wide dissemination of the outcomes and recommendations resulting from the INNOVCare project, at European and national level. In this scope, EURORDIS presented the **outcomes of INNOVCare at the meeting of the European Commission with the representatives of Member States** involved in the EU Programme for Employment and Social Innovation (EaSI Committee) and with the European Structural Fund Plus Working Group.



### 3.2.4 Promoting the recognition of rare diseases within the disability agenda

In 2018 EURORDIS strengthened its actions to raise awareness of the disabilities faced by people living with a RD and to advocate for the recognition of the needs of the RD community within the disability agenda.

In March, EURORDIS supported the contribution of the NGO Committee for Rare Diseases to the study of the United Nations Special Rapporteur on the Rights of People with Disabilities, Catalina Devandas Aguilar, on 'the right of persons with disabilities to the highest attainable standard of health'. The report of the study, presented at the 73<sup>rd</sup> Session of the General Assembly of the United Nations, referred to the RD community

as one of the most marginalised groups of persons with disabilities.

EURORDIS also presented at the 'Work Forum on the Implementation of the UN Convention on the Rights of Persons with Disabilities', organised by the European Commission, in May. EURORDIS' presentation, included in the session on 'Health, rehabilitation and habilitation', focused on demonstrating the disabilities faced by people living with a RD as well as on showing good practices of addressing health and rehabilitation for RD.

Throughout 2018, EURORDIS also continued to strengthen its cooperation with the European Disability Forum (EDF). A representative of EDF has joined a session of the European Conference on Rare Diseases & Orphan Products (ECRD, May 2019) on 'Disability: unveiling the invisible double-burden of rare diseases'.

## 3.3 Patient Engagement in Research

### 3.3.1 International Consortium for Rare Disease Research

*The International Rare Diseases Research Consortium (IRDiRC) unites national and international governmental and non-profit funding bodies, companies (including pharmaceutical and biotech enterprises), umbrella patient advocacy organizations, and scientific researchers to promote international collaboration and advance rare diseases research worldwide. Importantly, the coverage of the Consortium is global and involves stakeholders from Africa, Asia, Australia, North America, and Europe.*

*The vision 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*

EURORDIS has actively participated in the International Consortium for Rare Disease Research (IRDiRC) since its launch and in particular in 2018, with involvement of several staff members in the Consortium Assembly, the Operating Committee, the Patient Advocacy Constituent Committee (PACC) and the Therapies Scientific Committee (TSC). Since January 2018, Virginie Bros-Facer, Scientific Director is the official representative of EURORDIS in the Consortium Assembly and the PACC; since March 2017, Virginie Hivert, Therapeutic Development Director is Vice-Chair of the TSC in which Yann Le Cam, CEO of EURORDIS is also a Member and immediate past-Chair. As Vice-Chair of the TSC, Virginie Hivert is therefore also a member of the Operating Committee.

**Task Force 'Orphan Drug Development Guidebook':** In 2018, Virginie Hivert (TSC) was involved in the Task Force 'Orphan Drug Development Guidebook' with the aim of creating a handbook for academic, patient and industrial drug developers describing the available tools and initiatives specific for rare disease development and how best to use them.

This Taskforce led to the organization of a workshop gathering international experts contributing to achieving this work, i.e. to devise a development strategy for a



number of paradigmatic cases of development in rare disease indications and set the ground for the preparation of a guidebook for developers to navigate the incentives, initiatives, and practices available in the rare disease space and understand how to best integrate them in their development.

**Task Force 'Identification of barriers to patient participation in RD research and recommendations to remove them':** In 2018, Virginie Bros-Facer (PACC) was involved in this Task Force which has developed a phased approach to carry out its activities. It is important to note that due to the move of the scientific secretariat from Orphanet to the European Joint Programme on Rare Diseases in late 2018, the Task Force was momentarily put on hold and activities have been delayed. A re-launch will take place in 2019.

The first phase will focus on 1) mapping existing surveys, recommendations and other relevant materials to establish a benchmark and 2) start an internal consultation among IRDiRC and PACC members on the perceived barriers to patient participation in RD research. The second phase will include a wider consultation of the international RD patient community on the challenges and barriers of participation in RD research with questionnaires and survey based on the mapping and benchmark analysis performed in the first phase.

This Task Force will ultimately develop recommendations by i) determining alignment of current efforts with needs; ii) facilitating better patient engagement across geographic areas using shared resources; iii) determining strategic areas for new funding initiatives and iv) informing IRDiRC of its future activities.

## 3.4 Patient Engagement in lifecycle development

### 3.4.1 Patients creating their Community Advisory Boards to engage 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 their 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. The same group of patients advises several sponsors in their field. It avoids selection of patients' representatives by the sponsor. The agenda and secretariat are driven by the patients.*

This activity started in 2018 to support patient organisations in setting up and structuring a Community Advisory Board (CAB), which is a group of patients who offer their expertise to sponsors of clinical research for their

disease area through a transparent and effective process. EURORDIS support through the programme includes establishing a common framework for patient groups and sponsors: capacity building of patient advocates, peer-to-peer exchange of experiences across CABs, quality monitoring of the process and outputs of CABs, transparency and prevention of competing interests, promotion of the programme, and evaluation and possible eventual scientific publication.

During 2018, two guidelines were finalised: How to create and operate a CAB; Travel, subsistence and compensation of time spent whereas three more were under development: Code of Conduct for CAB members; Confidentiality undertaking and Insider Trading prevention; How to evaluate your CAB?.

A number of training webinars were organised throughout the year focusing on ethics, medical research, regulatory/EMA and HTA; and the EUROCAB programme was presented to the Council of European Federations (CEF) during the annual CEF meeting held in Paris in December.

By end of 2018, 7 CABs had been launched, with another 4 in preparation.

### 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).*

#### Identification of & support to RD patients participating in EMA Scientific Committees

Dedicated expert patient representatives contributed to the examination and scientific evaluation of dossiers in 2018 through the work of the scientific committees they belong to, as well as to the activities of several adhoc working groups all along the year.

EURORDIS supports the **work of patient representatives** in the COMP, PDCO, CAT; supports the **participation of regular/ad hoc experts** to the COMP, PDCO, CAT; promotes **ad hoc participation of Patient experts** on the COMP in the discussions on reassessment of the orphan status at the time of Marketing Authorisation; identifies & selects patient representatives to be appointed to EMA Scientific Committees. In 2018, EURORDIS selected and, proposed or endorsed, **patient representatives** to be appointed to the CAT and the PRAC.

Patients' representatives and staff involved with Scientific Committees at the EMA dedicate their time, experience and expertise to the **tasks** of evaluating dossiers of



EUROPEAN MEDICINES AGENCY  
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medicinal products applying for orphan designation and for orphan status reassessment at the time of Marketing Authorisation and contribute to Scientific Advice. In addition, they review Public Summaries of Opinion on orphan designation and Maintenance/Significant Benefit Assessment reports; contribute to Scientific Advice and Paediatric Investigation Plans; evaluate advanced therapies, contributing to Scientific Advice, classification and ensuring accurate, transparent and available information to patients on authorised medicinal products.

Patients' representatives and staff involved with Scientific Committees at the EMA also participate to **Working Groups** organised by the different Committees. In 2018, they participated to the **Strategic and Learning Review meetings** held under the EU Presidency and to **internal workshops** organised by the Committees (e.g. Multi-stakeholder workshop to further improve the implementation of the Paediatric Regulation; Workshop on haemophilia registries at EMA, 8 June 2018; Annual workshop of the European network of paediatric research at EMA, 7 June 2018). During the COMP SLRM in Amsterdam (May 2018), Virginie Hivert presented on Hospital preparations, it was a Joint presentation from EURORDIS/ANSM (FR), prepared by Virginie Hivert and Annie Lorence & colleagues from the French Agency.

### Identification of & support to RD patients participating in Protocol Assistance/Scientific Advice (SAWP - Scientific Advice Working Party)

Last year 41 patients were identified of which 40 were involved. We have reduced the drop-out, by strengthening the communication with the patients (follow-up emails, calls, helping them filling in the DOI, submitting their DOI, etc). Procedures with EMA Patient Relations Team have been strengthened and optimised by holding a monthly conference call following internal meeting between EMA patient relations team and scientific advice team (scientific officers and assistants). During this meeting the need for patient input is discussed and outcomes of the meeting are shared with in the monthly conference call. Discrepancies between scientific officers' input on the need for patient input and EURORDIS views are discussed on a case-by-case basis always reaching an agreed solution.

In summary, the time spent for the whole package per month is about 60 hours per month. This period time usually spreads over one month (from SAWP meeting to the next, more or less).

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 2018 applications responding to the **European Commission's Call for Expression of Interest** were submitted for the **CAT** membership. After an internal Call for Expression of Interest within its Members, EURORDIS has proposed Chris Sotirelis and Michele Lipucci and has issued one letter of endorsement for Mariette Driessens.

Applications responding to the **European Commission's Call for Expression of Interest** were submitted for the **PRAC** membership. EURORDIS decided to answer when the EC relaunched the call for the second time and given the time constraint, no internal call has been performed. EURORDIS Board of Directors has decided to propose Virginie Hivert.

The patients' representatives and staff involved with Scientific Committees and PCWP (Patients and Consumers Working Party) are Members of the EURORDIS' Therapeutic Action Group (TAG). The TAG is a **forum for discussion** composed of EURORDIS and non-EURORDIS patient representatives in the scientific committees and working party at the EMA. The monthly TC allows exchange of information and opinions on various topics pertaining to the activities at the EMA and to the field of regulatory affairs, therapeutic development and patient engagement. All the participants have signed a Confidentiality Agreement with the EMA.



### 3.4.3 Post-marketing authorisation

#### European Medicines Agency

The Patients' and Consumers' Working Party (PCWP), of which EURORDIS is a member, is a unique forum where all scientific committees of the Agency meet with patients and consumers. Two PCWP face-to-face meetings were held in 2018 (17&18 April, 25 September) combined with the Healthcare Professionals' Working Group and one e-meeting (13 December).

EURORDIS worked in particular on: a three-year work plan (joined with the work plan of healthcare professionals); a response to the EMA study supporting the Evaluation of the EMA Fee System; the review of the Annual report on Stakeholder involvement; the preparation of a response to the European Ombudsman inquiry into EMA's pre-submission activities

EURORDIS activities in pharmacovigilance in 2018 included participation in the following:

- + CIOMS working Group in Geneva, 19-20 April, with the initiation of guidelines on Patients Involvement in the Development and Evaluation of Medicines
- + WHO Coordinating Centre on Medicines, 17-18 May, Uppsala
- + Forum 2018 Pharmacovigilance, 30 May 2018, Paris, presenting on the Pharmacovigilant Patient in a Digital Era
- + AESGP Pharmacovigilance committee, 8 October 2018, Paris, presenting on "The pharmacovigilant patient in the digital era: How can we be useful?"
- + International Society of Pharmacovigilance, 11-14 November 2018, Geneva, with a keynote lecture "Patients modulation of risks relating to medicines" at the Bengt Erik Wiholm plenary session and presenting on "Risk communication with patients – clear questions, clear responses?" in another session

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

*The EURORDIS HTA Task Force is a group of EURORDIS volunteers, trained and experienced in HTA-related activities, both at national and at European level. The Task Force aims at sharing experience and knowledge between EURORDIS members and staff about all HTA-related aspects, and at feeding EURORDIS' positions. The objectives are to map HTA systems across Europe, to analyse current policies and practices, and to make proposals for the adequate engagement of patients in HTA. Other objectives of the TF are to raise awareness among the Patients and the HTA communities about the value of patient engagement, and to explore and discuss new methods of cost-effectiveness analysis.*

- + Meetings of the Web-RADR Management Board (composed of FDA, MHRA, EMA, WHO, EFPIA and EURORDIS) to plan the long-term sustainability of the Web-RADR mobile application to report adverse drug reactions

#### Involvement of patients in the benefit/risk evaluation

Since September 2014, the CHMP has decided to invite patients to an oral explanation with the marketing authorisation applicant in some situations where their participation can add to the scientific discussions, first as a pilot, and since 2017 as a more systematic policy. These consultations are now detailed in the EMA Annual Report on Stakeholder Involvement. In 2018, Eurordis was not invited to accompany patients to CHMP explanations with the marketing authorisation applicant, as now most of the patients invited to these meetings know the regulatory process (as having participated to scientific advice or scientific advisory group meetings).

A reflection paper to further improve patient involvement in benefit/risk evaluation has been initiated by Harald Henzmann, CHMP member and now CHMP chair. The PCWP is invited to continue the reflection and EURORDIS will contribute.

To improve the communication and exchange of information between patients and scientific advice, a meeting took place between EURORDIS and the EMA (Scientific Advice Working Party and Stakeholder Engagement Unit) on 10 December, where Community Advisory Boards were discussed.

For the renewal of patients as members of the Pharmacovigilance and Risk Assessment Committee (PRAC), EURORDIS informed members of the DITA task force and the Therapeutic Action Group of the call for candidates by the European Commission

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. EURORDIS launched a call for expression of interest in early 2018, leading to the appointment of 12 members to the HTA Task Force. Of the 12 members, 8 are patients or parents and 4 are staff or volunteers. 10 EU Member States represented.

A first face-to-face meeting of the TF was held on 28<sup>th</sup> November in Paris providing an opportunity for the members to discuss the EURORDIS knowledge base on HTA at EU level and the role of the Task Force



## 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 2018. One on 5 July where the main topics were the EC proposal for a Regulation on HTA Cooperation, the EMA Regulatory Science Strategy 2025, the Second medical use of pharmaceuticals, the EURORDIS programme for Community Advisory Boards. The second meeting was held on 5 December where the main topic was the use of medicines during pregnancy and the information available to women, with the participation of Dr Elisabeth ELEFANT, Reference Centre on Teratogenic Products (CRAT), Armand Trousseau hospital in Paris.

Members of the task force were also consulted on, among other topics: the EMA Patients' and Consumers' Working Party work plan; EMA Regulatory Science Strategy; EMA workshop on European Reference Networks; EMA data anonymisation approach; information they have on medicines shortages in their countries; the investigation of benefit/risks in pregnant women; EURORDIS position on second medical use of medicines / drug repurposing; the EURORDIS statement on the HTA cooperation. In addition, DITA task force members presented EURORDIS views/data in a number of conferences.

**Review of EMA documents for the public or contributions to EMA consultations:** European Public Assessment Reports for the Public now called Medicine Overviews (10), Package Leaflets (5).

Since 2007 when the procedure to review EPAR summaries and PL was established for authorised medicines in the EU, 76 EPAR summaries and 116 PL were reviewed, for a total of 192 documents



# 4. CROSS-CUTTING PRIORITIES

## 4.1 Governance

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### 4.1.1 Annual General Assembly

The EURORDIS General Assembly was held in Vienna on 10 May 2018. EURORDIS full members voted on four vacant positions on the Board of Directors, electing Alexandre Mejat, AFM-Telethon, France; Simona Bellagambi, UNIAMO - Rare Diseases Italy, Italy; Avril Daly, Rare Diseases Ireland, Ireland and Anne-Sophie Lapointe, Alliance Maladies Rares, France.

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.

Anne Sophie Lapointe resigned from the EURORDIS Board of Directors as of 1 October 2018, having taken up a new position with the French Ministry of Social Affairs and Health.

EURORDIS full members also voted on the new EURORDIS Membership Fees. The last increase in fees had taken place in 2010, at the General Assembly in Krakow. Since then, EURORDIS has significantly increased the scope of its activities and services to members, in terms of information, training, networking opportunities and advocacy impact.

The new scale of membership fees is more proportionate to member organisation's budgets, with a more gradient scale for greater equity, while continuing to maintain flexibility for the most vulnerable organisations.



#### 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



##### RVA

Rare Voices Australia



##### CORD

The Canadian Organization for Rare Disorders



##### RPU

Russian Patients' Union



##### JPA

The Japan Patients' Association



##### RADOIR

Rare Diseases Foundation of Iran



### MoU between EURORDIS & RDI

In May 2018, during the annual meeting of Rare Diseases International (RDI) in Vienna, Austria, the members of RDI overwhelmingly voted to legally establish RDI as an independent entity. EURORDIS' commitment to RDI and to rare disease patients around the world remains strong and will continue to grow thanks to a Memorandum of Understanding (MoU) signed by both organisations that sets out how they will work together over the next five years (2018-2022).



EURORDIS also has partnerships with a number of learned societies:

#### EFIM

European Federation of Internal Medicine



#### ESHG

European Society of Human Genetics



#### HOPE

European Hospital & Healthcare Federation



#### ISPOR

International Society for Pharmacoeconomics and Outcomes Research



#### IFSW-Europe

International Federation of Social Workers Europe



In 2018, EURORDIS signed an MoU with the European Connected Health Alliance (EChAlliance) to develop awareness on rare diseases, to promote initiatives aiming at accelerating innovation to improve access to information, quality diagnosis, treatment and multidisciplinary care and research for rare diseases.



## 4.2 Human resources



### 4.2.1 EURORDIS Staff

The team comprised 46 people as at 31 December 2018. 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. A further 9 employees are in the Barcelona office and 5 in Brussels. 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 2018 (in chronological order):

- + **Céline Schwob**, Corporate Relations Manager, joined EURORDIS
- + **Rob Camp**, Patient Engagement Senior Manager - CABs, joined EURORDIS
- + **Mathieu Boudes**, Operations & Projects Manager, left EURORDIS
- + **Inés Hernando**, ERN and Healthcare Director, joined EURORDIS
- + **Jill Bonjean**, Corporate & Foundations Relations Director, left EURORDIS
- + **Gulcin Gumus**, Research & Policy Project Manager, joined EURORDIS
- + **Nancy Hamilton**, Open Academy Manager, left EURORDIS
- + **Jean-Marc Sfeir**, Web Technology Manager, left EURORDIS
- + **Maria Cavaller**, Patient Engagement Junior Manager, joined EURORDIS
- + **Martina Bergna**, Events Junior Manager, joined EURORDIS
- + **Emilie Zingg**, Events Junior Manager, left EURORDIS
- + **Corina Puls**, Office Assistant, joined EURORDIS
- + **Anna Kole**, Public Health Policy Advisor - Rare 2030 Lead, joined EURORDIS
- + **Davor Duboka**, Web Technology Manager, joined EURORDIS
- + **Katarzyna Radwan**, Team Assistant and Brussels Office Manager, joined EURORDIS

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## 4.2.2 EURORDIS Volunteers

In 2018, EURORDIS was privileged to rely on **444 volunteers** including **100 dedicated volunteer patient advocates, 1 office support volunteer, and 343 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.

### Focus on EURORDIS volunteer patient advocates (100):

Since the creation of EURORDIS, these patient advocates have greatly contributed to shaping EU rare disease policies. Most of them are either patients or parents of patients living with rare diseases.

They are selected amongst the EURORDIS’ membership

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

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

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

### EPAC: European Public Affairs Committee

This internal committee plays an active and key role in EURORDIS’ advocacy activities. The EPAC members discuss all relevant advocacy issues for people living with rare diseases and their families. They can also provide their comments on EURORDIS’ positions on some specific issues. The EPAC is governed by Rules of Procedure. As of end 2018, it was composed of 49 members: **19 staff members** (CEO, Directors and managers) involved in advocacy as well as **30 volunteers**. The EPAC members have a mandate to represent EURORDIS.

### TAG: Therapeutic Action Group

The TAG includes **5 EURORDIS volunteers** who are member, alternate or observer on the Scientific Committees of the EMA (CAT, COMP, PDCO). This group is coordinated by 2 staff members.

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

The DITA is composed of **14 volunteers** and coordinated by 2 staff members. They are trained (via the EURORDIS

# 4.3 Finance & Support Services

Finance and support services’ activities in 2018 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.
- + Management of human resources activities, such as recruitment.
- + Management of office support: IT infrastructure, contact database, office supplies.
- + Management of legal and fiscal matters.

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## Contract Grants

### Renewed

- + Specific Grant Agreement (Operating Grant) for year 2018 (SGA FY2018), single beneficiary, DG Sante, 12 months

### Ongoing

- + Advocacy and core activities, AFM-Téléthon, 2014-2017 (renewed for 2018)
- + E-RARE 3: For the extension and strengthening of the transnational cooperation on rare disease research funding organisations, Horizon 2020, 2015-2019

Summer School) and active on issues concerning therapeutic development of medicines for rare diseases as well as access.

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 Santé).

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

Composed of **12 volunteers** and coordinated by 1 staff member, the PAC is acting to inform partners from **RD-Connect, EUrenOmics and NeuroOmics\*** of issues important to patients. The PAC ensures that all project activities have a patient-centric approach.

### Health Technology Assessment (HTA) Task Force

This Task Force was established in 2018. It is composed of **11 volunteers** and coordinated by 2 staff members. The Task Force advises EURORDIS on all aspects regarding Health Technology Assessment policies and procedures. Its role is to inform EURORDIS on how health technologies are assessed at the national level, how patients are involved in these assessments and share views on the future European Cooperation on HTA.

### SPAG: Social Policy Advisory Group

Composed of **11 volunteers** and coordinated by 1 staff member. The SPAG has been established to inform on

rare disease patients' and families' social challenges and to advise on social policy, provision of social care and related issues - such as holistic care, social services, social innovation, disability, special education, psychological support - guaranteeing the formulation of patient-centric approaches to the different social challenges faced by people living with rare diseases.

### ePAGs – EURORDIS volunteers

In the framework of the establishment of European Reference Networks (ERNs) for rare and complex diseases, EURORDIS launched in parallel the establishment of European Patient Advocacy Groups.

European Patient Advocacy Groups' representatives, also called "**ePAGs**", have an official permanent mandate to ensure true and equitable representation of the patient voice by participating in the Board and sub-clinical committees of their respective ERN.

EURORDIS has established a Steering Committee of ePAGs, composed of 2 ePAGs for each of the 24 ERNs. **33 members of this ePAGs Steering Committee have expressed their interest in becoming EURORDIS volunteers.** The ePAGs – EURORDIS volunteers are coordinated by 4 staff members.

They are working towards sharing experiences amongst ePAGs across ERNs and diseases with the objective to further strengthening patient advocates' involvement and raising awareness of ERNs amongst the wider rare disease community.

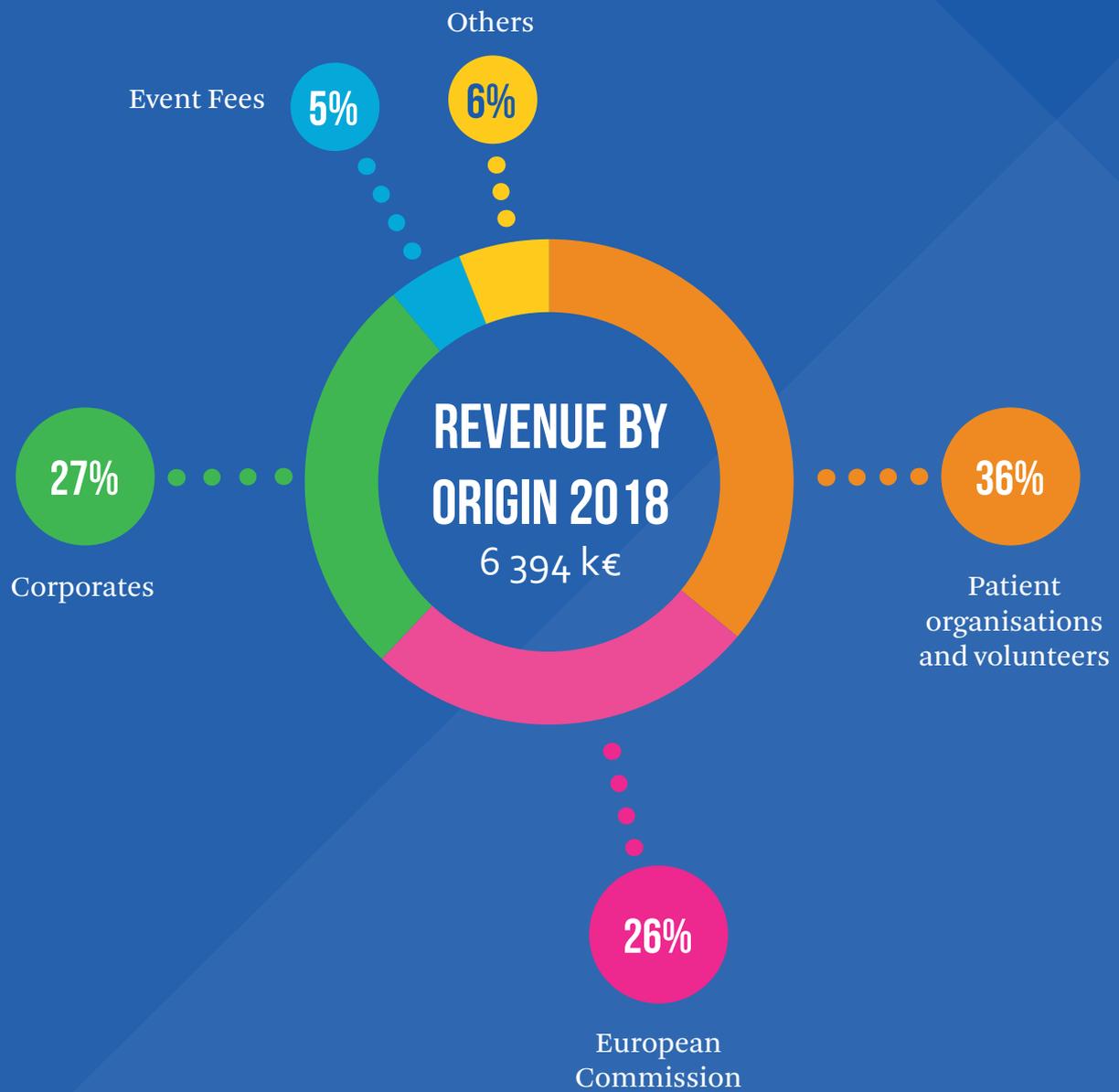
**EURORDIS is extremely grateful to this group of dedicated individuals who offer their time and expertise to improve the lives of people living with a rare disease and their families.**

- + Framework Partnership Agreement 2015-2017 (Operating Grant), single beneficiary, DG Sante, 2018-2021
- + 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

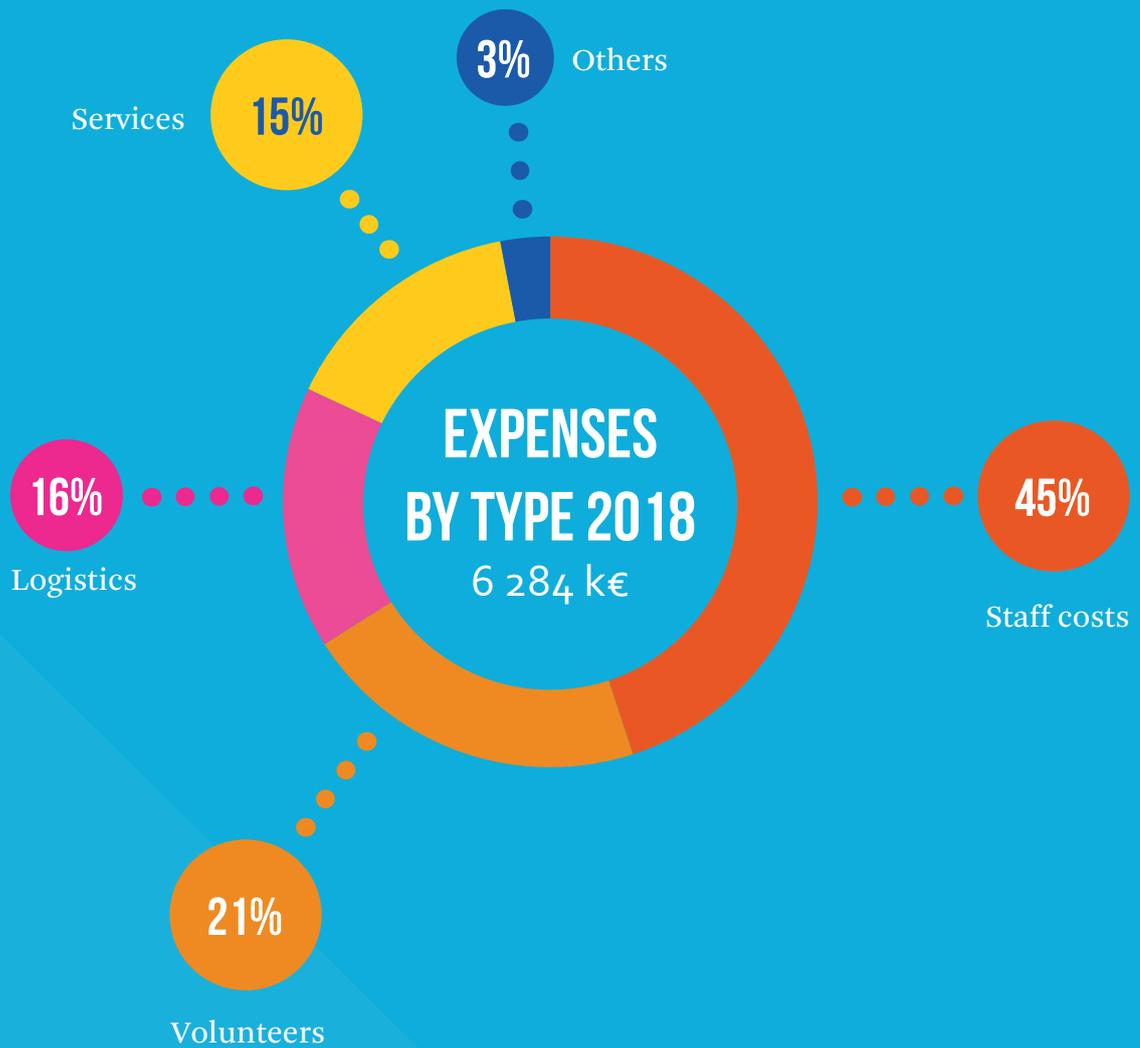
### New

- + PARADIGM: Patients Active in Research and Dialogues for an Improved Generation of Medicines: Advancing meaningful patient engagement in the life cycle of medicines for better health outcomes., beneficiary, IMI-JU2, 2018-2020
- + Solv-RD: Solving the unsolved Rare Diseases, beneficiary, DG Research, 2018-2022
- + c4c: connect4children (COLLABORATIVE NETWORK FOR EUROPEAN CLINICAL TRIALS FOR CHILDREN), beneficiary, IMI-JU2, 2018-2024

# REVENUE 2018



# EXPENSES 2018





# BOARD of Directors

May 2018 - May 2019

## 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** Rare Diseases Ireland  Ireland  
Vice-President

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

**Ms Birthe Byskov Holm** Rare Diseases Denmark  Denmark

**Ms Anne-Sophie Lapointe\*** Vaincre les Maladies Lysosomales  France

**Mr Alexandre Mejat** AFM-Téléthon  France

**Mr Dimitrios Synodinos** Tuberous Sclerosis Association  Greece  
Treasurer

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

**Ms Vlasta Zmazek** Rare Diseases Croatia  Croatia

\*Ms Anne Sophie Lapointe resigned from the EURORDIS Board of Directors as of 1 October 2018, having taken up a new position with the French Ministry of Social Affairs and Health.

# MEMBERS of EURORDIS

FULL MEMBER ..... :  
ASSOCIATE MEMBER ..... :

## ALBANIA

SHOQATA E SEMUNDJEVE TE 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 FÜR TAY-SACHS & PALLIATIVKINDER  
ICA ÖSTERREICH  
NF KINDER – VEREIN ZUR FÖRDERUNG DER  
NEUROFIBROMATOSEFORSCHUNG ÖSTERREICH  
PH AUSTRIA – INITIATIVE LUNGENHOCHDRUCK  
PRO RARE AUSTRIA, ALLIANZ FÜR SELTENEN ERKRANKUNGEN  
PULMONARY HYPERTENSION ASSOCIATION EUROPE  
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  
BE-TSCVZW  
CF EUROPE  
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  
FEDERATION OF EUROPEAN 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  
RADIÖRG - RARE DISEASE ORGANISATION BELGIUM  
RARE DISORDERS BELGIUM  
RELAIS 22 ASBL  
SIOP EUROPE - EUROPEAN SOCIETY FOR PAEDIATRIC ONCOLOGY  
SPIERZIEKTEN VLAANDEREN VZW  
VASCULAR ANOMALY PATIENT ASSOCIATION  
VLAAMS PATIËNTENPLATFORM VZW  
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 ASSOCIATION WILSON DISEASE  
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  
RETINA 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

ASOCIACION COLOMBIANA DE PACIENTES CON ENFERMEDADES DE  
DEPOSITO LISOSOMAL  
FOUNDATION DIANA GARCIA DE OLARTE FOR PID

## 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  
KLUB NEMOCNYCH CYSTICKOU FIBROZOU  
META, ASSOCIATION OF PATIENTS WITH LYSOSOMAL STORAGE  
DISEASES  
NARODNI SDRUZENI PKU A JINYCH DMP (CZECH PKU ASSOCIATION)  
RARE DISEASES CZECH REPUBLIC (CESKA ASCOCIACE PRO VZACNA  
ONEMOCNENI)

## DENMARK

22Q11 DANMARK  
ADDISON FORENINGEN I DANMARK  
BLÆREEKSTROFIFORENINGENS  
CCHS DANMARK  
DANISH APERT SYNDROME ASSOCIATION /DANMARKS  
APERTFORENING  
DANMARKS BLODERFORENING / DANISH HAEMOPHILIA SOCIETY  
DANSK FORENING FOR NEUROFIBRMATOSIS RECKLINGHAUSEN  
EHLERS-DANLOS FOREDANMARKNINGEN I  
FORENINGEN AF MÖBIUSSYNDROM I DANMARK  
FORENINGEN FOR ATAKSI / HSP  
ICHTHYOSIS ASSOCIATION IN DENMARK  
IMMUN DEFJEKT FORENINGEN  
LUPUS EUROPE  
MCADD-FORENINGEN  
MITOKONDRIE-FORENINGEN I DANMARK

PATIENTFORENINGEN HAE DANMARK  
PORFYRIFORENINGEN DANMARK - PORPHYRIA ASSOCIATION  
DENMARK  
RARE DISEASES DENMARK (SJAELDNE DIAGNOSER)  
THE DANISH ASSOCIATION OF VON HIPPEL LINDAU PATIENTS AND  
THEIR RELATIVES  
THE DANISH OSTEOGENESIS IMPERFECTA SOCIETY  
WILSON PATIENTFORENINGEN

## ESTONIA

ESTONIAN PRADER WILLI SYNDROME ASSOCIATION

## FINLAND

ASSOCIATION OF CANCER PATIENTS IN FINLAND (SUOMEN  
SYÖPÄPOTILAAT RY, )  
FINNISH ALLERGY, SKIN AND ASTHMA FEDERATION  
FINNISH ASSOCIATION FOR ULTRA-RARE DISEASES  
FINNISH ASSOCIATION OF PEOPLE WITH PHYSICAL DISABILITIES  
FINNISH NEURO SOCIETY  
HÄRSÖ-RARE DISEASE ALLIANCE FINLAND  
HÄRVINAISET-VERKOSTO - FINNISH NETWORK FOR RARE DISEASES  
INCLUSION FINLAND KVTL  
RINNEKOTI FOUNDATION, NORIO CENTRE  
SUOMEN VASKULIITTIYHDISTYS RY

## FRANCE

AFMKT-FRANCE  
AFM-TÉLÉTHON  
ALLIANCE MALADIES RARES  
ALLIANCE SYNDROME DE DRAVET  
ASS. INTERNATIONALE DE DYSTROPHIE NEURO AXONALE INFANTILE  
ASSEDEA  
ASSOCIATION ISIS  
ASSOCIATION «LES PETITS COURAGEUX»  
ASSOCIATION AMS-ARAMISE  
ASSOCIATION ANÉMIES DYSERYTHROPOÏÉTIQUES 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 SOUFFRANT D'ANGIO OEDÈME PAR  
DÉFICIT EN C<sub>1</sub> 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 DE LA FIEVRE MÉDITERRANÉENNE  
FAMILIALE  
ASSOCIATION FRANÇAISE DE LA MALADIE DE FANCONI  
ASSOCIATION FRANÇAISE DE L'ATAXIE DE FRIEDREICH  
ASSOCIATION FRANÇAISE DE L'OSTEODYSTROPHIE HÉRÉDITAIRE  
D'ALBRIGHT  
ASSOCIATION FRANÇAISE DE L'OSTÉOGENÈSE IMPARFAITE  
ASSOCIATION FRANÇAISE DE NARCOLEPSIE-CATAPLEXIE ET  
HYPERMOMNIE  
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 LOWE  
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 AUPÉTTIT  
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 ICTHYOSE 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 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  
CÛTIS LAXA INTERNATIONALE  
DEBRA FRANCE  
DRAVET SYNDROME EUROPEAN FEDERATION  
ENFANTS DE LA LUNE ASSOCIATION POUR LE XERODERMA  
PIGMENTOSUM  
EUROPEAN FEDERATION FOR HEREDITARY SPASTIC PARAPLEGIA  
EUROPEAN FEDERATION LESCH-NYHAN DISEASE  
EUROPEAN FEDERATION OF ASSOCIATIONS OF PATIENTS WITH  
HAEMOCHROMATOSIS  
FÉDÉRATION DES MALADES DRÉPANOCYTAIRES ET THALASSÉMIQUES  
SOS GLOBI  
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'CEIL  
LES ENFANTS DU JARDIN  
LES PETITS BOURDON  
LIGUE CONTRE LA CARDIOMYOPATHIE  
LIGUE NATIONALE CONTRE LE CANCER  
LUPUS FRANCE  
MOSAÏQUES - ASSOCIATION DES «X FRAGILE»  
NÆVUS GLOBAL  
OÙVRIR 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<sub>1</sub> NETZWERK EV  
 ARVC-SELBSTHILFE E.V.  
 BSHV FÜR KINDER, JUGENDLICHE UND ERWACHSENE MIT SELTENEN, CHRONISCHEN SKELETTERKRANKUNGEN E.V.  
 BUNDESVERBAND DER CLUSTERKOPFSCHMERZEN-SELBSTHILFE-GRUPPEN  
 BUNDESVERBAND FÜR ANGEBORENE GEFÄSSFEHLBILDUNGEN E.V.  
 BUNDESVERBAND KLEINWÜSCHIGE MENSCHEN UND IHRE FAMILIEN E.V.  
 BUNDESVERBAND POLIOMYELITIS EV  
 CHARGE SYNDROM EV  
 CHOLESTERIN & CO EV  
 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.  
 DEUTSCHE UVEITIS-ARBEITSGEMEINSCHAFT EV  
 DEUTSCHSPRACHIGE SELBSTHILFEGRUPPE FÜR ALKAPTONURIE E.V.  
 EHLERS-DANLOS SELBSTHILFE E. V  
 EUROPEAN ASSOCIATION OF PATIENT ORGANISATIONS OF SARCOIDOSIS (EPOS)  
 EUROPEAN CONGENITAL HEART DISEASE ORGANISATION  
 EUROPEAN MEN ALLIANCE  
 EUROPEAN NETWORK FOR ICHTHYOSIS - ENI  
 FAUN STIFTUNG  
 GAUCHER GESELLSCHAFT DEUTSCHLAND E.V.  
 GESELLSCHAFT FÜR MUKOPOLYSACCHARIDosen E.V.  
 HAE VEREINIGUNG E.V. (HEREDITARY ANGIOEDEMA)  
 HOFFNUNGSBAUM E.V.  
 HSP-SELBSTHILFEGRUPPE DEUTSCHLAND E.V.  
 ICA-DEUTSCHLAND E.V. FÖRDERVEREIN INTERSTITIELLE CYSTITIS  
 INTERESSENGEMEINSCHAFT EPIDERMOLYSIS BULLOSA - DEBRA DEUTSCHLAND  
 INTERESSENGEMEINSCHAFT FRAGILES-X E.V.  
 KINDER MIT DELETIONSSYNDROM 22Q11 (KIDS-22Q11) EV  
 KINDER-AUGEN-KREBS-STIFTUNG  
 KINDERNETZWERK E.V  
 KINDNESS FOR KIDS FOUNDATION  
 LEBEN MIT BEHCET - SÜDDEUTSCHLAND  
 LEBEN MIT BEHCET IN DEUTSCHLAND  
 LEONA E.V.  
 MARFAN EUROPE NETWORK  
 MORBUS-OSLER SELBSTHILFE EV  
 MPN-NETZWERK E.V.  
 MPS EUROPE  
 MULTINATIONAL INTERSTITIAL CYSTITIS ASSOCIATION (MICA)  
 MYELITIS E.V.  
 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  
 PEUTZ-JEGHERS-GERMANY E.V.  
 PRO RETINA DEUTSCHLAND E.V.  
 PULMONALE HYPERTONIE E.V.  
 SANFILIPPO INITIATIVE E.V.  
 SELBSTHILFE EPP E.V  
 SELBSTHILFE ICHTHYOSE E.V.  
 SELBSTHILFEGRUPPE EKTODERMALE DYSPLASIE E.V.  
 SELBSTHILFEGRUPPE FÜR PXE - ERKRANKTE DEUTSCHLANDS E.V.  
 SELBSTHILFEGRUPPE GLYKOGENOSE DEUTSCHLAND E.V.  
 SELBSTHILFEORGANISATION FÜR MENSCHEN MIT ANOREKTALFEHLBILDUNGEN  
 SKLERODERMIE SELBSTHILFE E.V.  
 SMA EUROPE  
 SOS-DESMOID E.V.  
 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 MONDSCHIEINKINDER  
 ZNM - ZUSAMMEN STÄRKE 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  
 KARKINAKI AWARENESS FOR CHILDHOOD AND ADOLESCENT CANCER  
 PANHELLENIC ASSOCIATION OF PATIENTS & FRIENDS WITH NEUROFIBROMATOSIS «LIFE WITH NF»  
 PANHELLINIK ASSOCIATION OF PATIENTS WITH LYSOSOMAL DISORDERS  
 PARENTS AND FRIENDS OF PEOPLE WITH RETT SYNDROME ASSOCIATION  
 PRADER WILLI SYNDROME ASSOCIATION HELLAS  
 SOCIETY OF PATIENTS AND FRIENDS OF PATIENTS WITH INHERITED METABOLIC DISEASE  
 TUBEROUS SCLEROSIS ASSOCIATION OF GREECE  
 VHLFA ALLIANCE IN GREECE

## GUATEMALA

ASOCIACIÓN NACIONAL GUATEMALTECA PARA LAS ENFERMEDADES DE DEPÓSITO LISOSOMAL  
 PROCREE

## HONG KONG, SAR OF CHINA

JOSHUA HELLMANN FOUNDATION FOR ORPHAN DISEASE

## HUNGARY

(HUNGARIAN HAEMOPHILIA SOCIETY) MAGYAR HEMOFÍLIA EGYESÜLET  
 MAGYÁORSZÁGI MITOCHONDRÁLIS 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  
 GUDRUN'S RETT SYNDROME RESEARCH TRUST  
 HYPOPARATHYROIDISM EUROPE (HPTH EUROPE)  
 THE ICELANDIC CHILDHOOD CANCER PARENT ORGANIZATION

## INDIA

INDIAN ORGANIZATION FOR RARE DISEASES  
 ORGANIZATION FOR RARE DISEASES INDIA

## IRAN

RARE DISEASE FOUNDATION OF IRAN

## IRELAND

22Q11 IRELAND SUPPORT GROUP  
 ALPHA ONE FOUNDATION  
 BARRETTSTOWN SERIOUS FUN  
 CYSTIC FIBROSIS IRELAND  
 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 PIETRELICINA ONLUS  
 ANGELI NOONAN  
 ASSOCIAZIONE PERSONE CON MALATTIE REUMATICHE - APMAR ONLUS  
 ASSOCIAZIONE NAZIONALE PEMFIGO/PEMFIGOIDE  
 ASSOCIAZIONE CONTO ALLA ROVESCIA  
 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. LOMBARDA)  
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 PER LE IMMUNODEFICIENZE PRIMITIVE ONLUS  
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 CDDP ITALIA ONLUS  
COLLAGENE VI ITALIA ONLUS - COL6  
COSTELLO.CFC - ASSOCIAZIONE ITALIANA SINDROME DI COSTELLO - RADIOFACIOCUTANEA - RASOPATIE - ONLUS  
DEBRA ITALIA ONLUS - ASSOCIAZIONE PER LA RICERCA SULL'EPIDERMOLISI BOLLOSA  
DRAVET ITALIA ONLUS  
FEDERAZIONE SINDROME DI PRADER WILLI ITALIA  
FOP ITALIA ONLUS  
GLI AMICI DI DANIELA  
GRUPPO «ITALIA - GLIOBLASTOMA MULTIFORME - CANCRO AL CERVELLO»  
GRUPPO ITALIANO PER LA LOTTA ALLA SCLERODERMIA ONLUS  
HHT EUROPE  
INCONTINENTIA PIGMENTI ASSOCIAZIONE ITALIANA ONLUS  
LA STRADA PER L'ARCOBALENO  
LEGA ITALIANA SCLEROSI SISTEMICA ONLUS  
LEGA PER LA NEUROFIBROMATOSI 2-ONLUS  
LND FAMIGLIE ITALIANE ONLUS  
P63 EEC SYNDROME INTERNATIONAL NETWORK 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  
WORLD FEDERATION OF INCONTINENCE PATIENTS  
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

ASSOCIATION FOR CHILDREN RARE DISEASES  
COMMUNITY OF ONCOHEMATOLOGICAL PATIENT ORGANISATIONS  
KRAUJAS

## LUXEMBOURG

ALAN - MALADIES RARES LUXEMBOURG  
DEN I ASBL SYNDROME DE MARFAN  
EEN HÄERZ FIR KRIBSKRANK KANNER ASBL  
FONDATIOUN KRIBSKRANK 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

COPIII PLOII

## MONTENEGRO

NATIONAL ORGANISATION FOR RARE DISEASES

## MOROCCO

ASSOCIATION MAROCAINE DE LA FIEVRE MEDITERRANÉENNE FAMILIALE ET DES AUTRES FIEVRES RÉCURRENTES  
SUN'HOP

## NEPAL

GBS/CIDP FOUNDATION NEPAL

## NETHERLANDS

ALS PATIENTS CONNECTED  
AUTOSOMAAL DOMINANT CEREBELLAIRE ATAXIE-VERENIGING NEDERLAND  
BIJNIERVERENIGING NVACP  
CHILDHOOD CANCER INTERNATIONAL  
CMTC-OVM  
CORNELIA DE LANGE SYNDROME WORLD FEDERATION  
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 LIVER PATIENTS ASSOCIATION  
NEUROFIBROMATOSE VERENIGING NEDERLAND  
OSCAR NEDERLAND  
PATIENTENORGANISATIE FIBREUZE DYSPLASIE  
PSC PATIENTS EUROPE  
SARCOIDOSE.NL  
STICHTING AA & PNH CONTACTGROEP  
STICHTING AMYLOIDOSE NEDERLAND SAN  
STICHTING CHRISTIANSON SYNDROME EUROPE  
STICHTING HART, ONDERZOEK / HEART, RESEARCH  
STICHTING HISTIOCYTOSE NEDERLAND  
STICHTING IJZERSTERK  
STICHTING NET-GROEP  
STICHTING SHWACHMAN DIAMOND SYNDROME SUPPORT HOLLAND  
STICHTING STOFWISSELKRACHT  
STICHTING TERRE - RETT SYNDROME FOUNDATION  
STICHTING VOOR AFWEERSTOORNISSEN  
STICHTING ZELDZAME BLOEDZIEKTEN  
THYROID CANCER ALLIANCE  
UNITED PARENT PROJECTS MUSCULAR DYSTROPHY  
VASCULITIS 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  
EUROPEAN HUNTINGTON ASSOCIATION  
FRAMBU - RESOURCE CENTRE FOR RARE DISORDERS  
MORBUS ADDISON ASSOCIATION NORWAY  
MPS-FORENINGEN I 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  
 RETT SYNDROME POLAND - OGÓLNOPOLSKIE STOWARZYSZENIE  
 POMOCY OSOBOM Z ZESPOŁEM RETTA  
 THE DINA RADZIWIŁLOWA 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)  
 RARÍSSIMAS - ASSOCIAÇÃO NACIONAL DE DEFICIÊNCIAS MENTAIS E  
 RARAS

**ROMANIA**

ASOCIATIA COPILUL MEU- INIMA MEA  
 ASOCIATIA PERSOANELOR CU TALASEMIE MAJORA  
 ASOCIATIA ROMÂNĂ DE CANCERE RAR  
 ASOCIATIA ROMÂNĂ SPINA BIFIDA ȘI HIDROCEFALIE  
 ASOCIATIA SMACARE  
 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  
 PATIENT'S 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 OJEDLINELÝCH GENETICKÝCH OCHORENIE

**SLOVENIA**

ASSOCIATION OF PATIENTS WITH BLOOD DISEASES - DRUSTVO  
 BOLNIKOV S KRVNIMI BOLEZNIMI  
 DEBRA SLOVENIA - DRUSTVO DEBRA SLOVENIJA  
 EAMDA - EUROPEAN ALLIANCE OF NEUROMUSCULAR DISORDERS  
 ASSOCIATIONS  
 FABRY PATIENTS ASSOCIATION SLOVENIA / DRUŠTVO BOLNIKOVA  
 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  
 ASOCIACIÓN ANDALUZA CONTRA LA FIBROSIS QUISTICA  
 ASOCIACIÓN ANDALUZA DE PACIENTES CON SÍNDROME DE TOURETTE  
 Y TRASTORNOS ASOCIADOS  
 ASOCIACIÓN ARTROGRIPOSIS MÚLTIPLE CONGÉNITA  
 ASOCIACION CHIARI Y SIRINGOMIELIA DEL PRINCIPADO DE ASTURIAS  
 ASOCIACIÓN DE AFECTADOS DE NEUROFIBROMATOSIS  
 ASOCIACIÓN DE AFECTADOS POR DISPLASIA ECTODÉRMICA  
 ASOCIACIÓN DE AFECTADOS POR HIPERINSULINISMO CONGÉNITO  
 ASOCIACION DE ATROFIA DE NERVIÓ OPTICO DE LEBER

ASOCIACIÓN DE EPIDERMOLISIS BULLOSA DE ESPAÑA (DEBRA  
 ESPAÑA)

ASOCIACIÓN DE ESCLERODERMIA CASTELLON  
 ASOCIACION DE FAMILIARES Y AFECTADOS POR LIPODISTROFIAS  
 ASOCIACION DE FAMILIARES Y ENFERMOS DE FPI Y PFP  
 ASOCIACIÓN DE HEMOGLOBINURIA PAROXÍSTICA NOCTURNA  
 ASOCIACIÓN DE HUESOS DE CRISTAL DE ESPAÑA  
 ASOCIACIÓN DE LAS MUCOPOLISACARIDOSIS Y SÍNDROMES  
 RELACIONADOS  
 ASOCIACIÓN DE NEVUS GIGANTE CONGÉNITO  
 ASOCIACIÓN ESPAÑOLA DEL SÍNDROME CDG, DEFECTOS CONGENITOS  
 DE LA GLICOSILACION  
 ASOCIACIÓN ESPAÑOLA ANIRIDIA  
 ASOCIACIÓN ESPAÑOLA DE AFECTADOS POR SARCOMA  
 ASOCIACIÓN ESPAÑOLA DE ANGIOEDEMA FAMILIAR  
 ASOCIACIÓN ESPAÑOLA DE ENFERMOS DE GLUCOGENOSIS  
 ASOCIACIÓN 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 PARAPARESIA ESPÁSTICA FAMILIAR  
 STRÜMPPELL-LORRAIN  
 ASOCIACIÓN ESPAÑOLA DE PORFIRIA  
 ASOCIACIÓN ESPAÑOLA DE RAQUITISMOS Y OSTEOMALACIA  
 HEREDADOS  
 ASOCIACIÓN ESPAÑOLA DE SÍNDROME DE POLAND  
 ASOCIACIÓN ESPAÑOLA DEL SÍNDROME DE JOUBERT  
 ASOCIACIÓN ESPAÑOLA SÍNDROME DE SJÖGREN  
 ASOCIACIÓN HHT ESPAÑA  
 ASOCIACIÓN 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  
 ASOCIACION STOP SANFILIPPO  
 ASOCIACIÓN XERODERMA PIGMENTOSUM  
 ASSOCIACIÓ CATALANA DE LA DELECCIÓ 22Q  
 ASSOCIACIÓ CATALANA DE SÍNDROME DE SJÖGREN  
 ASSOCIACIÓ D'APRÀXIA OCULAR I MALALTIES ASSOCIADES  
 ASSOCIACIÓN CATALANA DE ENFERMEDADES NEUROMUSCULARES  
 DE NEU - ASOCIACIÓN DE ENFERMEDADES DE LOS  
 NEUROTRANSMISORES  
 D'GENES ASOCIACIÓN DE ENFERMEDADES RARAS DE MURCIA  
 DRÄVET 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 MINORITÄRIES  
 FEDERACIÓN DE ASOCIACIONES DE RETINOSIS PIGMENTARIA DE  
 ESPAÑA  
 FEDERACIÓN DE ATAXIAS DE ESPAÑA  
 FEDERACIÓN ESPAÑOLA DE ENFERMEDADES NEUROMUSCULARES  
 FEDERACIÓN ESPAÑOLA DE FIBROSIS QUISTICA  
 FEDERACIÓN ESPAÑOLA DE HEMOFILIA  
 FEDERACIÓN ESPAÑOLA DE MALFORMACIÓN DE CHIARI Y PATOLOGÍAS  
 ASOCIADAS  
 FEDERACION ESPANOLA DE PADRES CON NINOS CON CANCER  
 FEDERACIÓN ESPAÑOLA DEL SÍNDROME X FRÁGIL  
 FUNDACIÓN ANDRÉS MARCIO, NIÑOS CONTRA LA LAMINOPATÍA  
 FUNDACION MENUDOS CORAZONES  
 FUNDACIÓN NIEMANN-PICK DE ESPAÑA  
 FUNDACIÓN SÍNDROME 5P MENOS  
 FUNDACIÓN SÍNDROME DE MOEBIUS  
 FUNDACION SINDROME DE WEST  
 FUNDACIÓN SÍNDROME WOLF HIRSCHHORN 4P-  
 HIPERTENSIÓN PULMONAR ESPAÑA ORGANIZACION DE PACIENTES -  
 HPE-ORG  
 INSTITUTO DE INVESTIGACIÓN Y DESARROLLO SOCIAL DE  
 ENFERMEDADES POCO FRECUENTES  
 MIQUÈL VALLS FOUNDATION (FUNDACION PRIVADA CATALANA DE  
 ESCLEROSI LATERAL AMIOTROFICA)

**SWEDEN**

AGRENSKA  
 AORTA DISSEKTION FÖRENINGEN SKANDINAVIEN  
 EUROPEAN DYSMELIA REFERENCE INFORMATION CENTRE  
 NEUROFÖRBUNDET / SWEDISH ASSOCIATION OF PERSONS WITH  
 NEUROLOGICAL DISABILITIES  
 PRÄDER 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  
THYROID FEDERATION INTERNATIONAL  
WILHELM FOUNDATION

## SWITZERLAND

ASSOCIATION ENFANCE ET MALADIES ORPHELINES  
ASSOCIATION SUISSE ROMANDE INTERVENANT CONTRE LES  
MALADIES NEURO-MUSCULAIRES  
BLACKSWAN FOUNDATION  
ESPERARE  
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  
MARAVAL - MALADIES RARES VALAIS - SELTENE KRANKHEITEN WALLIS  
MARFAN FOUNDATION SWITZERLAND (MARFAN STIFTUNG SCHWEIZ)  
PRADER WILLI SYNDROM VEREINIGUNG SCHWEIZ  
PRORARIS  
RETINA INTERNATIONAL  
SCHWEIZERISCHE GESELLSCHAFT FÜR FAMILIÄRE FORMEN DER  
HYPERCHOLESTERINAMIE  
SCHWEIZERISCHE GESELLSCHAFT FÜR PORPHYRIE  
SELBSTHILFE ICHTHYOSE SCHWEIZ  
SMA SCHWEIZ

## TAIWAN, PROVINCE OF CHINA

TAIWAN FOUNDATION FOR RARE DISORDERS

## THE FORMER YUGOSLAV REPUBLIC OF MACEDONIA

ASSOCIATION FOR HELP AND SUPPORT OF PATIENTS AND THEIR  
CAREGIVERS WITH HAEMATOLOGICAL DISEASES  
ASSOCIATION FOR PERSONS WITH SPINAL MUSCULAR ATROPHY- STOP  
SMA  
LIFE WITH CHALLENGES  
NATIONAL ALLIANCE FOR RARE DISEASES OF R. MACEDONIA  
SAVE LIVER ASSOCIATION OF PATIENTS WITH LIVER DISEASES  
ZDRUŽENIE ZA CISTIČNA FIBROZA (MACEDONIAN CF ASSOCIATION)

## TURKEY

KIFDER  
MÜKÖPOLISAKKARİDOZ VE BENZERİ LİZOZOMAL DEPO HASTALIKLARI  
DERNERGİ  
YUZÜMLÜ MUTLUYUM DERNEĞİ

## 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  
UKRAINIAN PARENT PROJECT «MIO-LIFE»  
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

AMYLOIDOSIS RESEARCH CONSORTIUM 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

BEHCET'S UK

BRITTLE BONE SOCIETY

CAMBRIDGE RARE DISEASE NETWORK (CRDN)

CANCER 52

CAVERNOMA ALLIANCE UK

CHILD GROWTH FOUNDATION

CHILD LUNG FOUNDATION

CHILDREN'S LIVER DISEASE FOUNDATION

CILIOPATHY ALLIANCE

CONTACT A FAMILY

CYSTINOSIS FOUNDATION OF THE UK

## DANCING EYE SYNDROME SUPPORT TRUST

DEGOS DISEASE SUPPORT NETWORK

ECTODERMAL DYSPLASIA SOCIETY

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 GAUCHER 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

METABOLIC SUPPORT UK

MPS SOCIETY

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

PTEN RESEARCH FOUNDATION

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 AARSKOG FOUNDATION

THE CHROMOSOME 18 REGISTRY AND RESEARCH SOCIETY (EUROPE)

THE CURE & ACTION FOR TAY-SACHS (CATS) FOUNDATION

THE EHLERS-DANLOS SOCIETY

THE FRAGILE X SOCIETY

THE MADDI FOUNDATION

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

ALAGILLE SYNDROME ALLIANCE

ALSTROM SYNDROME INTERNATIONAL

APS FOUNDATION OF AMERICA, INC (APSFA)

CHILDREN'S TUMOR FOUNDATION

CHORDOMA FOUNDATION

CURE PSP

CYSTINOSIS FOUNDATION

FMD CHAT

INTERNATIONAL FOXG1 FOUNDATION

INTERNATIONAL PEMPHIGUS & PEMPHIGOID FOUNDATION

INTERNATIONAL WALDENSTROM'S MACROGLOBULINEMIA FOUNDATION

MCT8-AHDS FOUNDATION INC.

NEUROMUSCULAR DISEASE 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

## VENEZUELA

FUNDACIÓN FURA



# PARTICIPATION OF EURORDIS' REPRESENTATIVES IN PUBLIC EUROPEAN / INTERNATIONAL CONFERENCES & Workshops 2018

**4<sup>th</sup> Conference on European Reference Networks: "ERNs in action", Brussels, Belgium, 21-22 November**

**Yann Le Cam:** The role and participation of patients representatives in the networks

**Matt Johnson:** ERNs' Main challenges for the future

**CARE 2018: "Equal access to diagnosis for all rare disease patients", under the auspices of the President of the French Republic, Paris, France, 12 November**

**Yann Le Cam:** L'accès au diagnostic dans les maladies rares, état des lieux européen et perspectives ?

**Virginie Bros-Facer:** Impasse diagnostique dans les maladies rares : Etats des lieux Européens

**National Conference on Rare Diseases, Helsinki, Finland, 22 October**

**Raquel Castro:** Why patient-centred care and patient engagement are essential: experiences from the EU-funded project INNOVCare

**Autumn School of the French National Alliance for Rare Diseases, Paris, France, 4 October**

**Raquel Castro:** « Les défis en terme d'accès aux services sociaux pour les personnes atteintes de maladie rare et leurs aidants & INNOVCare Project »

**Ariane Weinman:** Presentation of the European Reference Networks

**2018 Golden Helix Summer School, Syros, Greece, 29 September**

**Dimitris Synodinos:** "Rare Disease Patient Organisations and Healthcare Stakeholders: The experience of Eurordis"

**CHAM 2018: "Building the European Health System", Chamonix, France, 28-29 September**

**Terkel Andersen,** panellist in "Patients, towards transnational healthcare"

**20<sup>th</sup> Anniversary of ALAN – Rare Diseases Luxembourg, Mamer, Luxembourg, 26 September**

**Yann Le Cam:** "Enjeux locaux & Perspectives Européennes"

**Valentina Bottarelli & Ariane Weinman** also represented EURORDIS

**Rare Disease Briefing – session with Irish politicians, Dublin, Ireland, 25 September**

**Avril Daly:** Presentation on Rare Diseases

**MEPs Against Cancers (MAC): "Celebrating 20 Years of Progress in Paediatric Haemato-Oncology in Europe", European Parliament, Brussels, Belgium, 25 September**

**Ariane Weinman** represented EURORDIS

**"Rare Cancers Europe: 10 years of cooperation to improve care for cancer patients", European Parliament, Brussels, Belgium, 25 September**

**Ariane Weinman** represented EURORDIS

**5<sup>th</sup> Croatian National Conference and Symposium on Rare Diseases, Zagreb, Croatia, 21-22 September**

**Simone Boselli:** "Drugs availability, situation in Europe"

**Matt Johnson:** "European Reference Networks"

**47<sup>th</sup> Annual General Assembly of EAMDA (European Alliance of Neuromuscular Disorders Association), Prague, Czech Republic, 20-23 September**

**Alexandre Mejat:** "Presentation of EURORDIS"

**13<sup>th</sup> International Conference on Rare Diseases and Orphan Drugs (ICORD) Sweden, 20 September**

**Yann Le Cam:** "Unmet Needs Today in Rare Diseases"

**Paloma Tejada:** "The global alliance of rare disease patients"

**6<sup>th</sup> International Summer School -Rare Disease & Orphan Drug Registries - Istituto Superiore di Sanità - Rome, Italy, 10-14, September**

**Virginie Bros-Facer:** "Patients Registry in European Reference Networks: Roles of European Patient Advisory Groups"

**EUROSCIENCE OPEN FORUM – ESOE 2018: "Sharing Science: Towards New Horizons", Toulouse, France, 11 July**

**Virginie Bros-Facer:** "Life Beyond Academia" & "The values of biomedical innovation: the patient perspective"

**RIME Alliance Maladies Rares : Recherche et innovations thérapeutiques dans les maladies rares - Etats des lieux et perspectives, Paris, France, 27 June**

**Yann Le Cam:** "La Recherche dans les maladies rares, dynamiques nationales et européennes"

**Forum French Rare Diseases National Alliance, Nancy, France, 14 June**

**Virginie Bros-Facer:** "Dynamiques recherche maladies rares en Europe" (Rare Diseases research in Europe)

**"Heart Failure Patient Organisations Academy", Croi West of Ireland Cardiac Foundation, Galway, Ireland, 26-27 June**

**Avril Daly:** "Generating the Evidence of Disease Impact: The Rare Disease Example"

**2018 Work Forum on the implementation of the UN Convention on the Rights of Persons with Disabilities in the EU and its Member States (organised by EC – DG EMPL - Employment, Social Affairs and Inclusion), Brussels, Belgium, 29 May**

**Raquel Castro:** "Perspectives of people living with a rare disease & disabilities"

**Citizens Forum: Presentation of patient organisations' projects, INFARMED, Lisbon, Portugal, 22 May**

**Lesley Greene:** "Living With an Orphan Disease - Ordinary People Achieving Extraordinary Things"

**Rare Diseases Conference, Tirana, Albania, 4 May**

**Vlasta Zmazek:** "The role EURORDIS and rare disease patient organisations"

**Rare Disease Symposium, University College Dublin, Dublin, Ireland, 3 May**

**Raquel Castro:** "Social challenges of people living with a rare disease and their carers & INNOVCare Project - Innovative Patient-Centred Approach for Social Care Provision"

**World Orphan Drugs Congress, Washington D.C., USA, 27 April**

**Jill Bonjean:** "Scaling up rare disease patient engagement in Europe"

**MEDEV Piperska meeting, Athens, Greece, 24 April**

**Simone Boselli:** "Access and Affordability"

**MPE (Myeloma Patients Europe) Annual General Meeting 2018, Brussels, Belgium, 21 April**

**Simone Boselli:** "Access and Affordability"

**"LE MALATTIE RARE IN ITALIA: Epidemiologia, spesa sanitaria, assistenza, accesso all'innovazione" (RARE DISEASES IN ITALY: epidemiology, health expenditure, care, access to innovation), Ministry of Health, Rome, Italy, 19 April**

**Simona Bellagambi,** moderator of the Round Table: "Taking care of the patient with rare disease on the territory- local level"

**RD-Connect Annual Meeting, Athens, Greece, 16-18 April**

**Virginie Bros-Facer:** "Patient engagement throughout RD-Connect via the patient advisory council"

**9<sup>th</sup> Childhood Cancer International-Europe Regional Conference, Lisbon, Portugal, 13-14 April**

**Ariane Weinman:** "Training and Patient Empowerment"

**EUCOPE meeting, Brussels, Belgium, 20 March**

**Simone Boselli:** "Breaking the Access Deadlock: Looking for a "new deal?"

**EMA/EC multi-stakeholder workshop to further improve the implementation of the paediatric regulation, London, UK, 20 March**

**Virginie Hivert:** "Patients' perspective on identification of paediatric medical needs – Methodology"

**RaDiOrg.be (Rare Diseases – Belgium) workshop, Brussels, Belgium, 10 March**

**Matt Bolz-Johnson and Ines Hernando:** "ERN New Central European Infrastructure"

**Annual Conference on EU Law in the Pharmaceutical Sector 2018, Brussels, Belgium, 1 March**

**Simone Boselli:** "Should the EU legal framework on incentives for innovation be revised? A patient perspective"

**Rare Disease Symposium, University College Dublin, Dublin, Ireland, 1 March**

**Avril Daly:** "Innovation in Research for Rare Disease"

**Rare Disease Day Event, Aliança Portuguesa de Associações das Doenças Raras, Lisbon, Portugal, 27 February**

**Anja Helm:** "National Alliances for Rare Diseases in Europe"

**Workshop on Rare Diseases Clinical Research – organised by the Irish National Clinical Programme for Rare Disease, Dublin, Ireland, 23 February**

**Avril Daly:** "Innovation in Research for Rare Disease"

**Ethical Review for Biobank based research projects: towards a risk based ethical review (BBMRI –ERIC / EURORDIS / RD-Connect), Paris, France, 20 February**

**Virginie Bros-Facer:** "Patient's perspective on Ethical review for secondary uses"

**European Patients' Forum (EPF) Educational Workshop on Pharmaceutical Incentives, Brussels, Belgium, 19 February**

**Simone Boselli:** The Patient's perspective

**Solve-RD Kick-off Meeting, Tübingen, Germany, 29-30 January**

**Virginie Bros-Facer** presented EURORDIS' involvement in endeavours for undiagnosed patients

**BBMRI-ERIC Stakeholder Forum, Brussels, Belgium, 25 January**

**Virginie Bros-Facer** represented EURORDIS

# ACKNOWLEDGEMENTS

EURORDIS would like to thank the following organisations and companies for their financial support in 2018:

## 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 2018
- RD-ACTION – Rare Diseases Joint Action – Data and policies for Rare Diseases
- JARC – Joint Action on Rare Cancers



Co-funded by the Health Programme of the European Union

### EUROPEAN COMMISSION

DG Research and Innovation

- RD-Connect Project to improve connections among different clinical datasets such as registries, biobanks and clinical bioinformatics for rare disease research
- The European Clinical Research Infrastructures Network Integrated Activity (ECRIN-IA) project, which partners with and supports the EURORDIS Summer School
- The Solve-RD Project -Solving the Unsolved Rare Diseases
- The Innovative Medicines Initiative-Joint Undertaking (IMI-JU) projects:
  - PARADIGM, Patients Active in Research and Dialogues for an Improved Generation of Medicines: Advancing meaningful patient engagement in the life cycle of medicines for better health outcomes
  - conect4children, a Collaborative Network for European Clinical Trials For Children



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 corporates who support us. EURORDIS had 71 different health sector corporate donors in 2018. Companies have supported EURORDIS through the EURORDIS Round Table of Companies<sup>1</sup>, the European Conference for Rare Diseases and Orphan Products 2018 Vienna<sup>2</sup>, the EURORDIS Black Pearl Awards<sup>3</sup>, as well as international initiatives such as Rare Disease Day<sup>4</sup>, Rare Barometer<sup>5</sup>, RareConnect<sup>6</sup>, Rare Diseases International<sup>7</sup>, EURORDIS Open Academy<sup>8</sup>, NGO Committee for Rare Diseases<sup>9</sup>, and multi-lingual communications. 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 PFIZER



2 SHIRE BELGIUM  
BVBA



3 CELGENE B.V.



4 CSL BEHRING



5 NOVARTIS



5

GSK



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

<sup>2</sup> <https://www.rare-diseases.eu>

<sup>3</sup> <https://blackpearl.eurordis.org>

<sup>4</sup> <https://www.rare-diseaseday.org>

<sup>5</sup> <http://www.eurordis.org/voices>

<sup>6</sup> <https://www.rareconnect.org/en>

<sup>7</sup> <http://www.rare-diseases-international.org>

<sup>8</sup> <https://openacademy.eurordis.org>

<sup>9</sup> <https://www.ngocommitteerare-diseases.org>

Other Pharmaceutical & Biotechnology Companies & Health Sector Corporates



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## Other corporates

BURSON-MARSTELLER			DLA PIPER UK LLP
GOOGLE			MEDIAPLANET
PUBLICIS LIFE BRANDS			TERRAPINN
THE FINANCIAL TIMES			

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## Other contributors

ASSOCIATION OF INTERNATIONAL PHARMACEUTICAL MANUFACTURERS			BERTARELLI FOUNDATION
FOUNDATION DONAU-STIFTUNG			
KINDNESS FOR KIDS	 <p data-bbox="651 1435 994 1480">Foundation for children with rare diseases Moving mountains for kids in need.</p>		

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## Special Mention

EURORDIS would like to thank Luca Dotti for supporting the Remembering Audrey event that took place in London in October. The net proceeds of the event were donated to EURORDIS and Lymphoma Action via Square Rock.





**EURORDIS**  
RARE DISEASES EUROPE

# WORKPLAN

2019

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# ACTION PLAN

## 2019

## 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 2010-2027** that will fund EU policies and programmes.
- + Organise the Pledge for Rare campaign leading up to the European Parliamentary elections. Re-establish the Parliamentary Advocates for Rare Diseases network, inviting new MEPs to join.
- + Execute the **Rare 2030 Foresight Study on Rare Disease Policy**, to identify long term policy scenarios with participatory methodologies. Establish the Panel of Experts and knowledge base required to identify future trends and drivers; propose future scenarios based on different prioritizations of drivers.

### 1.3 PROMOTE THE SUSTAINABILITY OF RARE DISEASES AS A POLICY AND BUDGET PRIORITY IN THE EU PROGRAMMES FOR THE PERIOD 2021-2027:

- + Promote of RDs as a research priority in Horizon Europe 2021-2027, including expanded support to the European Joint Programme co-fund for research on rare diseases, clinical research networks for RDs and a possible dedicated Mission RDs;
- + Promote the needs of people with rare diseases as a priority in IMI 3 2021-2027
- + Promote RDs within Health strategies across EU programmes 2021-2027; public health, social and employment, digital, European social funds, European structural and regional funds

## 1.4 ADVOCATE FOR 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
- + Promote integration of ERNs into national healthcare systems in collaboration with National Alliances (NAs) by continuing to support NAs in the EU member states through local face-to-face meetings and the CNA Working Group of National Alliances in EU MS with small populations to identify specific needs, challenges and ad hoc strategy

## 1.5 ADVOCATE FOR HEALTH TECHNOLOGY ASSESSMENT METHODS AND A EUROPEAN COLLABORATION ON HTA

- + Advocate for HTA methods and European collaboration on HTA in coordination with National Alliances on EU legislation

## 1.6 ADVOCATE TO IMPROVE ACCESS TO RARE DISEASE THERAPIES

- + Organise the 3<sup>rd</sup> EURORDIS Multi-Stakeholder Symposium on Improving Patients' Access to Rare Disease Therapies, 13-14 February 2019 "Let's make a pact to ensure patients' sustainable access to rare disease therapies" including the development of a roadmap for the future
- + Lead the RARE-IMPACT initiative for the improvement of patient access to gene and cell therapies for rare diseases

## 1.7 VOICING THE SOCIAL NEEDS OF PEOPLE WITH A RARE DISEASE AND ADVOCATING FOR INTEGRATED HEALTH AND SOCIAL CARE

- + Organise the campaign for Rare Disease Day 2019 on 28 February on "Bridging Health and Social Care"
- + Launch the major EURORDIS position paper on Holistic Care, developed in collaboration with the National Alliances, European Federations and EURORDIS members, at the EMM 2019 Bucharest as well as subsequent dissemination to all relevant RD and social stakeholders and presentation at conferences
- + Continue to act on social policy and disability at the European level

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



### Rare Diseases International:

- + Support RDI as an established independent entity from January 2019. Provide operational and financial support as defined in the MoU signed between EURORDIS and RDI
- + Organise RDI annual meeting in New York back to back with the NGO Committee RDD Policy event
- + Develop a Collaborative Framework on Rare Diseases with the WHO

### NGO Committee for Rare Diseases:

- + Organise the 2<sup>nd</sup> Committee event at the UN HQ in New York on 21 Feb 2019 as a RDD policy event co-organised by EURORDIS, RDI and Agrenska.
- + Take first steps towards establishing a UN General Assembly Resolution on Rare Diseases
- + Establish further relationships with country permanent missions in New York and Geneva as well as relevant departments and agencies of UN
- + Contribute to relevant reports in the HR council, in the convention of disability and ECOSOC

### Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease:

- + Launch of Year 1 report events in New York and Brussels on 20 February
- + Explore the possibility of launching a 2<sup>nd</sup> Global Commission

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

- + Continue the Rare Barometer growth internationally. 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).
- + 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
- + Organise the EURORDIS Membership Meeting 2019 Bucharest

### 2.1.2 Capacity building of European Networks

- + Organise two meetings of the Council of National Alliances (CNA); work more closely with National Alliances on certain key activities through the CNA and CNA working groups
- + Organise one meeting of the Council of European Federations (CEF); continue the EURORDIS programme "Support to European Rare Disease Federations" and work more closely with European Federations on key items
- + Organise one meeting of the EU Network of Rare Disease Help Lines and work on materials to increase the network's visibility/awareness on cross-border care

### 2.1.3 RareConnect

- + Consolidate EURORDIS' new role at the heart of the new RareConnect governance structure
- + Explore new funding opportunities to ensure the long-term sustainability of the platform
- + Grow the use of the platform as a driver of research studies

## 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
- + Begin **development of a new eurordis.org website** with a user driven design and updated content in 7 languages
- + Consolidate EURORDIS webinar programme used within different EURORDIS activities.

### 2.2.2 EURORDIS Open Academy

- + Adopt and implement a cohesive strategy and assess feasibility of additional content
- + Continue to **improve and contribute to the online training resources that are** freely available via the EURORDIS



Website to anyone interested in building their capacities independently

- + Continue to build a relationship with alumni and provide further capacity building for them
- + Organise the **2019 Winter School on Scientific Innovation and Translational Research** in Paris in March 2019
- + Organise the **2019 Summer School on Medicines Research & Development** in Barcelona in June 2019
- + Develop the **EURORDIS Digital School** as part of the Open Academy for launch at the end of 2019
- + Develop the **EURORDIS Leadership School** as a capacity-building programme for ePAG patient advocates

## 2.3 RAISING AWARENESS & INFORMING

### 2.3.1 Rare Disease Day 2019



- + Organise Rare Disease Day 2019 in over 90 countries, continue the #ShowYourRare campaign
- + Organise a Rare Disease Day Policy Event at the NY UN Headquarters
- + **Complete the strategic review of RDD** with the purpose of creating further positive change for people living with rare diseases in the next decade

## 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 patient organisations, promote a meaningful patient engagement in ERN activities & provide regular support to ePAGs
- + **Revise the EPAG Constitution and Rules of Procedure** and support their implementation
- + Contribute to the promotion of **common methods to capture feedback on patient satisfaction & patient experience across the ERNs**
- + Deliver a comprehensive capacity-building programme (Leadership School) for ePAG patient advocates; organize a peer learning face-to-face meeting to share best practices

### 3.2 PATIENT ENGAGEMENT IN LIFECYCLE DEVELOPMENT

#### 3.2.1 Support patients creating their Community Advisory Boards (CABs) to engage with Industry (EUROCAB programme)

- + Flesh out **guidelines on how to organise and operate a CAB**
- + Provide key templates needed to start discussions with industry or other sponsors
- + Maintain a **mentoring programme for CAB members**, alongside ad hoc guidance to help preparing meeting
- + Provide metrics to support the evaluation of the impact of the CABs
- + Develop a **charter for collaboration** on research where companies make commitments to work with patients
- + Develop a sustainability programme for the CABs

#### 3.2.2 Support 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
- + Ensure that EURORDIS patient engagement practices and insight contributes to the workstream that's designing patient engagement tools
- + Support the project's efforts to build consensus from all stakeholders on the value and methods for patient engagement
- + **Successfully lead our work package on co-designing a sustainability model for patient engagement that demonstrates the 'return on the engagement' for all players**

### 3.3 PATIENT ENGAGEMENT IN THERAPEUTIC DEVELOPMENT

#### 3.3.1 Support Patient involvement in EMA activities

- + Participate in the **EMA Committee for Orphan Medicinal Products (COMP)**
- + Participate in the **EMA Pharmacovigilance and Risk Assessment Committee (PRAC)**
- + 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.

#### 3.3.1 Support patient involvement in European HTA Network, EUnetHTA JA 3 and related HTA activities

- + Represent patient organisations in the European HTA Network
- + Launch the work of the **EURORDIS HTA Task Force with a mandate to advise EURORDIS on all aspects regarding Health Technology Assessment policies and procedures**; one face-to-face meeting planned for 2019
- + Identify and mentor patients participating in HTA Joint Procedures

#### 3.3.1 Support patient involvement in quality information on medicines through the EURORDIS Drug Information Transparency & Access Task Force (DITA TF)

- + Organise one face-to-face meeting with regular e-meetings throughout the year

## 3.4 PATIENT ENGAGEMENT IN RESEARCH

### 3.4.1 Continue participation in the International Rare Disease Research Consortium (IRDiRC)

- + Take part in all relevant meetings and activities. EURORDIS is a member of the Consortium Assembly, Patient Advocacy Constituent Committee, Operating Committee, vice-chair of the Therapies Scientific Committee and involved in several Task Forces.



### 3.4.2 Take an active part in the European Joint Programme on Rare Diseases (EJP)



- + Participate in the Operations and strategic development of the EJP within the Operating and Executive Committee
- + Coordinate all training activities within Pillar 3
- + Develop guidance & guidelines to support meaningful patient involvement within the joint transnational calls through the establishment of a Working Group involving ePAGs and Winter School Alumni
- + Develop metrics to measure impact of patient involvement in research projects
- + Further develop the Winter School for patient representatives on scientific innovation and translational research by adding contents on the online pre-training specifically on basic concepts of genetics and working with a multi-stakeholder Programme Committee to improve contents and delivery of the F2F training sessions.

### 3.4.3 Participate in the collaborative network for European clinical trials for children (Conect4Children - c4c)



- + Development of education and training for clinical researchers & for children, young patients and parents

### 3.4.4 Continue coordinating the Community Engagement Task Force (Solve-RD)



- + Ensure that development of the activities within Solve-RD are patient-centered
- + Develop a toolkit to support follow up of genetic counselling for patient organisations and genetic counsellors.

### 3.4.5 Continue to represent the voice of RD patients in several networks and initiatives:

- + BBMRI Stakeholder forum meeting
- + Go FAIR RD Network
- + Undiagnosed Diseases Network International

## 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, EURORDIS Rare Barometer, major advocacy campaigns and new projects
- + Continue developing and collecting EURORDIS Indicators
- + Start the planning for the review and drafting of EURORDIS Strategy for the next five years (2021-2026)

#### 4.1.2 Finalise the EURORDIS By-laws

- + Consolidate all of EURORDIS internal governance documents into the EURORDIS by-laws, for presentation at the AGA 2019 Bucharest

#### 4.1.3 Strategic Partnerships (MoUs)

- + Maintain partnerships with international organisations and review and renew MoUs as needed, EURORDIS has signed MoUs with the following organisations: NORD (USA), CORD (Canada), the Japanese Patients Association - JPA (Japan), Rare Voices Australia (RVA), the Russian Patients Union (RPU) Chinese Organisation for Rare Diseases, in addition to being open to new partnerships with other international patient organisations
- + Develop strategic partnership with ORPHANET

### 4.2 RESOURCE DEVELOPMENT

- + Maintain activity to support current contributions from the health sector within the confines of the EURORDIS Policy of Relationship with Commercial Companies, EMA Policy on Prevention of Conflict of Interest and CHAFAEA rules
- + Pursue opportunities to deliver new sources of diversified income
- + Appoint a **Development Relations Senior Manager to take forward work with foundations**

### 4.3 EURORDIS EVENTS

- + Organise the **EURORDIS Black Pearl Awards** in February 2019 in Brussels
- + Plan the **10<sup>th</sup> European Conference on Rare Diseases & Orphan Products – ECRD 2020 Stockholm** (May 2020); carry out a **structured review of ECRD to reinforce the event's utility/the impact and better leverage external opportunities**

### 4.4 HUMAN RESOURCES

#### 4.4.1. Enhance the decentralised structure of EURORDIS and maintain and improve HR processes:

- + Expansion of EURORDIS offices in Brussels
- + Exploration of new management team structure with an improved internal coordination meeting structure
- + Maintain quality of video conferences and tool for remote working
- + Create and maintain a **new EURORDIS contact database**

#### 4.4.2 EURORDIS Volunteers

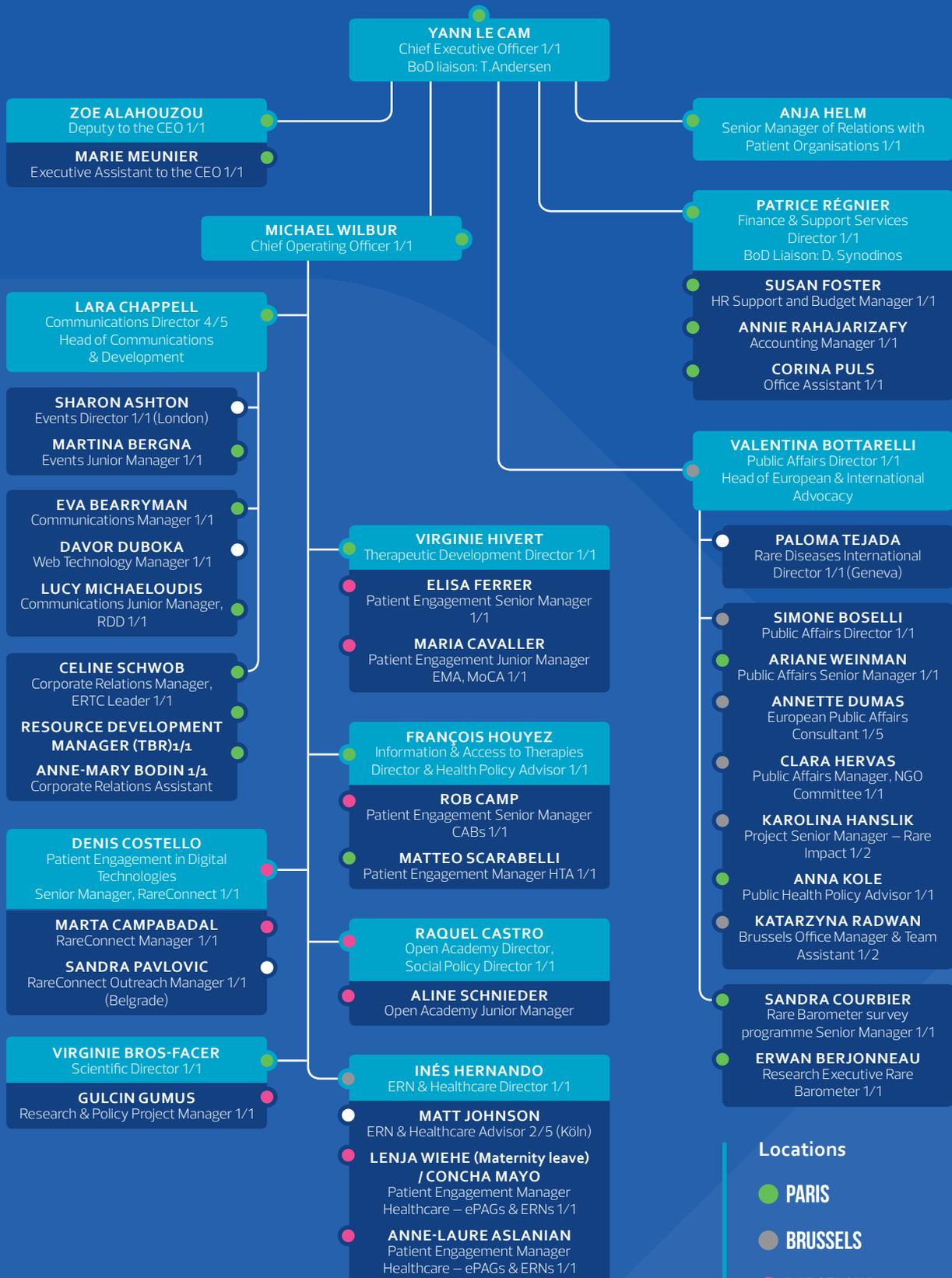
- + **Revise processes for effective and improved volunteer management** (improve volunteer acknowledgement)

#### 4.4.3 EURORDIS Staff

Appointment of following positions:

- + Chief Operating Officer
- + Development Relations Senior Manager
- + Project Senior Manager - Rare Impact (Brussels)
- + Team Assistant & Office Manager (Brussels)
- + Public Health Policy Advisor - Rare2030 Project Lead
- + Patient Engagement Manager Healthcare – ERN & ePAGs

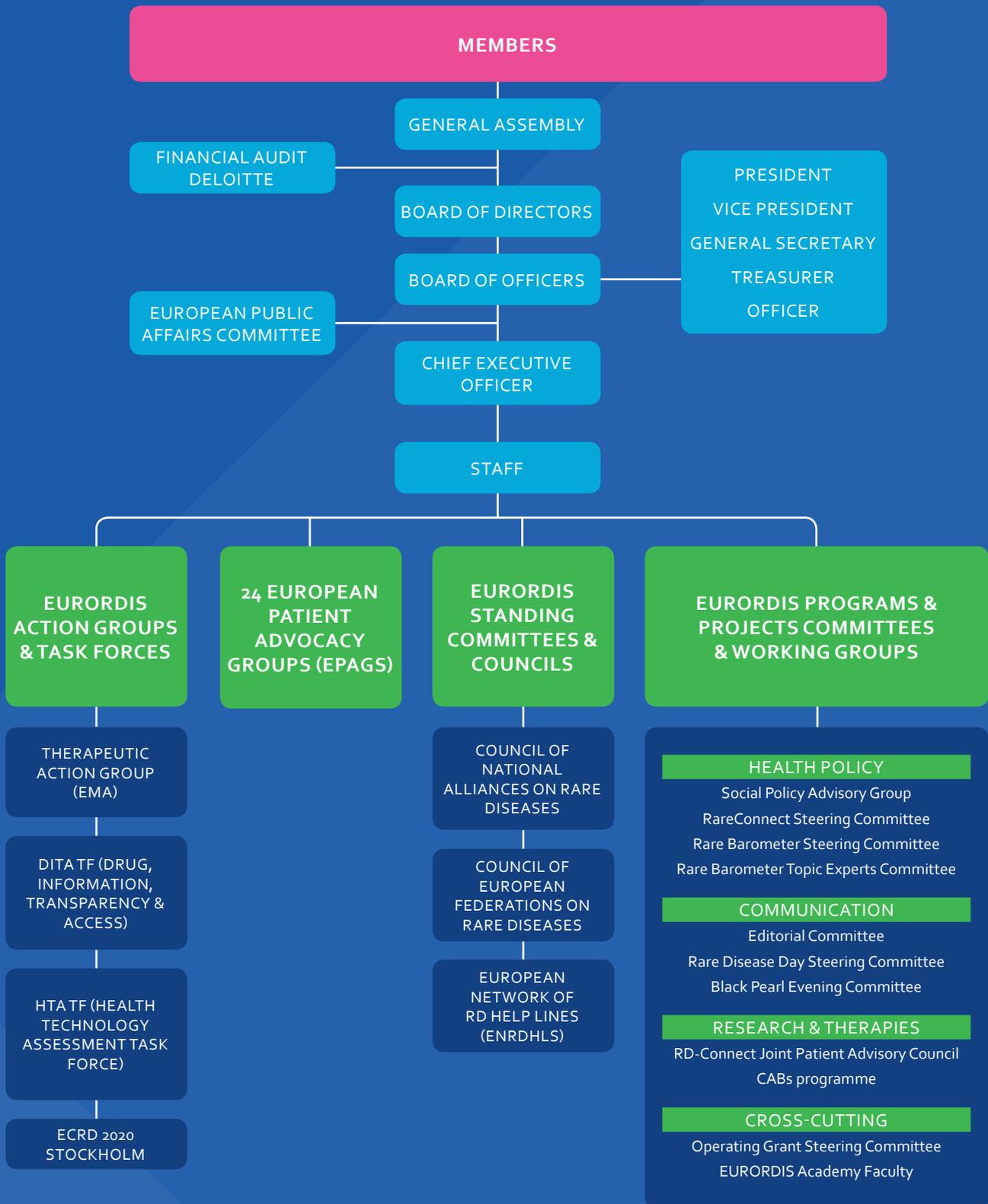
# TEAM Chart 2019



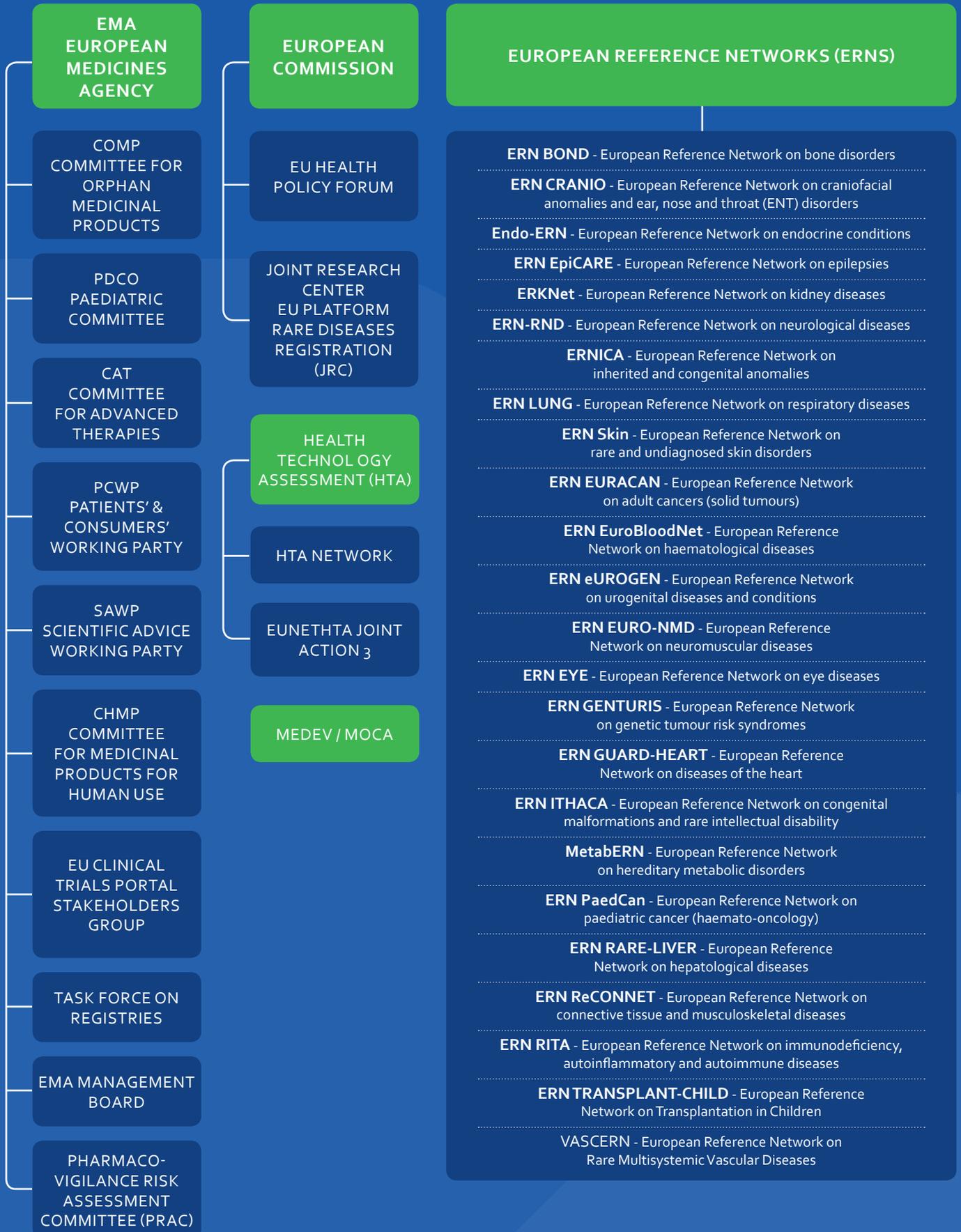
## Locations

- PARIS
- BRUSSELS
- BARCELONA
- OTHER

# GOVERNANCE Chart 2019



# EXTERNAL REPRESENTATION Chart 2019



## 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

WECAN: informal network of leaders of cancer patient umbrella organisations active in Europe

## EUROPEAN NETWORK OF PARLIAMENTARIAN ADVOCATES 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 to end the diagnostic odyssey for children

International partnerships (MoUs): NORD (USA), CORD (Canada), JPA (Japan), RVA (Australia), RPU (Russia), CORD (China) RADOIR (Iran)

## PARTNERSHIP LEARNED SOCIETIES

European Federation of Internal Medicine (EFIM)

European Hospital & Healthcare Federation (HOPE)

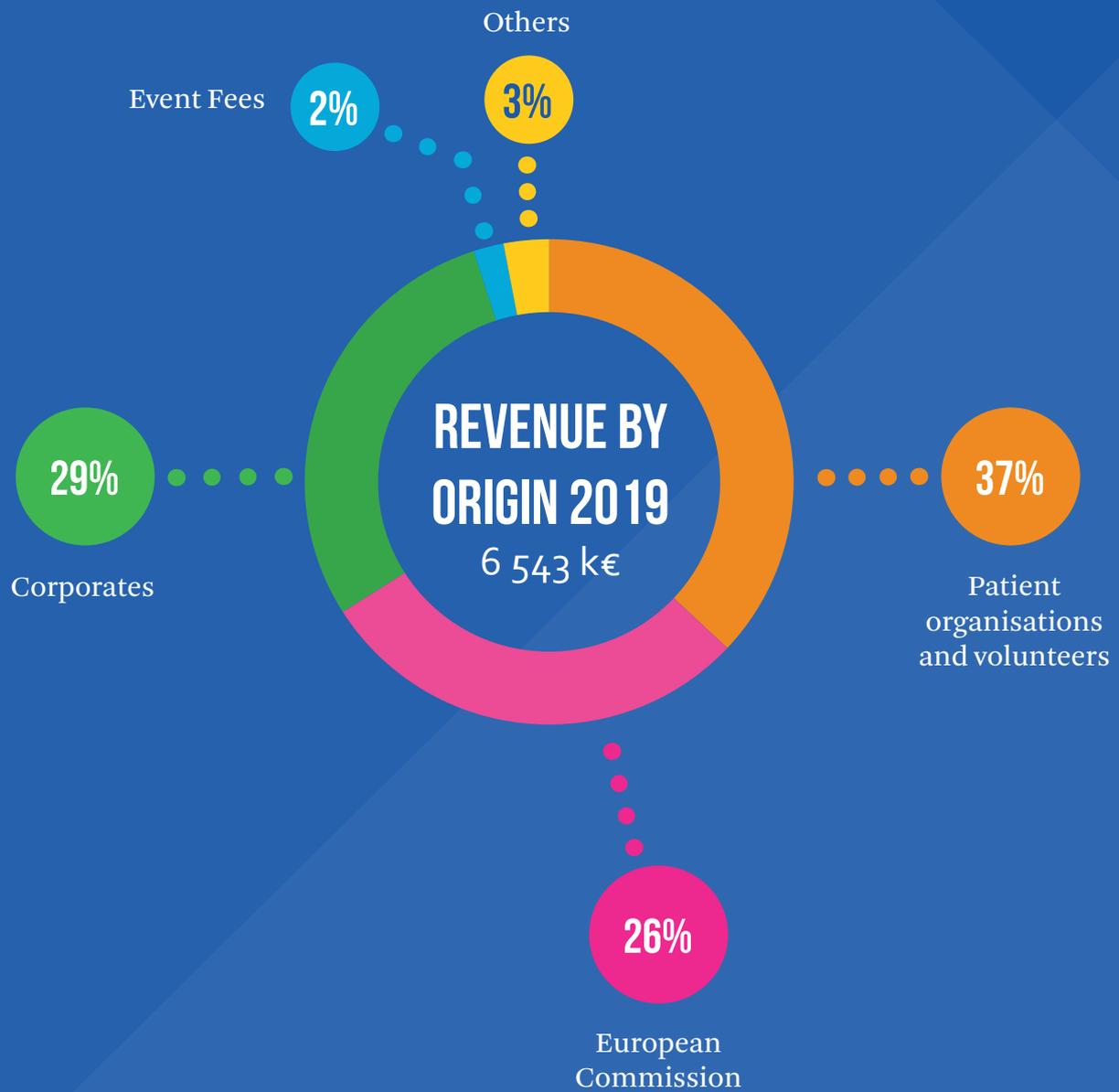
International Federation of Social Workers Europe (IFSW-Europe)

European Society of Human Genetics (ESHG)

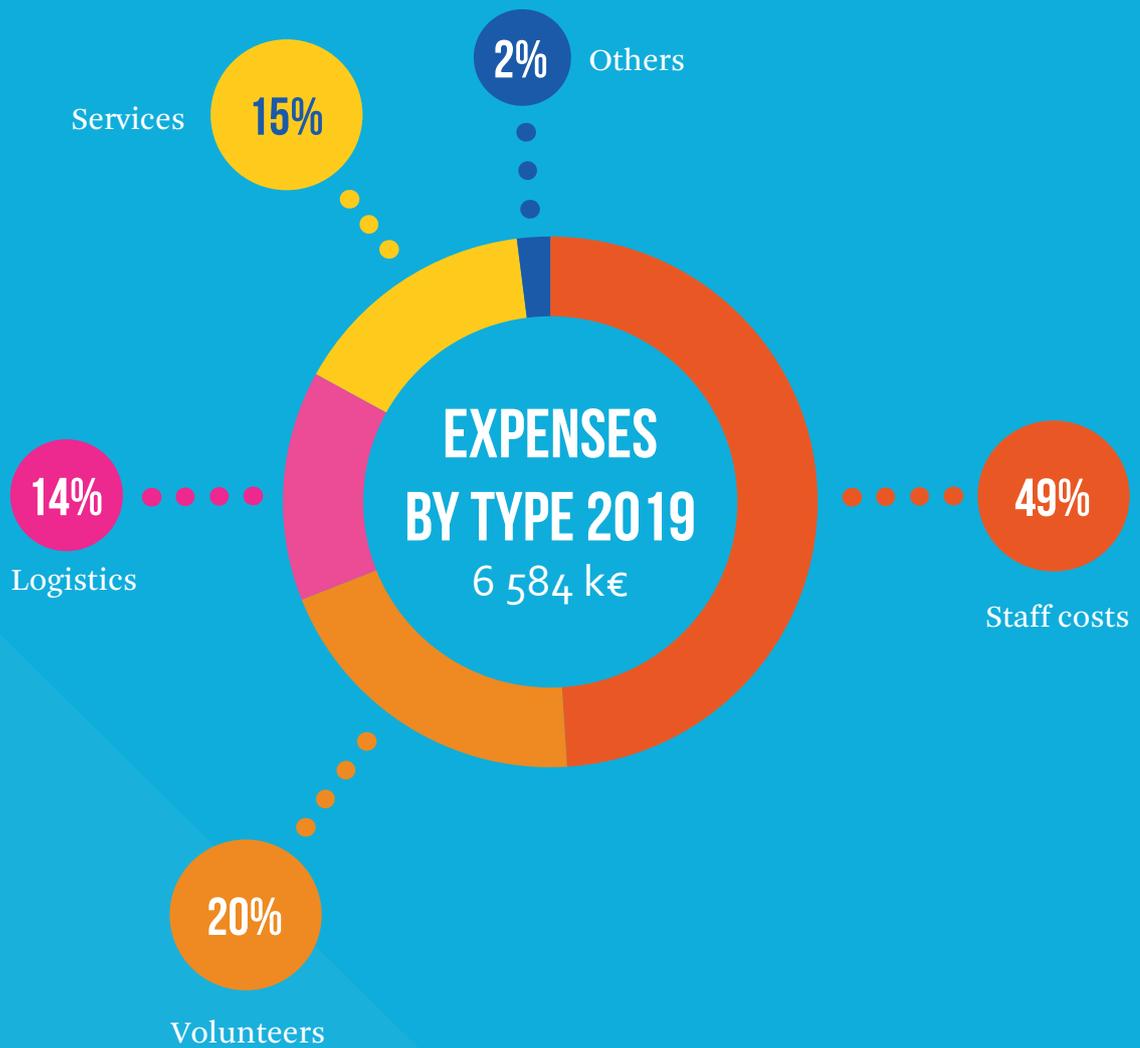
International Society for Pharmaco-economics and Outcomes Research (ISPOR)

European Connected Health Alliance - ECHAlliance

# REVENUE 2019



# EXPENSES 2019



# 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	Commission 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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Laura from Spain is living  
with arthrogryposis  
multiplex congenita.

