

EURORDIS
ACTIVITY REPORT 2019
& WORKPLAN 2020



Emily and her mother,
juvenile Batten disease

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YANN LE CAM

Chief Executive Officer



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President of the
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FOREWORD

by the President and the Chief
Executive Officer



In 2019, EURORDIS' activities focused on advocating for, empowering and engaging patients.

EURORDIS' membership base reached **884 members** by the end

of the year, including **39 new members** in EU Member States, in **72 countries** and all **28 EU member states**.

The **Council of National Alliances (CNA)** (37 members in 2019) held 2 CNA Workshops, in Bucharest in May and in Brussels in November, which was again organised partly in common with the **Council of European Federations (CEF)** (68 members in 2019). The CNA's main activities in 2019 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 **EURORDIS Membership Meeting (EMM 2019 Bucharest)** held in Bucharest in May 2019, focused on building members' capacities to promote holistic care. The EMM attracted 260 participants from 40 countries. 39 fellowships supported patient representatives from 18 countries.

In an evolving EU policy context, a focus from early 2019 was for EURORDIS and National Alliances to actively prepare the **election of the new Members of the European Parliament** and called on returning MEPs or candidates to **pledge for rare diseases (#Pledge4RD)**. The campaign successfully ended with **over 60 candidate MEPs supporting the Pledge4RD**, and with a number of them being re-elected. Following the elections, building on the relationship with re-elected MEPs, EURORDIS created contacts with the new elected MEPs, with the objective to form and launch the **new group of Parliamentary Advocates for Rare Disease**, both at the European and national level.

An advocacy achievement of 2019 is the **EURORDIS position paper on "Achieving Holistic Person-Centred Care to Leave No One Behind: A contribution to improve the everyday lives of people living with a rare disease and their families"** launched at the EMM in Bucharest. With this paper, EURORDIS and its members called upon

the EU, all European countries and all stakeholders within the health and social sector to take action based on ten recommendations for the **provision of holistic care for the 30 million Europeans living with a rare disease and their families by 2030**.

In the same dynamic, EURORDIS continued to **advocate within the legislative process toward the Work-life Balance Directive** until its adoption in June 2019, submitting amendments to the text concerning the **carers' leave and flexible working arrangements**. EURORDIS was also able to **successfully submit amendments to the first draft of the 'European Economic and Social Committee (EESC) Own-Initiative Opinion: Shaping the EU agenda for disability rights 2020'**.

EURORDIS continued to spearhead and actively support the efforts for the **development of an international rare disease movement** through **Rare Diseases International** which is since January 2019 operating independently as a legally incorporated organisation.

In December 2019, **RDI and the WHO signed a Memorandum of Understanding**.

EURORDIS continued to promote rare diseases as an international issue through the **NGO Committee for Rare Diseases** and a **Rare Disease Day Policy Event organised at the United Nations Headquarters in New York** which provided an opportunity to elevate **rare diseases within the UN 2030 Sustainable Development Agenda**.

The advocacy achievement of 2019 was the inclusion of **rare diseases within the UN Political Declaration on Universal Health Coverage (UHC)**. Rare Diseases International published its position paper on **'Rare Diseases: Leaving No One Behind in Universal Health Coverage'** and a joint EURORDIS-RDI campaign with 13 national alliances that advocated for the inclusion of rare diseases in the final text of the UN Political Declaration on UHC. In September, the **UN Political Declaration on UHC was adopted** by 193 Member States and endorsed by all Heads of State, engraving (Article 34) rare diseases as an action area for national UHC policies.

The organisation of **Rare Disease Day** is a key EURORDIS activity which in 2019 involved **over 100 participating**

countries for the first time ever. EURORDIS updated the **Show Your Rare** campaign video for 2019 in addition to producing new patient testimonials with 3 national alliance partners. The videos had over 138 000 views on social media and youtube and were translated into 35 languages.

A **strategic review was completed on the future of Rare Disease Day**, with key recommendations on new strategies for planning and carrying out the campaign for higher impact in the next decade.

The work carried out within the context of the **European Reference Networks** and the engagement of patients within them was a dominating feature of 2019. EURORDIS has promoted meaningful patient involvement in ERNs through the creation of the 24 European Patient Advocacy Groups (ePAGs) involving over **300 patient representatives, including non-members of EURORDIS**. In 2019, EURORDIS organised one **ePAG f2f meeting, one ePAG f2f Steering Committee meeting and a two-day f2f training session on leadership, network management, healthcare and research, as part of the EURORDIS Open Academy**. In addition, EURORDIS organised **more than 100 conference calls with the individual ePAG groups** and supported ePAG advocates participating in **15 ERN annual meetings**. This year we also supported the development of **"Patient Journeys"** as a tool to collect the needs and expectations of the patient community regarding their care. On the national level EURORDIS organised **workshops in 8 different countries** to help raise awareness and knowledge on ERNs and engage with national stakeholders (patient organisations, clinicians and health authorities) **to further embed the ERNs within the national healthcare systems**.

Building upon its longstanding experience in capacity-building programmes, an achievement of 2019 was consolidation of all **EURORDIS' training activities** into the **EURORDIS Open Academy**. 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 Summer School 2019** was held for the 12th year running and had an attendance of **27 patient advocates and 7 researchers, representing 15 countries and 25 rare diseases**.

The **2nd EURORDIS Winter School on Scientific Innovation and Translational Research** took place on 11-15 of March 2019, at the Imagine Institute, in Paris and brought together 30 patient advocates from 18 countries.

The **first edition of the EURORDIS Digital School** took place at the Ågrenska Resource Centre for Rare Diseases, in Gothenburg with the attendance of **20 patient representatives, from 14 countries**.

The **first EURORDIS Leadership School** launched as a comprehensive training programme for ePAG advocates on leadership, network management, healthcare and research with **56 ePAG advocates, representing 17 countries, 21 European Reference Networks and over 45 rare diseases**.

The **European Joint Programme Co-fund for Rare Diseases (EJP RD)** is a key note of 2019. An initiative, EURORDIS had long advocated for and was highly supportive of. The **EJP RD** brings over 130 institutions from 35 countries to create a comprehensive, sustainable

ecosystem allowing a virtuous circle between research, care and medical innovation, articulated with ERNs. EJP focusses on maximising the potential of already funded tools and programmes by supporting them further, scaling up, linking, and adapting them to the needs of end-users through implementation tests in real settings.

EURORDIS maintained a high level involvement 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 **162 meeting days** in EMA Committees over the year. A total of **100 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 patient representatives on the EMA Scientific Committees (CAT, PRAC, 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.

From early 2019, the deep running 360° and far reaching new project **Rare 2030**, a two-year **participatory foresight study, working with academic partners and a panel of 200 experts**, has focused on establishing today's knowledge base, so to identify the trends and derive possible future exploratory scenarios 2030-2040. This provides the ground in 2020 to develop the policy options over the next 10 years, in a back casting exercise to reach the preferred scenarios.

No foreword can be exhaustive and there are many EURORDIS' activities that have not been able to fit into these two pages such as the Rare Barometer survey programme or the EUROCabs Community Advisory Boards. For a detailed account of EURORDIS' activities in 2019 we invite you to read the report as well as explore other EURORDIS publications.

It goes without saying that none of the activities detailed in this report would be possible without the tireless dedication of the EURORDIS volunteers. In 2019, EURORDIS was privileged to rely on **437 volunteers**. EURORDIS volunteers have a unique insight into the complexity of different rare diseases across Europe and reinforces EURORDIS as a grassroots movement.

The **unexpected and unprecedented challenge** in the form of the COVID-19 pandemic affects EURORDIS, its constituencies and stakeholders, as much as our environment. EURORDIS however, is a resilient organisation that prides itself on its foresight and adaptability. We have reacted quickly to this new challenge and reviewed our activities in light of the crisis, moving most of our events online, so far with great success, but also reaching out to the rare disease community from the beginning to establish the particular needs and expectations of our community. **Building on the reality of the persons needs and on our collective expertise, we will stay the course to tackle together the new challenges lying ahead.**

”

EURORDIS IN BRIEF

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 900 rare disease patient organisations from more than 70 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.

924

member patient organisations



72 countries
(28 EU countries)

52 National Alliances of RD Patients Organisations

70 European Federations of specific rare diseases

 Founded in

1997

6 Million €
Budget

Outreach to over

2500

patient groups

 Over

470

Volunteers

93 patient advocates

382 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

2019

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HIGHLIGHTS 2019

By the end of 2019, EURORDIS had **884 members in 72 countries**, 43 of which are European countries, 28 being members of the European Union.



The **Council of National Alliances** had **37 members** in 2019. 2 CNA Workshops took place in Bucharest in May and Brussels in November, which was held partly in common with the **Council of European Federations (CEF)** (**68 members in 2019**), to allow cross cutting discussions on common issues. The main activities in 2019 were the preparation and coordination of the **Rare Disease Day 2019** and **Rare Disease Day Strategic Review**; the two CNA Working Groups: **Small EU countries and Western Balkans**; integration of ERN in national healthcare system; advocacy for the EU Legislation on HTA; new issues of patient access to potentially curative medicines.



Rare Disease Day 2019 involved over **100 participating countries worldwide**, including all 28 EU countries. EURORDIS updated the Show Your Rare campaign video for 2019 in addition to producing new patient testimonials with 3 national alliance partners. The videos had over 138 000 views on social media and youtube and were translated into 35 languages. A **strategic review in preparation of the future of Rare Disease Day**, was completed in March 2019 with key recommendations on new strategies for planning and carrying out the campaign.



EURORDIS continued its support to the **24 European Patient Advocacy Groups (ePAGs)** aligned to the scope of the ERNs, involving over **300 patient representatives** and including non-member organisations of EURORDIS. In 2019, EURORDIS organised one ePAG f2f meeting, one **ePAG f2f Steering Committee meeting** and a **two-day f2f training session** on leadership, network management, healthcare and research, as part of the EURORDIS Open Academy. In addition, EURORDIS organised more than **100 conference calls** with the individual ePAG groups and supported ePAG advocates participating in **15 ERN meetings**. This year we also supported the development of **Patient Journeys** as a tool to collect the needs and expectations of the patient community regarding their care. On the national level EURORDIS organised **workshops in 8 different countries** to help raise awareness and knowledge on ERNs and engage with national stakeholders (patient organisations, clinicians and health authorities).



EURORDIS Open Academy which consolidates all of EURORDIS' training activities greatly took off in 2019. Through the Open Academy, EURORDIS empowers patient

advocates to have the confidence and knowledge needed to **bring their expertise to discussions on health care, research and medicine development**. In 2019, EURORDIS delivered four training programmes via the EURORDIS Open Academy, in a blended format, with online courses and face-to-face training sessions.

- The **EURORDIS Summer School 2019** face-to-face training took place on the 10-14th of June 2019, in Barcelona with the attendance of **27 patient advocates and 7 researchers, representing 15 countries and 25 rare diseases**.
- The **EURORDIS Winter School** took place on 11-15 of March 2019, at the Imagine Institute, in Paris and brought together **30 patient advocates from 18 countries**, to cover important topics to **support patient engagement in research**.
- The **first edition of the EURORDIS Digital School** took place on 8-9 October, at the Ågrenska Resource Centre for Rare Diseases, in Gothenburg. **20 patient representatives, from 14 countries, participated in this innovative training course** to support patient organisations to use digital communication tools.
- **56 ePAG advocates, representing 17 countries, 21 European Reference Networks and over 45 rare diseases** took part in the **first EURORDIS Leadership School** launched as a comprehensive training programme for ePAG advocates on leadership, network management, healthcare and research.

At the end of 2019, **59 e-learning courses** were available on the **online platform of the EURORDIS Open Academy**. The platform had then reached over **1100 registered users, from more than 100 countries**.

The **European Joint Programme Co-fund for Rare Diseases (EJP RD)** was officially launched in January 2019. The EJP RD brings over 130 institutions from 35 countries to create a comprehensive, sustainable ecosystem allowing a virtuous circle between research, care and medical innovation. EJP focusses on maximising the potential of already funded tools and programmes by supporting them further, scaling up, linking, and adapting them to the needs of end-users through implementation tests in real settings. EURORDIS has been involved in the strategic development of the proposal and as such is a member of the Operating Group.

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 Pharmacovigilance Risk Assessment Committee (PRAC), the Committee for Advanced Therapies (CAT) and the Patients' and Consumers' Working Party (PCWP). EURORDIS spent a **total number of 162 meeting days in EMA Committees over the year**. A **total of 100 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 and seven project partners kicked off the **Rare 2030 project** in January 2019 to launch a two-year participatory

foresight study preparing for the next ten years of rare disease policy in Europe. Rare 2030 will guide a reflection on rare disease policy in Europe through the next ten years and beyond. Project Partners defined **4 different scenarios** around the combination of future trends to be presented at ECRD 2020 Stockholm.

EURORDIS and National Alliances prepared for the **election of the new Members of the European Parliament** in early 2019 and called on returning MEPs or candidates to **pledge for rare diseases (#Pledge4RD)**. Thanks to the active involvement of National Alliances, the campaign #pledge4RD successfully ended with **over 60 candidate MEPs supporting the Pledge4RD**, and with a number of them being re-elected. Following the elections, EURORDIS undertook to rebuild relations with MEPs engaged during the previous mandate and created contacts with the new elected MEPs, with the objective to prepare and launch the **new European Network of Parliamentary Advocates for Rare Disease**, both at the European and national level.

In May 2019, EURORDIS launched the position paper on **"Achieving Holistic Person-Centred Care to Leave No One Behind: A contribution to improve the everyday lives of people living with a rare disease and their families"**. With this paper, EURORDIS and its members called upon the EU, all European countries and all stakeholders within the health and social sector to take action based on ten recommendations for the **provision of holistic care for the 30 million Europeans living with a rare disease and their families by 2030**. Launched at the **EURORDIS Membership Meeting**, with dedicated plenary sessions and a workshop, the paper was also **widely disseminated** to all RD stakeholders and to the media.

EURORDIS continued to promote rare diseases as an **international public health priority** in 2019. Alongside the NGO Committee for Rare Diseases, a **Rare Disease Day Policy Event was organised at the United Nations Headquarters in New York** which provided an opportunity to elevate rare diseases within the UN 2030 Sustainable Development Agenda. The bulk of work with Rare Diseases International revolved around **Universal Health Coverage**, with the publication of a position paper on **'Rare Diseases: Leaving No One Behind in Universal Health Coverage'** and a joint EURORDIS-RDI campaign with 13 national alliances that advocated for the inclusion of rare diseases in the final text of the UN Political Declaration on UHC. In September, the **UN Political Declaration on UHC was adopted** by 193 Member States and endorsed by all Heads of State and **includes rare diseases** (Article 34) and engraves it as an area that needs to be addressed as part of UHC implementation in national policies. In December 2019, **RDI and the WHO signed a Memorandum of Understanding**.



1. PATIENT ADVOCACY

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

In the course of 2019, 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 2019 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 ceased existence of any rare disease Expert Groups and the end of the Joint Action for Rare Diseases (RD-ACTION) in 2018, EURORDIS continued several 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, it is clear that unmet needs are still to be addressed while new challenges emerge. New impetus at European level is necessary to ensure that they are addressed with adequate actions.

Therefore in 2019, 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. The adoption in 2019 of the European Court of Auditors Special Report on the implementation of the Cross-border Healthcare Directive (Dir. 2011/14/EU) marked a milestone in the process as a key recommendation of the report to include the assessment of the results of the 2008 rare disease strategy and decision by 2023 whether this strategy needs to be updated, adapted or replaced.

As 2019 was a year of change for the EU Institutions, EURORDIS with its members worked to prepare the political and institutional ground for this future work. With the election of the new European Parliament and then the approval of a new college of European Commissioners, a new political leadership established in Brussels. This changeover will affect directly how policies that have improved the life of people with rare disease can continue in the next electoral cycle.

Early in the year EURORDIS and its National Alliances campaigned to recruit a new group of parliamentary advocates of the rare disease cause, willing and able to commit to implement concrete actions in support of people living with rare diseases (see below point 1.3.2). EURORDIS also started to establish relations with designate-Commissioners, notably the future Commissioner for Health, by providing supporting and informative material on rare disease policy in view of their Hearings before the European Parliament. Following the approval by the European Parliament, EURORDIS was invited to meet the new Commissioner for Health Stella Kyriakides (meeting that took place in January 2020).

EURORDIS also pursued opportunities to continue the multi-stakeholder dialogue and the policy work that the dismantled Expert Groups for Rare Diseases secured for many years with key Joint Action partners. Exchanges with the European Commission continued on the feasibility of a "Stakeholder Network" for Rare Diseases hosted by the online EU Health Policy Platform, a virtual space coordinated by the European Commission (DG SANTE) to promote dialogue amongst stakeholders in the field of health policy. In the course of 2019 it became clear that such efforts should be coordinated and integrated with the activities of Rare 2030 and the creation of the Panel of Expert therein (see below 'Foresight Study on Rare Disease in 2030').



Throughout 2019, the Panel grouped nearly 200 stakeholders in the rare disease field, with the objective of building scenarios for rare disease policy and identifying policy recommendations for the next decade. Hence, in 2019, project partners started planning for the continuity of the Expert Panel within the Health Policy Platform when the project comes to an end (December 2020) as required in the project description.

Finally, EURORDIS continued to follow the discussions on the next budgetary period that led to the adoption of the new **EU Multiannual Financial Framework 2021-2027** 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 consultations that European Commission organised on specific funding programmes within the future budgetary framework, such the consultation on notably Horizon Europe, that will support research and innovation from 2021 to 2027.

1.3.1 Foresight Study on Rare Diseases in 2030



In January 2019, EURORDIS (as project coordinator) and seven project partners (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) kicked off the Rare 2030 project with the support of Belgian MEP Frederique Ries (Photo 1) to launch a two-year participatory foresight study preparing for the next ten years of rare disease policy in Europe. Continued gaps and new challenges for people living with rare diseases require the proposal of a new forward-looking policy framework since the adoption of the Council Recommendation on European Action in the field of Rare Diseases in 2009. Rare 2030 will guide a reflection on rare disease policy in Europe through the next ten years and beyond.

During this first year, project partners (Photo 2) recruited several consultative bodies to contribute to all four stages of the Rare 2030 foresight study (Figure below). These groups include:

- + a nearly 200-member Panel of Experts
- + a Research Advisory Board
- + a group of Young Citizens
- + 6 national Alliances (and surrounding countries) holding EU presidencies between 2020 and 2023: Croatia (and Slovenia and Romania), Germany, Spain (and Portugal), France (and Belgium, Luxembourg, Switzerland), Sweden, Czech Republic (and Slovakia)



With the help of these groups, project partners followed through with two of the four critical steps in a foresight process which included establishing 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.



Considering these trends, Project Partners defined 4 different scenarios around the combination of future trends related to our societal values toward solidarity and equity and how we apply these values to drive innovation.

The European Conference on Rare Diseases in Stockholm in 2020 will be the first opportunity to present these scenarios and ask stakeholders which is most likely to happen, which scenario they prefer and begin to propose the policy options to get there.

As such, the theme of the conference: 'The Patient Journey in 2030' has been set to encourage conference discussions to reflect and support the final step of Rare 2030, that is transforming preferred scenarios into policy recommendations. Where 2019 was a year of preparing the groundwork and setting the stage to accomplish this last step with all stakeholders including patients, 2020 will be the year of executing the workshops and conferences planned.

To best disseminate the reflections of the project partners and consultative bodies, several channels of communication including a website (rare2030.org), Twitter account (@rare2030) and Facebook group (@rare2030) have been established. In terms of sustainability, project partners have considered the opportunity to continue discussions across the panel of experts, project partners and research advisors to maintain activities via a "Stakeholder Network" hosted by the EU Health Policy Platform.

A new policy framework may take several years to prepare and adopt. It will also require the support of Member States. For this reason, EURORDIS has also initiated many bilateral conversations in 2019 with health ministries, patient umbrellas and other key stakeholders in national policy making positions to prepare for uptake of policy recommendations at the end of the project.

1.3.2 Parliamentary Advocates for Rare Diseases

The network of Parliamentary Advocates for Rare Diseases brings 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 of governance.

With the European Parliament five-year term coming to an end in early 2019, EURORDIS and its National Alliances prepared for the election of the new Members of the European Parliament (23 to 26 May 2019). To ensure that the 25-30 million people living with a rare disease in Europe and their families are not left behind, EURORDIS and its member associations called on returning MEPs or candidates to pledge for rare diseases (#Pledge4RD).



PEOPLE WITH RARE DISEASES AND THEIR REPRESENTATIVES MUST BE ENGAGED IN ALL FORA WHERE DECISIONS THAT AFFECT THEIR LIVES ARE MADE.

The election campaign aimed at showing how rare diseases are a successful example of European action at political level in the spirit of solidarity and cooperation, that Europe has helped tremendously to advance the cause of people living with a rare disease in the last 20 years and called for candidates willing, in new legislative term, to continue working together to address challenges and unmet needs.

Thanks to the active involvement of EURORDIS National Alliances, the campaign #pledge4RD successfully ended with over 60 candidate MEPs supporting the Pledge4RD, and with a number of them being re-elected. Following the elections, EURORDIS undertook to rebuild relations with old supporters and created contacts with the new elected MEPs, with the objective to prepare the formal creation of the new group of Parliamentary Advocates for Rare Disease, both at the European and national level, and thus consolidate a new core group of advocates of the rare disease cause and committed to implement concrete actions in support of people living with rare diseases within the political agenda, as outlined in the #Pledge4RD manifesto.

In July, a group of MEPs attended the Summer Reception organised on the occasion of the 'Intimate Audrey' Exhibition in Brussels, when MEP Tilly Metz gave a keynote address.



In autumn, EURORDIS provided parliamentarians with a list of suggested questions that MEPs could ask during the confirmatory Hearings of the designate Commissioners. The questions covered a number of areas of relevance for people living with rare diseases that are or should appear on the EU policy agenda, from the adoption of the HTA Regulation to cross-border healthcare.

Finally, in view of the official launch of the network of Parliamentary Advocates in February 2020, as from September EURORDIS established one-to-one contacts and held meetings with new and old MEPs, preparing a programme that would cover crucial areas where action by politicians is required, from access to medicines to holistic care, from cross border care to research.

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

The EU Joint Action on Rare Cancers (JARC) was a multi-stakeholder collaboration between 21 Member States, Norway, Switzerland and the European Commission, coordinated by the Fondazione IRCCS Istituto Nazionale dei Tumori of Milan. There were altogether 61 partners involved in the JARC including Ministries of Health, universities, public health institutions, cancer registries, oncological institutes, research societies and three patient organisations: ECPC – European Cancer Patient Coalition, CCI-E – Childhood Cancer International – Europe and EURORDIS. The partnering patient organisations were involved across all work packages.

The JARC aims to prioritise all rare cancers on the agenda of the EU and Member States with regard to national cancer plans, harmonisation of practices and funding of research, and support to the development of European Reference Networks for rare cancers, namely:

- + EURACAN (rare solid tumours in adults);
- + PaedCan (paediatric cancers);
- + EuroBloodNet (rare haematological diseases including haematological malignancies in adults);
- + GENTURIS (Genetic Tumour Risk Syndrome).

In September 2019, the JARC concluded three years of work with the publication of its book entitled: **"RARE CANCER AGENDA 2030: Ten Recommendations from the EU Joint Action on Rare Cancers"**. The recommendations were presented at the European Parliament on Wednesday 11 September:

- 1 Rare cancers are the rare diseases of oncology
- 2 Rare cancers should be monitored
- 3 Health systems should exploit networking
- 4 Medical education should exploit and serve healthcare networking
- 5 Research should be fostered by networking and should take into account an expected higher degree of uncertainty
- 6 Patient-physician shared clinical decision making should be especially valued
- 7 Appropriate state-of-the-art instruments should be developed in rare cancers
- 8 Regulation on rare cancers should tolerate a higher degree of uncertainty
- 9 Policy strategies on rare cancers and sustainability of interventions should be based on networking
- 10 Rare cancers patients should be engaged (...in all crucial areas such as disease awareness and education, healthcare organization, state-of-the-art instruments, regulatory mechanisms, clinical and translational research)

ECPC, CCI-E and EURORDIS contributed to each chapter of the JARC book. A section on paediatric cancers is included at the end of each chapter to emphasise their specific needs as compared to rare cancers in adults.

The three partner patient organisations developed together the last chapter on patient engagement (recommendation N°10), highlighting patients, families and carers' recommendations to EU institutions and Member States for improving policies for research, care and follow up of cancer survivors, and supporting the full deployment of ERNs for rare cancers.

Focus on National Cancer Control Programmes

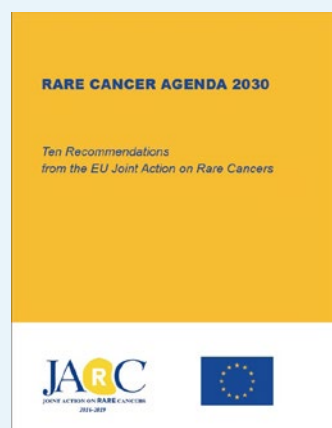
In addition, the JARC, under the leadership of the Catalan Institute of Oncology (ICO), undertook in 2017-2018 "a comparative analysis of the priorities and recommendations on rare and paediatric cancers based on National Cancer Control Programmes (NCCPs), National

Rare Disease Plans (NRDPs), EU funded initiatives and priorities for patients' organisations". EURORDIS actively participated in this study together with SIOPe (the European Society for Paediatric Oncology) and CCI-E.

The results of this study are published in the Journal of Cancer Policy (12 February 2020): "Rare cancers are hardly addressed in NCCPs and not addressed in NRDPs. Of the 15 NCCPs analysed, only 8 contained some elements on RCs [Rare Cancers], and only 3 of these described specific measures to address this disease group or took a comprehensive approach. The cross-cutting analysis of the 8 NCCPs allowed identifying 14 critical issues necessary to reach a comprehensive approach to RCs' policy."

This study will help support future advocacy work on the inclusion of rare cancers (paediatric cancers and rare cancers in adults) in EU MS's NCCPs as well as in the future 'Europe Beating's Cancer Plan', making relevant synergies with national rare disease plans.

The work produced in the JARC serves to lay the groundwork for actions to be developed and implemented over these next ten years with a view to further improve research, care and quality of life of children, adolescents and adults affected by a rare cancer in Europe. The partners of the JARC are receiving the support of Rare Cancers Europe to pursue this mission.



As part of the Awareness Campaign on the four ERNs on Rare cancers at ESMO 2019 (27 Sep-1 Oct 2019), Kathy Oliver, Chair of International Brain Tumour Alliance (IBTA), Muriel Rogasik, EURACAN Network Manager and EURORDIS developed a joint leaflet of the four ERNs relevant to rare cancers, namely EURACAN (rare solid tumours in adults), PaedCan (paediatric cancers), EuroBloodNet (rare haematological diseases including haematological malignancies in adults) and GENTURIS (genetic tumour risk syndromes). The content of the joint leaflet was reviewed by respective ERN coordinators, managers and ePAGs. A copy of this joint leaflet was inserted in the bag of each of the **27 000 participants** attending the annual congress of the European Society for Medical Oncology (ESMO).



1.3.4 Advocate to improve the regulatory process for orphan medicinal products

The debate on how to effectively shape an adequate regulatory and incentives system for the development of therapies for rare diseases continued to be a very contested issue, certainly in European political circles. The so-called Regulation for Special Populations (the Regulations on Orphan Medicinal Products and on Paediatric Medicines) have been under the microscope for quite some time, and in 2019 it was no different, owing it in particular to the high prices of several new therapies coming to market during the year.

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. In this context, the European Commission (DG SANTE) organised

in June 2019 a workshop "Medicines for Rare Diseases and Children: Learning from the Past, Looking to the Future" to gather stakeholder input into the ongoing evaluation. EURORDIS participated, together with some 150 experts from across the EU, representing national governments and health authorities, academia, patient and health professionals' organisations and the pharmaceutical industry.

Unfortunately, this evaluation is yet to be completed, primarily due to the changeover in the European Commission as well as shifting priorities, and it is now forecasted for summer 2020, together with the launch of a new European Pharmaceutical Strategy.

1.3.5 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).

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.

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 third and final Multistakeholder Symposium on Improving Patients’ Access to Rare Disease Therapies. The four pillar approach presented in the paper constituted the cornerstone of the two-day meeting, which took place in February 2019.

Over 250 people gathered at this event, including rare disease patients, patient advocates, healthcare industry, consultants, investors, ERN clinical representatives, academia, healthcare professionals, researchers, payer bodies, HTA agencies, National Competent Authorities, regulators, policy makers, and government workers.

Some of the important take-home messages include the recognition that multi-stakeholder collaboration is a fundamental condition to achieving patient access to rare disease medicines; that there is a need for a more trusted and transparent cooperation between payers and companies; and that the expectations for the European Reference Networks (ERNs) to be an important facilitator in evidence generation building is also an area of high interest that will be monitored closely over the coming years.

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.

Real-world data (RWD) collection and real-world evidence (RWE) generation can play a critical role in assessing the value of highly innovative treatments for patients and healthcare systems, as RWD brings deeper insights into lifestyle patterns and 'real life' settings. This is essential when 'highly innovative technologies', which are potentially transformative (such as immunotherapies and cell & gene therapies), come to market early via expedited regulatory approvals to benefit patients, and often only limited evidence from traditional clinical development programmes is possible.

In 2018 and 2019, the Belgian National Institute for Health and Disability Insurance (INAMI/RIZIV) worked together with stakeholders comprising policy makers, HTA bodies, payers, regulatory agencies, clinicians, patient groups, industry and academic experts to consider the use of RWE. Two practical guidance papers have been commissioned by INAMI/RIZIV and developed through multi-stakeholder collaboration (facilitated by FIPRA), to which EURORDIS participated actively.

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 continued throughout 2019 with analysis of key challenges and the co-creation of European and national solutions. The multi-stakeholder consortium went full steam ahead and completed reports assessing the situation in regards to affordability, availability, assessment and accessibility of gene and cell therapies in ten European countries. Preliminary results have been presented at a number of conferences across Europe, gathering along the way the crucial input of the rare disease community. The initiative is due to be completed by Summer 2020, with reports already published and available for consultation on the RARE IMPACT website (www.rareimpact.eu). The reports will constitute the evidence base to advocate for improved processes to allow for the broadest access to potentially transformative treatments. These therapies as well as the outcomes of RARE IMPACT will feature in two sessions at the next European Conference on Rare Diseases and Orphan Products.

To prepare the reimbursement decision: the HTA momentum

HTA Patients' Involvement & Mentoring Programme in EUnetHTA

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

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.

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 Union³, conducted by Nivel, the National Institute for Public Health and Environment (Netherlands) and EPHA, on behalf of the European Commission.

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

1.3.6 Advocate to improve Access to Care for rare disease patients

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

permanently available on line: <https://www.eurordis.org/access-campaign-participate>.

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



1.3.7 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 2019, 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 3rd 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.

³ https://ec.europa.eu/health/sites/health/files/files/documents/2017_02_28_final_study_report_on_off-label_use_.pdf

- + After the adoption of the European Commission proposals, EURORDIS has been following through 2018 and 2019 the legislative work of the other EU Institutions, notably the European Parliament and the Council of the EU.
- + 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. In 2019, EURORDIS participated to a large stakeholder consultation on the priorities of the future research and innovation funding programme, by preparing an extensive contribution on all areas of rare disease research. EURORDIS also participated to the Open Day workshop on rare diseases organised by the European Commission, DG Research and Innovation, contributing to the shaping up of the EU research priorities on the upcoming years.
- + 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.

1.3.8 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 regularly 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.

EURORDIS therefore continue monitoring the implementation of the Cross Border Healthcare Directive (Dir. 2011/24/EU), including by supporting the European Patient Forum (EPF) action 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.

In January 2019, the **European Parliament** adopted a **Resolution** aimed to shed light on the **implementation of the 2011 Directive**. The so called ‘Belet Report’, from the name of the MEP leading on the file, brought political attention to issues such as its implementation, cross-border healthcare funding, ERNs and eHealth. EURORDIS proposed amendments that, tabled by MEP Frédérique Ries, were adopted and brought improvements to the text of the Resolution by emphasising, amongst others, the shortcomings of the prior authorisation system and upfront payments in cross-border care for patients; and the importance of integrating ERNs into national healthcare systems and providing them with adequate funding.

A turning point on healthcare patients’ rights implementation was the release, on 4th June 2019, of the **European Court of Auditors’** (ECA) special report **“EU actions for cross-border healthcare: significant ambitions but improved management required”**.

In preparation of this landmark report, the auditors examined whether the European Commission has monitored the implementation of the EU cross-border healthcare Directive and supported Member States in informing patients of their rights. They assessed the results achieved on exchanges of health data across borders and checked key actions on rare diseases.

The report concluded that the Directive’s implementation falls short of its ambition and EU citizens still do not benefit enough from the actions set out in the Cross-Border Healthcare Directive.

The report also put forward three key recommendations to improve support to facilitate rare disease patients’ access to healthcare, in particular by:

- + Assessing the results of the 2008 rare disease strategy (including the role of the ERNs) and decide whether this strategy needs to be updated, adapted or replaced (by 2023);
- + Setting out ways forward to address the challenges faced by the ERNs (by 2020);
- + Work towards a simpler structure for any future EU funding to the ERNs (by 2022).

Prior to the publication of the report, EURORDIS has been among the key stakeholders that have been contacted and interviewed since 2017 to provide opinion and evidence. Both the European Commission and the EU Council have agreed to implement these recommendations by the timeline indicated by the ECA in its report.

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

In 2019, EURORDIS continued to support Rare Disease National Alliances to help raise awareness and knowledge on ERNs at national level. We organised 8 workshops in different countries targeted at engaging with national stakeholders (patient organisations, clinicians and health authorities) to help them understand how they can

participate and benefit from this new structure. As part of this effort, in 2019 we have also engaged with some National Alliances on specific topics linked to rare disease healthcare delivery such as accreditation of centres of expertise and national networks of rare disease centres of expertise.

1.3.10 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 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 programme of work and activities of the brand new EJP for RD started in January 2019. The proposed joint programme of activities ranging from research to coordination and networking activities, including training, demonstration and dissemination activities, is 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 and Executive Committee. 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. Furthermore, EURORDIS Public Affairs Director is a member of the Policy Board of the EJP.

1.3.11 Advocate to improve access to quality rare disease diagnosis

Throughout 2019, EURORDIS continued to advocate for improved access to and quality of rare disease diagnosis.

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 translational 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).

EURORDIS is also a member of the Steering Committee for the work package that addresses dissemination of the results of the Solve-RD project as well as ensuring the proper engagement of stakeholders.

Capacity building programme for patient representatives on scientific innovation and translational research (EURORDIS Winter School)

The EURORDIS Winter School aims to deepen patient representatives' understanding of how pre-clinical research translates into real benefits for rare disease patients. The 4,5-day onsite 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 main objective of the Winter School is to improve RD research and innovation and to enhance the uptake of research results by building the capacity of the patient community and promoting the inclusion of patients in rare disease research.

The specific objectives of the Winter school include:

- + Providing the knowledge and skills required for RD patients to become legitimate collaborators in RD scientific & translational research;
- + Empowering the RD patient community and specifically patient representatives in their roles as equal, valued, and efficient partners in research and scientific project / ERNs governance bodies;

The second edition of the Winter School took place over 5 days in March 2019 at the Imagine Institute for Genetic Diseases in Paris with 30 patient representatives from 18 countries, representing over 25 rare diseases. Training was delivered by expert speakers and researchers from across Europe covering a broad range of topics on scientific innovation and translational research. Specific topics covered included genetics and diagnosis, genome editing tools, translational research, pre-clinical models, drug repurposing, IRDiRC activities, EJP RD, patient participation in research projects and ERNs. The pre-training, based on the 5 online training unities and 3 webinars, took place from January to March.

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 counsellors, 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. In 2019, EURORDIS continued to participate in UDNI annual meetings and share patients' perspectives



by specifically being involved in two working groups on data sharing and genetic counselling.

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, genetic counsellors 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 Community Engagement Task Force (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).

In 2019, EURORDIS organised 2 teleconferences and a face to face meeting where CETF members worked on creating a resource map to outline the journey of the undiagnosed patient. CETF members also discussed issues addressing specific needs of undiagnosed rare disease patients and their families in order to establish the engagement of stakeholders within and across initiatives and networks in the field of diagnosis at European and international levels. Moreover 3 CETF members have participated as speakers in the EURORDIS Winter School on Scientific Innovation and Translational Research as speakers on topics relevant to their area of expertise.

Patient advocacy and involvement in Biobanks & Registries

Biobanks:

EURORDIS actively participated in the BBMRI Stakeholder forum 2019, contributing to the discussion and approval of Rules of Procedure of the Stakeholder forum at the face to face and online meetings.

EURORDIS also played an active role at the Europe Biobank Week 2019 by being a member of the scientific committee for the selection of abstracts and by chairing the session "Rare Diseases: The Next Big Step" where projects on rare diseases and biobank research were presented.

Registries:

+ **International Summer School for rare diseases and orphan drug registries:** EURORDIS participated on 23-25 September 2019 in the 7th International Summer School for rare diseases and orphan drug registries organized by ISS (the Italian Institute for Health) in Rome and funded by the EJP on RD since 2019. EURORDIS presented past and current EURORDIS activities on patient registries highlighting the specific roles of the European Patient Advocacy Groups (ePAGs) involved in the 24 ERNs and the activities of the ePAG Cross-Working Group on Research & Registries as well as the infographics on how to improve qualities of RD patient registries developed with the Patient Advisory Council within the RD-Connect project.

+ **Rare Diseases (RDs) Global Open FAIR Implementation (RDs GO FAIR) Network:** EURORDIS is a member of the seed group of the RDs GO FAIR network, ensuring that RD patients and patient representatives are actively engaged in each phase of planning and implementation of rare disease research and that FAIR sharing in the RD community is respectful and responsible towards RD patients. RD patients can contact EURORDIS for

GO FAIR members to analyse data in their registry and get advice on organising a FAIR implementation for their registry in order to make their data usable across resources. Moreover, RD patients can contribute to the RDs GO FAIR network to help organise a FAIR network and help GO FAIR with advocacy.

- + **Transversal working group for ePAG on Research and Registry:** EURORDIS launched this dedicated working group in October 2018. The main goals are to 1) inform, guide and support ePAGs work on research and registries activities for ePAG Patient Advocates; 2) bring together the collective understanding of relevant ERN clinical and basic research topics and patient registries to inform the overall research & registry strategy and its implementation in terms of contents and priorities and 3) exchange between group members and registry experts working and/or involved in RD research

activities. EURORDIS organised 3 teleconferences in 2019 that mainly focused on RDregistries initiatives and implementation of FAIR principles in registries.

- + **Registries For Rare Endocrine Conditions Workshop:** EURORDIS participated on 13th December in a workshop organised by EuRECa, the Rare Endocrine Registries Project to explain the value of registries from the patient perspective. As EURORDIS hold RD patient registries as an advocacy priority since 2006, the presentation also illustrated how EURORDIS activities have influenced EU policies in the field of registries.
- + **Short series of EURORDIS Webinars on Registry initiatives:** In June 2019, EURORDIS organised the first webinar in a short series of webinars dedicated to present and inform RD patients and the wider community on ongoing projects, platforms and initiatives related to registries.

1.3.12 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 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. 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.

RDI was incorporated under French law as an Association Loi 1901 on December 7, 2018 and has been operating as a legally-registered organisation since January 2019. RDI continues to benefit from EURORDIS financial and in-kind support as spelled out in the Memorandum of Understanding signed by both organisations in May 2018 and that is valid until 2022.

RDI's 6th annual meeting, New York, USA

RDI's 6th Annual Meeting took place on February 20th, 2019 at the Microsoft Global Headquarters in New York, USA back to back to the Launch of the Recommendations Report of the Global Commission to End the Diagnosis Odyssey of Children with a Rare Disease and the day before the NGO Committee for Rare Diseases High Level Event which was on the occasion of Rare Disease Day at the United Nations Headquarters in New York.

40 delegates from 32 member organisations from 26 countries attended the General Assembly and Membership Meeting and 119 delegates registered to attend the RDI Global Meeting, which was open to member and non-member patient groups, as well as all other stakeholders.

RDI Position Paper on Universal Health Coverage

In April 2019 RDI adopted and released the Position paper 'Rare Diseases: Leaving No One Behind in Universal Health Coverage'. The paper, which included input and featured country case studies from RDI members, argues that UHC shall never be attained or realised if persons living with rare diseases are left behind and their needs left unmet.

Rare Diseases highlighted at the 72nd World Health Assembly

On May 23rd 2019, RDI organised two events on rare diseases in the context of the WHO 72nd World Health Assembly, gaining further momentum towards formal relations with WHO and the inclusion of Rare Diseases in the UN Political Declaration on UHC.

First Formal WHA Side Event on Rare Diseases

The formal side event: «How transformational digital technologies can contribute to leave no one behind in UHC: the case of rare diseases » was held inside the Palais des Nations, at the UN Headquarters in Geneva, and was part of the official World Health Assembly (WHA) agenda. The event was sponsored by EU, Romania, Kuwait and was co-sponsored by an additional nine Member States.

Rare Diseases included in landmark UN Political Declaration on Universal Health Care

In July-August 2019, RDI and EURORDIS led a joint campaign with 13 national rare disease alliances to reach out to national ministries of health and foreign affairs; and to Permanent Missions to the UN – to advocate for the inclusion of rare diseases in the final text of the UN Political Declaration on UHC.

On September 23, 2019 the UN Political Declaration on UHC was adopted by 193 Member States and endorsed by all Heads of State at High-Level Meeting on UHC during UNGA 74th Session. The Declaration includes rare diseases (Article 34) and engraves it as an area that needs to be addressed as part of UHC.



Start of formal collaboration with World Health Organization

On 18 March 2019, Yann Le Cam and Durhane Wong-Rieger met with Dr. Tedros Adhanom Ghebreyesus, Director-General of the WHO, in Geneva. The meeting was a highlight in the journey to carve a space for rare disease within the WHO programme of work and served to identify areas of collaboration in view of a Memorandum of Understanding between the two organisations, which was signed in December 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.

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).

Rare Disease Day Policy Event at the United Nations

On 21 February 2019, NGO Committee for Rare Diseases organised a Rare Disease Day Policy Event at the United Nations Headquarters in New York (the Second High-Level Event of the NGO Committee for Rare Diseases). The event, which was hosted by the Permanent Mission of Estonia to

the UN, and co-hosted by 14 other Permanent Missions to the UN, was an opportunity to elevate rare diseases within the UN 2030 Sustainable Development Agenda.

The event which was organised in close collaboration with EURORDIS and RDI, was attended by 100 participants from the international NGO community, UN agencies, national governments, academic institutions, the private sector and the rare disease patient community came together at the United Nations Headquarters in New York.

At the landmark event, Yann Le Cam, Chief Executive Officer of EURORDIS, launched a call for rare diseases to be included in universal health coverage (UHC) and also called on UN Member States to support a UN resolution on rare diseases. There was an overwhelming sense of support from Member States and UN and WHO speakers present.

Rare Diseases on the sidelines of the WHO World Health Assembly

On May 23rd RDI, in collaboration with the NGO Committee for Rare Diseases, hosted an informal rare disease flagship Side Event at the International Museum of the Red Cross, on the sidelines of the 73rd WHA. The meeting under the theme "UHC: Including rare diseases to leave no one behind" gave the opportunity to show that integration of rare diseases in policies and legislations is possible in all countries whatever their level of development (ex. Philippines)

The event was attended by 65 representatives of Member States and the rare disease community and was endorsed by the Permanent Mission to the UN of France, Georgia, Malta and the Netherlands.

Key speakers included: Dr Rüdiger Krech, Director, Universal Health Coverage and Health Systems, Office of the Assistant Director-General, World Health Organization and Todd Howland, Chief of the Development and Economic and Social Issues Branch, Office of the United Nations High Commissioner for Human Rights.

Rare diseases recognised by UN Human Rights

Throughout 2018 and 2019, the NGO CfRD advocated for rare diseases to be recognised in the human rights agenda. In July 2019, this resulted in the inclusion of rare diseases in the 2019 Annual Report of the Office of the High Commissioner for Human Rights (OHCHR) "The Right to Health: how human rights framework can contribute to

implementation of UHC." The report refers to persons living with a rare disease as a particular focus group to take into account when implementing UHC within a human rights framework (paragraphs. 28, 29, 47 and recommendation 51).

1.4 Voicing the social needs of people with a rare disease and advocating for holistic care



In May 2019, EURORDIS launched the position paper on "Achieving Holistic Person-Centred Care to Leave No One Behind: A contribution to improve the everyday lives of people living with a rare disease and their families" (<https://www.eurordis.org/carepaper>).

With this paper, EURORDIS and its members call for the provision of holistic care for the 30 million Europeans living with a rare disease and their families by 2030.

The strategy to ensure holistic and integrated care for rare diseases is structured around three pillars:

- + **Pillar 1:** Quality and adequate social services and policies
- + **Pillar 2:** Integrated care: bridging health and social care
- + **Pillar 3:** Equity of rights and opportunities

In April 2019, the new Social Policy Action Group (SPAG) was launched, as a group of 11 volunteer patient advocates who disseminate and contribute to the positions of EURORDIS and its members, advocating for holistic and integrated care for people living with a RD and their families.

Throughout the year, 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), conducted via the RareBarometer Programme.

With this paper EURORDIS and its members call upon the EU, all European countries and all stakeholders within the health and social sector to take action based on ten recommendations:

Our recommendations to achieve holistic care for people living with a rare disease and their families, by 2030:



1. Making full use of EU instruments and European networks
2. Creating a supportive political environment at national level
3. Gathering and disseminating knowledge and good practices
4. Implementing specific mechanisms that ensure integrated care
5. Meaningful engagement in design and implementation of policies/services
6. Ensure access to adequate social services and social protection
7. Ensuring the recognition and compensation of disabilities
8. Creating conditions for access to adapted and sustainable employment
9. Implementing specific mechanisms for empowerment
10. Eliminating all types of discrimination

43 specific recommendations & action points.
Full paper at www.eurordis.org/carepaper

EURORDIS.ORG



Launched at the EURORDIS Membership Meeting, with dedicated plenary sessions and a workshop, the paper was also widely disseminated to all RD stakeholders and to the media. Throughout the year, EURORDIS and its members seized all the opportunities to present this position paper.

Earlier in 2019, EURORDIS contributed to the work of the European Commission on Long Term Care, by participating in a dedicated EC-led workshop, held in January. EURORDIS brought forward the unmet long term care needs of people living with a RD and the outcomes of the INNOVCare project.

EURORDIS also continued to follow the discussions on the Work-life Balance Directive until its adoption in June 2019, after contributing to the related European Parliament discussions in 2018, submitting amendments to the text concerning the carers' leave and flexible working arrangements.

1.5 Advocate to improve access to disability rights

With the approach of the conclusion of the European Disability Strategy 2010-2020, EURORDIS took the opportunity to get involved in the ongoing discussions on the EU Disability Strategy post-2020, raising awareness of the disabilities faced by people living with a RD and advocating for the disability agenda to recognise and address the needs of people living with a RD.

In October, EURORDIS was able to successfully submit amendments to the first draft of the 'European Economic and Social Committee (EESC) Own-Initiative Opinion: Shaping the EU agenda for disability rights 2020', during a Public Hearing held in Brussels.

As a result, the EESC Own-initiative opinion, released in December, included specific language that takes into account the needs of people living with a RD. The report recommends that the EC provides guidance to MS on disability assessment, to ensure persons with rare conditions or multiple impairments are not overlooked. It also recommends that the EC pushes for persons with disabilities to be entitled to the same flexible working arrangements and leave of absence provided to parents by the Work-life Balance Directive.

Throughout 2019, EURORDIS also continued to strengthen its cooperation with the European Disability Forum (EDF), engaging in regular exchanges about common priorities.

1.6 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 2019, **Rare Barometer Voices**, the panel of rare disease patients who answer on a regular basis to EURORDIS' surveys reached more than 11000 patients.

A new survey on rare disease patients' experience of treatment was disseminated from March to May 2019. 7494 respondents in total answered the survey, representing 1354 diseases. This survey aimed to measure access and availability of treatment from a patient's point of view and to see how the situation on access to therapies has evolved as compared to 2017. Patients were also invited to share their experience and satisfaction regarding treatments. Insights from this survey helped us to build and strengthen our positions to contribute to the evaluation of EU orphan drug regulation and the wider EU regulatory framework and to propose practical and implementable solutions to accelerate the development of, and timely access to all, rare disease treatments and therapies. Public report of the survey will be available 2020 mid-year.

The programme also initiated a pilot project in collaboration with four European Reference Networks

to develop a centralised patient feedback mechanism across the 24 European Reference Networks. This project aimed to measure the evolution of patients' and caregivers' experience with the care they receive over time and across Europe and to provide information in order to align strategic decisions and operational delivery of the ERNs with patients' needs and experiences, specifically the development of healthcare pathways and treatment protocols. Developing a common mechanism to gather patient's feedback across ERNs will help to avoid duplication of efforts through pooling of resources (human, financial, tools such as software, etc.) which will result in economies of scale while ensuring robust, comparable and independent validated data and results by using the same survey methodology across ERNs. This project also tests the possibilities to develop a scientifically validated questionnaire that captures patients' experience with care for the over 8000 rare and complex diseases and understand how to best recruit patients and administer the survey across the 24 European Reference Networks and across countries. Results of this pilot project will be publicly available second half of 2020.

In July 2019, the programme published **the results from the first multi-country survey on rare disease data sharing and protection in the Orphanet Journal of Rare Diseases** in collaboration with the School of Social Sciences of Cardiff University. Over 2,000 rare disease patients, family members and carers from 66 countries representing 600+ diseases responded to the survey. The published article also sets out a series of recommendations to inform and support policy makers, researchers, funders and patient organisations so they take into account patients' preferences when creating and implementing data-sharing initiatives.

2. PATIENT EMPOWERMENT:

Building the network & building capacities

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

2.1.1 Membership

73 new members joined EURORDIS in 2019. At the end of 2019, EURORDIS had 884 members in 72 countries, 43 of which are European countries, 28 being members of the European Union.

2.1.2 EURORDIS Membership Meeting 2019 Bucharest

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 2019 Bucharest took place 17-18 May 2019. It comprised the General Assembly, a plenary session and 5 capacity building workshops

- 1 How to make the best use of the Social position paper?
- 2 Why start a Community Advisory Board (CAB)
- 3 European Reference Networks – The Eastern European Countries Dimension
- 4 Rare 2030 Foresight Study (half day workshop)
- 5 Share and protect our health data! (half day workshop)

The day before the EMM, two satellite meetings took place: CNA meeting and ePAG workshop.

EURORDIS offered 39 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 52 national alliances, 37 of which constitute the CNA.

The CNA's main activities in 2019 were:

- + a) the preparation and coordination of the Rare Disease Day 2019 and Rare Disease Day Strategic Review
- + b) Meetings of two Working Groups: Small EU countries and Western Balkans
- + c) Integration of ERN at National level

In 2019, 2 CNA Workshops took place in Bucharest in May and Brussels in November, back to back with the Rare2030 Panel of Experts meeting

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 Brussels in November 2019, back to back with the Rare2030 Panel of Experts meeting to learn about and discuss the following items

- + ERN and ePAG update
- + Orphanet Prevalence Paper
- + Horizon Europe – DG Research

- + New European Commission & Parliament, EU Presidencies,
- + EU Fund – Table of negotiations
- + Rare Impact – better access to advanced therapies in Europe

EURORDIS continued for the 10th 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 2019, EURORDIS gave 18 European RD Federations financial support to help them organise their different meetings. A total of 34850 € was granted.

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 2019, **18 help lines participated in the activities, from 13 countries:** Belgium (RadioOrg), 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), Norway (Norwegian National Advisory Unit on Rare Disorders), Portugal (Linha Rara), Romania (NORO, Myasthenia Gravis Romania), Spain (SIO-Feder), Switzerland (Info Maladies Rares, Help Line Seltene Krankheiten), Hungary (Huferdis, 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)

The network conducted its 12th Annual Caller Profile Analysis in October 2019 and participated in discussions on how to announce results of a genetic test to the family, and on the governance of the network. EURORDIS organised a face to face meeting held in Paris in November 2019 with a main focus on the General Data Protection Regulation.

2.1.6 European Patient Advocacy Groups (ePAGs): Training Programme

In 2019, EURORDIS launched the EURORDIS Leadership School, delivered within the EURORDIS Open Academy, as comprehensive training programme for ePAGs on leadership, network management, healthcare and research (see more under section on EURORDIS Open Academy).



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 215 disease-specific communities created in partnership with 1000 patient groups and managed with the support of 420 volunteer moderators.

RareConnect has improved during 2019 the members' experience in different ways with new features like: a new badge for verified patient organizations, an improved search tool, and new ways for members to sort and save important information for them. The platform has also followed the steps to become GDPR-compliant with the implementation of the "right to be forgotten", and new protection and security measures. In addition to that, in 2019, the research platform of RareConnect, My Studies, was launched. The launch of My Studies was accompanied by the formal acceptance of the My Studies governance framework. Two pilot studies were launched. One was completed successfully, the second is an ongoing longitudinal study. The platform has gained momentum with one new study set to be launched in April 2020, one study to be launched in the summer of 2020, and two additional studies in REB submission.

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 the smooth running of webinars.

Webinars focus on providing policy updates, involving patients in consultations, providing capacity-building trainings for patient advocates. 13 webinars were organised in 2019 on a number of topics such as Rare Disease Day; the EURORDIS campaign on the European elections; using the results of the EURORDIS data sharing survey and others.

2.1.9 Training for patient advocates

EURORDIS OPEN ACADEMY

Through the Open Academy, EURORDIS empowers patient advocates to have the confidence and knowledge needed to bring their expertise to discussions on health care, research and medicine development.

In 2019, EURORDIS delivered four training programmes via the EURORDIS Open Academy, in a blended format, with online courses and face-to-face training sessions.

144 patient advocates, from 30 countries, and over 60 trainers participated in these training programmes in 2019.

At the end of 2019, 59 e-learning courses were available on the online platform of the EURORDIS Open Academy. The platform had then reached over 1100 registered users, from more than 100 countries.

EURORDIS Winter School on Scientific Innovation & Translational Research



Launched in 2018, the EURORDIS Winter School consists of a one 4,5-day face-to-face training, preceded by an online pre-training. 23 e-learning courses are also available. Provided by over 20 expert trainers, the Winter School deepens patient advocates' understanding of how pre-clinical research translates into real benefits for people living with a RD. Winter school alumni are empowered to effectively participate in discussions with researchers, policy makers and companies responsible for research.

30 patient advocates from 18 countries participated in the 2019 edition of Winter School, which covered important topics to support patient engagement in research.

"My patient advocacy genome was definitely edited this week. I am no longer the patient advocate who arrived last Monday", said one of the participants of this second edition of the EURORDIS Winter School.

The face-to-face training took place on 11-15 of March 2019, at the Imagine Institute, in Paris. The pre-training,

composed of e-learning courses and 2 webinars, took place from January to March.

Specific topics covered included genetics and diagnosis, genome editing tools, translational research, pre-clinical models, drug repurposing, IRDiRC and European Joint Programme activities, patient participation in research projects and European Reference Networks.

100% of the participants who responded to the evaluation questionnaires (rate of response of 83%) have found that the Winter School effectively builds the capacity of patient advocates on scientific innovation and translational research.

A Winter School Programme Committee was established in September 2019 to support the upcoming editions, from 2020 to 2022. This Programme Committee, composed of expert faculty members on diagnosis and research topics, will support the annual programme reviews aimed at improving the programme.

EURORDIS Summer School on Medicines Research & Development



The EURORDIS Summer School, initiated in 2008, aims to provide patient advocates and researchers with the knowledge and skills that they need to become experts in medicines research and development. It is made up of a 4,5-day face-to-face training session, preceded by an online pre-training. 36 e-Learning courses are also available online. The training covers all the topics of the medicines research and development pathway, bringing together patients, researchers and an expert faculty of over 20 trainers.

The Summer School 2019 face-to-face training took place on the 10-14th of June 2019, in Barcelona. The pre-training, from March to May 2019, included e-learning courses and 3 webinars.

27 patient advocates and 7 researchers attended this edition, representing 15 countries and 25 rare diseases.

100% of the participants who responded to the feedback questionnaires (rate of response of 88%) have found the topics of the Summer School relevant for their activity as patient advocate or researcher.



The knowledge I gained at the Summer School helped me see how to proceed in my advocacy efforts in the best way possible and helped me influence change.



Denica, North Macedonia

EURORDIS Summer School alumni

This year the Summer School was reviewed to include more interactive sessions during the onsite training as well as a half-day dedicated to actions that can be taken by the participants after the training.

The training topics in 2019 included therapeutic development, clinical trials, European Medicines Agency, Health Technology Assessment, pricing, reimbursement and the role of patient advocates.

In October 2019, a new Programme Committee has been established for the EURORDIS Summer School, to support the upcoming editions, from 2020 to 2022.

EURORDIS Digital School on Social and Digital Media



The pilot edition of the EURORDIS Digital School consisted of two days of face-to-face training. Provided by experts in digital and social media, this training aimed at empowering patient advocates to use digital communication tools to improve the strategic outreach and community-building capacities of their organisations.

The first edition of the EURORDIS Digital School took place on 8-9 October, at the Ågrenska Resource Centre for Rare Diseases, in Gothenburg.

20 patient representatives, from 14 countries, participated in this innovative training course to support patient organisations to use digital communication tools.

The training topics covered included understanding social and digital landscapes in a patient organisation context, creating empowered communities, building effective channels and campaigns, goal-setting for social media, creating videos and reaching the right people.

100% of the participants who responded to the feedback questionnaires (rate of response of 95%) **recommend the Digital School to other patient representatives.**



This Digital School pilot was reviewed at the end of 2019 based on the feedback of participants. Participants suggested that online and e-learning courses as well as more examples from the community would be of great value. These suggestions were taken into account to develop the 2020 edition.

EURORDIS Leadership School on Healthcare & Research





The EURORDIS Leadership School was launched in 2019, as a comprehensive training programme for ePAG advocates on leadership, network management, healthcare and research. This pilot blended training programme included 6 months of online training and a 2 day face-to-face training. Over 20 expert trainers in leadership and communication were involved in this first edition of the Leadership School.

56 ePAG advocates, representing 17 countries, 21 European Reference Networks and over 45 rare diseases took part in the first EURORDIS Leadership School.

93% of the participants who responded to the feedback questionnaires (rate of response of 90%) **found that it effectively builds their capacity on leadership and on topics related to ERNs.**

"It made me think a lot about my style of positioning myself as a representative of an ERN", said one of the ePAG advocates who attended the Leadership School.

The online training, composed of a total of 20 webinars, took place April to October 2019. The face-to-face training took place on the 26th-27th of November 2019, in Barcelona.

The training topics included communicating with impact, creating authority and legitimacy, healthcare pathways, clinical guidelines, digital health, research and therapeutic development.

The participants were divided in two groups, with distinct training pathways: the New Leaders Group, of ePAG patient advocates who represent the wider patient community in ERNs; and the Advocates Group, of ePAG patient advocates active in their disease area.

This pilot programme was reviewed in December 2019, based on the feedback of the participants in view of preparing the 2020 edition. The suggestions from participants included reducing the number and frequency of the webinars and advancing the date of the face-to-face training.

2.2 Raising Awareness & Informing

2.2.1 Rare Disease Day 2019

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 12th edition of the day, Rare Disease Day events took place in over 100 countries and regions on every corner of the globe. 8 new countries had events this year – Dominican Republic, Lesotho, Montenegro, Namibia, Niger, Qatar, Sri Lanka and Zambia. 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 'Bridging health and social care'. The slogan was: Show Your Rare, Show You Care. The Rare Disease Day website (rarediseaseday.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 for Rare Disease Day was launched in 2018 and continued in 2019. 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 with the hashtag #ShowYourRare. The campaign's simple and interactive call to action was popular around the world. #RareDiseaseDay trended in 7 countries on Rare Disease Day.

EURORDIS updated the Show Your Rare campaign video for 2019 in addition to producing 3 new patient testimonials with 3 national alliance partners in order to share stories from the daily life on families affected by a rare disease. The 3 videos featured Karlo from Croatia, Filip from Romania and Lorena from Spain. The videos had over 138 000 views on social media and youtube and were translated into 35 languages.

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 Alliances 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.



Rare Disease Day Strategic Review

EURORDIS carried out a strategic review for Rare Disease Day in preparation for the next 10 years to maintain a campaign that creates positive change for people living with a rare disease. The strategic review comprised three phases and included internal and external audits, presentations, and meetings based on research and behavioural science methods.

The results and recommendations from the strategic review were presented to EURORDIS in March 2019 and to

the Council of National Alliances in a face-to-face meeting in May 2019 and via an online webinar.

Since March 2019 EURORDIS and the partner National Alliance have been working to define the long-term cause of the campaign. Through a wide consultation with rare disease patient organisations around the world it was collectively decided that the campaign will advocate for increased equity for people living with a rare disease.

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 550.000 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 2019 we completed the user research phase of the new eurordis.org website and moved into the graphic design phase. The agency built a set of functional front end templates which is going to be followed by backend development in 2020.

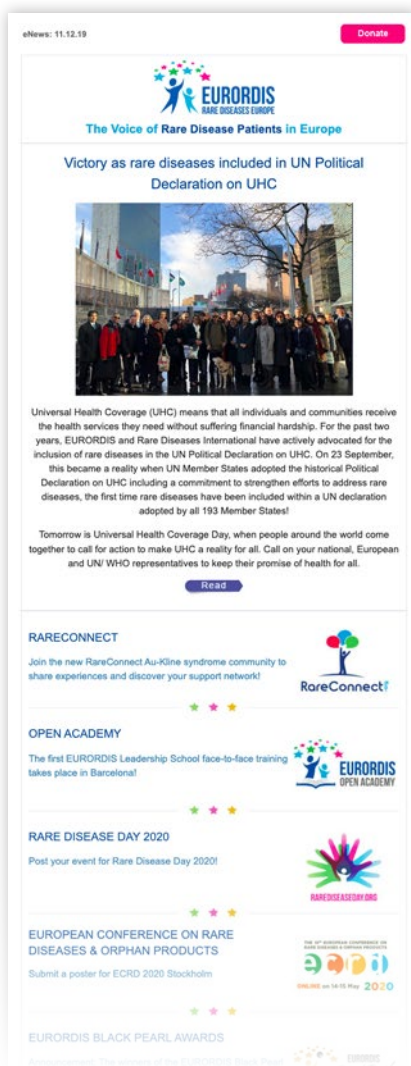
2.2.3 eNews & Member News

eNews

The EURORDIS eNews is a 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. Content is also made available via EURORDIS Facebook and Twitter.

Throughout 2019, 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 2019 included: Victory as rare diseases included in UN Political Declaration on UHC; Carpe diem - Noémie's story of living with amyotrophic lateral sclerosis; Pledge support for rare diseases ahead of the 2019 European elections.



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 2019, 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.



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 2019 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.



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 2019. There were hundreds of nominations, representing over 30 different countries worldwide.

The winners of the EURORDIS Awards 2019 were:

+ Young Patient Advocate Award: Laëtitia Ouillade (France)

For her exceptional advocacy work, in particular through the AFM-Téléthon in France, to raise awareness of the needs of people living with a rare disease among a wider audience.

+ European Rare Disease Leadership Award: Prof. Till Voigtländer (Austria)

For his outstanding leadership and commitment to the rare disease community and the positive impact he has made on rare disease policy both in supporting national advocacy actions in Austria with Pro Rare Austria and on a European level as Co-Chair of the European Reference Networks Board of Member States.

+ Policy Maker Award: Dr Edmund Jessop (UK)

For his outstanding work and support of the rare disease community through his dedication to patient advocacy and tackling rare diseases as a public health issue. As public health lead of the National Commissioning Group and author for the reform of highly specialised care in the UK, his role was of huge value in protecting the rare disease services at a time of important reform with the establishment of NHS England.

+ Scientific Award: Prof. Philip Van Damme (Belgium)

For his exceptional achievements in the field of amyotrophic lateral sclerosis (ALS) research as well as his outstanding support and care for ALS patients through his collaborative work with the patient organisation ALS Liga Belgium.

+ EURORDIS Volunteer Award: Richard West, Behçet's Syndrome Society (UK)

For his admirable commitment of over 20 years as a dedicated advocate of rare disease issues on behalf of the Behçet's community and his longstanding supportive partnership with EURORDIS.

+ EURORDIS Volunteer Award: Russell Wheeler, Leber's Hereditary Optic Neuropathy (LHON) Society (UK)

For his exceptional work as a patient advocate for all rare eye conditions through LHON Society and as a patient board member of ERN-EYE, and for all rare diseases as one of EURORDIS' longstanding volunteers.



+ EURORDIS Members Award: ALS Liga Belgium (Belgium)

For ALS Liga Belgium's great work in providing services and support that have significantly enhanced the quality of life of people living with amyotrophic lateral sclerosis (ALS) across Belgium and Europe.

+ Company Award for Innovation: Chiesi (Italy)

For Chiesi's strong commitment to rare diseases, its support to policy development, as well as its strong pipeline for bringing to market a wide number of treatments, including products for extremely rare diseases.

+ Company Award for Patient Engagement: The HERCULES Project (Pfizer Inc, PTC Therapeutics, Roche, Sarepta Therapeutics, Solid Biosciences, Summit Therapeutics, Wave Life Sciences and Duchenne UK)

For the collaborative effort of all the Companies involved in the HERCULES Project, bringing together patient organisations and industry to support access to new treatments for Duchenne Muscular Dystrophy (DMD). The Award also recognises how the HERCULES Project has set a model example and has the potential to encourage similar initiatives across other rare diseases.

+ Company Award for Health Technology: Air Liquide Medical Systems (France)

For Air Liquide's longstanding commitment to the development of life-changing respiratory devices and services, benefiting the lives of many people living with a rare disease at an international level.

+ International Media Awards

Visual and Audio Media Award: Anne-Dauphine Julliand (France)

For her inspiring documentary film, Et Les Mistrals Gagnants, which raises awareness through following the lives of five children each living with a different rare disease.

Written Media Award: Bojana Miroslavljević (Serbia)

For her specialised journal for rare diseases 'Word for Life', which has brought the needs of people living with a rare disease to the attention of a wider audience in Serbia and beyond.

+ Lifetime Achievement Award: Michael Griffith (Ireland)

For his exceptional work and vast achievements as co-founder of Fighting Blindness, founder of Debra Ireland and the Medical Research Charities Group, IPPOSI and Rare Disease Ireland. He has been a central figure in making a true and lasting difference for people living with a rare disease through his collegiate approach to advancing and funding medical research and providing platforms that represent the patient voice and improve patient access.



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.

In 2019, EURORDIS has worked with the ePAG advocates to develop a shared understanding of their role in the ERNs and has worked closely with the ERN project managers, Coordinators and advocates to increase the impact of ePAG advocates involvement in the different activities.

A priority area in 2019 has been to facilitate the exchange of good practices on ePAG advocates engagement in the ERNs. The advocates were invited first to describe their good practices filling in a template, with a view to register the experience so that other advocates could eventually replicate them. 7 different good practices were presented by the ePAG advocates in teleconferences and in a face to face meeting that was organised in May in Bucharest. The factsheets of the good practices and recordings are available [here](#).

This year we have also started to support the ePAG advocates in the development of Patient Journeys as a tool to collect and summarise the care needs and expectations of the patient community. These journeys allow them to share relevant information with the ERN clinicians and engage with them in discussing their needs with a view to inform ERN activities such as the development of standards

of care, clinical decision support tools, care pathways, training and education, etc. In 2019, we supported the development of patient journeys for 46 different rare diseases.

Finally, in 2019 we have launched quarterly ePAG induction calls targeted at providing an overview of ERNs and the role of ePAG advocates to the ePAG advocates. The first induction call was held in September 2019 and the calls will continue to be organised on a quarterly basis throughout 2020.

EURORDIS ePAG managers have organised more than 100 conference calls with the individual ePAG groups. We also supported onsite the ePAG advocates participating in 15 ERN meetings throughout 2019. In addition, the ePAG Steering Committee has had 6 calls to discuss strategic topics of relevance to the 24 groups and the EURORDIS transversal ePAG Research WG has had 3 calls to get updates and discuss developments in this area. In addition, we have organised an all-ePAG face-to-face meeting and an ePAG f2f Steering Committee meeting.

The all-ePAG face-to-face meeting took place on the 16th of May in Bucharest. The meeting served various purposes; it was the occasion for ePAG advocates to learn from each other and build their network with patient advocates active in other European Reference Networks, but it was also an opportunity for the ePAG advocates to interact with members from the RD National Alliances during the coffee and lunch breaks. The meeting was structured around five good practices that were developed and presented by eight ePAG advocates. 54 ePAG advocates from 20 European Reference Networks (ERNs) attended this meeting.

The ePAG f2f Steering Committee meeting took place on the 20th September 2019 at the EURORDIS offices in Brussels. 20 ePAG advocates from 18 ERNs attended this meeting. The main topic discussed was the development of an impact assessment framework to measure the impact of ePAG advocates involvement in the ERNs. Participants developed the change narrative linked their role in the ERNs and identified the final goals, intermediate

objectives and the activities required to achieve them. To continue with the work initiated, a Task and Finish Working Group was appointed. This group identified and developed a set of measures that they have presented to the larger ePAG Steering Committee in subsequent calls. The work to refine this set of measures to be collected across all 24 ePAGs will continue in 2020 with a view to have the assessment framework ready by the end of 2020.

3.1.2 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' role in IMI-PARADIGM has led the development of important deliverables such as recommendations to help stakeholders and their organisations identify the required capabilities for patient engagement. These recommendations aim at enhancing system readiness across stakeholder organisations and ease the path towards operationalizing meaningful engagement.

In addition, EURORDIS is involved in the co-development of a monitoring and evaluation (M&E) framework to evaluate patient engagement practices and towards demonstrating the return on engagement and the value that patient engagement brings to product development for developers, patients and society. This M&E framework



is intended to be used by all stakeholders who can adapt it to their own specific needs. The EURORDIS EUROCRAB programme is serving as a case study to develop suitable metrics indicators for both patients and industry in a Community Advisory Board (CAB) context.

In October, EURORDIS organised the 28th Round Table of Companies on a New PARADIGM of Meaningful Patient Engagement in the Life Cycle of Medicines. This workshop provided valuable information to its participants with the following objectives:

- + **Understand** which good practices EURORDIS promotes and implements for working with patients to develop therapies for rare diseases
- + **Understand** the objectives of PARADIGM and how to enable meaningful patient engagement in the product life-cycle as a whole
- + **Reflect** on the return on engagement as a win-win process between developers and patients
- + **Learn** about patient expectations with regards to the value of patient engagement (financial value, quality, representativeness)
- + **Learn** how to implement a Monitoring and Evaluation Framework in your patient engagement activities
- + **Understand** and use metrics for evaluating Community Advisory Boards (CABs)
- + **Reflect** and raise awareness on the management of competing interests in a multi-stakeholder environment

3.2 Patient Engagement in Holistic Care

EURORDIS reinforced its focus on holistic care, mainly through the dissemination of its **position paper on "Holistic Person-Centred Care"**, published in May, and the launch of its **new Social Policy Action Group (SPAG)**.

The SPAG, launched in April 2019, is a group of **volunteer patient advocates** who disseminate and contribute to the positions of EURORDIS and its members, **advocating for holistic and integrated care**.

EURORDIS also continued to **support the European Network of Resource Centres for Rare Diseases – RareResourceNet**, which aims at **advancing holistic high quality care** for people living with RD across Europe. In **February 2019, EURORDIS joined the Board of Directors of RareResourceNet**, at its general assembly, at the Frambu Resource Centre in Norway. Raquel Castro, EURORDIS Social Policy Director, was nominated by the EURORDIS Board of Directors to undertake this role.

3.2.1 Promote adequate social services and policies

EURORDIS continued to engage with social services and social policy makers, to **promote the access of people living with a RD to quality and adequate social services and policies**. This was further reinforced with the launch of the position paper on "Holistic Person-Centred Care", which includes a specific pillar on this priority.

In September, Dorica Dan, member of the EURORDIS Social Policy Action Group (SPAG) **presented at the European Commission Seminar on Social Innovation for Social Services**, bringing forward results of the INNOVCare project and raising awareness for the importance of adequate and quality social services for people living with a RD.

A week later, Gabor Pogány, also member of the EURORDIS SPAG, presented at the **e-Rare Workshop on "How Social and Human Sciences research can improve health care implementation and everyday life of people living with a RD and their families"**. The presentation included **EURORDIS' recommendations on socio-economic research** (from the Position Paper on Holistic Care) and a **list of key social research needs**, based on EURORDIS' position paper, surveys and social projects.

3.2.2 Promoting integrated care: bridging health and social care

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

In April 2019, EURORDIS **presented the results of the INNOVCare project at the 19th International Conference on Integrated Care**, attended by 1300 experts from 60 countries. On behalf of the project's consortium, EURORDIS **shared the case management good practices** tested in the project and **its successful results**.

The case management pilot implemented within the INNOVCare project, at the NoRo resource centre in Romania, has indeed **increased the coordination between care providers, beneficiaries' level of information on their disease, their rights and available services as well as their capacity to manage their own care**. Additionally, there was a **reduction in the burden faced by carers**.

Throughout the year, EURORDIS continued to work in **collaboration with the International Foundation for Integrated Care (IFIC)** as volunteer moderator of its **Self-Management and Co-production Special Interest Group**. EURORDIS **attended and spoke at the Special Interest Group workshop** held during the 19th International Conference on Integrated Care, in April.

2019 was also a year for EURORDIS to **continue seeking opportunities to promote key actions in integrated care within European projects and initiatives**, through the involvement in European calls for proposals.

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-

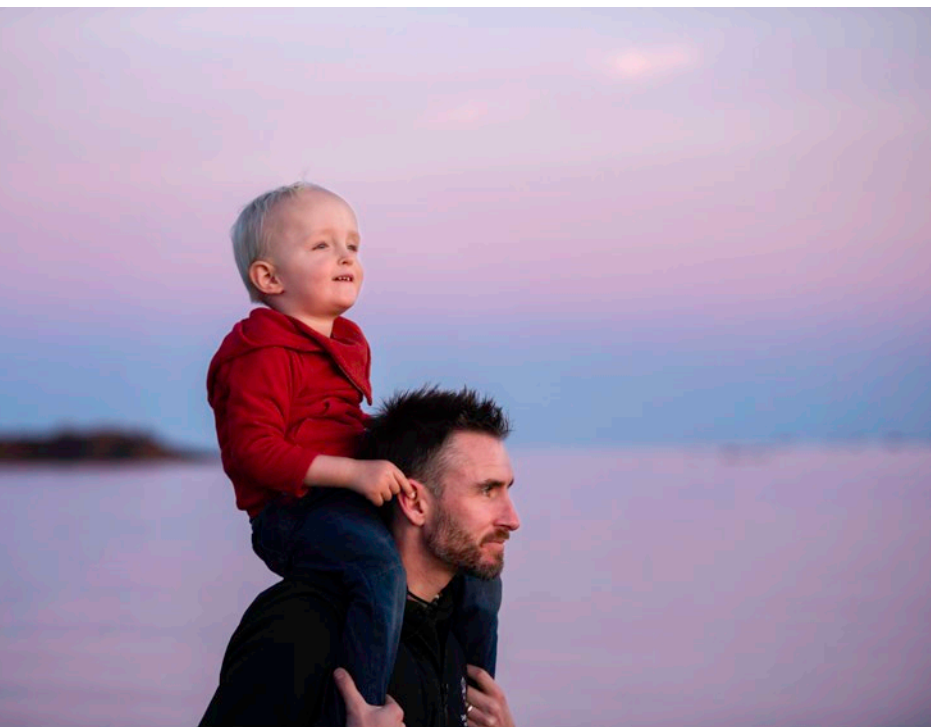


Chair of the TSC in which Yann Le Cam, CEO of EURORDIS, is also a Member and immediate past-Chair. As Vice-Chair of the TSC, Virginie Hivert is therefore also a member of the Operating Committee. EURORDIS will continue to actively promote opportunities for patient representatives to get involved as Task Forces and Working groups are being launched and open for expression of interest during the year.

Task Force 'Orphan Drug Development Guidebook': In 2019, 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 'Clinical Research Networks for Rare Diseases': The objective of the Task Force are threefold: 1) to map and analyse the existing ecosystem of national/ supranational clinical research networks, 2) to develop policy recommendations on guiding principles for an international framework of collaboration of these networks in respect to best practices, interoperability, tools and common goals and 3) to develop relevant recommendations for funders based on gaps identified through the mapping exercise. Following a public call for expressions of interest, EURORDIS supported the nomination of a patient representative to join this TF which was launched in October 2019 and started to discuss the scope of its activities via 2 teleconferences.

Task Force 'Identification of barriers to patient participation in RD research and recommendations to remove them': The task force, led by the Patient Advocacy Constituent Committee of IRDiRC, was approved back in July 2018 with the general goal of exploring barriers to patient participation in RD research and develop recommendations to remove them. The TF has held six conference calls since its launch but several issues are holding back its progress. Therefore, following the last PACC meeting that took place in November 2019 during the IRDiRC Consortium Assembly, it was decided to transform the TF into a working group which will first conduct a literature review on the topic in order to produce a state of play document. This will serve as a preparatory work to determine the need for further work and future directions.

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

Within the Joint Translational Calls of the European Joint Programme on Rare Diseases, patient organisations are eligible to apply as partners within the consortia submitting a research proposal since 2019. Indeed, patient involvement is strongly encouraged with the possibility for patient organisation to request a budget in line with their respective roles and responsibilities within the proposed research project. Topics for these annual calls vary. For example, the 2019 call asked for "Research projects to accelerate diagnosis and/or explore disease progression and mechanisms of rare diseases". 22 consortia have been funded in this first call involving 26 countries, 140 research teams and 17 patient advocacy organisations. The list of funded projects in 2019 are available here : <https://www.ejprarediseases.org/index.php/funded-projects/>.



In view of the experience of the first call, it became evident that a working group should be established to better describe the role and expectations of patient involvement in research proposals, provide guidance to applicants based in part on concrete examples of successful patient engagement in research. EURORDIS communicated an expression of interest to take part in this new working group that was launched in January 2020. The guidance to be developed will support fruitful partnerships between scientists and patient organisations, leading the way for a systematic patient-centered research, fostering a culture of partnerships and contributing to an improved understanding of the added value of patient engagement in research for the whole RD community.

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.

The EURORDIS EuroCAB programme supports 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.

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 ad hoc working groups all along the year.

EURORDIS supports the **work of patient representatives** in the COMP, PDCO, PRAC; supports the **participation of regular/ad hoc experts** to the COMP, PDCO, PRAC; 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 2019, EURORDIS selected and, proposed or endorsed, **patient representatives** to be appointed to the PDCO.

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



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

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, the **strategic and learning review meetings** held under the EU Presidency and to **internal workshops** organised by the Committees such as the workshop on the role of registries monitoring cancer therapies based on genetic molecular features.

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

Last year 46 patients were identified, finally, 41 were involved. Since mid-2019 the EMA has incorporated 'mentors' who can participate together with the identified patient representative. This allows experienced patient representatives (mentors) to help, assist, and complement other patient representatives. Since then, 5 mentors have been involved giving more representativeness and broader views. We have only had 5 drop out, and the main reasons were delays in submission of paperwork (CV, DOI, Expert form), procedure withdrawals by the company and conflict of interest. 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 Public Engagement Team have been strengthened and optimised by holding a monthly conference call following an internal

meeting between the EMA public engagement team and scientific advice team (scientific officers and assistants).

In summary, the time spent for the whole package per month is about 60 hours per month.

Of note, on several occasions, COMP Members who are also SAWP Members have expressed satisfaction and noticed an increase in the number of patients involved in Protocol Assistance dossiers and the quality of their input in the procedures.

In 2019: Applications responding to the **European Commission's Call for Expression of Interest** were submitted for the **PDCO** membership, after an internal Call for Expression of Interest within its Members. Dimitrios Athanassiou (who is currently a Member of PDCO) has

been proposed as EURORDIS candidate. Her letter of endorsement has also been issued for Tomasz Grybek.

The patients' representatives and staff involved with Scientific Committees and PCWP (Patients and Consumers Working Party) are Members of the **EURORDIS' Therapeutic Action Group**. 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. François Houyez, member, and Russell Wheeler (volunteer, Leber Hereditary Optic Neuropathy UK Society,) appointed as alternate by EURORDIS Board in September 2019. In 2018 and 2019, due to the EMA relocation, most PCWP face-to-face meetings were cancelled or replaced by e-meetings. Two PCWP face-to-face meetings were held (24 September and 20 November with all eligible organisations) combined with the Healthcare Professionals' Working Group and two e-meetings (3 May and 26 June).

EURORDIS worked in particular on the STAMP initiative on drug repurposing, and whether patients and/or healthcare professionals could be Champions, with questions to the EMA regarding possible conflicts this could create; The European Regulatory Science Strategy 2020-2025; The European web portal on vaccines with e-CDC; The response to the European Ombudsman inquiry into EMA's pre-submission activities.

EURORDIS activities in pharmacovigilance in 2019

- + August 2019: EMA survey on public communication of the nitrosamine contamination in sartan medicines
- + CIOMS working Group in Geneva, 1-2 May, and on 16-17 October in Basel, on guidelines on Patients Involvement in the Development and Safe Use of Medicines
- + What does the future hold for patients? 12th Pharmacovigilance Conference, Medicines for Europe, 30 January 2019, London
- + The Vigilant patient - Moving Forward in EU Pharmacovigilance, DIA 2019 Global meeting, San Diego, 24 -25 June 2019
- + Causes and Effects of Therapeutic Interventions: a propensity-based approach. Erice, 12-15 September 2019
- + 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

In 2019, 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 (having participated to scientific advice or scientific advisory group meetings).

In addition, the EMA can contact EURORDIS when organising Scientific Advisory Group meetings with external expert, to inform the benefit/risks evaluation.

This is when the EMA cannot find an expert patient with its own resources.

In 2019, the EMA asked EURORDIS on three occasions:

- 1 scientific advisory group meeting on severe aplastic anaemia 7 October
- 2 scientific advisory group meeting on Sickle Cell Anaemia 11 September 2019
- 3 scientific advisory group meeting on seizures associated with Lennox-Gastaut syndrome or Dravet syndrome mid-June 2019

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.

Two face-to-face meetings of the EURORDIS HTA Task Force were held in 2019. The Task Force provided four responses to EUnetHTA consultations, and participated in the EUnetHTA Annual Stakeholder Forum in Amsterdam.

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

The Task Force represent a group of 18 volunteers who are trained (via the EURORDIS Summer School) and active in issues concerning therapeutic development of medicines for rare diseases. The Task Force supports and/or advises the EURORDIS representatives who participate in EMA Scientific Committees and Working Parties, or in the European Network of Health Technology Assessment (EUnetHTA) and the HTA Network (DG Sante). It is consulted on papers prepared by EURORDIS.

early access programmes; comments on the European Vaccination Information Portal. DITA task force members also presented EURORDIS views/data in the following conferences (not exhaustive): What does the future hold for patients? 12th Pharmacovigilance Conference, Medicines for Europe, 30 January 2019, London; CIOMS working Group XI on Patient involvement in the development and safe use of medicines, 1-2 May 2019, Geneva; Time to access? Or time to assess? Making Early Access for Patients



One meeting of the DITA TF was held on 10 December where the main topic was the launch of the new task force and setting of priorities for its mandate 2019-2022. The agenda included Drug Repurposing and the European Commission initiative by STAMP, an update on medicines shortages, the EMA Regulatory Science Strategy 2025, EMA training activities for patient advocates, ICH E8 guidelines on General Considerations for Clinical Trials, a progress report on EUROCAE programme (community Advisory Boards) and information on the proposed Regulation for European Cooperation on HTA.

Members of the task force were also consulted on, among other topics: HMA-EMA Joint Big Data Taskforce; HMA subgroup on Timely Access Compassionate Use Programmes Survey regarding information provided on

Happen and The Vigilant patient - Moving Forward in EU Pharmacovigilance, DIA 2019 Global meeting, San Diego, 24-25 June 2019; Shortages of medicines, how do they affect patients? Cost Action 15105, Vilnius 10-11 September 2019.

Review of EMA documents for the public or contributions to EMA consultations:

European Public Assessment Reports for the Public now called Medicine Overviews (1), Package Leaflets (4). Since 2007 when the procedure to review EPAR summaries now called Medicine Overviews and Package leaflets was established for authorised medicines in the EU, 77 EPAR summaries and 120 PL were reviewed, for a total of 197 documents.

4. CROSS-CUTTING PRIORITIES

4.1 Governance

4.1.1 Annual General Assembly

The EURORDIS General Assembly was held in Bucharest on 17 May 2019. EURORDIS full members voted on five vacant positions on the Board of Directors. For the first time ever, EURORDIS was faced with the unprecedented situation of having a tie in the votes. In order to resolve the situation, the Board of Directors decided to add one more position to the Board thus welcoming the following members of the Board: Elizabeth Vroom, World Duchenne Organisation, UPPMD (Netherlands), Jayne Spink, Genetic Alliance UK (United Kingdom), Terkel Andersen, Danish Haemophilia

Society (Denmark), Lieven Bauwens, International Federation for Spina Bifida and Hydrocephalus (Belgium), Geske Wehr, Selbsthilfe Ichthyose e. V. (Germany), and Vlasta Zmazek, Rare Diseases Croatia (Croatia).

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



4.1.2 Partnerships with international organisations (MoUs)

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

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

NORD

The US Organization for Rare Disorders



CORD

The Canadian Organization
for Rare Disorders



JPA

The Japan Patients' Association



RVA

Rare Voices Australia



RPU

Russian Patients' Union



RADOIR

Rare Diseases Foundation of Iran



EURORDIS also has partnerships with a number of learned societies:

European Federation of Internal
Medicine (EFIM)



European Hospital & Healthcare
Federation (HOPE)



International Federation of Social
Workers Europe (IFSW-Europe)



European Society of Human Genetics (ESHG)



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



European Connected Health
Alliance (ECHAAlliance)





4.2 Human resources

4.2.1 EURORDIS Staff

The team comprised 47 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 6 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 2019.

6 new positions were created in 2019 (in order of appearance):

- + Project Senior Manager - Rare Impact, **Karolina Hanslik**, Brussels (limited contract)

- + Patient Engagement Manager - ERN & Healthcare / ePAGs, **Anne-Laure Aslanian**, Barcelona
- + Chief Operating Officer, **Michael Wilbur**, Paris
- + Corporate Relations Assistant, **Hlawulani Mkhabela**, Paris
- + Development Relations Senior Manager, **Brian Howard**, Paris
- + Patient Experience Survey Research Executive, **Jessie Dubief**, Paris (limited contract)

2 positions were stopped:

- + Social Policy Senior Advisor, **Annette Dumas**, Brussels (limited contract)
- + Web Communications Senior Manager & Rare Connect Leader, **Denis Costello**, Barcelona



4.2.2 EURORDIS Volunteers

In 2019, EURORDIS was privileged to rely on **437 volunteers** including **93 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 (93):

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

They are selected 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. Some of them can belong to two task forces, and sometimes three:

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 2019, it was composed of 47 full members: **28 volunteers** as well as 19 staff members (CEO, Directors and managers) involved in advocacy. The EPAC members have a mandate to represent EURORDIS.

TAG: Therapeutic Action Group

The TAG includes **2 EURORDIS volunteers** who are respectively member of the European Medicines Agency’s committee for orphan medicinal products and for paediatric drugs. These committees hold meetings every month over two to three days. The expertise required and involvement in terms of time are both significant.

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

In 2019, **23 volunteers** contributed to the DITA's work: 14 from January to August 2019, and 16 from September to December 2019 (7 being the same volunteers). The mandate of DITA volunteers was renewed in September 2019 with a new call for expression of interest.

Selected volunteers are trained (via the EURORDIS 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). The DITA Task Force is coordinated by two staff members.

Health Technology Assessment (HTA) Task Force

It is composed of **11 volunteers** and coordinated by two staff members, who also manage the DITA Task Force in order to ensure a good synergy between these two task forces. The HTA 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 one 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' advocates, 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 two ePAGs for each of the 24 ERNs. In 2019, **33 members of this ePAGs Steering Committee have expressed their interest in becoming EURORDIS volunteers**. The ePAGs–EURORDIS volunteers are coordinated by four 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.



4.3 Finance & Support Services

Finance and support services' activities in 2019 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.

Contract Grants

Renewed

- + Specific Grant Agreement (Operating Grant) for year 2019 (SGA FY2019), single beneficiary, DG Sante, 12 months

Ongoing

- + Advocacy and core activities, AFM-Téléthon, 2018-2021 (renewed for 2019)
- + E-RARE 3: For the extension and strengthening of the transnational cooperation on rare disease research funding organisations, Horizon 2020, 2015-2019

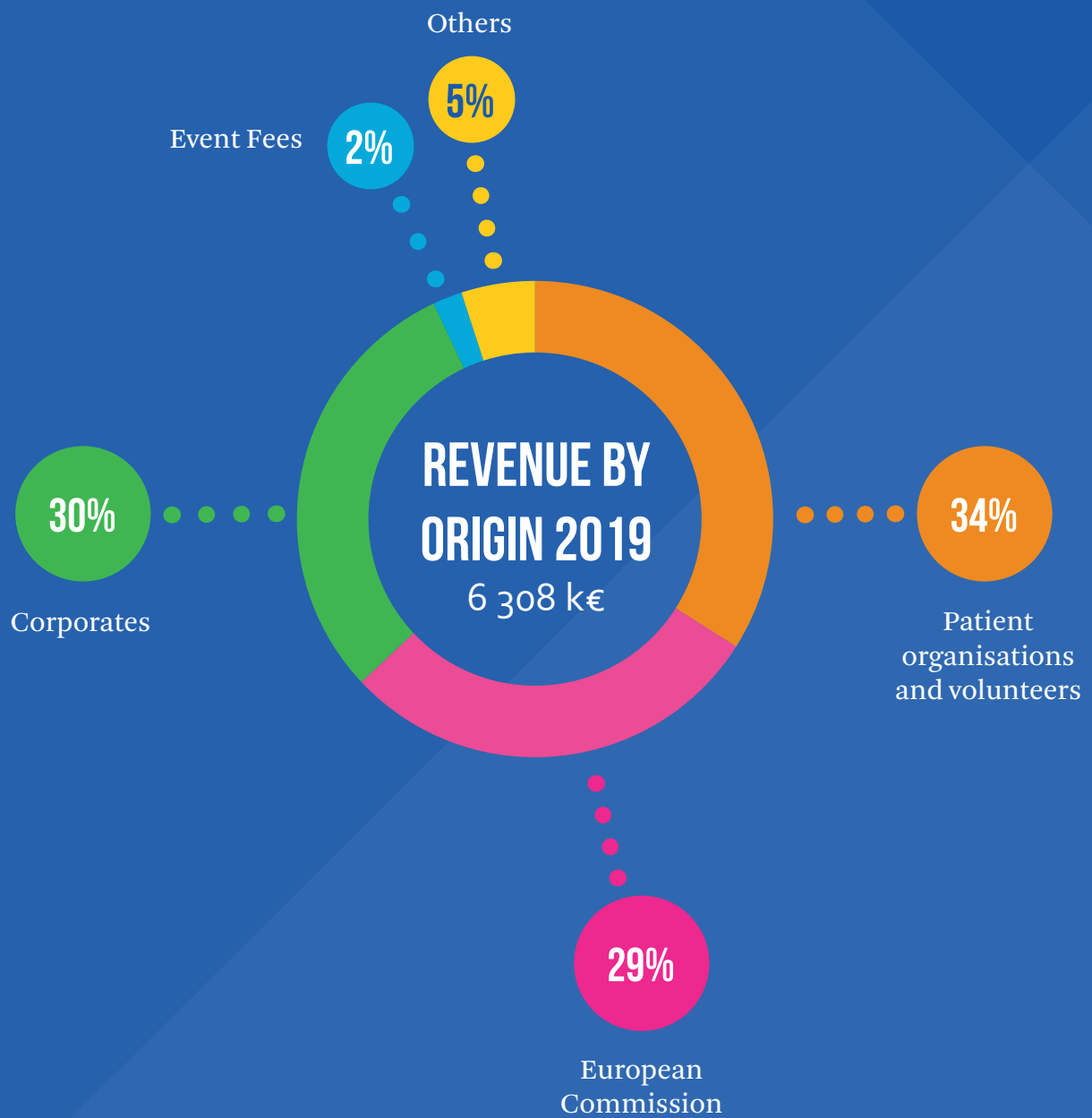
- + Framework Partnership Agreement 2018-2021 (Operating Grant), single beneficiary, DG Sante, 2018-2021
- + JARC: Joint Action on Rare Cancers, DG SANTE, 2016-2019
- + PARADIGM: Patients Active in Research and Dialogues for an Improved Generation of Medicines: Advancing meaningful patient engagement in the life cycle of medicines for better health outcomes., beneficiary, IMI-JU2, 2018-2020
- + Solve-RD: Solving the unsolved Rare Diseases, beneficiary, DG Research, 2018-2022
- + c4c: conect4children (Collaborative Network for European Clinical Trials For Children), beneficiary, IMI-JU2, 2018-2024

New

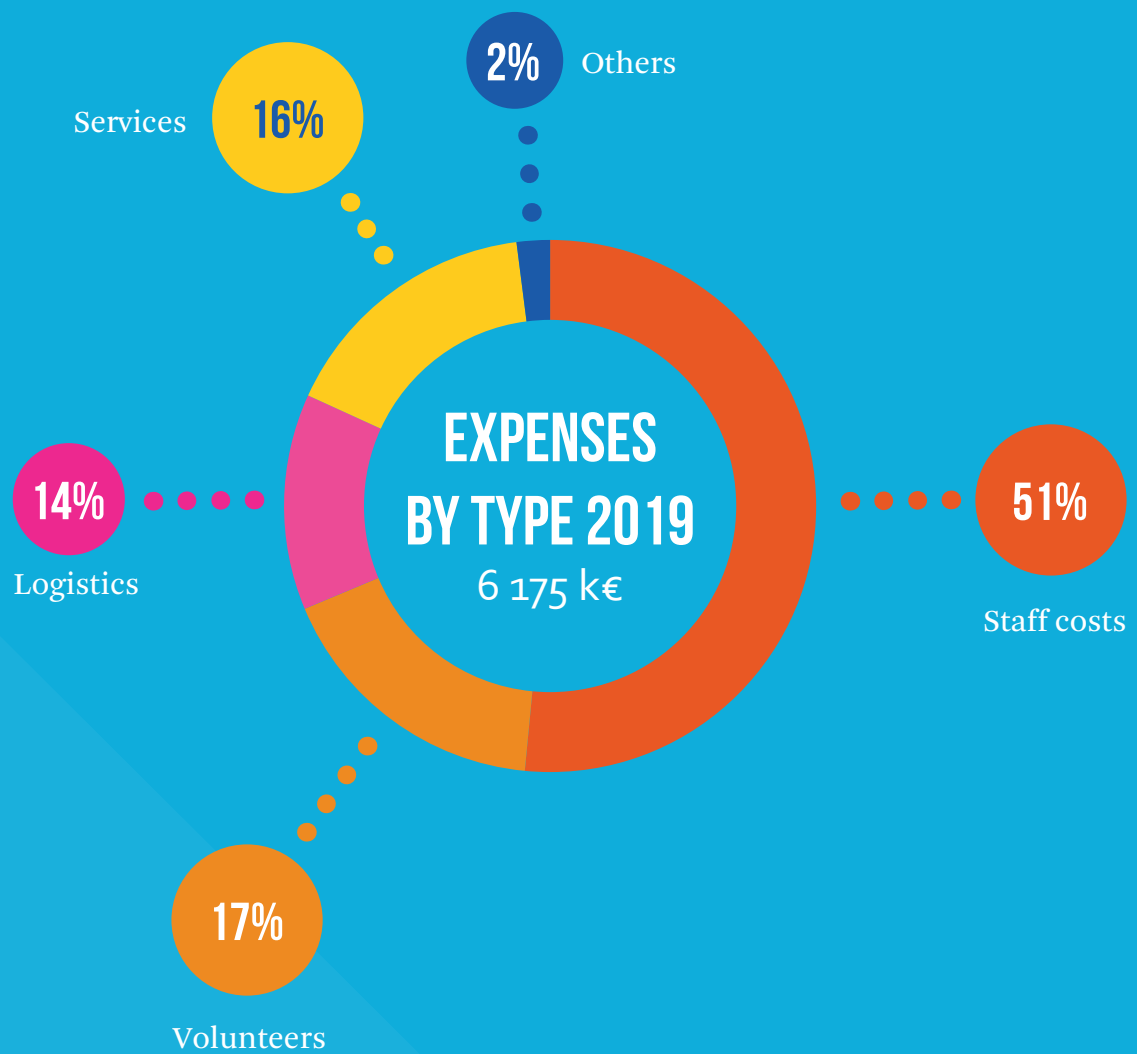
- + EJP RD: European Joint Programme Cofund, Horizon 2020, 2019-2024
- + Rare 2030: Foresight in Rare Disease Policy, European Union Pilot Projects and Preparatory Actions Programme, 2019 – 2021
- + RD-Code: Rare Diseases Code, EU Third Health Programme, 2019-2021
- + HTx: Next Generation Health Technology Assessment (HTA), Horizon 2020, 2019-2024



REVENUE 2019



EXPENSES 2019



BOARD of Directors

May 2019 - May 2020

PRESIDENT

Mr Terkel Andersen Danish Haemophilia Society

 Denmark

DIRECTORS

Ms Alba Ancochea Spanish Federation of Rare Diseases (FEDER)

 Spain

Mr Lieven Bauwens International Federation for Spina Bifida and Hydrocephalus

 Belgium

Ms Simona Bellagambi UNIAMO - Rare Diseases Italy

 Italy

Ms Avril Daly
Vice-President Rare Diseases Ireland

 Ireland

Ms Dorica Dan
Officer Romanian Prader Willi Association

 Romania

Ms Birthe Byskov Holm Rare Diseases Denmark

 Denmark

Mr Alexandre Mejat AFM-Téléthon

 France

Ms Jayne Spink Genetic Alliance UK

 UK

Mr Dimitrios Synodinos
Treasurer Tuberous Sclerosis Association

 Greece

Ms Elizabeth Vroom World Duchenne Organisation

 Netherlands

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

 Germany

Ms Vlasta Zmazek Rare Diseases Croatia

 Croatia

MEMBERS of EURORDIS

FULL MEMBER●
ASSOCIATE MEMBER●

ALBANIA

RARE DISEASE ASSOCIATION ALBANIA (SHOQATA E SEMUNDJEVE TE RRALLA)

ALGERIA

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

ARGENTINA

FUNDACION GEISER

ARMENIA

DOCTORS AND CHILDREN HEALTH CARE
NEUROHEREDITARY DISEASES CHARITY ASSOCIATION

AUSTRALIA

CYSTIC FIBROSIS AUSTRALIA
GENETIC ALLIANCE AUSTRALIA
GENETIC SUPPORT NETWORK OF VICTORIA
RARE VOICES AUSTRALIA

AUSTRIA

ANGELMAN VEREIN ÖSTERREICH
DEBRA INTERNATIONAL
HAND IN HAND FÜR TAY-SACHS & PALLIATIVKINDER
ICA ÖSTERREICH
NF KINDER – VEREIN ZUR FÖRDERUNG DER
NEUROFIBROMATOSEFÖRDERUNG ÖSTERREICH
NF PATIENTS UNITED
PHA AUSTRIA – INITIATIVE LUNGENHOCHDRUCK
PHA EUROPE
PRO RARE AUSTRIA, ALLIANZ FÜR SELTENEN ERKRANKUNGEN
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-TSC VZW
CONTACTGROEP MYELOOM EN WALDENSTRÖM PATIËNTEN
DEBRA BELGIUM VZW
EUROPEAN CHROMOSOME 11 NETWORK
EUROPEAN CMT FEDERATION
EUROPEAN HAEMOPHILIA CONSORTIUM
EUROPEAN IDIOPATHIC PULMONARY FIBROSIS & RELATED DISORDERS FEDERATION
EUROPEAN NETWORK FOR RESEARCH ON ALTERNATING HEMIPLEGIA
EUROPEAN POLIO UNION
EYE HOPE FOUNDATION
FEDERATION OF EUROPEAN SCLERODERMA ASSOCIATIONS
FEDERATION OF EUROPEAN WILLIAMS SYNDROME
FEDERG - FEDERATION OF EUROPEAN ASSOCIATIONS OF PATIENTS AFFECTED BY RENAL DISEASES
ICHTHYOSE BELGIQUE - ICHTHYOSIS BELGIË
INTERNATIONAL FEDERATION FOR SPINA BIFIDA AND HYDROCEPHALUS
MARFAN EUROPE NETWORK
MYELOMA PATIENTS EUROPE
RADIORG - RARE DISEASE ORGANIZATION BELGIUM ASBL/VZW
RARE DISORDERS BELGIUM
RELAIS 22 ASBL
SIOP EUROPE - EUROPEAN SOCIETY FOR PAEDIATRIC ONCOLOGY
SPIERZIEKTEN VLAANDEREN VZW
STEUNPUNT KINDEREPILEPSIE VZW
VASCULAR ANOMALY PATIENT ASSOCIATION
VLAAMS PATIËNTENPLATFORM VZW
VLAAMSE VERENIGING VOOR ERFELIJKE BINDWEEFSELAANDOENINGEN

BENIN

ALBINOS SANS FRONTIÈRES

BOSNIA AND HERZEGOVINA

ALLIANCE FOR RARE DISEASES OF REPUBLIC OF SRPSKA, BOSNIA AND HERZEGOVINA

BRAZIL

ASSOCIAÇÃO BRASILEIRA DE ENFERMIDADES RARAS
ASSOCIAÇÃO BRASILEIRA DE PARAMILOIDOSE
INSTITUTO VIDAS RARAS

BULGARIA

ASSOCIATION OF PEOPLE SUFFERING BY ACROMEGALY IN BULGARIA
BULGARIAN ASSOCIATION WILSON DISEASE
BULGARIAN CYSTIC FIBROSIS ASSOCIATION
BULGARIAN HUNTINGTON ASSOCIATION
BULGARIAN NATIONAL ALLIANCE OF PEOPLE WITH RARE DISEASES
INFORMATION CENTRE FOR RARE DISEASES AND ORPHAN DRUGS
NAS - NATIONAL ASSOCIATION SARCOIDOSIS BULGARIA
NATIONAL ASSOCIATION FOR CHILD SUPPORT CONGENITAL HYPOTHYROIDISM
NATIONAL ASSOCIATION OF THE PATIENTS WITH GROWTH HORMONE DEFICIENCY
NATIONAL ASSOCIATION OF SYRINGOMYELIA
NATIONAL GAUCHER ORGANIZATION
NATIONAL SCLEROSIS 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
PVNH SUPPORT & AWARENESS

CHINA

CHINESE ORGANIZATION FOR RARE DISORDERS

COLOMBIA

ASOCIACIÓN COLOMBIANA DE PACIENTES CON ENFERMIDADES DE DEPÓSITO LISOSOMAL
FOUNDATION DIANA GARCIA DE OLARTE FOR PID

CROATIA

DEBRA CROATIA
DRAVET SYNDROME 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

CZECH REPUBLIC

ASSOCIATION OF A ATYPICAL PARKINSONIAN SYNDROMES
CZECH ASSOCIATION OF MARFAN SYNDROME
CZECH HUNTINGTON ASSOCIATION
KLUB NEMOCNYCH CYSTICKOU FIBROZOU
META, ASSOCIATION OF PATIENTS WITH LYSOSOMAL STORAGE DISEASES
NARODNÍ SDRUŽENÍ PKU A JINÝCH DMP (CZECH PKU ASSOCIATION)
RARE DISEASES CZECH REPUBLIC (ČESKÁ ASOCIACE PRO VZÁCNÁ ONEMOCNĚNÍ)

DENMARK

22Q11 DENMARK
ADDISON FORENINGEN I DANMARK
BLÆREEKSTROFIFORENINGENS
CCHS DANMARK
DANISH APERT SYNDROME ASSOCIATION / DANMARKS APERTFORENING
DANMARKS BLODERFORENING / DANISH HAEMOPHILIA SOCIETY
EHLERS-DANLOS FORENINGEN I DANMARK
FORENINGEN AF MÖBIUSSYNDROM I DANMARK
FORENINGEN FOR ATAKSI / HSP
ICHTYOSIS ASSOCIATION IN DENMARK
IMMUN DEFEKT FORENINGEN
LUPUS EUROPE
MCADD-FORENINGEN
MITOKONDRIE-FORENINGEN I DANMARK
PATIENTFORENINGEN HAE DANMARK
PORFYRIFORENINGEN DANMARK - PORPHYRIA ASSOCIATION DENMARK
RARE DISEASES DENMARK (SJAELDNE DIAGNOSER)
THE DANISH OSTEOGENESIS IMPERFECTA SOCIETY

WILSON PATIENTFORENINGEN

XLH, ARVELIG RAKITIS (HEREDITARY RICKETS PATIENT ASSOCIATION)

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

HARSO - RARE DISEASE ALLIANCE FINLAND

HARVINAISET-VERKOSTO - FINNISH NETWORK FOR RARE DISEASES

INCLUSION FINLAND KVTL

RINNEKOTI FOUNDATION, NORIO CENTRE

SUOMEN VASKULIITTIYHDISTYS RY

FRANCE

AFMKT-FRANCE

AGIR POUR LA MALFORMATION LYMPHATIQUE EN ALLIANCE

ALLIANCE MALADIES RARES

ALLIANCE SYNDROME DE DRAVET

AMIS DE ADNP FRANCE

ASS. INTERNATIONALE DE DYSTROPHIE NEURO AXONALE INFANTILE ASSEDEA

ASSOCIATION ISIS

ASSOCIATION «LES PETITS COURAGEUX»

ASSOCIATION AMS-ARAMISE

ASSOCIATION ANÉMIES DYSERYTHROPOIÉTIQUES CONGÉNITALES

ASSOCIATION CONTRE LES MALADIES MITOCHONDRIALES

ASSOCIATION CRANIOPHARYNGIOME SOLIDARITÉ

ASSOCIATION DE PREFIGURATION DE LA FONDATION DENISE PICARD

ASSOCIATION DE SOUTIEN ET D'INFORMATION A LA LEUCÉMIE

LYMPHOÏDE CHRONIQUE ET LA MALADIE DE WALDENSTRÖM

ASSOCIATION DES AMIS D'ANNE-LORÈNE - SYNDROME D'AICARDI

ASSOCIATION DES GROUPES AMITIÉ TURNER

ASSOCIATION DES MALADES DU SYNDROME DE MCCUNE-ALBRIGHT

ASSOCIATION DES MALADES SOUFFRANT D'ANGIO OEDÈME PAR DÉFICIT

EN C1 INHIBITEUR

ASSOCIATION DES PANCRÉATITES CHRONIQUES HÉRÉDITAIRES

ASSOCIATION DES PATIENTS DE LA MALADIE DE FABRY

ASSOCIATION DES PERSONNES CONCERNÉES PAR LE TREMBLEMENT

ESSENTIEL

ASSOCIATION DES POIC

ASSOCIATION DU LOCKED-IN SYNDROME

ASSOCIATION DU NAEVUS GÉANT CONGÉNITAL

ASSOCIATION FRANÇAISE CONTRE L'AMYLOSE

ASSOCIATION FRANÇAISE CONTRE LES MYOPATHIES - TÉLÉTHON

ASSOCIATION FRANÇAISE DE LA FIÈVRE MÉDITERRANÉENNE FAMILIALE

ASSOCIATION FRANÇAISE DE LA MALADIE DE FANCONI

ASSOCIATION FRANÇAISE DE L'ATAXIE DE FRIEDREICH

ASSOCIATION FRANÇAISE DE L'OSTÉODYSTROPHIE HÉRÉDITAIRE

D'ALBRIGHT

ASSOCIATION FRANÇAISE DE L'OSTÉOGENÈSE IMPARFAITE

ASSOCIATION FRANÇAISE DE NARCOLEPSIE-CATAPLEXIE ET

HYPERSONNIE

ASSOCIATION FRANÇAISE DES DYSPLASIES ECTODERMiques

ASSOCIATION FRANÇAISE DES HÉMOPHILES

ASSOCIATION FRANÇAISE DES MALADES ATTEINTS DE PORPHYRIES

ASSOCIATION FRANÇAISE DES MALADIES HÉRÉDITAIRES DU RYTHME

CARDIAQUE

ASSOCIATION FRANÇAISE DES PATIENTS EN AUTO-TRAITEMENT

ASSOCIATION FRANÇAISE DES SYNDROMES D'EHLERS-DANLOS

ASSOCIATION FRANÇAISE DU GOUGEROT-SJÖGREN

ASSOCIATION FRANÇAISE DU LUPUS ET AUTRES MALADIES AUTO-

IMMUNES

ASSOCIATION FRANÇAISE DU SYNDROME DE CORNELIA DE LANGE

ASSOCIATION FRANCAISE 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 NIEMANN PICK

ASSOCIATION FRANÇAISE POUR LE SYNDROME DE GILLES DE LA

TOURETTE

ASSOCIATION FRANCAISE STURGE WEBER «VANILLE-FRAISE»

ASSOCIATION FRANCE VASCULARITES

ASSOCIATION FRANÇOIS AUPÉTT

ASSOCIATION FRANCOPHONE CONTRE LA POLYCHONDRIE CHRONIQUE

ATROPHIANTE

ASSOCIATION FRANCOPHONE DES GLYCOGÉNOSES

ASSOCIATION FRANCOPHONE DU SYNDROME D'ANGELMAN

ASSOCIATION GRANDIR

ASSOCIATION HEMOCHROMATOSE FRANCE

ASSOCIATION HISTIOCYTOSE FRANCE

ASSOCIATION HUNTINGTON FRANCE

ASSOCIATION HYPOPARATHYROIDISME FRANCE

ASSOCIATION ICTHYOSE FRANCE

ASSOCIATION INTERNATIONALE MALADIES KYSTES TARLOV

ASSOCIATION KOURIR

ASSOCIATION MARFANS

ASSOCIATION MÉDICALISTES

ASSOCIATION MICROPTALMIE 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 SURRÉNALES

ASSOCIATION SYNDROME DE KLEINE-LEVIN

ASSOCIATION SYNDROME DE MOEBIUS FRANCE

ASSOCIATION SYNDROME DE ROKITANSKY - MRKH

ASSOCIATION TANGUY MOYA MOYA

CHARCOT-MARIE-TOOTH FRANCE

CUTIS 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

EUROPEAN PATIENT ORGANISATION FOR DYSIMMUNE AND

INFLAMMATORY NEUROPATHIES

FÉDÉRATION DES MALADES DRÉPANOCYTAIRES ET THALASSÉMIQUES

SOS GLOBI

FITIMA EUROPE - FONDATION INTERNATIONAL TIERNO ET MARIAM

FOP FRANCE

FRANCE LYMPHANGIOLEIOMYOMATOSE

FRANCE LYMPHOME ESPOIR

GÉNÉRATION 22

GENESPOIR: ASSOCIATION FRANÇAISE DES ALBINISMES

GENIRIS

HTAP FRANCE

HYPOPHOSPHATASIE EUROPE

INCONTINENTIA PIGMENTI FRANCE

INFLAM'CEIL

LES AMIS DE REMI

LES ENFANTS DU JARDIN

LES PETITS BOURDONS

LIGUE CONTRE LA CARDIOMYOPATHIE

LIGUE NATIONALE CONTRE LE CANCER

LUPUS FRANCE

MED13L SYNDROME ASSOCIATION

MOSAÏQUES - ASSOCIATION DES «X FRAGILE»

NAEVUS GLOBAL

OUVRIR LES YEUX - OLY

PIERRE ROBIN EUROPE FOUNDATION

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 LYOSOMALES

VALENTIN - ASSOCIATION DES PORTEURS D'ANOMALIES

CHROMOSOMIQUES

VHL FRANCE

WHITE SUTTON FRANCE

X FRAGILE FRANCE

XTRAORDINAIRE

GEORGIA

GEORGIAN FOUNDATION FOR GENETIC AND RARE DISEASES

GERMANY

AHC-DEUTSCHLAND E.V.

AKTION BENNI & CO E.V.

ALLIANZ CHRONISCHER SELTENER ERKRANKUNGEN E.V.

ALPHA 1 NETZWERK EV

ARVC-SELBSTHILFE E.V.

ASOCIATIA ROMANA SPINA BIFIDA SI HIDROCEFALIE

BSHV FÜR KINDER, JUGENDLICHE UND ERWACHSENE MIT

SELTENEN, CHRONISCHEN SKELETTERKRANKUNGEN E.V.

BUNDESVERBAND DER CLUSTERKOPFSCHMERZEN-SELBSTHILFE-

GRUPPEN

BUNDESVERBAND FÜR ANGEBORENE GEFÄSSFEHLBILDUNGEN E.V.

BUNDESVERBAND KLEINWÜSCHSIGE MENSCHEN UND IHRE FAMILIEN E.V.
 BUNDESVERBAND POLIOMYELITIS E.V.
 CHARGE SYNDROM E.V.
 CHOLESTERIN & CO E.V.
 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 E.V.
 DEUTSCHSPRACHIGE SELBSTHILFEGRUPPE FÜR ALKAPTONURIE E.V.
 EHLERS-DANLOS SELBSTHILFE E.V.
 EUROPEAN ASSOCIATION OF PATIENT ORGANISATIONS OF SARCOIDOSIS
 EUROPEAN MEN ALLIANCE
 EUROPEAN NETWORK FOR ICHTHYOSIS
 FAUN STIFTUNG
 GAUCHER GESELLSCHAFT DEUTSCHLAND E.V.
 GESELLSCHAFT FÜR MUKOPOLYSACCHARIDOSEN E.V.
 HAE VEREINIGUNG E.V. (HEREDITARY ANGIOEDEMA)
 HAND IN HAND GEGEN TAY-SACHS UND SANDHOFF E.V.
 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 E.V.
 KINDER-AUGEN-KREBS-STIFTUNG
 KINDERNETZWERK E.V.
 KINDNESS FOR KIDS FOUNDATION
 LEBEN MIT BEHÇET IN DEUTSCHLAND
 LEONA E.V.
 MARFAN HILFE DEUTSCHLAND E.V.
 MORBUS-OSLER SELBSTHILFE E.V.
 MPN-NETZWERK E.V.
 MPS EUROPE
 MULTINATIONAL INTERSTITIAL CYSTITIS ASSOCIATION
 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 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
 SSADH-DEFIZIT E.V.
 STIFF-PERSON VEREINIGUNG DEUTSCHLAND E.V.
 THE FEDERATION OF ESOPHAGEAL ATRESIA AND TRACHEO-ESOPHAGEAL FISTULA SUPPORT GROUPS E.V.
 TOM WAHLIG STIFTUNG
 VEREIN AHc18+
 VEREIN VHL (VON HIPPEL - LINDAU) BETROFFENER FAMILIEN E.V.
 XP - FREU(N)DE MONDSCHNEINKINDER
 ZNM - ZUSAMMEN STARK! E.V.

GREECE

"95" - RARE ALLIANCE GREECE
 ASSOCIATION OF GREEK FRIENDS FOR PAEDIATRIC IMMUNOLOGY PRIMARY IMMUNODEFICIENCIES «HARMONY»
 CHILD'S HEART
 GREEK ALLIANCE FOR RARE DISEASES
 HELLENIC CYSTIC FIBROSIS ASSOCIATION
 HELLENIC LEAGUE AGAINST RHEUMATISM
 HELLENIC MYASTHENIA GRAVIS ASSOCIATION
 KARKINAKI AWARENESS FOR CHILDHOOD AND ADOLESCENT CANCER
 MUSCULAR DYSTROPHY ASSOCIATION HELLAS
 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
 TO MELLON- ASSOCIATION OF PEOPLE WITH GENETIC DISORDERS
 TUBEROUS SCLEROSIS ASSOCIATION OF GREECE
 VHLFA ALLIANCE IN GREECE

GUATEMALA

ASOCIACIÓN NACIONAL GUATEMALTECA PARA LAS ENFERMEDADES DE DEPÓSITO LISOSOMAL
 PROCRECE

HONG KONG, SAR OF CHINA

JOSHUA HELLMANN FOUNDATION FOR ORPHAN DISEASE

HUNGARY

HUNGARIAN HAEMOPHILIA SOCIETY - MAGYAR HEMOFÍLIA EGYESÜLET
 MAGYÁORSZÁGI MITOCHONDRÁLIS BETEGEK ALAPÍTVÁNYA
 PRIMER IMMUNHIANYOS BETEGEK EGYESÜLETE
 RARE DISEASES HUNGARY - HUFERDIS

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
 THE ICELANDIC CHILDHOOD CANCER PARENT ORGANIZATION

INDIA

ORGANIZATION FOR RARE DISEASES INDIA

IRAN (ISLAMIC REPUBLIC OF)

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
 THE ULTRA RARE DISEASES, DISORDERS & DISABILITIES FOUNDATION

ISRAEL

GRIN DISORDERS RESEARCH FOUNDATION
 ISRAEL ADULT POLYGLUCOSAN BODY DISEASE
 LITTLE STEPS ASSOCIATION

ITALY

ABC ASSOCIAZIONE BAMBINI CRI DU CHAT
 ACONDROPLASIA - INSIEME PER CRESCERE - ONLUS
 AIBWS ONLUS
 AMICI DELLA PORFIRIA - SAN PIO DA PIETRELcina ONLUS
 ANGELI NOONAN
 ASSOCIAZIONE PERSONE CON MALATTIE REUMATICHE
 ASSOCIAZIONE NAZIONALE PEMFIGO/PEMFIGOIDE
 ASSOCIAZIONE CONTO ALLA ROVESCIA
 ASSOCIAZIONE FAMIGLIE DI SOGGETTI CON DEFICIT DELL'ORMONE DELLA CRESCITA ED ALTRE PATOLOGIE
 ASSOCIAZIONE ITALIANA ADRENOLEUCODISTROFIA ONLUS
 ASSOCIAZIONE ITALIANA CISTITE INTERSTIZIALE
 ASSOCIAZIONE ITALIANA DEI CARDIOPATICI CONGENITI ADULTI - ITALIAN GUCH ASSOCIATION
 ASSOCIAZIONE ITALIANA ESTROFIA VESICALE-EPISPADIA ONLUS
 ASSOCIAZIONE ITALIANA GAUCHER ONLUS
 ASSOCIAZIONE ITALIANA GLICOGENOSI
 ASSOCIAZIONE ITALIANA LARYNGECTOMIZZATI
 ASSOCIAZIONE ITALIANA LEUCODISTROFIE UNITE
 ASSOCIAZIONE ITALIANA LINFOISTIOCIOTOSI EMOFAGOCITICA
 ASSOCIAZIONE ITALIANA MALATI DI ALKAPTONURIA
 ASSOCIAZIONE ITALIANA MALFORMAZIONE DI ARNOLD-CHIARI CHILD
 ASSOCIAZIONE ITALIANA MIASTENIA E MALATTIE IMMUNODEGENERATIVE - AMICI DEL BESTA ONLUS
 ASSOCIAZIONE ITALIANA MIASTENIA ONLUS
 ASSOCIAZIONE ITALIANA MORBO DI HIRSCHSPRUNG
 ASSOCIAZIONE ITALIANA MUCOPOLISACCARIDOSI E MALATTIE AFFINI
 ASSOCIAZIONE ITALIANA NIEMANN PICK E MALATTIE AFFINI - ONLUS
 ASSOCIAZIONE ITALIANA PER LA LOTTA ALLE PHTS
 ASSOCIAZIONE ITALIANA PER LE MALFORMAZIONI ANORETTALI
 ASSOCIAZIONE ITALIANA SCLEROSI LATERALE AMIOTROFICA (SEZ. LOMBARDIA)
 ASSOCIAZIONE ITALIANA SINDROME DI EHLERS-DANLOS
 ASSOCIAZIONE ITALIANA SINDROME DI POLAND
 ASSOCIAZIONE ITALIANA SINDROME E MALATTIA DI BEHÇET
 ASSOCIAZIONE ITALIANA SINDROME X FRAGILE ONLUS
 ASSOCIAZIONE ITALIANA SIRINGOMIELIA E ARNOLD CHIARI
 ASSOCIAZIONE ITALIANA SOSTEGNO MALATTIE METABOLICHE EREDITARIE ONLUS
 ASSOCIAZIONE ITALIANA TELEANGECTASIA EMORRAGICA ITALIANA
 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
CIDP ITALIA ONLUS
COLLAGENE VI ITALIA ONLUS - COL6
COSTELLO.CFC - ASSOCIAZIONE ITALIANA SINDROME DI COSTELLO -
CARDIOFACIOCUTANEA - RASOPATIE - ONLUS
DEBRA ITALIA ONLUS - ASSOCIAZIONE PER LA RICERCA
SULL'EPIDERMOLISI BOLLOSA
DRAVET ITALIA ONLUS
EUROPEAN CONGENITAL HEART DISEASE ORGANISATION
EUROPEAN TUBEROUS SCLEROSIS COMPLEX ASSOCIATION
FEDERAZIONE SINDROME DI PRADER WILLI ITALIA
FOP ITALIA ONLUS
GLI AMICI DI DANIELA
GLI EQUILIBRISTI HIBM
GRUPPO «ITALIA - GLIOBLASTOMA MULTIFORME - CANCRO AL CERVELLO»
GRUPPO FAMIGLIE DRAVET ASSOCIAZIONE ONLUS
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
LND FAMIGLIE ITALIANE ONLUS
MITOCON - INSIEME PER LO STUDIO E LA CURA DELLE MALATTIE
MITOCONDRIALI - ONLUS
P63 EEC SYNDROME INTERNATIONAL NET WORK WORD COMMUNICATION -
MALATTIE RARE ONLUS
PANDAS ITALIA
PARENT PROJECT ONLUS
PKS KIDS ITALIA ONLUS
RETINA ITALIA ONLUS
RING 14 INTERNATIONAL
SOD ITALIA - ASSOCIAZIONE ITALIANA DISPLASIA SETTO OTTICA E
IPOPLASIA DEL NERVO OTTICO
UN FILO PER LA VITA ONLUS
UNIAMO- RARE DISEASES ITALY
UNIONE ITALIANA ITTIOSI
UNIONE TRAPIANTATI POLMONE DI PADOVA
UNITED ONLUS
WORLD FEDERATION OF INCONTINENCE PATIENTS
XLPDR INTERNATIONAL ASSOCIATION

JAPAN

JAPAN PATIENT ASSOCIATION

KAZAKHSTAN

PATIENTS WITH CANCER AND RARE DISEASES SUPPORT ASSOCIATION

KOSOVO

RARE DISEASES KOSOVO (SHOQATA E EMUNDJEVE TE RRALLA KOSOVE)

LATVIA

ASSOCIATION OF PEOPLE WITH SPECIAL NEEDS «MOTUS VITA»

EMPTY NOSE SYNDROME INTERNATIONAL ASSOCIATION

LATVIAN ALLIANCE FOR RARE DISEASES

RARE DISEASE ASSOCIATION «CALADRIUS»

LEBANON

LEBANESE ASSOCIATION FOR NEUROMUSCULAR DISEASES

LITHUANIA

COMMUNITY OF ONCOHEMATOLOGICAL PATIENT ORGANISATIONS

KRAUJAS

RARE DISEASES LITHUANIA

LUXEMBOURG

ALAN - MALADIES RARES LUXEMBOURG

DEN I ASBL SYNDROME DE MARFAN

EEN HÄERZ FIR KRIIBSKRANK KANNER ASBL

FONDATIOUN KRIIBSKRANK KANNER

RETT SYNDROME EUROPE

MALAYSIA

MALAYSIA METABOLIC SOCIETY

MALAYSIAN RARE DISORDERS SOCIETY

MALTA

NATIONAL ALLIANCE FOR RARE DISEASES SUPPORT - MALTA

MEXICO

PROYECTO PIDE UN DESEO MEXICO IAP

RED SANFILIPPO

MONTENEGRO

NATIONAL ORGANISATION FOR RARE DISEASES

MOROCCO

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

SUN'HOP

NEPAL

GBS/CIDP FOUNDATION NEPAL

NETHERLANDS

ALS PATIENTS CONNECTED

AUTOSOMAAL DOMINANT CEREBELLAIRE ATAXIE-VERENIGING
NEDERLAND

BIJNIEVERENIGING NVACP

CF EUROPE

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 LYMFEOEDEEM EN LIPOEDEEM

NEDERLANDSE HYPOFYSE STICHTING (DUTCH PITUITARY FOUNDATION)

NEDERLANDSE LEVERPATIENTEN VERENIGING

NEDERLANDSE PHENYLKETONURIE VERENIGING / DUTCH PKU
ASSOCIATION

NEPHCEUROPE

NETHERLANDS HAEMOPHILIA SOCIETY

NEUROFIBROMATOSE VERENIGING NEDERLAND

OSCAR NEDERLAND

PATIENTENORGANISATIE FIBREUZE DYSPLASIE

PATIËNTENVERENIGING VOOR BLAASEXTROPHIE NEDERLAND

PITT HOPKINS UK

PSC PATIENTS EUROPE

SARCOIDOSE.NL

STICHTING AA & PNH CONTACTGROEP

STICHTING AMYLOIDOSE NEDERLAND SAN

STICHTING CHRISTIANSON SYNDROME EUROPE

STICHTING HART4ONDERZOEK / HEART4RESEARCH

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

VASCULITIS STICHTING

VERENIGING SPIERZIEKTEN NEDERLAND - DUTCH ASSOCIATION FOR
NEUROMUSCULAR DISEASE

VOLWASSENEN, KINDEREN EN STOFWISSELINGSZIEKTEN

VSOP - VERENIGING SAMENWERKENDE OUDER EN

PATIËNTENORGANISATIES

WORLD DUCHENNE ORGANIZATION

NEW ZEALAND

NEW ZEALAND ORGANISATION FOR RARE DISORDERS

NORTH 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 NORTH MACEDONIA

SAVE LIVER ASSOCIATION OF PATIENTS WITH LIVER DISEASES

ZDRUZENIE ZA CISTICNA FIBROZA (MACEDONIAN CF ASSOCIATION)

NORWAY

ANIRIDI NORGE

ANIRIDIA EUROPE

EUROPEAN HUNTINGTON ASSOCIATION

FRAMBU - RESOURCE CENTRE FOR RARE DISORDERS

HYPOPARA NORGE

INTERNATIONAL HUNTINGTON ASSOCIATION

MORBUS ADDISON ASSOCIATION NORWAY

MPS-FORENINGEN I NORGE

NORSK FORENING FOR ARVELIG SPASTISK PARAPARESE / ATAKSI

NORSK FORENING FOR EHLERS-DANLOS SYNDROM

NORSK FORENING FOR OSTEOGENESIS IMPERFECTA

NORSK FORENING FOR TUBEROS SKLEROSE

NORWEGIAN FEDERATION OF ORGANISATIONS OF DISABLED PEOPLE
(FUNKSJONSHEMMEDES FELLESGRANISASJON)

NORWEGIAN ORGANISATION FOR PRADER WILLIS SYNDROME

OSLERFORENINGEN NORGE

POLAND

DEBRA POLSKA

FOUNDATION OF BORYS THE HERO /FUNDACJA BOHATERA BORYSA

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 RADZIWIŁŁOWA CHILD'S HEART FOUNDATION

PORTUGAL

ALIANÇA PORTUGUESA DE ASSOCIAÇÕES DAS DOENÇAS RARAS

ASSOCIAÇÃO NACIONAL DE DISPLASIAS ÓSSEAS

ASSOCIAÇÃO NACIONAL PARA DIVULGAR E ORIENTAR PARA COMBATER E

ENFRENTAR A TAY SACHS E OUTRAS GANGLIOSIDOSES

ASSOCIACAO PORTUGUESA CDG E OUTRAS DOENÇAS METABOLICAS

ASSOCIAÇÃO PORTUGUESA DE CHARCOT-MARIE-TOOTH

ASSOCIAÇÃO PORTUGUESA DE DOENTES NEUROMUSCULARES

ASSOCIAÇÃO PORTUGUESA DE INSUFICIENTES RENAIIS

ASSOCIAÇÃO PORTUGUESA DE LEUCEMIAS E LINFOMAS

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EUROPEAN CAVERNOMA ALLIANCE
EUROPEAN DYSMELIA REFERENCE INFORMATION CENTRE
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NEUROLOGICAL DISABILITIES
PRADER WILLI SYNDROME ASSOCIATION IN SWEDEN
PRIMÄR IMMUNBRIST ORGANISATIONEN
RARE DISEASES SWEDEN (RIKSFÖRBUNDET SÄLLSYNTA DIAGNOSER)
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SVENSKA MARFANFÖRENINGEN - SWEDISH MARFAN ASSOCIATION
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KİFDER
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PARTICIPATION OF EURORDIS' REPRESENTATIVES IN PUBLIC EUROPEAN / INTERNATIONAL CONFERENCES & Workshops 2019

Human Genetics and Genomics Foresight Workshop,
London, UK, 18 January

Virginie Bros-Facer represented EURORDIS

Newborn Screening for Rare Diseases, Brussels, Belgium,
30 January

Simone Boselli: What is at stake for people living with a rare disease

European Joint Programme for Rare Diseases: Executive Committee Kick Off, Paris, France, 30-31 January

Virginie Bros-Facer: Capacity Building and Empowerment; Development and adaptation of training activities; Capacity building and training of patients and researchers; training on data management and quality (acknowledgement); Rare disease research challenges

Journée des membres de RaDiOrg, Brussels, Belgium, 2 February

Ariane Weinman: Discussion : le rôle de l'information afin d'aider le patient et le rôle des organisations de patients dans ce cadre

Shaping the Future of Rare Disease Together Summit, San Diego, USA, 4 February

Yann Le Cam, Virginie Bros-Facer represented EURORDIS

Solving the unsolved Rare Diseases: Annual Meeting 2019, Nijmegen, The Netherlands, 7-8 February

Virginie Bros-Facer: Winter School highlights: brand new training for patient organisations on genetics, diagnosis and data sharing

Virginie Bros-Facer: Community Engagement Task Force

7th Accelerate Paediatric Oncology Conference, Brussels, Belgium, 14-15 February

Ariane Weinman represented EURORDIS

European Reference Network Workshop (ERN-RND) on Trial readiness and registries in RND, Amsterdam, The Netherlands, 18 February

Gulcin Gumus represented EURORDIS

Rare Diseases across the borders: Europe and the Euregio Meuse-Rhine, Maastricht, The Netherlands, 28 February

Matt Johnson: Cross border health care & cooperation

European Policy and Expert Summit on Preconception Health, The Hague, The Netherlands, 8 March

Yann Le Cam: Plenary instructional presentation: How to advocate for the good sake?

Meeting on Data sharing for Duchenne, Amsterdam, The Netherlands, 21-22 March

Gulcin Gumus represented EURORDIS

19th International Conference on Integrated Care, San Sebastian, Spain, 1-3 April

Raquel Castro: Bridging the gaps between health, social and local services, to improve care for people living with rare and complex conditions: key findings of the EU-funded INNOVCare project and its case management pilot

World Orphan Drug Congress, Washington, USA, 10-12 April

Yann Le Cam: Rare Disease Patients' perspectives on data sharing and data protection

7th UDNI Conference, New Delhi, India, 16 April

Virginie Bros-Facer: Undiagnosed Community Engagement Task Force Solve-RD

International Summit on Patient and Public Partnership, Montreal, Canada, 2 May

Yann Le Cam: Patient advocacy and the influence of patient-driven associations, a pan-European perspective

RDI-CORD A Rare International Dialogue, Toronto, Canada, 10-12 May

Yann Le Cam: Celebration of Rare Disease Heroes (and their impact)

Yann Le Cam: WHO Universal Health Coverage Policies and Practices to Insure "no patient is left behind" regardless of social, economic and political circumstances: Inclusion of Rare Diseases

C4c General Assembly Meeting, Rome, Italy, 23-24 May

Virginie Bros-Facer represented EURORDIS

Side event to the 72nd World Health Assembly: How transformational digital technologies can contribute to leave no one behind in universal health coverage: the case of rare diseases, Geneva, Switzerland, 23 May

Yann Le Cam: How digital technology can accelerate time of access to diagnostic and help patients find the right experts and specialised services

EHA Congress, Amsterdam, The Netherlands, 13-16 June

Ariane Weinman represented EURORDIS

European Joint Programme on Rare Diseases: Policy Board Meeting, Brussels, Belgium, 4 July

Virginie Bros-Facer: Rare disease research challenges

Virginie Bros-Facer: Capacity building and training of patients and researchers

Seminar on Social Innovation for Social Services, September

Dorica Dan: Bridging the gaps between health and social care: results of the EU-funded project INNOVCare

E-Rare Strategic Workshop, Gdansk, Poland, 20 September

Gábor Pogány: SHS research: improvement of health care implementation & everyday life of people living with a rare disease – Perspective of patient

Virginie Bros-Facer: Chair of session 1: setting the scene

EJP RD General Assembly: Consortium meeting, Gdansk, Poland, 16-19 September

Virginie Bros-Facer: Transversal: Patient Engagement

Alexandre Mejat: EJP RD in the eyes of stakeholders

The International Summer School on Rare Disease Registries and FAIRification of Data, Rome, Italy, 23-27 September

Virginie Bros-Facer: Roles of RD patients in registries & research

NAKSE 2019, Berlin, Germany, 26 September

Yann Le Cam: A European vision for better healthcare for people with rare diseases

2014-2020 EU Health Programme Conference, Brussels, Belgium, 30 September

Yann Le Cam: Facilitate access to better and safer healthcare for Union citizens

Europe Biobank Week 2019, Lübeck, Germany, 8-11 October

Gulcin Gumus represented EURORDIS

Académie des technologies: Séminaire annuel 2019, Paris, France, 9 October

Yann Le Cam: Les technologies de la santé : quelle perception de l'impact pour les bénéficiaires, quelles attentes et enjeux

Université d'Automne de l'Alliance Maladies Rares, Paris, France, 11 October

Ariane Weinman: Réseaux européens de référence pour les maladies rares

XX Telethon Scientific Convention, Riva del Garda, Italy, 28 October

Sandra Coubier: Science and Society – whose data are my data? Sharing and protecting personal health data

Youth Cancer Europe Cross Border Health Roundtable, Brussels, Belgium, 6 November

Ariane Weinman: European Reference Networks: where information, rather than patients, travel

European Joint Programme on Rare Diseases Info Day and Networking Event, Istanbul, Turkey, 7 November

Gulcin Gumus: Patient Advocacy – Where is POAs in EJP-RD Programme and Their Integration to Research Projects?

The 14th Meeting of the International Conference on Rare Diseases and Orphan Drugs (ICORD): Rare Diseases Are Not Orphans, Rishon LeZion, Israel, 12 November

Virginie Bros-Facer: Capacity building programme in the European Joint Programme on Rare Diseases to address unmet needs in rare disease research education and training

World Orphan Drug Congress, Barcelona, Spain, 12-14 November

Yann Le Cam represented EURORDIS

1st ARRIGE meeting, Paris, France, 14 November

Virginie Bros-Facer represented EURORDIS

QualityOfLife4OI, Amsterdam, The Netherlands, 23 November

Raquel Castro: Supporting patient engagement: EURORDIS Open Academy

ERN EYE Annual Workshop, Strasbourg, France, 22 November

Simone Boselli: The Rare Impact initiative

Better health and wellbeing - Increasing the impact of health research to improve health and health promotion, Brussels, Belgium, 26 November

RCE-ESMO-ESO Training Course for Rare Cancer Patient Advocates 2019, Milan, Italy, 29-30 November

Ariane Weinman: The JARC Recommendations from rare cancer adult patients and their representatives on ERNs for rare cancers

Ariane Weinman: Opportunities provided by ERNs: Rare Cancer adult patients ePAGs' perspectives

TREAT-NMD International Conference 2019, Leiden, The Netherlands, 11 December

Yann Le Cam: EURORDIS visions and proposals to address current challenges of access to new treatments

Registries for Rare Endocrine Conditions Workshop, Glasgow, UK, 13 December

Virginie Bros-Facer: Perspectives from rare diseases patients



JOURNAL PUBLICATIONS 2019

Share and protect our health data: an evidence based approach to rare disease patients' perspectives on data sharing and data protection – quantitative survey and recommendations, Orphanet Journal of Rare Diseases, July

Contributing authors:
Sandra Courbier, Virginie Bros-Facer

Priorities on rare cancers' policy in National Cancer Control Plans (NCCPs): A review conducted within the framework of EU-JARC Joint-Action, Journal of Cancer Policy, July

Contributing authors:
Ariane Weinman, Yann Le Cam

Defining orphan conditions in the context of the European orphan regulation: challenges and evolution, Nature Reviews Drug Discovery, July

Contributing authors:
Virginie Hivert

Evaluating the “return on patient engagement initiatives” in medicines research and development: A literature review, Health Expectations, September

Contributing authors:
Elisa Ferrer, Virginie Hivert

Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database, European Journal of Human Genetics, September

Contributing authors:
Yann Le Cam

The use or generation of biomedical data and existing medicines to discover and establish new treatments for patients with rare diseases – recommendations of the IRDiRC Data Mining and Repurposing Task Force, Orphanet Journal of Rare Diseases, October

Contributing authors:
Virginie Hivert

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- The Operating Grant for year 2019
- JARC – Joint Action on Rare Cancers
- Rare 2030, Foresight in Rare Disease Policy
- RD-Code project



Co-funded by the Health Programme of the European Union

EUROPEAN COMMISSION

DG Research and Innovation

- The European Joint Programme Rare Diseases (EJP)
- The European Clinical Research Infrastructures Network Integrated Activity (ECRIN-IA) project, which partners with and supports the EURORDIS Summer School
- The Next Generation Health Technology Assessment (HTx)
- The Solve-RD Project -Solving the Unsolved Rare Diseases
- The Innovative Medicines Initiative-Joint Undertaking (IMIJU) 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 Horizon 2020 Framework Programme of the European Union



Health Sector Corporates

EURORDIS appreciates the donations received from health sector companies and other private funders. Ensuring a sustained variety of funding is key to minimise potential conflicts of interest. EURORDIS had 70 different health sector corporate donors in 2019. Health sector companies have supported EURORDIS through the EURORDIS Round Table of Companies¹, the Membership Meeting in Bucharest², the EURORDIS Black Pearl Awards³, as well as international initiatives such as Rare Disease Day⁴, Rare Barometer⁵, RareConnect⁶, Rare Diseases International⁷, EURORDIS Open Academy⁸, NGO Committee for Rare Diseases⁹, 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 TAKEDA



2 PFIZER



3 NOVARTIS



4 CELGENE



5 CSL BEHRING



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

² <https://www.eurordis.org/content/presentations-eurordis-membership-meeting-2019-bucharest>

³ <https://blackpearl.eurordis.org>

⁴ <https://www.rarediseaseday.org>

⁵ <http://www.eurordis.org/voices>

⁶ <https://www.rareconnect.org/en>

⁷ <http://www.rarediseasesinternational.org>

⁸ <https://openacademy.eurordis.org>

⁹ <https://www.ngocommitteerarediseases.org>

Other Pharmaceutical & Biotechnology Companies & Health Sector Corporates making Donations to EURORDIS



Other corporates providing direct and in-kind support



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BERTARELLI FOUNDATION

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IMAGINE INSTITUTE

We also would like to sincerely thank all the generous individuals and organisations from across countries who made a gift to EURORDIS in 2019.

Special Mention

EURORDIS would like to Sean Hepburn Ferrer for his support for EURORDIS via the Intimate Audrey exhibitions.





EURORDIS
RARE DISEASES EUROPE

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ACTION PLAN

2020

EURORDIS Action Plan 2020 and the COVID-19 crisis

This Action Plan 2020 was reviewed before its formal adoption at the General Assembly in May 2020 to take into account the impact of the COVID-19 crisis.

Overall, the planned actions for 2020 are being reduced by 5 to 10%. A good anticipation and preparedness has limited the impact on the continuity of activities. Thanks to a series of measures, the staff capacities have been maintained at 75-80% during March-April-May. Though some actions are being deferred to 2021, such as the sustainability plans for the European Conference on Rare Diseases (ECRD), the Community Advisory Boards (EuroCAB) and Rare Disease Day; the RareBarometer Voices' survey on Diagnosis; the content revision and development for the new; the Open Academy Alumni communication actions; and several leads on the diversification of private resources from major donors and foundations.

Our decision has been to maintain all core activities and main projects as planned and usually for the same dates, with the exception of the Foresight Study Rare 2030 which has been extended from December 2020 to March 2021. All events from March to July have been moved online such as the European Conference on Rare Diseases and Orphan Products (ECRD 2020 Stockholm), the EURORDIS Membership Meeting, the Board meetings, the EURORDIS General Assembly, the Council of National Alliances Meeting, the Winter School in March, the Summer School in June. The Rare 2030 events are planned online for the Young Citizen Conference, most national workshops, the ERN & Stakeholder conference. The annual Steering

Committee meeting and the annual all-ePAG meeting will both be held online in Oct-Nov. Other meetings during the second semester are being planned face to face (F2F) with a contingency plan online (OL) which applies to the HTA Task Force, DITA Task Force, European Network of Rare Diseases Help Lines (ENRDHL), Council of National Alliances and Council of European Federations, and more.

The budget impact is significant. Some budget has been saved on travel, accommodation and meeting expenses, although often these have been cancelled while losing the resource attached to it. Money has been lost on venue renting, travels and services already contracted. New expenses have been incurred when switching major events online. Some expected significant private resources have been lost or are uncertain.

New actions are being taken to specifically address the challenges of the COVID-19 crisis. An Internal COVID-19 Task Force has been created to propose and implement these actions. The Board of Directors has had additional meetings. A dedicated website section was created with curated information on COVID-19 for our members, partners and stakeholders providing reliable sources of information, clinical guidelines, clinical trials and studies. Several statements have been published to publicly convey the concerns of people living with rare diseases. A survey on the impact of the COVID-19 in the daily life of the persons living with a rare disease has been conducted through RareBarometer, the EURORDIS survey programme.

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 therapies for rare diseases
- + Promoting cross-border rare disease expertise and knowledge generation and sharing to improve quality of care, diagnostics, medical care & social care at local level
- + Promoting access to cross-border healthcare and making possible patient mobility
- + Promoting research and bridging patient's perspective and researcher activities
- + Addressing the 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

- + Promote a more integrated and ambitious EU Public Health Strategy in areas of action of high European added value and high economic impact such as health threats, cancers, Alzheimer, mental illnesses, obesity and diabetes, and rare diseases.
- + Prepare for the next decade of rare disease legislative & policy framework to take the necessary steps to requalify rare diseases as a public health issue.
- + Execute the **Rare 2030 Foresight Study on Rare Disease Policy**, to create a new policy framework for rare diseases. This entails carrying out the following activities: Drafting Rare 2030-2040 Future Exploratory Scenarios with thorough contribution from the project's partners, 200-member Panel of Experts, Research Advisory Board; Presenting proposed Rare 2030 Scenarios at the 2020 European Conference on Rare Diseases and Orphan drugs (ECRD) and voting on stakeholder preferences; Using participative methods to elaborate and discuss online meaningful policy options to integrate in the preferred policy scenario 2030, through the ECRD, 6 national-regional workshops and a European workshop with ERNs and stakeholders; Gathering patients and families perspective through a survey with the Rare Barometer Voices platform; organising a Young Citizen Conference to bring the perspective of the next generation; Consolidating

all proposals and presenting them in the form of recommendations to the European Commission, EU Council and European Parliament in February 2021.

- + Based on Rare2030 recommendations, and in line with the 2019 Court of Auditors special report on cross border healthcare, explore and consolidate political options with EU Institutions to update, review or replace the overall EU rare disease strategy.
- + Engage with upcoming EU Presidencies to support the EU policy strategic review.
- + Re-launch and expand the European Parliamentary Advocates for Rare Diseases network, inviting MEPs elected for the term 2019-2024 to join, in coordination with the National Alliances, and preparation of the first Rare Disease Week in Brussels for rare disease advocates on the occasion of Rare Disease Day 2021.

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

- + Continue focusing on the negotiations that lead by the end of 2020 to an agreement on the **EU Multiannual Financial Framework 2010-2027** that will fund EU policies and programmes and propose specific proposals on ERNs and access to medicines.
- + Consolidate the position 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 Rare Diseases** embedded within ERNs and rare cancers research within the Mission of Cancer.
- + Promote RDs within health, social and digital strategies across EU funding programmes 2021-2027, including **European Structural and Investment Funds**, notably the **European Social Fund Plus**.
- + Promote the needs of people with rare diseases as a priority in the Innovative Health Initiative-IHI (IMI's successor) programme 2021-2027.

1.4 ADVOCATE FOR THE INCLUSION OF RARE CANCERS IN EUROPE'S BEATING CANCER PLAN

- + Ensure inclusion of the estimated 200 rare cancers and 4 million people affected by them in Europe in the forthcoming Europe's Beating Cancer Plan.
- + Continue to advocate for the inclusion of rare cancers in the EU policy framework and actions both of rare diseases and of cancers, while advocating for the inclusion of rare cancers in the national cancer plans.
- + Build on the in-depth policy preparedness forged over the past years thanks to the Joint Action on Rare Cancers and other collaborative platforms.

1.5 ADVOCATE FOR THE SUSTAINABILITY AND INTEGRATION OF ERNS WITH WIDER NATIONAL HEALTHCARE SYSTEMS AND RESEARCH INFRASTRUCTURES



- + Promote the deployment of ERNs, support the implementation of their key functions and ensure their sustainability by: developing EURORDIS advocacy messages around ERNs post 2022; advocating for an ERN health data strategy; ensuring that patients' interests are well represented in key areas of ERNs development through our involvement in key tenders; supporting ePAG advocates and clinicians to strengthen their partnership in the ERNs; building our institutional relationship with the group of hospitals managers and contributing to shape the development of a model of collaboration with industry.
- + Promote integration of ERNs into national healthcare systems in collaboration with National Alliances (NAs) and ePAG advocates by continuing to support National Alliances to play an active role on the integration of ERNs into national healthcare systems through local face-to-face meetings with national stakeholders and online webinars tailored to their country needs and health systems specificities.

1.6 ADVOCATE FOR THE CURRENT PATIENTS' PERSPECTIVE ON ORPHAN & PAEDIATRIC LEGISLATION AND HTA

- + Advocate in favour of the EU Regulation on European Cooperation on HTA, in coordination with National Alliances, focusing on the priority scope, national re-use of common assessment reports, HTA methods, and engagement of patients.
- + Foster through advocacy actions, following the evaluation of the EU legislation on medicines for special populations (orphan medicinal products and paediatric medicines), an adequate environment for the development of therapies for people with rare diseases.
- + Promote proposals within the pharmaceutical legislation review to better address unmet medical needs, enforce patient engagement in the decision-making process, grasp scientific and technological advancements to support innovation and/or repositioning of existing treatments, accelerate development, and address the challenges of patients' access.

1.7 ADVOCATE TO IMPROVE SUSTAINABLE ACCESS TO RARE DISEASE THERAPIES

- + Conclude the RARE-IMPACT initiative on the improvement of patient access to gene and cell therapies for rare diseases and ensure adequate follow up at country level with publication of reports on challenges, as well as filling potential gaps through adequate analysis
- + Promote a set of bold proposals to improve access to new rare disease therapies: EU Table of Negotiation, EU co-fund of post marketing authorisation evidence generation, innovative payment models for medicines (e.g. differential pricing, payment on outcomes, discount for uncertainties) and reiterate solutions to the current shortfalls in patient access to therapies
- + Ensure alignment of actions on topics related to better access to therapies within EURORDIS and between EURORDIS and European Federations as well as National Alliances
- + Advocate to address critical shortages of medicines in Europe, building on the recommendations to industry, to Member States, to EU Institutions laid out in the Common Position (co-led by EURORDIS and signed by 45 organisations back in 2013)

1.8 VOICING THE SOCIAL NEEDS OF PEOPLE WITH A RARE DISEASE AND ADVOCATING FOR HOLISTIC CARE

- + Continue to disseminate the EURORDIS position paper on Holistic Care, developed with the EURORDIS members, to all relevant health and social stakeholders.
- + Promote an integrated approach to medical and social care amongst healthcare providers and ERNs.
- + Promote a holistic and integrated health and social care approach within Europe's beating cancer plan, for people living with rare cancers and cancer survivors.
- + Monitor the transposition of the Directive on Work Life Balance in MSs legislations so to support National Alliances' advocacy on the key measures to advocate for.

1.9 ADVOCATE TO IMPROVE ACCESS TO DISABILITY RIGHTS

- + Advocate for the European Disability Strategy Rights Agenda 2020-2030 to address the needs of people living with a rare disease, based on the EURORDIS position paper on Holistic Care, regarding the access to adequate disability assessment and disability rights.
- + Partnering with the European Disability Forum (EDF), of which EURORDIS is a new full member, to amplify the voice of people living with a rare disease and disability in EU forums and in disability issues.

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

Rare Diseases International:

- + Support to RDI as an established independent entity as defined in the EURORDIS-RDI MoU signed for the period 2019-2023 to provide operational and financial support.
- + Building on the success of the **UN Political Declaration on Universal Health Coverage (UHC)** adopted in 2019 with the inclusion of rare diseases, participate in UHC 2030 and develop advocacy tools to promote its implementation in Europe and other regions.
- + Implement the activities included in the MoU signed with WHO End 2019: a) developing an operational description of rare diseases based on key prevalence and incidence figures, and b) proposing a conceptual and methodological framework for the establishment of a **WHO Collaborative Global Network for Rare Diseases (CGN4RD)**; a network of highly specialised multi-disciplinary university research hospitals – Initial work supported by EURORDIS
- + Convey the political messages on rare diseases in the Human Right Convention session in Geneva in February 2020 through a photo exhibition and Member state support
- + Support RDI and its members in developing the Key Asks, drafting the UN Resolution on Rare Diseases, and strategizing the advocacy with Member States

NGO Committee for Rare Diseases:

- + EURORDIS to take full advantage of its ECOSOC status to advocate within the UN system
- + EURORDIS, RDI and Agrenska within the NGO Committee to consult stakeholders, draft the outline of a UN General Assembly Resolution on Rare Diseases, and start advocating to the Permanent Missions to the UN in New York to provide political support.
- + Establish further relationships with country Permanent Missions in New York and Geneva as well as relevant departments and agencies of the UN through the newly established EURORDIS International Advocacy Volunteers based in NYC.

International Rare Diseases Research Consortium (IRDIRC) : see 3.4.1

- + As one of the founding members, fully participate as a member of the Consortium Assembly, of the Patient Advisory Committee and scientific committee
- + Take active part as a member and bring the patient perspective in the Task Forces

World Economic Forum (WEF):

- + EURORDIS is part of the World Economic Forum Shaping the Future of Health and HealthCare
- + Take part in the new WEF Board of Stewards of Future Health and HealthCare
- + Continue to be part of and contribute in the WEF Global Precision Medicines Council
- + Contribute to the agenda on Diagnostic & Rare Diseases, Genomics & Gene Editing
- + Start bridging with WEF Digital Economy and New value Creation, and Digital Health

1.11 EURORDIS RARE BAROMETER PROGRAMME: GENERATING DATA FROM PATIENT EXPERIENCE



Rare Barometer

A EURORDIS INITIATIVE

- + Scale up the collection of patient experiences and perspectives to enrich a patient evidence based policy development building on the 11 000 patients and families who have joined Rare Barometer Voices and continue the growth internationally.
- + Continue dissemination of reports of previous surveys and infographics of surveys in 23 languages.
- + The pilot survey aiming to develop a common mechanism to collect patients' experience through the ERNs will be on-going until third quarter of the year; Provide the results to the ERNs involved in the project and report on the outcomes and conclusions of the pilot for the next phase.
- + Harness the results of the survey on access to treatments for rare diseases, by developing targeted analyses and reports in support of national and EU advocacy on the development and access to treatment of people living with a rare disease.
- + Explore the feasibility and plan a research project to develop validated scale(s) on the experience of patients and families in the healthcare system.
- + Carry out and analyse a survey on the impact of COVID-19 on rare disease patients and draw recommendations that highlight patients' needs within the crisis based on the survey results.
- + Carry out and analyse a survey on rare disease patients' perspectives on the scenarios of the Rare 2030 project to explore the potential impact of the scenarios on patients and identify preferred scenarios.
- + Carry out a survey on how the COVID-19 pandemic is affecting the rare disease community.
- + A significant part of the year will be dedicated to **develop the sustainability of the Rare Barometer programme**, find funding to scale up the ERN survey pilot, and have the possibility to develop a survey on diagnosis - identified as the next priority. The programme will apply for foundation and EC grants as well as developing support from corporate partners.

2. Patient Empowerment

2.1 BUILDING THE COMMUNITY & NETWORKING

2.1.1 Membership

- + Maintain EURORDIS' Membership at over 900 members and ensure regular interaction
- + Maintain process of regular membership reassessment
- + Enrich the information in the patient organization database for targeted outreach
- + Organise the EURORDIS Membership Meeting 2020 Online 13 May 2020

2.1.1 Capacity building of European Networks

Council of National Alliances (CNA)



- + Organise two meetings of the Council of National Alliances (CNA), Online in March and F2F/OL November 2020
- + Enhance the work between NAs through the Common Goals & Mutual Commitments with regular webinars on specific topics of common interest; Various Working Groups such as the WG on Small Countries and the WG on the Western Balkans; The creation of a monthly CNA information tool for national alliances to plan and anticipate as much as to guide and support their advocacy and capacity building
- + Reinforce the collaboration and integrate more the advocacy actions between EURORDIS and National Alliances; Dedicate a new Public Affairs Manager based in Brussels on EU&National Integrated Advocacy; empowering capabilities through regular bilateral meetings and webinars; Establish a WG for the Rare Disease Week 2021 enabling full participation of CNA into the design of the programme and selection of participants; EU&National coordination through the year to build relationship with MEPs.

Council of European Federations (CEF)

- + Reposition the Council of European Federations to address the current main challenges of European Federations on the different aspects of access to medicines
- + Organise one meeting of the Council of European Federations (CEF) in November F2F/OL, ensure regular

communication (emails, webinars) with the network or directly with relevant federations, and work more closely with European Federations on key items, in particular on access to medicines and newborn screening.

- + Continue the EURORDIS programme "Support to European Rare Disease Federations" to help European federations organise their network meetings; adapt the programme to the COVID-19 crisis by enlarging the type of activities supported, scaling up the amount of the grants, increasing the total amount of support.

Rare Cancers

- + Create a stronger sense of shared interest and solidarity across the + 60 Rare Cancer EURORDIS member organisations, to fully engage in the EU's Beating Cancer Plan.
- + Dedicate an experienced Public Affairs Senior Manager based in Paris, to maintain a high level of engagement of rare cancer patient advocates in all public consultations, notably through the Working Group composed of 30 ePAGs of the 4 relevant ERNs covering rare cancers. The WG serves as advisory group on advocacy issues in the field of (rare) cancer.
- + Ensure a close coordination and collaboration with European Cancer Patient Coalition, the network WE CAN, the European federations for specific rare cancers and in the USA the NORD Rare Cancer Coalition.

European Network of Rare Diseases Help Lines

- + Organise one meeting F2F/OL of the ENRDHL including a training on how to use the rare disease classification; governance and election of ENRDHL Steering Committee.
- + Develop a common Code of Conduct for Helplines steering up the ENRDHL to GDPR standard beyond the current obligation to provide a declaration from national data protection authority.
- + Implement the Caller Profile Analysis in Fall 2020.
- + Reflect on how genetic testing results are communicated to patients, the role of genetic counsellors, and how helplines can help patients informing family members.

2.1.1 RareConnect

- + Consolidate EURORDIS' role at the heart of the new RareConnect governance structure
- + Promote RareConnect through all relevant EURORDIS activities
- + Support the creation of new online communities for very rare diseases and undiagnosed disease patients
- + 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

- + Finalise the technical development of the new eurordis.org website with a user driven design; progress in 2020 the content update in 7 languages toward a launch in 2021

- + Maintain and regularly evaluate and update EURORDIS' communications tools such as the EURORDIS website, eNews, dedicated Member News, EURORDIS' social media, webinars programme;
- + Consolidate EURORDIS webinar programme used within different EURORDIS activities and different target audiences
- + Maintain and cultivate relations with media covering the policies of the European Union

2.2.2 EURORDIS Open Academy & other trainings

- + Review the vision and the strategy of EURORDIS on the Open Academy, to ensure that the programme developments in the most meaningful, cohesive and financially sustainable manner; Design a **multiannual training programme**, with a relevant and flexible training curriculum delivered within a blended



approach, including face-to-face and online training (webinars and e-learning).

- + Organise the third edition of the **EURORDIS Winter School on Scientific Innovation and Translational Research**, including one week of online training, in March 2020, preceded by pre-training webinars and e-learning courses.
- + Organise the thirteenth edition of the **EURORDIS Summer School on Medicines Research & Development**, including one week of online training, in June 2020, preceded by pre-training webinars and e-learning courses. A post-training online module will also take place in September.
- + Organise the **second edition of the EURORDIS Leadership School**, for ePAG advocates, including 8 webinars, from April to November, and two-days of intensive training, in October 2020.
- + Organise the **EURORDIS Digital School**, as a fully online programme, with webinars and e-learning courses;
- + For each of the Open Academy schools, coordinate the contents of the course with the respective multi-stakeholder Programme Committee, continuously updating and improving the content and the delivery methods;
- + Further develop the **Open Academy's e-learning platform**, increasing the availability and outreach of online training materials to more patient advocates,

free of cost; improve the Summer and Winter School's e-learning courses, curating and updating the content.

- + Within the European Joint Programme for Rare Diseases, co-organise an **"EJP Leadership Training"** for ePAG advocates and other patient advocates who wish to be involved in ERNs or research, in Gdansk, in November 2020.

2.3 RAISING AWARENESS & INFORMING

2.3.1 Rare Disease Day 2020 & 2021

- + Implement the recommendations of the strategic review performed in 2019 to the International Rare Disease Day to create further positive change for people living with a rare disease over the next decade: champion the logo, targeted audiences, simpler messages, more visuals less words, more social media, common features
- + Coordinate the international Rare Disease Day 2020 in over 100 countries; focus the campaign on reframing the basic key messages about rare diseases: rare is many, rare is proud, rare is strong / 5% of the population, rare is 6000 diseases
- + Extend the 2020 campaign throughout the year with social media and stories from the global campaign
- + Co-create and produce Rare Disease Day 2021 campaign materials (visuals, website update, video) building on strategic review recommendations and a call to action
- + Prepare the EURORDIS' specific European events for RDD 2021:
 - Plan the 1st Rare Disease Week in Brussels in February 2021 for 50 patient advocates selected from most EU Member States, as a capacity building and advocacy week, with trainings on EU and national policy making ; interactions with decision-makers in the EU Institutions and Permanent Representations, including meetings with a wide range of MEPs, in connection with the European Network of Parliamentarian Advocate for Rare Diseases
 - Plan the final conference of the Foresight Study Rare 2030 at the European Parliament



3. Patient Engagement

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

- + Promote a meaningful patient engagement in ERN activities & provide regular support to ePAGs to ensure that they bring into the Networks activities the perspective of the wider patient community in the development of clinical decision support tools, outcomes measures, research and other patients' relevant activities
- + Define and pilot an ePAG impact assessment framework to evaluate the impact of the ePAGs engagement in the ERNs. Build the evidence around patient engagement in the ERNs
- + Promote and facilitate exchange of ePAG good practices and improve the management of knowledge assets to enable ePAGs to share, re-use and access relevant information and contents
- + Further build ERN team members capacities on patient engagement methodologies
- + Review the 24 ePAGs governance models to inform the process for the renewal of ePAG advocates mandates in 2021 ahead of the next ERN 5 years cycle; Review the ePAG Constitution building on experience gained to ensure it continues to be fit for purpose, and support the ePAG groups in its implementation.
- + Contribute to the promotion of common methods to capture feedback on patient satisfaction & patient experience across the ERNs (including through Rare Barometer)
- + Deliver a comprehensive capacity-building programme Leadership School for ePAG patient advocates within EURORDIS Open Academy; organise an all-ePAG online workshop 5th and 6th of November on and an online 1-day Steering Committee meeting on the 30th of October.

3.2 PATIENT ENGAGEMENT IN THERAPEUTIC DEVELOPMENT

3.2.1 Support Patient involvement in EMA activities



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

- + 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)
- + Engage patients as experts for CHMP consultations in writing, in Scientific Advisory group meetings, during

oral explanation with the marketing authorisation applicant,

- + Review Herbal Monographs prepared by the Herbal Medicinal Products Committee (HMPC)
- + 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, one alternate, and one representative of the PDCO and one representative of the PRAC in PCWP.
- + 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.
- + Collaborate with EMA to identify and support the participation of patients in Protocol Assistance dossiers at the Scientific Advice Working Party (SAWP)

3.2.2 Support patient involvement in European HTA Network, EUnetHTA JA 3, related HTA activities and through the new EURORDIS HTA Task Force (HTA TF)

- + Represent patient organisations in the European HTA Network
- + Support 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 and several online meetings planned for 2020
- + Identify and mentor patients participating in HTA Joint Procedures
- + Take part in the HTAx project, a Horizon 2020 project on innovative HTA methods

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

- + Organise one annual F2F/OL meeting with regular on-line meetings throughout the year
- + Revise the Common Position on Shortages of Medicines, coordinated by EURORDIS, between 45 patients', consumers' and healthcare professional's organisations
- + Contribute to the Concept Paper on the Prevention of Shortages with the EMA/HMA Task Force on availability of medicines
- + Contribute to the EMA consultations, the EU Register for Clinical Trials and on the Regulatory Strategy 2020-2025 with specific reflections on compassionate use, big data, access to individual patient data
- + Review the CIOMS' chapters on Patient Engagement in the Development and Safe Use of Medicines
- + Contribute to ICH Guideline on Clinical Trials E6 – Ethical aspects and GCP

3.2.4 Support patients' Community Advisory Boards (CABs) to engage with Industry (EUROCAP programme)

- + Maintain a mentoring programme for existing CABs, alongside ad hoc guidance to help prepare meetings
- + Assist European federations or networks creating a CAB
- + Revise the EUROCAP programme key features based on experience gained so far
- + Promote the EUROCAP programme to patient groups and health corporates
- + Provide metrics to support the evaluation of the impact of the CABs
- + Collect and analyse relevant feedback from CABs within or outside the CAB programme, from European federations and networks, from health corporates and regulators to develop a programme sustainability plan in 2021

3.2.5 Support patient engagement in medicines life-cycle (project PARADIGM)



- + Take part in the last year of the 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 and Member of the Steering Committee
- + Ensure that EURORDIS patient engagement practices and insight contributes to the workstream that is designing patient engagement tools
- + support the project's efforts to build consensus from all stakeholders on the value and methods for patient engagement; Disseminate broadly these methods
- + Successfully lead our work package on co-designing a sustainability road map for patient engagement that demonstrates the 'return on the engagement' for all players
- + Coordinate the EURORDIS Pool of Patient experts created in the context of the PARADIGM project

3.3. PATIENT ENGAGEMENT IN DIAGNOSIS

3.3.1 Shape position statements from the RD patient community on Newborn Screening:

- + Create a Newborn Screening (NBS) Working Group, chaired by EURORDIS and composed of various stakeholders, as NBS emerges as an issue more important than ever due to new diagnostic capacities and new or future potentially transformative treatments.
- + Review current policy and practice in the field of NBS, in order to develop a set of recommendations in 2020 for harmonious uptake of NBS programmes across the Member States, with a view to delivering maximum benefit and improving outcomes for babies born with rare diseases.

3.3.2 Co-lead and participate in the Global Commission to End the Diagnostic Odyssey for Children with Rare Diseases:



GLOBAL COMMISSION

to End the Diagnostic Odyssey for Children with a Rare Disease

- + Contribute to the Vision, Mission and Goals of the Global Commission
- + Contribute to extend partners – corporate and NGO – and members – expertise
- + Engage in the Patient Empowerment Education & Awareness Campaign
- + Promote engagement in relevant pilots and policy recommendations
- + Support communication of the Global Commission

3.3.3 Participate in the EU-funded project Solve-RD



- + Lead the work package on Impact and Engagement
- + Participate in the steering committee of the project
- + Participate in the Ethics Independent Advisory Board of the project
- + Continue coordinating the Community Engagement Task Force:
 - Ensure that development of the activities within Solve-RD are patient-centered
 - Develop a comprehensive map of resources and tools to support undiagnosed rare disease patients as well as follow up of genetic counselling for patient organisations and healthcare professionals.

3.3.4 Participate in the Undiagnosed Diseases Network International

- + Continue representing the EU RD patients' perspectives and voices the priority of this community.
- + Contribute to white papers, conferences and other statements.
- + Participate in the Data Sharing working Group.
- + Participate in the Patient Engagement Working Group in partnership with NORD.

3.4. PATIENT ENGAGEMENT IN RESEARCH

3.4.1 Participate in the International Rare Disease Research Consortium (IRDiRC)

- + Contribute as a member of the **Consortium Assembly, Patient Advocacy Constituent Committee, Therapies Scientific Committee** (vice-chair) and Operating Committee and being involved in the current and forthcoming **Task Forces and working groups** – Clinical Research Networks, Orphan Drug Development Guidebook (see below), Shared Molecular Aetiologies, Access to RD Treatment, Integrating New Technologies for the Diagnosis of Rare Diseases.



IRDiRC

INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

- + In particular, leadership role in the development of the Orphan Drug Development Guidebook with a multi-stakeholder group of experts. The Guidebook is aimed at public and private developers in order to help them navigate the therapeutic development ecosystem and describes the available tools and initiatives specific for rare disease development and how to best use them (the Guidebook is available at <https://irdirc.org/activities/task-forces/orphan-drug-development-guidebook-task-force/>)
- + Participate in the **2 annual consortium assembly meetings** (online in March and F2F in Milan in October). Prepare the meeting organised back to back with the RE(ACT)'s the International Congress of Research on Rare and Orphan Diseases in Berlin, postponed to January 2021.
- + A new **road map** for 2020 IRDiRC activities has recently been approved by the consortium assembly. EURORDIS will actively promote opportunities for **patient representatives** to get involved as Task Forces and Working groups are being launched and open for expression of interest during the year.

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 Committees.
- + Represent EURORDIS within the Policy Board and the French national mirror group.
- + Coordinate all training activities within Pillar 3 (10 different training activities in 2020, most online due to the COVID-19 situation).
- + **Develop guidance to support and further encourage meaningful patient involvement within the joint**

transnational calls through the establishment of a multi-stakeholder Working Group chaired by EURORDIS and involving ePAGs, Summer and Winter School Alumni and research funders.

- + Support the implementation of a new public private funding partnership, the **Rare Diseases Research Challenges** after successfully initiating its framework in 2019.
- + Organise the various patient trainings supported by the EJP, including the EURORDIS Winter School, the EURORDIS Summer School and the EJP Leadership Training (see section 2.2.2 EURORDIS Open Academy & other trainings).

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



- + Provide the patient perspective across all work packages including:
 - Sustainability - **development of a business plan to sustain and expand paediatric clinical trials**
 - **Development of education and training on clinical trials in children** for clinical researchers & for children, young patients, parents, and patient organisations
- + Ensure that EURORDIS patient engagement practices and insight contributes to the design of the patient engagement framework within the network for European clinical trials for children
 - Provision of support to develop parent/patient representation at European level
 - Establishment of a collaborative framework with all the stakeholders to ensure standardized procedures of patient involvement
- + Identify and promote of best practice to enhance the **value of data collected in clinical trials**

3.4.4 Participate in the HTX project, Next Generation HTA (IMI2)

- + Provide the patient perspective across all work packages including:
 - Treatment pathways in specific therapeutic areas (diabetes, multiple sclerosis, myelodysplastic syndromes, head and neck cancer)
 - Using real world data (RWD) for evidence synthesis
 - Using artificial intelligence (AI) to forecast individualised treatments
 - Implementation into systems and processes
 - Transferability and dissemination
- + Inform patients' organisations in rare diseases and beyond on HTx objectives and results
- + Empower and engage patient representatives in HTX implementation

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

- + **BBMRI Stakeholder forum meeting:**
 - Continued active engagement at the Europe Biobank Week 2020, ensuring the proper involvement and engagement of patients in all steps of biobank research.
 - Contribute in producing a work programme for the patients' pillar including education resources for BBMRI National Nodes regarding biobanking
- + **Go FAIR RD Network:**
 - Serve as a member of the GO FAIR RD Seed Group in order to improve the dialogue between GO FAIR RD network and patient representatives
 - Promote and support the adoption of FAIR data principles amongst rare disease patient organizations
- + **Partner in the project application for Clinical Research Networks on Rare Diseases:**
 - A Coordination and Support Action of the H2020 programme
 - Partner with all 24 ERNs for Rare Diseases to initiate a robust policy and operational framework to articulate European Clinical Research Networks within ERNs

3.5 PATIENT ENGAGEMENT IN HOLISTIC CARE

- + **Continue the EURORDIS Social Policy Action Group**, to disseminate and contribute to the positions of EURORDIS and its members and to engage with all stakeholders to promote holistic care, including social services, integrated care and disability rights;
- + **Support the work of the European Network of Resource Centres for Rare Diseases** - RareResourceNet, as part of its Board of Directors;
- + **Carry on promoting the results and good practices of the INNOVCare project**, ensuring the wide dissemination of the results of the project to support integrated care for rare diseases;
- + **Seek opportunities** to develop key actions in holistic and integrated care **within European projects and initiatives**;
- + **Continue to work in collaboration with the International Foundation for Integrated Care (IFIC)** as a moderator of its Self Management and Co-production Special Interest Group.

4. Cross-cutting priorities

4.1 GOVERNANCE

4.1.1 EURORDIS Strategy 2015-2020 Implementation

- + Respond and adapt to the COVID-19 crisis through 2020 and anticipate the long term impact
- + Continue collecting EURORDIS Indicators
- + Prepare the Strategic Revision 2021-2025, based on the future exploratory scenarios 2030-2040 and the back casting policy options scenario 2030 produced by the Foresight project Rare 2030, as well as on a comparable strategic review methodology used in 2009-2010, for adoption at the AGA 2021

4.1.2 Maintain the EURORDIS By-laws

- + Regularly review and update the EURORDIS internal governance documents that comprise the EURORDIS by-laws. The by-laws are publicly available on the EURORDIS website.

4.1.3 Develop the sustainability of EURORDIS programmes

- + Develop sustainability plans for two of EURORDIS' major programmes: Open Academy and Rare Barometer

4.1.4 Strategic Partnerships (MoUs)

- + Establish or maintain partnerships with international organisations and review and renew MoUs as needed, in addition to being open to new partnerships with other international patient organisations
- + Renew strategic partnerships with key international organisations, such as NORD (USA) and CORD (Canada)

4.2 RESOURCE DEVELOPMENT

- + Support the **EURORDIS Round Table of Companies (ERTC)**, **engaging over 70 health companies** in a bilateral dialogue as well as a collective dialogue through regular webinars and **two major workshops**
- + 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 the handling of competing interests of scientific committee members and experts and CHAFAEA rules
- + Pursue opportunities to deliver new sources of diversified income from foundations; **Appoint a new Resource Development Senior Manager to take forward work with Donors and Foundations**
- + **Focus team efforts and allocate resources to innovative project development in 2020 in strategic areas of research H2020, IMI, Health Tender, and other opportunities, toward 2021 & beyond**



4.3 EURORDIS EVENTS

- + Organise the **EURORDIS Black Pearl Awards Dinner** in February 2020 in Brussels
- + Organise the **10th European Conference on Rare Diseases & Orphan Products – ECRD 2020 Stockholm** in May 2020 “The journey of living with a rare disease in 2030”, fully online, using a highly professional platform, an adapted programme, adjusted target audiences and revised revenue streams; ensure a high engagement for quality inputs from the Programme Committee and Theme Leaders
- + Organise two major workshops of the ERTC with relevant stakeholders, dedicated to the repositioning of medicinal products – February, Brussels – and newborn screening – October, Barcelona.
- + Organise online the Young Citizen Conference and F2F/OL 6 national-regional workshops as part of the Rare 2030 project

4.4 HUMAN RESOURCES

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

- + Support the emergence of a group of team members who have a 360 degree view of the organisation in order to enhance transversal work strategically, streamline operations, optimise time and fund allocation
- + Reorganise and enhance new management team structure with an improved internal coordination meetings structure (operations meetings, leadership

meetings, management team meetings, unit team meetings, advocacy meetings)

- + Maintain, expand and support the implementation and usage of the new EURORDIS Contact Database; additional development; staff training on data inputs & outputs; implement actions for GDPR compliance
- + Upgrade the EURORDIS central server architecture and equipment for simultaneous remote work / work-from-home for all staff as well as all equipment and services for quality online meetings
- + Develop a Business Continuity & Contingency Plan for the Finance & Support Function
- + Increase the space of the Brussels office

4.4.2 EURORDIS Volunteers

- + Improve **volunteers’ visibility** in EURORDIS’ communication and acknowledgements on the EURORDIS website
- + **Revise processes for effective and improved volunteer support and management**
- + Prepare for 2021 a series of leaflets and other information that help EURORDIS volunteers and potential volunteers to understand the opportunities to get involved with EURORDIS work, the rules of engagement and support, the integration and dynamic within EURORDIS action

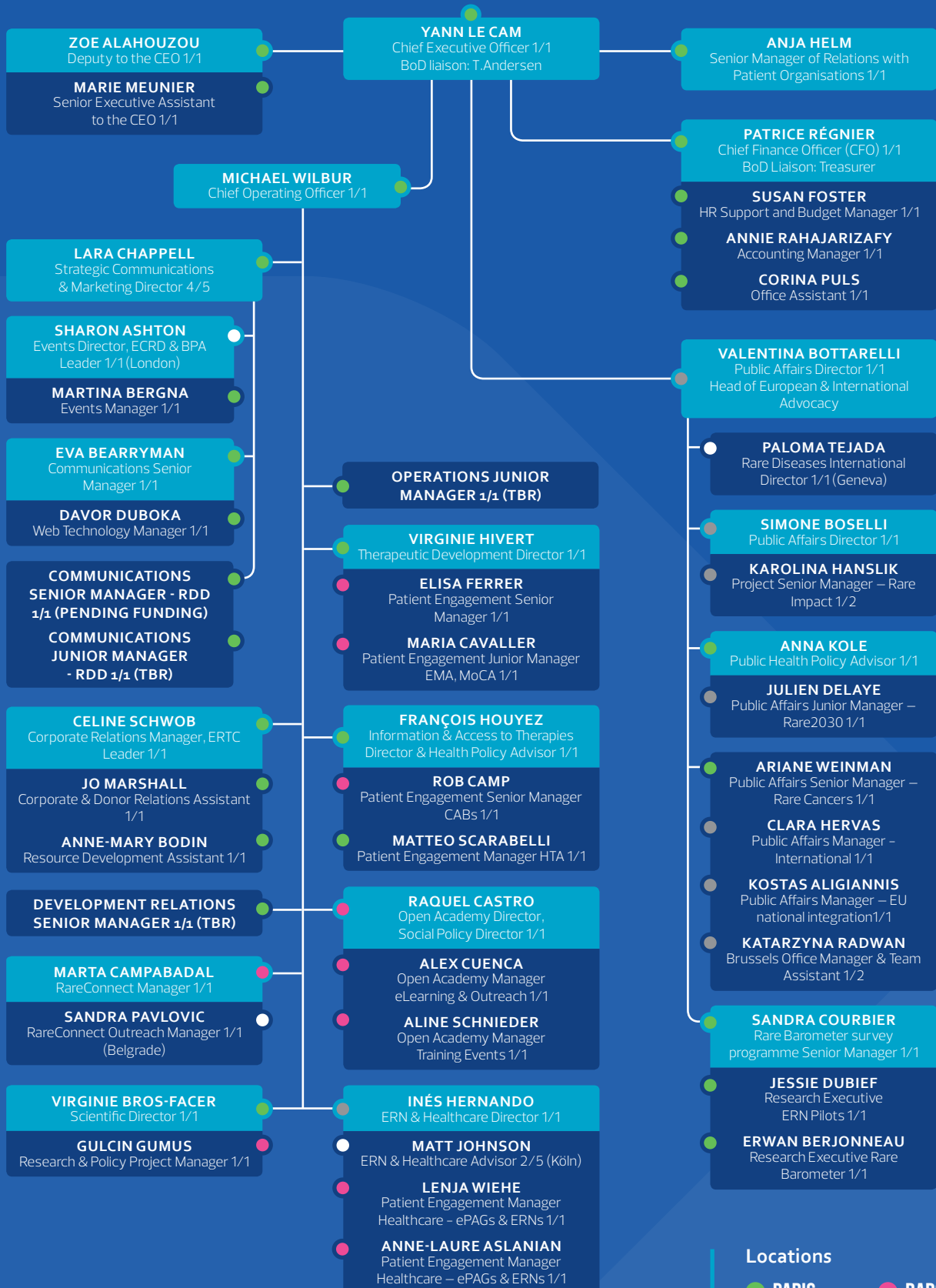
4.4.3 EURORDIS Staff

- + **Appointment of following positions created in 2019:**
 - Public Affairs Manager, EU & National Integrated Advocacy (Brussels)
 - Open Academy Manager, eLearning and Outreach Coordinator (Barcelona)

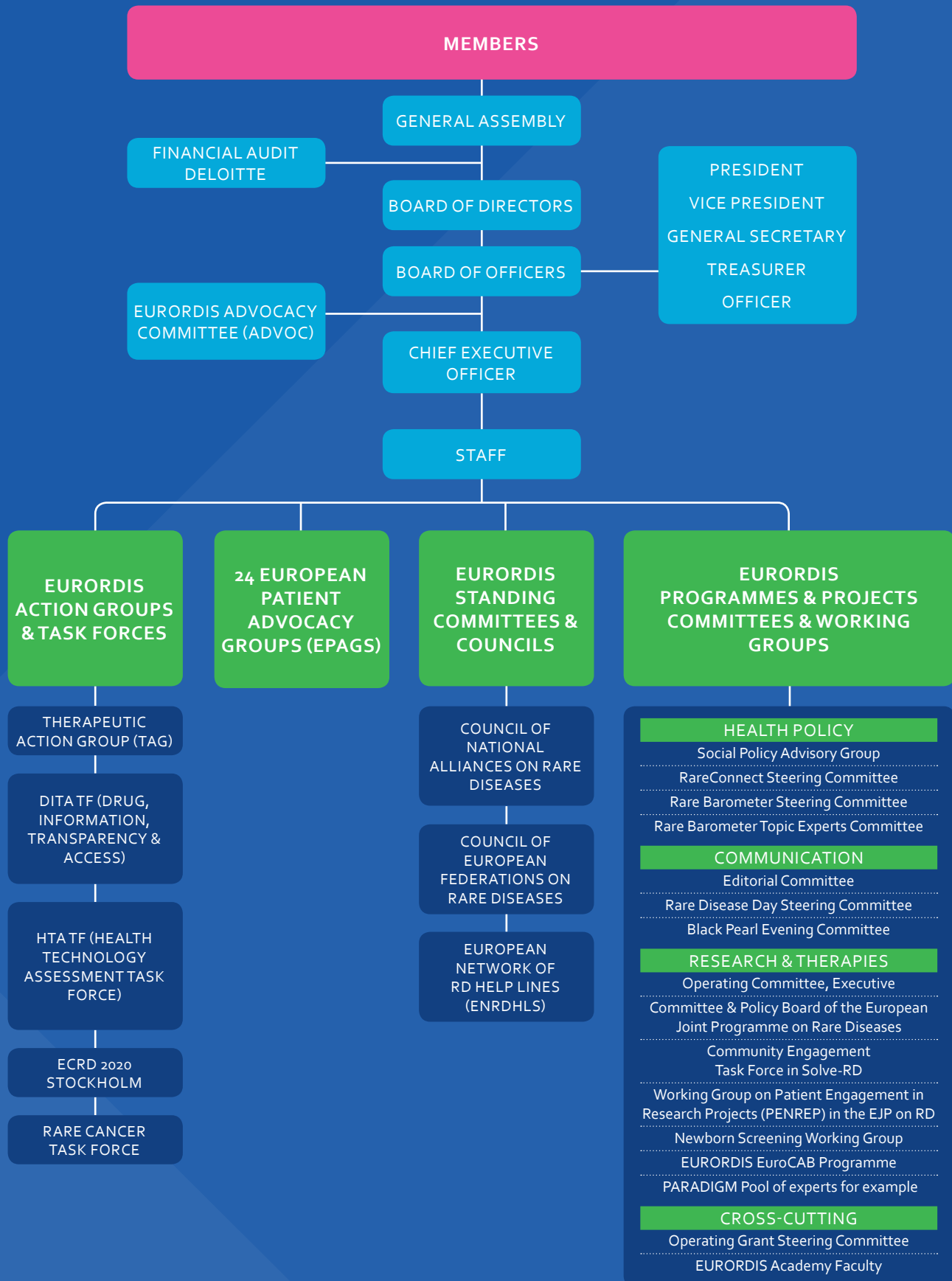
- Corporate and Donor Relations Assistant (Paris) – postponed late 2020
- + **Potential permanent positions created in 2020 pending resources:**
 - Digital Health & Data Director (postponed until 2021)
 - International Public Affairs Director
 - Operations Junior Manager
- + **Temporary positions in 2020 linked to a specific project:**
 - Public Affairs Junior Manager, project Rare 2030 (Brussels, 2020)
 - Social Research Advisor, project ERN Pilot Survey (Paris, 2020)
 - Patient Engagement Manager, ERN & HealthCare & ePAG, Maternity leave replacement
 - ECRD On Line Planning Assistant (Paris, 2Q2020)
 - Rare Disease Day Senior Manager (Paris, 2S2020)



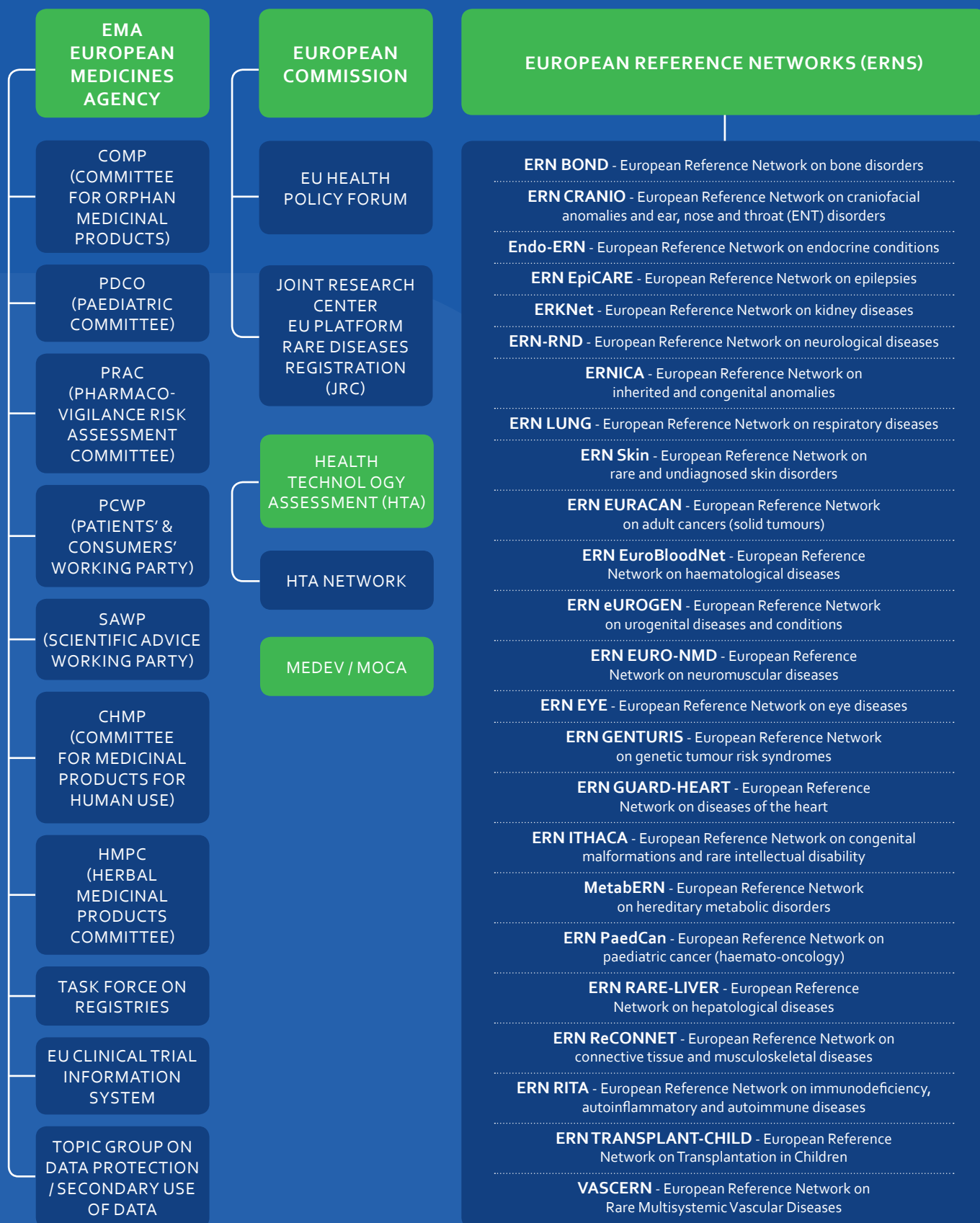
TEAM Chart 2020



GOVERNANCE Chart 2020



EXTERNAL REPRESENTATION Chart 2020



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

WEKAN: 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

BBMRI Stakeholders Forum

RD-Connect

HTx, Next Generation HTA

OpenMedicine

EUPATI

COST ACTION 15105 (drug shortages)

PARADIGM

C4C (Connect 4 Children)

CORBEL – MIUF

Solve – RD

reCOVID consortium IML2

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)

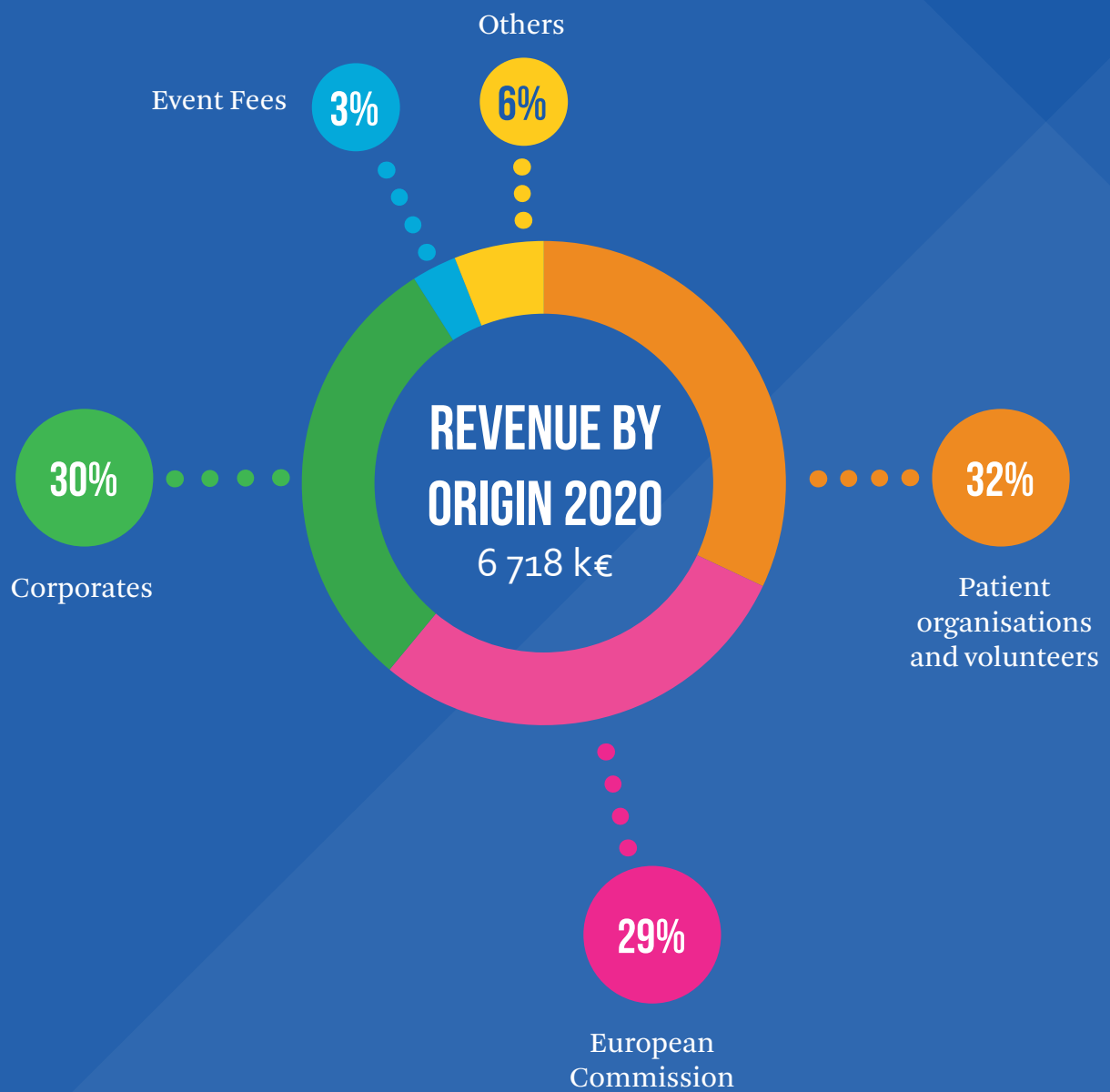
International Federation of Social Workers Europe (IFSW-Europe)

European Society of Human Genetics (ESHG)

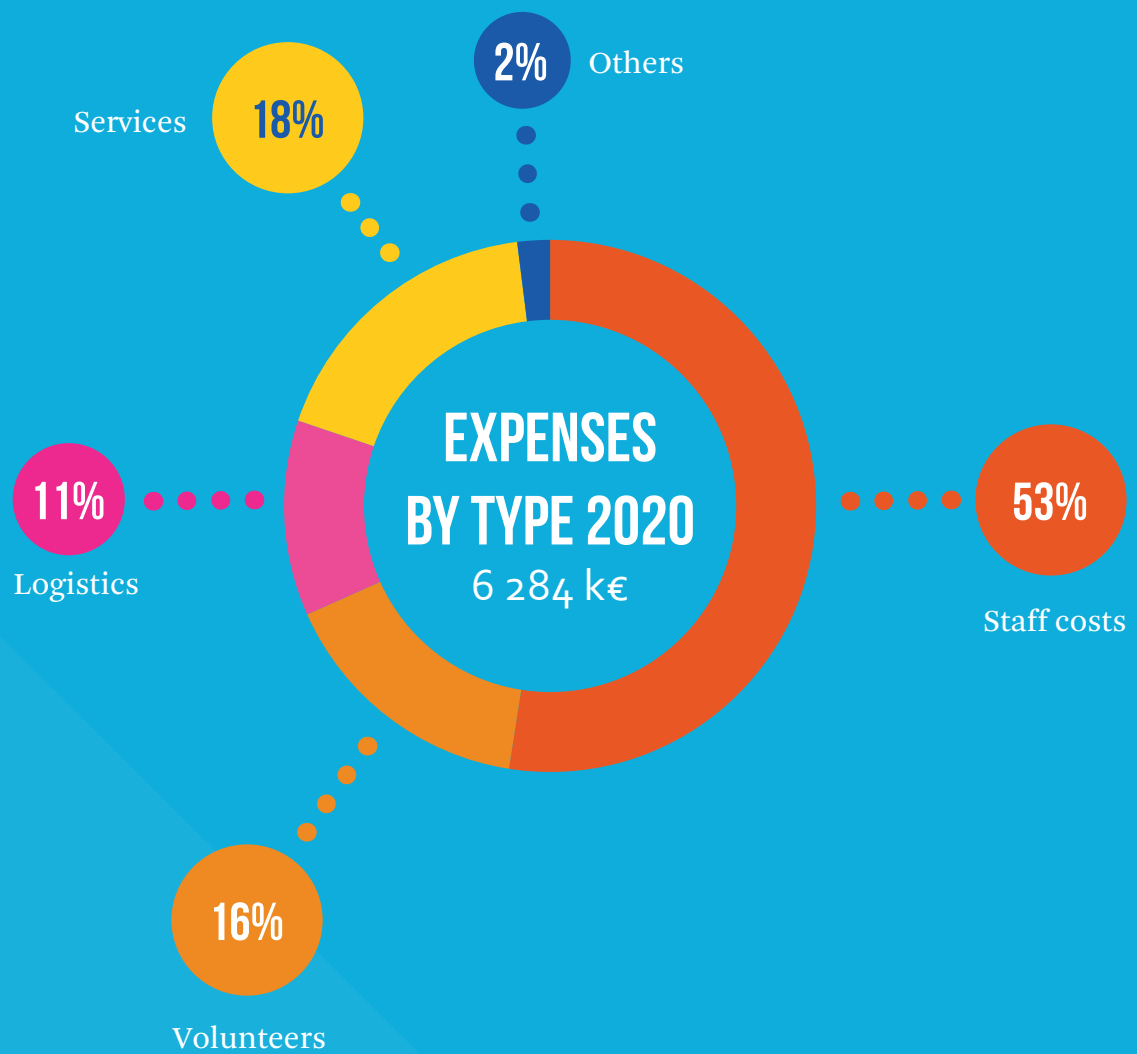
International Society for Pharmaco-economics and Outcomes Research (ISPOR)

European Connected Health Alliance - ECHalliance

REVENUE 2020



EXPENSES 2020



ACRONYMS & DEFINITIONS

EURORDIS INTERNAL COMMITTEES & TASK FORCES

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

PROJECTS OF EURORDIS OR IN WHICH EURORDIS IS INVOLVED

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

EURORDIS & EUROPEAN REGULATORY NETWORK

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

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

EUROPEAN COMMISSION

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

EURORDIS & EUROPEAN COMMISSION

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

EURORDIS & NON GOVERNMENTAL PARTNERS

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

MISCELLANEOUS

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

NOTES

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EURORDIS-RARE DISEASES EUROPE

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Calle Sant Antoni M^a Claret 167 ♦ 08025 Barcelona ♦ Spain

EURORDIS.ORG

Desislava, Dean & Vladi
from Bulgaria, Huntington disease

