


EURORDIS
ACTIVITY REPORT 2020
& WORKPLAN 2021



Robert Juríček and his wife
Alena Juríčková, living with
Proximal Spinal Muscular
Atrophy Type 2

TABLE of Contents

04

EURORDIS
in brief

05

Strategic
approach
2015-2020

07

Activity
Report 2020

HIGHLIGHTS 2020	8
PATIENT ADVOCACY	10
PATIENT EMPOWERMENT: BUILDING THE NETWORK & CAPACITIES	21
PATIENT ENGAGEMENT: ROLES IN DECISION-MAKING	29
CROSS-CUTTING PRIORITIES	36
REVENUE & EXPENSES 2020.....	40
BOARD OF DIRECTORS MAY 2020-MAY 2021	42
MEMBERS OF EURORDIS.....	43
CONFERENCES & WORKSHOPS 2020	50
JOURNAL PUBLICATIONS 2020	52
ACKNOWLEDGEMENTS	53

57

Workplan
2021

ACTION PLAN 2021.....	58
TEAM CHART 2021	68
GOVERNANCE CHART 2021	69
EXTERNAL REPRESENTATION CHART 2021.....	70
REVENUE & EXPENSES 2021.....	72

74

Acronyms & definitions





YANN LE CAM

Chief Executive Officer



TERKEL ANDERSEN

President of the
Board of Officers

FOREWORD

by the President and the Chief
Executive Officer



In 2020, EURORDIS' activities focused on advocating for, empowering and engaging people living with a rare disease, their carers and patient advocates, all the while addressing the many challenges of the coronavirus pandemic. EURORDIS' membership has continued to grow in 2020 reaching **949 members** by the end of the year, in **73 countries of which 27 are EU member states**.

The **Council of National Alliances (CNA)** (36 members in 2020) held 2 CNA Workshops that took place online, in March and in November, which was again organised partly in partnership with the **Council of European Federations (CEF)** (79 members in 2020). The CNA's main activities in 2020 were the preparation and coordination of Rare Disease Day 2021; Integration of ERNs at national level, Newborn Screening and Rare Disease Week 2021.

At the Policy Event organised at the European Parliament in Brussels in February 2020, EURORDIS re-launched the **Network of Parliamentary Advocates for Rare Diseases**, a group of European and national members of parliament advocating to improve the lives of the 30 million people living with a rare disease in Europe. With its relaunch, this group of MEPs committed to bring about a new EU policy framework on rare disease and stronger EU-wide action in health, research, social affairs and other relevant policies.

The **European Conference on Rare Diseases (ECRD) 2020**, originally planned to be held on site in Stockholm, was moved online in response to the impact and challenges of the COVID-19 pandemic on people with rare diseases. The new format was a great success with 28 online sessions: 4 plenary and 24 parallel sessions spread over 6 themes, over a two-day online conference. The ECRD welcomed 1509 participants from 57 countries, providing the **opportunity to co-design policy options today, to better conditions for people living with rare diseases for the years ahead**. Among other important conclusions, the ECRD attendees spent the course of the conference laying the foundation for the Rare2030 foresight study recommendations.

The **Rare2030 foresight study** was extended from December 2020 to March 2021, due to the COVID-19

pandemic. The project aimed to propose policy recommendations leading to improved **policy and a better future for people living with a rare disease in Europe**. It saw great progress in 2020 with a Rare Barometer Rare2030 survey, 6 regional conferences and a Young Citizen's Conference held in October. This work set the framework for the 2021 campaign **#30millionreasons** for Europe's Action Plan for Rare Diseases.

A focus for our advocacy work carried out in 2020 was to prepare for the evaluation and revision of numerous upcoming regulations and strategies including **the EU Regulation on Orphan Medicinal Products, the EU Pharmaceutical strategy particularly on Access and Unmet Medical Needs, the future EU Regulation on Health Technology Assessment, and the European Digital Health Strategy**.

Rare Disease Day 2020, one of the key EURORDIS activities, took place in **over 100 participating countries**. The **official hashtag #RareDiseaseDay** trended in a record 13 countries across 5 continents including Australia, Canada, France, India, and Mexico. **World famous landmarks** including the world's tallest building, the Burj Khalifa in the United Arab Emirates, and the Colosseum were **illuminated in the colours of Rare Disease Day** as a symbol of support for the rare disease community. The Strategic Review from 2019 helped reset the role of EURORDIS as a leader of the campaign, coordinating and serving the National Alliances to co-create and champion the campaign in their country, **building a global momentum**.

EURORDIS in 2020 has played a critical role in amplifying the patient voice and creating the conditions for engagement within **European Reference Networks** so that their activities remain driven by patients' needs. EURORDIS continued its support to the European Patient Advocacy Groups (ePAGs) aligned to the scope of the 24 ERNs. In addition, in 2020 EURORDIS along with representatives of patient organisations and ePAG advocates, developed a vision that a **mature European Reference Network (ERN) system** should be one which **leaves no person living with a rare disease in uncertainty** regarding their diagnosis, care and treatment, and published the **"Recommendations to achieve a mature ERN system in 2030"**.

In 2020, **EURORDIS Open Academy** continued to build on its success as a capacity-building programme. 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**. Due to the COVID-19 pandemic all of the programmes were adapted and provided online instead, in a blended format with **online** live sessions and e-learning courses.

The **EURORDIS Summer School 2020** was attended by **34 participants from 16 countries, representing more than 20 different rare diseases**. This year, great effort was put on optimising the programme for a suitable online delivery, while maintaining the interactivity and hands-on exercises.

The **3rd EURORDIS Winter School** on Scientific Innovation & Translational Research was held between 9-13 March 2020. The 2020 edition of Winter School brought together **30 patient advocates and 20 trainers from 13 countries** and covered important topics to support patient engagement in research.

The **EURORDIS Digital School** on Social & Digital Media is a fully online programme, composed of webinars and e-learning courses, available for free. 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 second edition of the **EURORDIS Leadership School** took place online, from April to November 2020. It was **attended by 24 ePAG advocates, representing 12 countries, over 30 rare diseases and 16 ERNs** and consisted of **9 webinars** covering topics such as self-awareness, emotional leadership, communicating with impact, power and influence, conflict resolution and negotiation.

RARE IMPACT completed its initial mission, shared in a report presented at a multi-stakeholder online event, and addressed challenges across four identified areas in the **accessibility, assessment, availability and affordability of gene and cell therapies across the European Union**.

Throughout 2020, EURORDIS, alongside the Council of National Alliances, Council of European Federations and member organisations worked towards developing **Key Principles to support a harmonised European approach to Newborn Screening**.

EURORDIS maintained a high level of involvement in the **EMA's activities related to the provision of information to patients and the public about medicines authorised via the centralised procedure**. A total of **125 EMA documents for public information were reviewed** by EURORDIS staff or volunteers in 2020. We also helped **include 32 patients (out of 39 requests) in protocol assistance procedures**, meaning 82% of patients are now involved in the procedures needing patient input.

2020 saw the end of the IMI project **PARADIGM**, the 30-month public-private partnership launched on 1 March 2018 led by the European Patients' Forum (EPF) and EFPIA. EURORDIS was a PARADIGM partner and leader of the work package developing a sustainability **roadmap to optimise patient engagement** and was involved in the co-development of a **monitoring and evaluation (M&E) framework to evaluate patient engagement practices**.

With the EURORDIS' Strategy 2015-2020 coming to an end, EURORDIS commissioned an external strategic review to develop its strategy from 2021-2030. The strategic review commenced in 2020 collecting input from EURORDIS members and stakeholders and will be discussed during the General Assembly 2021 for final adoption by the EURORDIS BoD by end of 2021.

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 2020, we invite you to read the full report as well as explore other EURORDIS publications.

None of the activities detailed in this report would be possible without the tireless dedication of the **EURORDIS volunteers**. In 2020, EURORDIS was privileged to rely on **over 450 volunteers**, our volunteer cohort comprises **81 patient advocates and 371 moderators**. EURORDIS volunteers have a unique insight into the complexity of different rare diseases across Europe and reinforce EURORDIS as a grassroots movement.

The year 2020 also came with the **unique challenges of COVID-19**. Following the outbreak of the pandemic, we reached out to the rare disease community to identify the particular needs and expectations of our community through a survey conducted by **RareBarometer on the impact of COVID-19 in the daily lives of people living with a rare disease**. New actions specifically addressing the challenges of the COVID-19 pandemic on people living with a rare disease were taken, including a) an **Internal COVID-19 Task Force** to propose and implement actions, b) the **Board of Directors** convening more regularly to closely monitor the situation, c) the **creation of a dedicated website section** with up-to-date, curated information on COVID-19, and d) finally several **statements to convey the concerns of people living with rare diseases during these difficult times**.

While the pandemic has certainly affected EURORDIS, its constituencies and stakeholders, EURORDIS is a resilient organisation that prides itself on its foresight and adaptability. In response to the challenge of operating in the pandemic, our organisation put in place a series of measures to ensure staff capacities were maintained at a high standard and the impact on activities was minimised, particularly by moving the grand majority of our events online.

”

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.

949

member patient organisations



73

countries
(27 EU countries)

53

National Alliances of RD Patients Organisations

79

European Federations of specific rare diseases



Founded in

1997

6

Million €
Budget

Outreach to over

2500

patient groups



Over

450

Volunteers

81

patient advocates

371

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 the European Union and in Europe at large 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 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 and 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 diagnosis, 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-centred 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 e.g. Rare Disease Day and European Year on Rare Diseases; EURORDIS has reached out to PLWRDs in the EU if not in Europe at large 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 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

2020

HIGHLIGHTS 2020	8
PATIENT ADVOCACY	10
PATIENT EMPOWERMENT: BUILDING THE NETWORK & CAPACITIES.....	21
PATIENT ENGAGEMENT: ROLES IN DECISION-MAKING	29
CROSS-CUTTING PRIORITIES	36
REVENUE & EXPENSES 2020.....	40
BOARD OF DIRECTORS MAY 2020-MAY 2021	42
MEMBERS OF EURORDIS.....	43
CONFERENCES & WORKSHOPS 2020	50
JOURNAL PUBLICATIONS 2020	52
ACKNOWLEDGEMENTS	53

HIGHLIGHTS 2020

2020 was a year marked by the **COVID-19 pandemic**. EURORDIS was swift to react to the challenge and thanks to a series of measures, staff capacities were maintained at high standards and the impact on our activities was limited as much as possible. All core activities and main projects were maintained as planned and all events from March onwards were moved online.

- In addition, new actions were specifically taken to address the challenges of the COVID-19 crisis on people living with a rare disease. An **Internal COVID-19 Task Force** was created to propose and implement these actions and the Board of Directors convened more regularly to closely monitor the situation and activities. 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 were 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** was also conducted with great success through RareBarometer, the EURORDIS survey programme, and was presented in various fora throughout the year.

Due to the COVID-19 pandemic, the **Rare2030 foresight study** was extended from December 2020 to March 2021 with the final conference planned for February 2021. Notwithstanding this, the project which aimed to gather the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations that will lead us to improved policy and a better future for people living with a rare disease in Europe, greatly progressed in 2020 with 6 regional conferences held, a Rare Barometer Rare2030 survey developed, disseminated and analysed and a Young Citizen's Conference held in October.

A lot of the advocacy work carried out in 2020 was to prepare for the evaluation and revision of a number of upcoming regulations and strategies such as: the EU Regulation on Orphan Medicinal Products & EU Regulation on Paediatric Use of Med & Incentives, the EU Pharmaceutical strategy particularly on Access and Unmet Medical Needs, the future EU Regulation on HTA, and the European Digital Health Strategy.

EURORDIS' membership base continued to grow in 2020 reaching **949 members** by the end of the year, including **69 new members in EU Member States**. EURORDIS has members in 73 countries and all 27 EU member states.

ECRD 2020, originally planned to be held on site in Stockholm was moved online. **The new online event proved to be a wise decision and was a big success with 28 online sessions: 4 plenary and 24 parallel sessions spread over 6 themes.** The Conference had 1509 participants registered from 57 countries; 120+ expert speakers, panellists and moderators; 3 European Commission representatives and 2 Ministers of Health. All sessions have been recorded and are available on demand for up to one year.

The **Council of National Alliances** had **36 members** in 2020. 2 CNA Workshops took place online, one in March and one in November, which were again held partly in common with the Council of European Federations (CEF) (**79 members in 2020**), in order to allow cross-cutting discussions on common issues. The CNA's main activities in 2020 were: the preparation and coordination of the Rare Disease Day 2021; Integration of ERNs at National level; Newborn Screening; Rare Disease Week 2021.

Rare Disease Day 2020 involved **over 100 participating countries**. The Strategic Review from 2019 helped reset the role of EURORDIS as a leader of the campaign, coordinating and serving the National Alliances to co-create and champion the campaign in their country, building a global momentum. In 2020 for the first time, infographics were created with a graphic designer and made available on the website. The graphics displayed key numbers for rare diseases. Finally, world-famous landmarks including the world's tallest building, the Burj Khalifa in the United Arab Emirates, the Colosseum and the Tower of Pisa in Italy were illuminated in the colours of Rare Disease Day as a symbol of support for the rare disease community.

EURORDIS continued its support to the **24 European Patient Advocacy Groups (ePAGs)** aligned to the scope of the ERNs. EURORDIS and representatives of patient organisations in addition to ePAG advocates developed a vision and **recommendations to achieve a mature ERN system in 2030**, published in December 2020.

EURORDIS delivered 4 training programmes via the **EURORDIS Open Academy**: the Summer School (medicine research and development); the Winter School (Scientific Innovation and Translational Research); the Leadership School and the Digital School. Due to the COVID-19 pandemic there were no face-to-face sessions for any of these trainings in 2020. The programmes were provided online instead, in a blended format with online live sessions and e-learning courses. 34 participants from 16 countries, representing over 20 rare diseases attended the 13th EURORDIS Summer School. **The total number of alumni since 2008 is now 542.**

◆
The **EURORDIS Newborn Screening Working Group (NBS-WG)** was set up to review current policy and practice in the field of NBS, in order to develop principles for harmonious uptake/ adoption of the NBS programs across the Member States with a view to delivering maximum benefit and improving outcomes for babies born with rare diseases. Members of NBS-WG include representatives from patient organisations, international screening societies and international and national federations with a focus on NBS. The work of the NBS-WG alongside the Council of National Alliances, Council of European Federations and EURORDIS members culminated in the development of the **11 Key Principles for Newborn Screening** to support a harmonised European approach.

2020 saw the end of **the project PARADIGM**, a public-private partnership, co-led by the European Patients' Forum and EFPIA, whose mission is to provide a unique framework that enables structured, effective, meaningful, ethical, innovative, and sustainable patient engagement (PE) and demonstrates the 'return on the engagement' for all players. Building on advances at the international level, PARADIGM integrated the needs, perspectives and expectations of all stakeholders involved **to co-produce a set of tools and recommendations to plan, conduct and evaluate patient engagement in medicines development in a meaningful and sustainable manner.** The toolbox includes a monitoring and evaluation framework and metrics to measure the impact of patient engagement for all stakeholders involved in medicines development. EURORDIS was a PARADIGM partner and leader of the work package developing a sustainability roadmap to optimise patient engagement.

◆
RARE IMPACT, a multi-stakeholder consortium working to improve patients' access to gene and cell therapies (advanced therapies) in which EURORDIS acted as Chair with the support of Dolon Consulting came to an end in 2020. The work culminated with the publication of the **RARE IMPACT European report** which presents access to advanced therapies through a European Union lens, and supplements ten detailed reports on country-specific challenges and solutions, based on two years of analysis and dialogue that has allowed us of better understand the challenges to access to therapies for rare disease patients.





1. PATIENT ADVOCACY

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

In the course of 2020, 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 & Newborn 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 2020 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 for rare diseases as a priority in the next decade 2020-2030

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 2020, 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 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.

With the election of the new European Parliament in 2019 and then the approval of a new college of European Commissioners, a new political leadership has been established in Brussels. EURORDIS continued its work to recruit new parliamentary advocates willing and able to commit to implementing concrete actions in support of people living with a rare disease. EURORDIS was invited to meet the new Commissioner for Health Stella Kyriakides in January 2020.

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. In July 2020 EURORDIS released a statement calling for the negotiators of the European Parliament, and the German Presidency of the European Council to take action to increase the European Council's funding to the EU4Health programme.



1.3.1 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.

At a Policy Event organised at the European Parliament in Brussels in February 2020, EURORDIS relaunched the Network of Parliamentary Advocates for Rare Diseases, a group of European and national members of parliament advocating to improve the lives of the 30 million people living with a rare disease in Europe.

Hosted by MEPs Tanja Fajon, Stelios Kypouropoulos and Frédérique Ries and held to mark the occasion of Rare Disease Day, the EURORDIS 'Reframe Rare' Policy Event brought together 70 participants to discuss current priorities for rare disease policy at the EU level.

Other MEPs including Kateřina Konečná and Tomislav Sokol also hosted conversations with patient representatives Claudia Crocione (HHT Europe), Mencia de Lemus Belmonte (SMA Europe) and Rebecca T. Skarberg



(Osteogenesis Imperfecta Federation Europe), on topics including research and innovation, access to medicines, holistic care and cross-border healthcare and access to specialised care.

With the relaunch of the Network of Parliamentary Advocates for Rare Diseases, this group of MEPs commits to bringing about a new EU policy framework on rare diseases and stronger EU-wide action in health, research, social affairs and other relevant policies.

1.3.2 Foresight Study on Rare Diseases in 2030 (Rare2030)

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 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 a 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. Due to the COVID-19 pandemic, the Rare2030 foresight study was extended from December 2020 to March 2021 with the final conference planned for February 2021.

The European Conference on Rare Diseases 2020, which was held online due to the pandemic, was the first opportunity to present the 4 different scenarios around the combination of future trends related to our societal values and ask stakeholders what is most likely to happen, which scenario they prefer and open a debate on the policy options needed to get there.

Throughout May to November 2020 a series of Regional Conferences were held throughout Europe (France, Spain, Croatia, and Italy). These Conferences provided an opportunity for National Rare Disease Alliances to highlight with key stakeholders on how the trends apply to regional contexts; to debate which scenarios should be favoured and to reflect on how policies may help shape a sustainable future for people living with a rare disease in their respective country.



In parallel, seeking to engage young citizens interested in the field of rare diseases in the policy debate, EURORDIS organised a series of meetings, conferences and capacity building opportunities to prepare young patient advocates to provide their opinion on the future of rare disease policy. Young citizens presented their work and policy recommendations to a selected panel of experts in the online Young Citizens Conference Debate held in October 2020.

The final conference for the purpose of presenting the Rare 2030 recommendations will be held in February 2021.

1.4 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) has been under the microscope for quite some time.

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 was 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 as well as the pharmaceutical industry.

The results of the report were published in August 2020 and indicated that both regulations fostered the development and availability of new drugs for patients with rare diseases including children. Nevertheless, the evaluation also highlighted that the drug development had not been efficiently supported in those rare and paediatric areas where the need for new medicines is the highest, while also indicating that European citizens did not have the same access to authorised treatments.

Following a public consultation on the European Commission Inception Impact Assessment (IIA) on Paediatric Medicines and Orphan Medicinal Products, EURORDIS provided comprehensive feedback for future improvements including some of the following suggestions: early-stage multi-stakeholder identification of unmet needs and subsequent priorities and investments; a graduated system of incentives, rewarding earliest dialogue in the areas with no therapeutic options available; a strengthened mandate for the Committee on Orphan Medicinal Products at the European Medicines Agency (EMA); a functional and efficient EU Health Technology Assessment (HTA) Framework and in the interim increased uptake of joint EMA/HTA assessment at the European level.

1.5 Advocate to improve 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. EURORDIS published its Position Paper "[Early Access to Medicines in Europe: Compassionate use to become a reality](#)" in March 2017 suggesting policy proposals.

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 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 published the paper "[Breaking the Access Deadlock to Leave No One Behind](#)" in late 2017 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.

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. EURORDIS' staff is involved in the MoCA Steering Group, in order to ensure the building of a sustainable framework for patient engagement in these dialogues with industry and payers.

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:



1.5.1 Collaborative Efforts on Equity of Access and Sustainable Approaches to the Financing of Innovative Pharmaceuticals (RARE IMPACT)

EURORDIS has been involved in the RARE IMPACT initiative since 2018. RARE-IMPACT aims to identify and validate the challenges to patients' access to gene and cell therapies through engagement with HTA agencies, regulatory bodies, payers, patient groups, clinicians, manufacturers and other experts across Europe. RARE IMPACT is a consortium of manufacturers of gene and cell therapies and umbrella organisations such as the European Federation of Pharmaceutical Industries and Associations (EFPIA), the European Confederation of Pharmaceutical Entrepreneurs (EUCOPE) and the Alliance for Regenerative Medicine (ARM). EURORDIS-Rare Diseases Europe chairs the consortium with the knowledge and support from Dolon Ltd.

RARE IMPACT came to an end in late 2020. The final RARE Impact report was presented at a multi-stakeholder online event, and addresses challenges across four identified areas in the accessibility, assessment, availability and affordability of gene and cell therapies across the European Union. The report highlights seven solutions including calls for:

- + Greater collaboration between EMA, HTA bodies and Heads of Medicines Agencies on guidance on HTA assessment of advanced therapies, as the complexity of these therapies as much as the new type of uncertainties require long term follow-up;
- + A coordinated approach on the development and use of registries serving multiple purposes (e.g. the follow-up of patients, assessment and reimbursement), as the limited set of data and today's fragmented approach needs to be addressed;
- + Greater cooperation and clarity on use of the cross border healthcare provisions, as many advanced therapies are delivered in only a few highly specialised hospital centres across Europe and cannot be delivered in all countries;
- + More informative and technical discussions to grasp the specifics on advanced therapies' cost and value;
- + Payment based on outcomes and payment over time, though innovative payment mechanisms will be pre-empted by the removal of barriers.

1.5.2 Preparing the reimbursement decision: the HTA momentum

Patients' Involvement in EUnetHTA

Although EUnetHTA hasn't defined a framework of interaction with patient organisations, EURORDIS has been supporting its work advising on the principles of patient engagement, identifying patients to be involved in specific procedures, discussing methodologies, guidelines, and tools, disseminating their deliverables, and introducing patient community to EUnetHTA.

EURORDIS contributed to all EUnetHTA public consultations and the preparation of the various EUnetHTA Annual Stakeholder Forum (attending as a chair or speaker).

In 2020, EUnetHTA obtained an extension of its mandate until May 2021 and restructured its activities by mainly focusing on diagnostics and treatments for Covid-19.

Patients' HTA Network and HTA Network Stakeholder Pool

The HTA Network is the forum chaired by DG SANTE composed by national HTA authorities and selected stakeholders that advises the European Commission on the European Cooperation on HTA, in line with the Commission implementing decision 2013/329/EU and the Multi-Annual work programme. It is also supported by the HTA Network Stakeholder Pool, composed, among other categories of stakeholders, of 9 Patients and Consumers Organisations.

EURORDIS assumed the secretariat of the Patients and Consumers group within the HTA Network Stakeholder Pool. EURORDIS also represents Patients and Consumers at the HTA Network, together with the Bureau Européen des Consommateurs, and coordinates common initiatives across the Stakeholder Pool.

In 2020, the HTA Network reconvened online after two years of inactivity.

1.5.3 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 reasons 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.

1.6 Advocate for progress in patients' 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 continues 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.

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". 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).

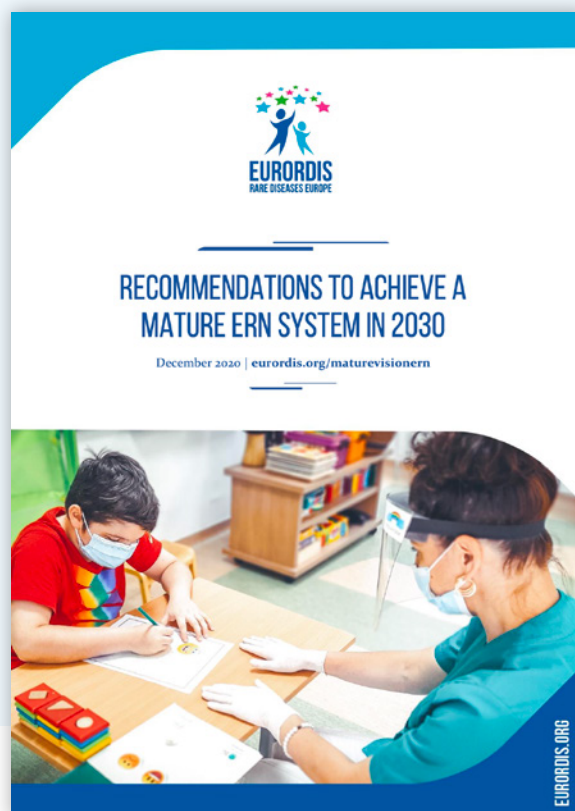
Prior to the publication of the report, EURORDIS has been among the key stakeholders that have been contacted and interviewed since 2017 to provide opinion and evidence. Both the European Commission and the EU Council have agreed to implement these recommendations by 2023, the timeline indicated by the ECA in its report.

Advocate for the development of a mature ERN system

EURORDIS has played a critical role in amplifying the patient voice and creating the conditions for engagement within ERNs so that their activities remain driven by patients' needs.

In 2020, the "[Recommendations to achieve a mature ERN system by 2030](#)" were published. These recommendations were developed by EURORDIS, our member organisations and ERN ePAG patient advocates. The paper, and the accompanying Policy Brief, reviews progress achieved so far and presents our vision of a mature ERN system. It calls for patient organisations and experts to unify within the Networks, and for ERNs to be embedded within healthcare systems across the European Union.

Our vision is for a mature European Reference Network (ERN) system that leaves no person living with a rare disease in uncertainty regarding their diagnosis, care and treatment.



1.7 Advocate to improve access to quality rare disease diagnosis

Throughout 2020, EURORDIS continued to advocate for improved access to and quality of rare disease diagnosis.

1.7.1 Collaborative H2020-funded projects on diagnostic characterisation of rare diseases (Solve RD)



Solve-RD - solving the unsolved rare diseases is a research project funded by the European Commission, aiming to solve large numbers of rare diseases for which a molecular cause is not known yet. 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.

To date, the Solve-RD Project is analysing 8,463 datasets (8,152 whole exome sequencing and 311 whole genome sequencing datasets) including datasets from 5,205 individuals from 4,862 families. The project has already solved 130 rare disease cases for which a molecular cause was not previously known.

EURORDIS is 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.

EURORDIS is also 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 EURORDIS-led CETF has created an infographic setting out the patient journey to diagnosis. The infographic demonstrates the diagnostic odyssey many people experience on a daily basis and presents existing resources from CETF member organisations to support patients on this journey.

1.7.2 Undiagnosed Community

EURORDIS participates in the Undiagnosed Diseases Network International (UDNI), an international network of clinical centres that 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. Patient organisations around the world can officially apply to join the UDNI as members as long as they can demonstrate that their organisation is a certified not-for-profit organisation, 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.



1.7.3 Newborn Screening

Newborn screening is the process of systematically testing newborns just after birth for certain diseases. Ideally, this practice is part of a larger programme that includes confirmatory diagnosis, immediate care, treatment and follow-up. Early diagnosis leads to a better life for people living with a rare disease. Newborn screening is a way to provide this: families are able to plan better for their child's care and treatment and make informed decisions about future pregnancies. In many cases, this early intervention prevents severe disabilities from developing and can save lives.

Throughout 2020, EURORDIS, alongside the Council of National Alliances, Council of European Federations and EURORDIS members, worked towards developing Key Principles to support a harmonised European approach to Newborn Screening. The vast inequalities across Europe, coupled with technological and scientific advances highlight the urgent need to move forward from the status quo as across Europe today there are significant discrepancies between the policies and programmes for newborn screening.

The 11 Key Principles for Newborn Screening will be published in early 2021.



1.8 Promote rare diseases as an international public health priority

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.



Rare Diseases International

Rare Diseases International is a 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 75 member organisations from over 30 countries, that in turn represent rare disease patient groups in more than 100 countries worldwide. The relationship between EURORDIS and Rare Diseases International is regulated by an MoU signed by both organisations.



Rare Disease Day Policy Event



On 25 February 2020, RDI organised "Rare Diseases, leaving no one behind" – a photo exhibit honouring life with a rare disease to mark the occasion of Rare Disease Day. Hosted at the United Nations headquarters in Geneva, on the margins of the 43rd Session of the Human Rights Council, the event was supported by the Permanent Missions of Cyprus and Brazil.

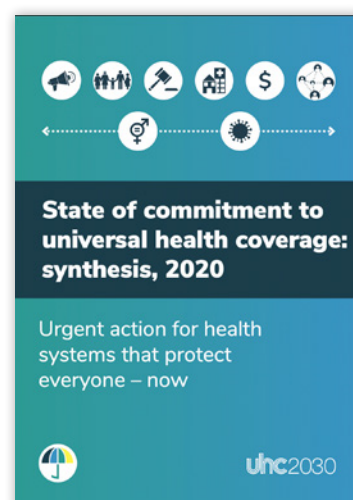
Universal Health Coverage for Rare Diseases (#UHC4RareDiseases) Campaign

In October 2020, RDI launched the 'Universal Health Coverage for Rare Diseases' (#UHC4RareDiseases) campaign, in a joint effort with EURORDIS-Rare Diseases Europe and members of both umbrella organisations. The campaign aimed to create awareness and ask policy makers to safeguard equity and consider the needs of Persons Living With a Rare Disease in national UHC strategies and essential health service packages in the lead up to Universal Health Coverage Day (December 12).

To mark UHC Day 2020, RDI co-organised a webinar with EURORDIS on the 11th December 2020 on 'Addressing Rare Diseases through UHC'. The webinar was attended by 76 participants and included testimonials from the Asia Pacific Organisation for Rare Disorders (APARDO) and the Botswana Organization for Rare Diseases (BORDIS); case studies on advocating for UHC and using the UHC4RareDiseases toolkit by the Federación Mexicana de Enfermedades Raras (FEMEXER), the Finnish Network for Rare Diseases, and the International Prader-Willi Syndrome Organisation (IPWSO); as well as a Keynote Closing Address by Dr. Rüdiger Krech, Director of the Department of Ethics and Social Determinants of Health at the WHO.

Rare Diseases in the UHC2030 Synthesis report

As a member of UHC2030, a multi-stakeholder platform convened by the WHO and the World Bank, RDI continued to raise awareness and represent the voice of Persons Living With a Rare Disease within this forum. A key milestone in 2020 included the inclusion of the challenges of the rare disease community within the UHC2030's "State of Commitment to UHC" synthesis, the first edition of an annual report monitoring action towards Universal Health Coverage in all UN Member states.



Call for a United Nations General Assembly (UNGA) Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families

In 2020, RDI set up the bases for the launch of an official advocacy campaign calling for the adoption of a [UNGA Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their families](#) in 2021. By the end of 2020, RDI was in touch with a core group of UN Member States from different regions of the world and was in a good place to obtain their official support by early 2021.

Memorandum of Understanding between WHO and RDI: 1st year of execution

In December 2019, RDI and WHO signed a Memorandum of Understanding based on ambitious goals and long-term framework of collaboration that will contribute to the WHO 13th General Programme of Work. In the first year of its implementation, activities under the Memorandum of Understanding (MoU) focused on a description framework to define rare diseases internationally and on laying the ground for the development of a global network of centres of excellence for rare diseases.

1.9 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 November, EURORDIS provided feedback in the public consultation of the roadmap of the European Commission for the EU Strategy on the Rights of Persons with Disabilities 2021-2030. Our key recommendations for the European Strategy on the Rights of Persons with Disabilities 2021-2030 included: providing guidance to Member States on disability assessment procedures to ensure persons with all types of disability, including

persons with rare conditions or multiple impairments, are not overlooked and are provided with adequate levels of disability allowance, social protection schemes, community services and independent living arrangements; providing guidelines for Member States on how to ensure reasonable accommodation for persons with disabilities in the workplace; providing Member States with the necessary support to ensure the full implementation of Directive (EU) 2019/1158 on work-life balance for parents and carers of persons with disabilities. The Strategy will be adopted in early 2021.

Throughout 2020, EURORDIS also continued to strengthen its cooperation with the European Disability Forum (EDF), engaging in regular exchanges about common priorities.

1.10 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 2020, the Rare Barometer panel of rare disease patients who answer on a regular basis to EURORDIS' surveys has reached more than 15000 patients.

The H-Care survey pilot aiming to measure rare disease patients experience of their healthcare has been finalised. This pilot survey was initiated by four European Reference Networks (ERKnet, eUROGEN, GENTURIS, ERN LUNG) in collaboration with EURORDIS and Rare Barometer to test the development of a feedback mechanism enabling rare disease patients to assess the care they receive in their hospitals. The pilot also tested the possibility to develop a scientifically validated questionnaire that captures patients' experience with care for the over 8000 rare and complex diseases. It also aimed to test how to best recruit patients and administer the survey across the 24 European



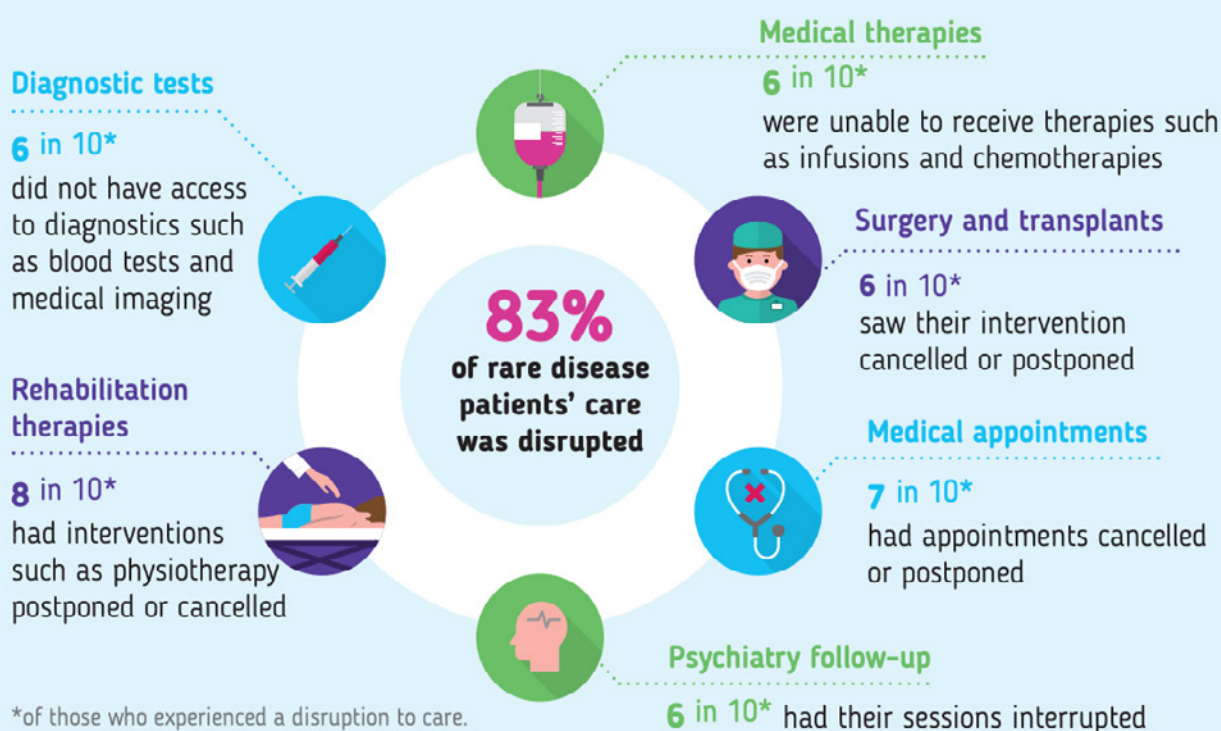
Reference Network. The four participating ERNs have disseminated the survey on site in their hospitals and EURORDIS coordinated the dissemination. Dashboard showing the results of the survey were created for each participating ERNs and hospitals. Overall analysis of the results showed higher satisfaction rates regarding care provided in hospitals affiliated to ERNs.

Results of the survey 'Share and protect our health data' were presented on several occasions, including at the [March PCWP meeting](#) and contributed to feed our positions on this topic in a number of consultations (European health data space consultation in December 2020 for example).

A large quantitative survey aiming to assess the impact of COVID-19 on people living with a rare disease was conducted and disseminated through our patients organisation network. Dashboards showing the number of respondents and the first results of the survey enabled patients organisations to follow the outcome of their dissemination work. More than 8500 responses were received internationally and almost 7000 in Europe. The survey showed that the COVID-19 pandemic has exacerbated the many challenges that people living with a rare disease face. Members of EURORDIS have been using the results of the survey as a basis for their advocacy campaign across Europe and also worldwide.

A significant part of the year was dedicated to discuss the outcomes of this survey with EURORDIS members and external stakeholders through webinars and working groups and to disseminate these outcomes. The survey was used as a basis to define [EURORDIS' positions](#) during the pandemic. An infographic summarising the results of the survey was released in November.

At the end of the year, a new quantitative survey conducted within the Rare 2030 project that aimed to gather rare disease patients' opinions on the future of rare disease on topics such as remote consultations and newborn screening was designed and launched. A new feature enabling to sort the results of Rare Barometer surveys based on the prevalence of the disease of the respondents was developed for this survey and implemented on a permanent basis into the Rare Barometer database.



2. PATIENT EMPOWERMENT:

Building the network & building capacities

2.1 Community Building, Networking & Capacity Building of Patient Advocates

2.1.1 Membership

85 new members joined EURORDIS in 2020. At the end of 2020, EURORDIS had 949 members in 73 countries, 43 of which are European countries, 27 being members of the European Union.

2.1.2 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 53 national alliances, 36 of which constitute the CNA.

The CNA's main activities in 2020 were:

- + a) the preparation and coordination of the Rare Disease Day 2021
- + b) Integration of ERN at National level
- + c) Newborn Screening
- + d) Rare Disease Week 2021

In 2020, two CNA workshops took place online. The first, in March, was a one day workshop for CNA members only. The second, in November was organised over a 3 day gathering of CNA and CEF representatives. Topics included COVID-19, Rare 2030, Newborn Screening, and a new pharmaceutical strategy for Europe amongst others.



2.1.3 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 online in November 2020, together with the CNA to learn about and discuss some of the following items

- + Advocating toward a new European policy framework
- + Recommendations to progress towards a mature ERN system
- + RDI and international activities
- + New pharmaceutical strategy for Europe
- + Manufacturing of Orphan Drugs in Europe

EURORDIS continued for the 11th 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 2020, EURORDIS granted support to 20 European RD Federations to help them organise their different meetings. However, due to the COVID-19 Pandemic, most of the meetings had to be cancelled. In light of the situation, EURORDIS is reviewing the support programme for 2021 in order to expand the grant to the organisation of online meetings as well.

2.1.4 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 19 help lines which are members of the European Network.

In 2020, **19 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, Telefono Verde Malattie Rare, and SAIO), Norway (Norwegian National Advisory Unit on Rare Disorders), Portugal (Linha Rara), Romania (NORO, Myastenia Gravis Romania), Spain (SIO-Feder), Switzerland (Portail Romand d'Informations sur les 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 13th Annual Caller Profile Analysis in October 2020 and participated in discussions on activities of the helplines in the context of COVID-19 and their responses to patient needs. The network also worked on a Code of Conduct to protect enquirers in the context of the Global Data Protection Regulation. EURORDIS organised an online meeting in November 2020 with a main focus on COVID-19.

2.1.5 European Patient Advocacy Groups (ePAGs): Leadership School

The EURORDIS Leadership School empowers European Patient Advocacy Groups (ePAGs) advocates and other rare disease patient advocates to be valued partners in European Reference Networks (ERNs) and when engaging with healthcare providers and other rare disease stakeholders.

For information on the 2020 Leadership School please refer to the Open Academy section.

2.1.6 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.



2.1.7 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 and updates on EURORDIS upcoming events. Webinars comprised an important communication tool during 2020 when face to face meetings were not possible. More than 10 webinars were organised on a number of topics including COVID-19 and its implications on the rare disease community.

2.1.8 Training for patient advocates

EURORDIS OPEN ACADEMY

The EURORDIS Open Academy which was established in 2018 encompasses the EURORDIS Summer School, Winter School, Digital School and Leadership School. 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. The goal is to build the capacity of rare disease patient advocates at large, as well as a select number of researchers and clinicians, so that they can go on to advocate for rare diseases at both local or international levels.

In 2020, EURORDIS delivered 4 training programmes via the EURORDIS Open Academy: the Summer School (medicine research and development); the Winter School (Scientific Innovation and Translational Research); the Leadership School and the Digital School. Due to the COVID-19 pandemic there were no face-to-face sessions for any of these trainings in 2020. The programmes were provided online instead, in a blended format with online live sessions and e-learning courses.

Across the 4 EURORDIS Open Academy programmes organised in 2020, there were over 100 trainees, from more than 35 countries, and 64 trainers.

An open access training platform, the EURORDIS Open Academy e-learning platform is freely available through the EURORDIS website and by the end of 2020 reached 1500 registered users and 50h of training.

EURORDIS Winter School on Scientific
Innovation & Translational Research

Launched in 2018, the EURORDIS Winter School consists of 4.5 days of 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.

The 3rd EURORDIS Winter School on Scientific Innovation & Translational Research was held between 9-13 March 2020. Although initially planned as a face-to-face meeting, the programme was adapted to an online format due to the pandemic, maintaining all the speakers and most content.

30 patient advocates and 20 trainers from 13 countries participated in the 2020 edition of Winter School, which covered important topics to support patient engagement in research. 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.

EURORDIS Summer School on Medicines Research & Development

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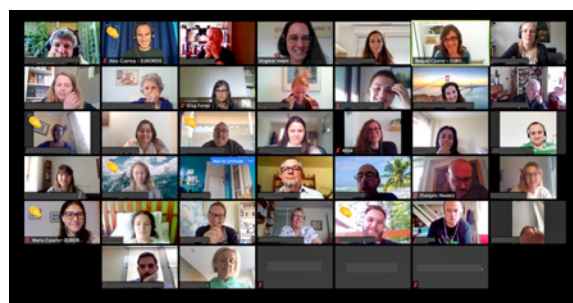


The EURORDIS Summer School, initiated in 2008, aims to provide patient advocates and researchers with the knowledge and skills that they need to become experts in medicines research and development. It is made up of 4.5 days of face-to-face training sessions, preceded by an online pre-training. 36 e-Learning 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 20 trainers.

The pre-training of the EURORDIS Summer School 2020 took place from March to May 2020. The 5-day training took place online, due to the COVID-19 pandemic, on the 8-12th of June 2020. An additional half-day training took place on the 21st September 2020.

This year, great effort was put on optimising the programme for a suitable online delivery, while maintaining the interactivity and hands-on exercises. A new session on “Patient engagement” was also added on the last day of the intensive training. This session aimed at discussing participants’ particular experiences and plans to engage with regulators, companies and researchers, based on a survey sent to the participants ahead.

The Summer School 2020 was attended by 34 participants from 16 countries, representing over 20 rare diseases.



EURORDIS Digital School on Social & Digital Media

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 EURORDIS Digital School on Social & Digital Media is a fully online programme, composed of webinars and e-learning courses, available for free.

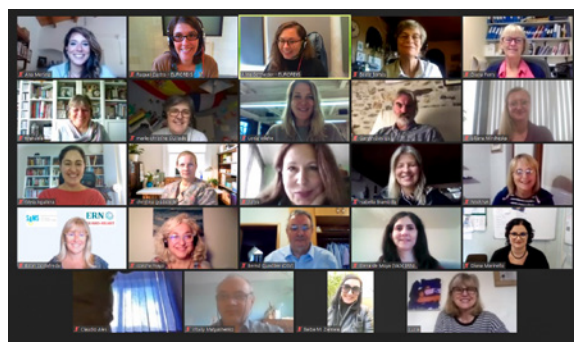
The training objectives range from understanding digital landscapes to learning about creating empowered communities, building effective channels and reaching the right people. The trainers include social and digital media experts as well as representatives of patient organisations with invaluable experience in digital media.

EURORDIS Leadership School on Healthcare & Research

The EURORDIS Leadership School empowers European Patient Advocacy Groups (ePAGs) advocates and other rare disease patient advocates to be valued partners in European Reference Networks (ERNs) and when engaging with healthcare providers and other rare disease stakeholders. Expert trainers from across Europe deliver the training webinars and sessions which normally consists of a series of webinars, from June to November, and an intensive 3-day training in October.

This second edition of the EURORDIS Leadership School took place online, due to the COVID-19 pandemic, from April to November 2020. The 9 webinars took place between April and November whereas the 3-day intensive training days took place on the 6-8th October 2020. The topics of the Leadership School included self-awareness, emotional leadership, communicating with impact, power and influence, conflict resolution and negotiation.

24 ePAG advocates attended the EURORDIS Leadership School 2020, representing 12 countries, over 30 rare diseases and 16 ERNs.



2.2 Raising Awareness & Informing

2.2.1 Rare Disease Day 2020

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.

After the Strategic Review carried out in 2019, more emphasis was put on the co-creation of RDD with the national champions of the campaign, the National Alliances both on the international and European level. Through surveys, virtual meetings and discussions, the



purpose, cause and call to action for the decade ahead were decided and adopted by all partners involved.

In 2020, for the 13th edition of the day, Rare Disease Day events took place in over 100 countries and regions on every corner of the globe. Thousands of events took place in over 100 countries. We welcomed 4

new countries to the campaign: Gabon, Iraq, Kosovo and Rwanda.

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.

Global Chain of Lights

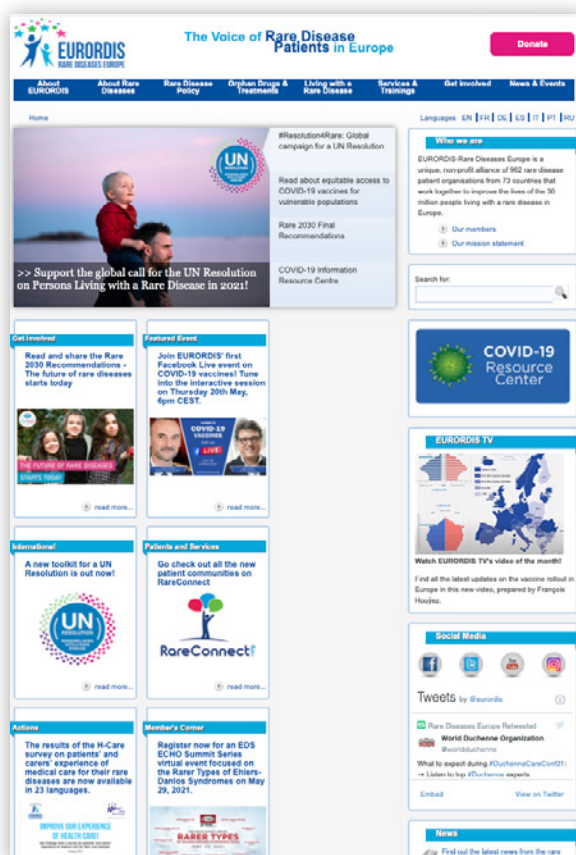
There was a fantastic variety of awareness events including a march to raise awareness in Argentina, a family day in Lebanon, and a discussion on health and educational policies on rare diseases in Tanzania. World famous landmarks including the world's tallest building, the Burj Khalifa in United Arab Emirates, the Colosseum and the Leaning Tower of Pisa in Italy and several monuments in Australia were illuminated in the colours of Rare Disease Day as a symbol of support for the rare disease community.



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.

In 2020 we moved ahead on the backend development of our new website. We began translations and uploading the content. Having a website which is translated in 7 languages is technically challenging and requires several technical and development specifics. The expected launch will be in 2021.



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 2020, 12 eNews issues were distributed to 14,525 subscribers and translated in 7 languages. This publication is year-round at a frequency of once a month. 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 2020 included: Making universal health coverage a reality for people living with a rare disease; The Rare2030 Foresight Study - building a better future for people living with a rare disease; Parliamentary Advocates for Rare Diseases working towards a new EU policy framework for rare diseases.

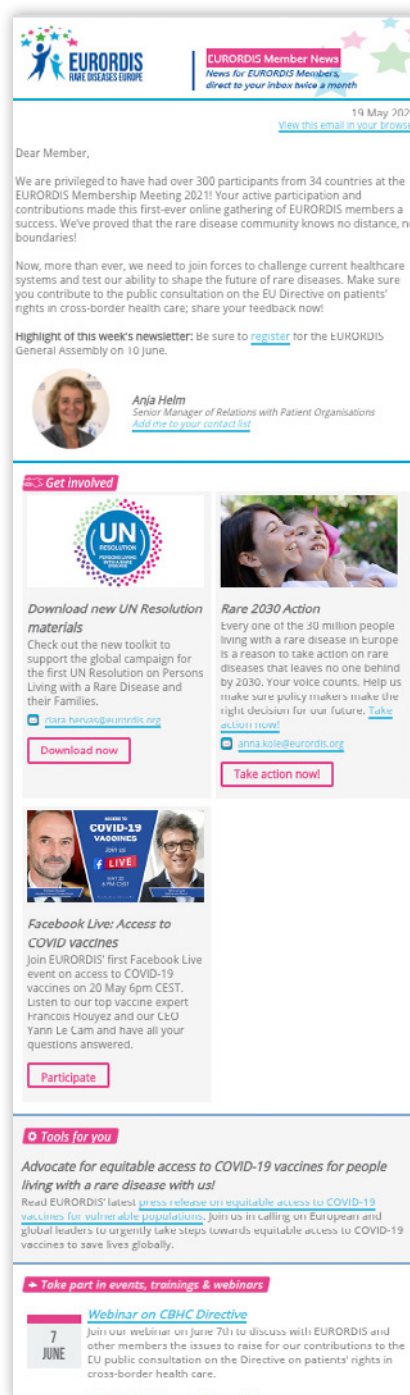


Member News

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 2,400 contacts.

In 2020, our Member News was distributed twice a month to 2,400 contacts at member organisations. In 2020 we launched the new design of the Member News – we have new Get Involved, Tools for you, and Policy Update sections. It also has a calendar of events to be sure our Members don't miss what is happening.

We also launched a new Council of National Alliances specific e-newsletter to better target these important patient organisation members, heavily active in advocacy activities nationally.



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 were celebrated on 18 February 2020 in Brussels and marked the occasion of Rare Disease Day 2020.

The winners of the EURORDIS Awards 2020 were:



Young Patient Advocate Award

Jana Popova (Bulgaria)

For her exceptional awareness raising of issues faced by the SMA community, as well as rare diseases as a whole.



European Rare Disease Leadership Award

Dr. Daria Julkowska (France/Poland)

For the outstanding leadership and dedication Dr. Julkowska has shown to the rare disease community and the positive impact she has had on rare disease research and partnerships in Europe and beyond.



Policy Maker Award

Dr. Vytenis Andriukaitis (Lithuania)

For his outstanding work and support of the rare disease community in his role as European Commissioner for Health and Food Safety 2014-2019.



EURORDIS Volunteer Award

Claudia Croccone (Italy)

For her exceptional work as a patient advocate for HHT and the wider rare disease community, as well as her outstanding contribution to EURORDIS on a volunteer basis for many years.



Scientific Award

Professor Annemieke Aartsma-Rus (The Netherlands)

For her exceptional achievements and dedication in the field of Duchenne Muscular Dystrophy (DMD).



EURORDIS Members Award

ALAN Maladies Rares Luxembourg (Luxembourg)

For their outstanding work in providing empowering information and support to patients living with a rare disease and their families, giving patients greater autonomy and significantly improving their quality of life.

healx

Company Award for Innovation

Healx (UK)

For Healx's promising alternative model for drug discovery, integrating Artificial Intelligence and emerging technologies to identify treatments for rare diseases.



Company Award for Patient Engagement

Boehringer Ingelheim (Germany)

For Boehringer Ingelheim's longstanding commitment to collaborative and comprehensive engagement with rare disease patients and patient organisations, including the Scleroderma, Idiopathic Pulmonary Fibrosis and Cystic Fibrosis Communities.



Company Award for Health Technology

Tobea (Greece)

The Company Award for Health Technology recognises TOBEA's unique SEATRAC device, which facilitates access to outdoor water activities for people with limited mobility. Rare disease and accessibility requirements are often closely linked, and this award celebrates TOBEA's technological solution which promotes well-being and equal access by allowing people to enjoy autonomous access to the sea.



Visual and Audio Media Award

Tomasz Śliwiński & Magda Hueckel (Poland)

For the important and impactful documentary film, 'Our Curse', telling the story of the first six months of their son Leo's life with Congenital Central Hypoventilation Syndrome (CCHS).



Written Media Award

Dr Lisa Sanders (USA)

For her prestigious New York Times Magazine column 'Diagnosis', which has recently inspired a Netflix documentary series of the same name. Her writing has brought the needs and experiences of people living with a rare disease to the attention of a much wider global audience.



Holistic Care Award

EB Haus (Austria)

For EB Haus' holistic approach to delivering outstanding multi-disciplinary care for patients living with Epidermolysis Bullosa; from treating patients with the latest therapies, to educating clinicians and researching potential treatments for the years to come.



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 a 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 2020, EURORDIS worked with the ePAG advocates to develop a shared understanding of their role in the ERNs and worked closely with the ERN project managers, Coordinators and advocates to increase the impact of ePAG advocates involvement in the different activities.

This year we continued 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.

The ePAG Steering Committee meeting took place on the 29 & 30 October 2020 online. 25 ePAG advocates from 20 ERNs attended this meeting. The main topics of discussion were the integration of ERNs into national health systems in addition to areas of common interest to improve patient engagement in the ERNs.



**European
Reference
Networks**

The all-ePAG meeting took place online on 5 & 6 of November. 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. 6 sessions were organised on topics such as Patient-centred outcome measures; Communication Strategy; Governance and Patient engagement in clinical practice guidelines. 155 ePAG advocates from 24 European Reference Networks registered this meeting.

For the second year in a row EURORDIS organised the Leadership School which empowers ePAG advocates to be valued partners in European Reference Networks. This second edition of the EURORDIS Leadership School took place online from April to November 2020 with 9 webinars taking place between April and November an intensive 3-day training organised on 6-8th October 2020. The topics of the Leadership School included self-awareness, emotional leadership, communicating with impact, power and influence, conflict resolution and negotiation. 24 ePAG advocates attended the EURORDIS Leadership School 2020, representing 12 countries, over 30 rare diseases and 16 ERNs.

2020 saw the publication of the paper "Recommendations to achieve a mature ERN system by 2030". These recommendations were developed by EURORDIS, our member organisations and ERN ePAG patient advocates. The paper, and the accompanying Policy Brief, reviews progress achieved so far and presents our vision of a

mature ERN system. It calls for patient organisations and experts to unify within the Networks, and for ERNs to be embedded within healthcare systems across the European Union.

Throughout 2020, EURORDIS and ePAG advocates also worked on the development of a preliminary set of measures to evaluate patient engagement in ERNs. Building a partnership culture to systematically

involve patients in ERNs activities and decision-making structures is challenging, partly because the role of patient representatives and the value of this collaboration is not always understood. The objective of this project was to develop an evaluation framework to assess the impact of patient engagement in the ERNs, to provide evidence of the value of patient-clinician partnership. The ePAG impact assessment framework will be published in 2021.

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

PARADIGM was a 30-month public-private partnership launched on 1 March 2018 and led by the European Patients' Forum (EPF) and EFPIA. Its mission to advance a structured, meaningful and ethical patient engagement in medicines development and aimed 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.

EURORDIS' role in IMI-PARADIGM led to 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 operationalising meaningful engagement.

In addition, EURORDIS was 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.

2020 saw the end of **project PARADIGM**. Building on advances at international level, PARADIGM integrated the needs, perspectives and expectations of all stakeholders involved **to co-produce a set of tools and recommendations to plan, conduct and evaluate patient engagement in medicines development in a meaningful and sustainable manner**. The toolbox includes a monitoring and evaluation framework and metrics to measure the impact of patient engagement for all stakeholders involved in medicines development. The Patient Engagement Toolbox is available online at <http://imi-paradigm.eu/petoolbox/>.

EURORDIS was a PARADIGM partner and leader of the work package developing a sustainability roadmap to optimise patient engagement.

3.2 Patient Engagement in Holistic Care



EURORDIS reinforced its focus on holistic care, mainly through the continued dissemination of its position paper on "Holistic Person-Centred Care", published in May in 2019, and the work of the 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.

3.3 Patient Engagement in Diagnosis

3.3.1 EURORDIS Newborn Screening Working Group

The EURORDIS Newborn Screening Working Group (NBS-WG) was set up to review current policy and practice in the field of NBS, in order to develop principles for harmonious uptake/adoption of the NBS programs across the Member States with a view to delivering maximum benefit and improving outcomes for babies born with rare diseases.

Members of NBS-WG include representatives from patient organisations, international screening societies and international and national federations with a focus on NBS. In 2020 the NBS WG working worked alongside the Council

of National Alliances, Council of European Federations and EURORDIS members, towards developing Key Principles to support a harmonised European approach to Newborn Screening. The vast inequalities across Europe, coupled with technological and scientific advances highlight the urgent need to move forward from the status quo as across Europe today there are significant discrepancies between the policies and programmes for Newborn Screening. The resulting 11 Key Principles for Newborn Screening will be published in early 2021.

3.4 Patient Engagement in Research

3.4.1 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 organisations, 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. 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': EURORDIS 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. The Guidebook was completed in 2020 and for its creation, different types of documents were created: A list of all Building Blocks; A fact sheet form for each Building Block; A power point presentation of the Guidebook, including different figures; A tutorial, explaining the Guidebook in detail.

+ 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 'Rare Disease treatments Access Working Group': Treatments are often unavailable for rare disease patients, especially in **low-and-middle-income Countries...** The goal of this Working Group, **leaving no one behind**, requires that **access to treatments be available for rare disease patients**. This WG was created in 2020 with the objectives of: creating a list of standard-of-care products for rare diseases and make the list available to countries throughout the world. The list should be updated periodically; identifying the barriers to accessing rare disease drugs, particularly in low-and-middle income populations.



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

The European Joint Programme on Rare Diseases (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. The European Joint Programme on Rare Diseases (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.

The EJP RD demonstrates how the centralised collaboration between different stakeholders advances rare disease (RD) research for the benefit of patients. The bridges built between expanded RD research community and the European Reference Networks allow the advancement of RD Virtual Platform and common approach to standards, registries, data and FAIRification. Integration of patients in all activities and close collaboration with funders leads to the long-expected mindset changes on patient-centred research.



EJP RD has two major objectives:

- 1 To improve the integration, the efficacy, the production and the social impact of research on RD through the development, demonstration and promotion of Europe/worldwide sharing of research and clinical data, materials, processes, knowledge and know-how.
- 2 To implement and further develop an efficient model of financial support for all types of research on RD (fundamental, clinical, epidemiological, social, economic, health service) coupled with accelerated exploitation of research results for benefit of patients.

EURORDIS has been involved in the strategic development of the proposal and as such is a member of the Operating Group, which includes the leaders of the different pillars. Indeed, EURORDIS is co-leader of Pillar 3 (capacity building and empowerment training courses for all relevant stakeholders including RD patient representatives) which in itself includes 45 partners across 5 work packages. EURORDIS has coordinated efforts to pull together a coherent programme of training courses and support activities within an adequate budget. EURORDIS is also involved in the transversal activities within Pillar 0 as well as Pillar 1 and Pillar 4.

3.4.3 Collaborative Network for European Clinical Trials for Children (C4C)

The C4C research network brings together pharmaceutical companies, paediatric national networks as well as EU multinational sub-specialty networks, large patient advocacy groups, children's hospitals and other public research organisations from across Europe. The project consortium is a novel collaboration between academic and



private sectors that includes 35 academic and 10 industry partners from 20 European countries, as well as more than 50 third parties and around 500 affiliated partners. C4C (conect4children) aims to facilitate the development of new drugs and other therapies for the entire paediatric population.

3.5 Patient Engagement in lifecycle development

3.5.1 Patients creating their Community Advisory Boards to engage with Industry (EuroCAB programme)

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.

3.5.2 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); the Patients' and Consumers' Working Party (PCWP); and the Pharmacovigilance and Risk Assessment Committee (PRAC).

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

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 2020, EURORDIS selected and proposed or endorsed, patient representatives to be appointed to the COMP.

Patients' representatives and staff involved with Scientific Committees at the EMA dedicate their time, experience and expertise to the tasks of: Discussing and assessing dossiers of medicinal products applying for orphan designation and for orphan status reassessment at the time of Marketing Authorisation; Contribution to Scientific Advice / Protocol Assistance; Reviewing Public Summaries of Opinion on orphan designation and Maintenance/Significant Benefit Assessment reports upon request; Evaluation of medicines for use in paediatric patients; contributing to Scientific Advice and Paediatric Investigation Plans; Contributing to discussions and decisions related to pharmacovigilance and risk-assessment procedures (Referrals, Risk management plans, Signals detection) with special attention to Patient



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

Information on Safety; Ensuring accurate, transparent and available information to patients on authorised medicinal products upon request.

Patients' representatives and staff involved with Scientific Committees at the EMA are also participating to Working Groups organised by the different Committees (e.g. COMP Protocol Assistance Working Group). Patients' representatives and staff involved with Scientific Committees at the EMA are also participating to the Strategic and Learning Review meetings held under the EU Presidency and to workshops organised by the Committees, e.g. Workshop on support for orphan medicines development where Virginie Hivert presented the IRDiRC Orphan Drug Development Guidebook co-developed by EURORDIS.

The patients' representatives and staff involved with Scientific Committees and PCWP (Patients and Consumers Working Party) are Members of the EURORDIS' Therapeutic Action Group.

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

Last year 38 patients were identified, Finally, 32 were involved. Since 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. During 2020, 3 mentors have been involved giving more representativeness and broader views. We have only had 6 drop-out, and the main

reasons were delays in submission of paperwork (CV, DOI, Expert form), procedure withdrawals by the company, and existence of a conflict of interest. This year, we also notice the effect of the COVID-19 pandemic where POs have experienced operational difficulties and shifted priorities due to the increased demand to support their community. Additional workload linked to COVID has also been reported on the EMA side.

We have reduced the drop-out rate, 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 internal meeting between EMA public engagement team and scientific advice team (scientific officers and assistants). During this meeting the need for patient input is discussed and outcomes of the meeting are shared in the monthly conference call.

3.5.3 Post-marketing authorisation

European Medicines Agency

Patients' and Consumers' Working Party (PCWP)

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 2020, due to COVID-19 locked down, one face-to-face meeting took place in the new EMA location in Amsterdam. All other meetings were virtual. The EMA training for patient advocates took place on 23 October (online). Total number of meeting days over the year: **17**

Scientific Committees' Questionnaires to patient

In 2019, the EMA initiated a new procedure to collect the opinions of a large group of patients, for example on the benefit/risks, via a written questionnaire. In 2019, EMA contacted EURORDIS to disseminate the survey to patients on three occasions (for achondroplasia (PDCO), Systemic sclerosis with interstitial lung disease (CHMP), and x-linked retinis pigmentosa (PDCO)) and 269 responses were obtained. In 2020, some European Reference Networks were contacted to improve the outreach to patients: Written consultation procedures: 1 (PDCO); Rare condition: long-chain fatty acid oxidation disorder. Numbers of responses: **62**

Scientific Advisory Group Meetings (SAG)

At the time of the marketing authorisation evaluation, the CHMP might have specific questions for experts and convene scientific advisory group meetings with scientists and patients. EURORDIS contributes to the identification and mentoring of patients.

In 2020, patients were looked for, for 7 SAG meetings.

Pharmacovigilance and Risk Assessment Committee (PRAC)

Virginie Hivert, EURORDIS Therapeutics Development Director has been nominated as PRAC Alternate by the EC for a 3-year mandate starting on March 2019.

Discrepancies between scientific officers' input on the need for patient input and EURORDIS views are discussed on a case-by-case basis always reaching an agreed solution. Follow-up of registration of patients as experts has been improved by including EURORDIS in all communications from EMA to patients and within the scientific advice team.

After reviewing the data from 2020, it was estimated that the time spent per dossier ranges from 3 to 9 hours, depending whether the patient representative was "experienced" or "new". In summary, the time spent for the whole package per month is about 60 hours per month. This period time usually spreads over one month (from SAWP meeting to the next, more or less).

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

EURORDIS activities in pharmacovigilance in 2020: Contribution to EMA report on "Lessons learnt from presence of N-nitrosamine impurities in sartan medicines"; Suspension of ranitidine medicines in the EU; CIOMS working Groups on 1 April, 26 June, 11 September, 20 October on guidelines on Patients Involvement in the Development and Safe Use of Medicines; Presentations made at the PCWP/HCPWP; Patients' challenges when supporting medicine's safety monitoring – how can we improve – Presentation at DIA EuroMeeting - Tuesday, 30 June 2020; Comprehending scientific information: what is the patient perspective? Science Communication of Pharmaco-epidemiology for Patient and Public Health – Presentation at ISPE 17 September 2020

Review of Public Information to Medicines

The European Medicines Agency (EMA) is responsible for providing information about medicines authorised via the centralised procedure that includes information directed to patients and the public. During the preparation of this information, the Agency interacts with patients' and consumers' organisations to ensure that it is appropriately worded and comprehensible to the target audience.

EURORDIS is extensively involved in these activities. In 2020 EURORDIS staff or volunteers reviewed a total of 125 EMA documents for public information.

Documents that are destined for the general public include:

- + Public Summary of Opinion (PSO) of orphan drug designations explain in lay terms the disease, the number of individuals potentially affected in the EU, the medicinal product and the stage of development. A link to the Sponsor and to EURORDIS and Orphanet are provided in the PSOs to help patients obtain more information.
- + Medicine Overviews (previously called EPAR summaries) are shorter documents based on the European Public Assessment Reports that are published at the time of Marketing Authorisation. The EPARs contain information about the development of the product and how the committee reached its recommendations.

- + The Package Leaflet (PL) contains information on what the drug is, what it is used for, how to take the drug, possible side effects and how to store the drug. It is important that this information is easily understandable for the general public.
- + Other documents to be disseminated to the public and EURORDIS members: EMA monthly highlights, Safety alerts, Information on referrals, Information on medicines with black symbol, Risk management plan summaries.

The EMA evaluates yearly the added value of the review of these documents by patients and consumers, and this analysis, described in a report, is approved by the EMA Management Board.

A discussion has taken place with EMA about Public Summary of Opinions: EURORDIS has been asked whether these documents are still of relevance for patients. The answer was positive as PSOs, together with the recently created Orphan Maintenance Assessment Reports, are valuable sources of information for the patients. The relevance of the various sections within the PSOs has also been discussed, including how to make sure that some information can remain up-to-date over time, e.g. by creating links in the document towards relevant websites.

EURORDIS Task Force on Health Technology Assessment (HTA Task Force)

The EURORDIS HTA Task Force is a group of 11 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.

Due to the COVID-19 pandemic the two annual face to face meetings were replaced by 4 online meetings. Representatives of the HTA TF participated in the EUnetHTA Stakeholder Meeting held online on the 4 December 2020. The TF also contributed to the preparation of training materials on patient engagement in HTA in national systems (19 April 2020; 29 October 2020).

The HTA TF contributed to two public consultations: Pharmaceutical Strategy for Europe, European Commission public consultation (July-September 2020); Engaging Stakeholders in EUnetHTA, public consultation EUnetHTA WP4 and 5 (December 2020 – January 2021).

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

The Task Force represents a group of 14 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.

Due to COVID-19, the task force reorganised its work in 2020 with intensive discussions on measures taken to protect people living with rare diseases, and possible consequences. A series of 17 online meetings throughout the year substituted to the two usual face-to-face meetings.

Members of the task force contributed to the following consultations: European Regulatory network strategy 2020-2025; EMA/HMA Discussion paper on the prevention



on shortages; European Parliament report on possible measures to address shortages in the EU; Guidance on the Management of Clinical Trials during the COVID-19 (Coronavirus) pandemic; European Pharmaceutical strategy; Inception Impact Assessment on blood and plasma-derived products; Guidance for registry studies.



4. CROSS-CUTTING PRIORITIES

4.1 Governance

4.1.1 Annual General Assembly

The EURORDIS Annual General Assembly was held online on the morning of 13 May. This was the first time that a General Assembly of EURORDIS was held online and voting was conducted remotely. EURORDIS full members voted on five vacant positions on the Board of Directors. The Board welcomed the following new members to the BoD: Alain Cornet, Lupus Belgium (Belgium) and Maria Montefusco, Rare Diseases Sweden (Sweden). Three Board members were re-elected: Alba Ancochea, FEDER (Spain);

Birthe Byskov Holm, Rare Diseases Denmark (Denmark); Dorica Dan, Romanian Prader Willi Association (Romania).

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: Alain Cornet, Belgium; and Officer: Dorica Dan, Romania.

4.1.2 EURORDIS Strategic Review

On the occasion of EURORDIS' Strategy 2015-2020 coming to an end and with the Rare2030 Foresight project proposing recommendations for 2030 and beyond, EURORDIS commissioned an external strategic review for the purpose of developing its strategy from 2021-2030.

The strategic review commenced in 2020 collecting input from EURORDIS members and stakeholders and will be discussed during the General Assembly 2021 for a final adoption by the EURORDIS BoD by end of 2021.

4.1.3 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



RADOIR

Rare Diseases Foundation of Iran



EURORDIS also has partnerships with a number of learned societies:

European Federation of Internal Medicine (EFIM)



European Society of Human Genetics (ESHG)



European Hospital & Healthcare Federation (HOPE)



International Society for Pharmacoeconomics and Outcomes Research (ISPOR)



International Federation of Social Workers Europe (IFSW-Europe)



European Connected Health Alliance (ECHAAlliance)





4.2 Human resources

4.2.1 EURORDIS Staff

The team comprised of 49 staff members as of 31 December 2020. 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 9 in Brussels. The Chief Executive Officer shares his time between the Paris and Brussels offices. The following were the main changes in human resources in 2020.

4 new positions were created in 2020 (in order of appearance):

- + Public Affairs Manager - EU & National Integrated Advocacy, Kostas Aliogiannis, Brussels

- + Open Academy Manager - eLearning & Outreach, Alejandro Cuenca, Barcelona
- + Operations Junior Manager, Adrien Ruez, Paris
- + Campaign & Digital Communications Senior Manager, Pamela Marinou, Paris (limited contract)

1 position was stopped:

- + Patient Engagement Manager, Elisa Ferrer, Barcelona

1 position has been temporarily interrupted since February 2020:

- + Development Relations Senior Manager, Brian Howard, Paris

4.2.2 EURORDIS Volunteers

In 2020, EURORDIS was privileged to rely on **452 volunteers** including **81 dedicated volunteer patient advocates** and **371 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 in addition to 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 EURORDIS' commitments towards its volunteers.

Focus on EURORDIS volunteer patient advocates (81):

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.

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 / now ADVOC: EURORDIS Advocacy 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 2020, it was composed of 45 full members: **27 volunteers** as well as 18 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 **volunteers** who represent patients on the European Medicines Agency's scientific committees. 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 2020, **16 volunteers** contributed to the DITA's work.

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).

Health Technology Assessment (HTA) Task Force

It is composed of **10 volunteers** and coordinated by two staff members, who also manage the DITA Task Force in order to ensure 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 **9 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 2020, **31 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 2020 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

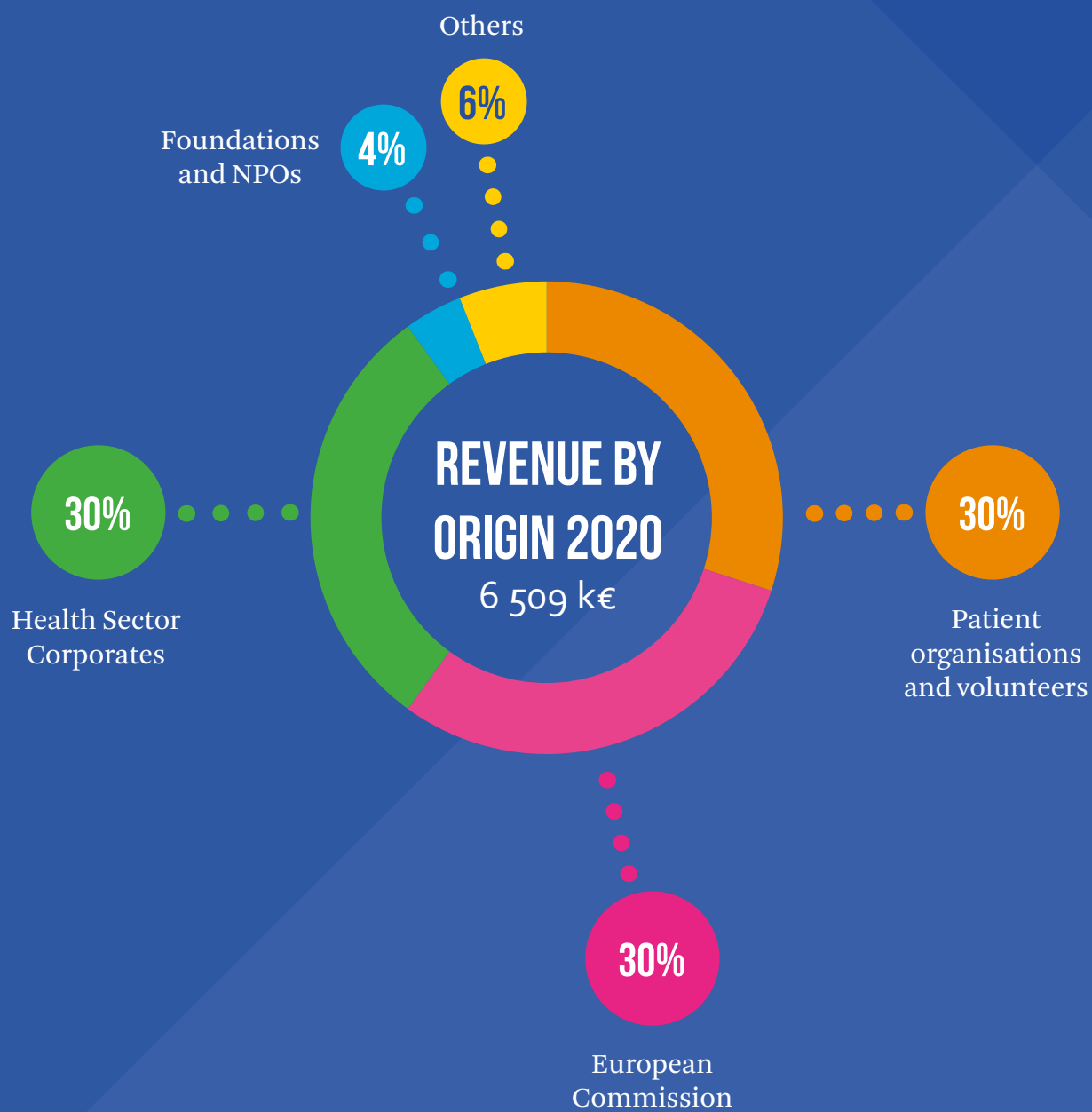
Specific Grant Agreement (Operating Grant) for year 2020 (SGA FY2020), single beneficiary, DG Sante, 12 months

Ongoing

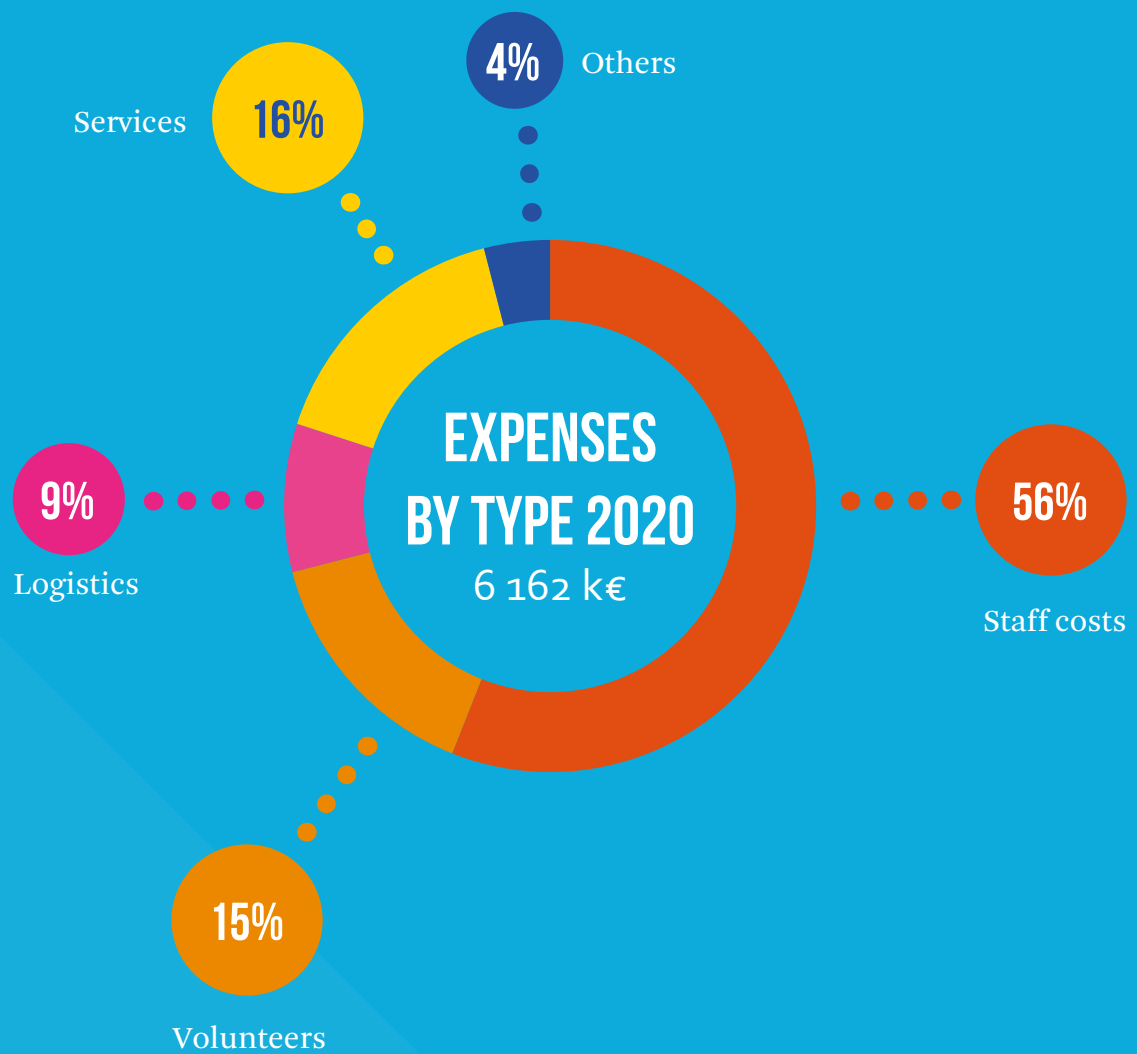
- + Advocacy and core activities, AFM-Téléthon, 2018-2021

- + Framework Partnership Agreement 2018-2021 (Operating Grant), single beneficiary, DG Sante, 2018-2021
- + 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
- + 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 2020



EXPENSES 2020



BOARD of Directors

May 2020 - May 2021

PRESIDENT



Mr Terkel Andersen
Danish Haemophilia Society
 Denmark



Ms Birthe Byskov Holm
Rare Diseases Denmark
 Denmark

DIRECTORS



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




Mr Alexandre Mejat
AFM-Téléthon
 France



Ms Maria Montefusco
Rare Diseases Sweden
 Sweden



Mr Lieven Bauwens
International Federation for Spina Bifida and Hydrocephalus
 Belgium




Ms Jayne Spink
Genetic Alliance UK
 UK




Ms Simona Bellagambi
UNIAMO - Rare Diseases Italy
 Italy




Ms Elizabeth Vroom
World Duchenne Organisation
 Netherlands




Mr Alain Cornet
Treasurer
Lupus Belgium
 Belgium



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



Ms Avril Daly
Vice-President
Rare Diseases Ireland
 Ireland



Ms Vlasta Zmazek
Rare Diseases Croatia
 Croatia



Ms Dorica Dan
Officer
Romanian Prader Willi Association
 Romania

MEMBERS of EURORDIS

FULL MEMBER

ASSOCIATE MEMBER

ALBANIA

SHOQATA E SEMUNDJEVE TE RRALLA / RARE DISEASE ASSOCIATION
ALBANIA

ARGENTINA

ALIANZA ARGENTINA DE PACIENTES
FEDERACIÓN ARGENTINA DE ENFERMEDADES POCO FRECUENTES

ARMENIA

DOCTORS AND CHILDREN HEALTH CARE
NEUROHEREDITARY DISEASES CHARITY ASSOCIATION

AUSTRALIA

CYSTIC FIBROSIS AUSTRALIA
GENETIC ALLIANCE AUSTRALIA
GENETIC SUPPORT NETWORK OF VICTORIA
MUSCULAR DYSTROPHY WA
RARE VOICES AUSTRALIA
SAVE OUR SONS

AUSTRIA

ANGELMAN VEREIN ÖSTERREICH
DEBRA INTERNATIONAL
HAND IN HAND FÜR TAY-SACHS & PALLIATIVKINDER
IC-ÖSTERREICH
NF KINDER – VEREIN ZUR FÖRDERUNG DER
NEUROFIBROMATOSEFORSCHUNG ÖSTERREICH
NF PATIENTS UNITED
PH 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
ALS LIGA BELGIË
ALPHA-1 PLUS ASBL
ASSOCIATION BELGE DU SYNDROME DE MARFAN ASBL
ASSOCIATION LUPUS ERYTHEMATOSUS
ASSOCIATION POUR L'INFORMATION ET LA RECHERCHE SUR LES MALADIES
RÉNALES GÉNÉTIQUES
BE-TSC VZW
BELGISCHE ORGANISATIE VOOR KINDEREN EN VOLWASSENEN MET EEN
STOFWISSELINGSZIEKTE
BELGISCHE VERENIGING VOOR LONGFIBROSE VZW
CF EUROPE
CHILDREN'S TUMOR FOUNDATION EUROPE
CONTACTGROEP MYELOOM EN WALDENSTRÖM PATIËNTEN
DEBRA BELGIUM VZW
EURO ATAXIA - EUROPEAN FEDERATION OF HEREDITARY ATAXIAS
EUROPEAN CMT FEDERATION
EUROPEAN CHROMOSOME 11 NETWORK
EUROPEAN FEDERATION OF WILLIAMS SYNDROME
EUROPEAN HAEMOPHILIA CONSORTIUM
EUROPEAN IDIOPATHIC PULMONARY FIBROSIS & RELATED DISORDERS
FEDERATION
EUROPEAN NETWORK FOR RESEARCH ON ALTERNATING HEMIPLEGIA
EUROPEAN POLIO UNION
EYE HOPE FOUNDATION
FEDERG - FEDERATION OF EUROPEAN ASSOCIATIONS OF PATIENTS
AFFECTED BY RENAL DISEASES
FAMILIAL ADENOMATOUS POLYPOSIS ASSOCIATION
FEDERATION OF EUROPEAN SCLERODERMA ASSOCIATIONS
HTAP BELGIQUE ASBL
ICHTHYOSE BELGIQUE - ICHTHYOSIS BELGIË
INTERNATIONAL FEDERATION FOR SPINA BIFIDA AND HYDROCEPHALUS
MYELOMA PATIENTS EUROPE
RADIOG - 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 PBC, PSC PATIËNTENVERENIGING VZW
VLAAMSE VERENIGING VOOR ERFELIJKE BINDWEEFSELAANDOENINGEN
ZEBRAPAD VZW
VZW GEN

BENIN

ALBINOS SANS FRONTIÈRES

BOSNIA AND HERZEGOVINA

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

BRAZIL

ASSOCIACAO BRASILEIRA DE ENFERMEDADES RARAS
ASSOCIAÇÃO BRASILEIRA DE PARAMILOIDOSE
INSTITUTO VIDAS RARAS

BULGARIA

BULGARIAN ASSOCIATION WILSON DISEASE
BULGARIAN CYSTIC FIBROSIS ASSOCIATION
BULGARIAN HUNTINGTON ASSOCIATION
BULGARIAN NATIONAL ALLIANCE OF PEOPLE WITH RARE DISEASES
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
LYMPHOMA COALITION
PVNH SUPPORT & AWARENESS

CHINA

CHINESE ORGANIZATION FOR RARE DISORDERS
ILLNESS CHALLENGE FOUNDATION

COLOMBIA

ASOCIACIÓN COLOMBIANA DE PACIENTES CON ENFERMEDADES 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 ATYPICAL PARKINSONIAN SYNDROMES
CZECH ASSOCIATION OF MARFAN SYNDROME
CZECH HUNTINGTON ASSOCIATION

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

DENMARK

22Q11 DANMARK
ADDISON FORENINGEN I DANMARK
BLÆREEKSTROFIFORENINGENS
CCHS DANMARK
DANISH APERT SYNDROME ASSOCIATION /DANMARKS APERTFORENING
DANMARKS BLODERFORENING / DANISH HAEMOPHILIA SOCIETY
EHLERS-DANLOS FORENINGEN I DANMARK
FORENINGEN FOR ATAKSI / HSP
HAE SCANDINAVIA
ICHTYOSIS ASSOCIATION IN DENMARK
IMMUN DEFEKT FORENINGEN
LUPUS EUROPE
MCADD-FORENINGEN
MITOKONDRIE-FORENINGEN I DANMARK
MÖBIUS SYNDROM FORENINGEN / MOEBIUS SYNDROME ASSOCIATION IN DENMARK
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 ACOUSTIC NEUROMA ASSOCIATION
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 / FINNISH VASCULITIS ASSOCIATION

FRANCE

ACO₂ GENE ASSOCIATION
AFM-TÉLÉTHON
AFMKT-FRANCE
AVML VIVRE MIEUX LE LYMPHŒDÈME
AGIR POUR LA MALFORMATION LYMPHATIQUE EN ALLIANCE
ALLIANCE MALADIES RARES
ALLIANCE SYNDROME DE DRAVET
AMIS DE ADN P FRANCE
ASS. INTERNATIONALE DE DYSTROPHIE NEURO AXONALE INFANTILE ASSEDEA
ASSOCIATION ISIS
ASSOCIATION «LES PETITS COURAGEUX»
ASSOCIATION «TURNER ET VOUS»
ASSOCIATION AMS-ARAMISE
ASSOCIATION ANÉMIES DYSEYTHROPOIÉTIQUES CONGÉNITALES
ASSOCIATION BPAN FRANCE
ASSOCIATION CONTRE LES MALADIES MITOCHONDRIALES
ASSOCIATION CRANIOPHARYNGIOME SOLIDARITÉ
ASSOCIATION FRANÇAISE STURGE WEBER «VANILLE-FRAISE»
ASSOCIATION FRANÇAISE DU SYNDROME DE LOWE
ASSOCIATION FRANCE VASCULARITES
ASSOCIATION FRANCOPHONE CONTRE LA POLYCHONDRIE CHRONIQUE ATROPHIANTE
ASSOCIATION FRANCOPHONE DES GLYCOGÉNOSES
ASSOCIATION FRANCOPHONE DU SYNDROME D'ANGELMAN
ASSOCIATION FRANÇAISE LESCH-NYHAN ACTION
ASSOCIATION FRANÇAISE NIEMANN PICK
ASSOCIATION FRANÇAISE CONTRE L'AMYLOSE
ASSOCIATION FRANÇAISE DE LA MALADIE DE FANCONI
ASSOCIATION FRANÇAISE DE NARCOLEPSIE-CATAPLEXIE ET HYPERSOMNIE
ASSOCIATION FRANÇAISE DE L'ATAXIE DE FRIEDREICH
ASSOCIATION FRANÇAISE DE L'OSTÉODYSTROPHIE HÉRÉDITAIRE D'ALBRIGHT
ASSOCIATION FRANÇAISE DE L'OSTÉOGENÈSE IMPARFAITE
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 PHELAN-MCDERMID
ASSOCIATION FRANÇAISE DU SYNDROME D'ONDINE
ASSOCIATION FRANÇAISE DU SYNDROME DE CORNELIA DE LANGE
ASSOCIATION FRANÇAISE DU SYNDROME DE KLIPPEL-FEIL
ASSOCIATION FRANÇAISE DU SYNDROME DE RETT
ASSOCIATION FRANÇAISE POUR LE SYNDROME DE GILLES DE LA TOURETTE
ASSOCIATION FRANÇOIS AUPÉTIT
ASSOCIATION GRANDIR
ASSOCIATION HEMOCHROMATOSE FRANCE
ASSOCIATION HISTIOCYTOSE FRANCE
ASSOCIATION HUNTINGTON FRANCE
ASSOCIATION HYPOPARATHYROIDISME FRANCE
ASSOCIATION ICHTYOSE FRANCE
ASSOCIATION INTERNATIONALE MALADIES KYSTES TARLOV
ASSOCIATION KOURIR
ASSOCIATION MALADIES FOIE ENFANTS
ASSOCIATION MARFANS
ASSOCIATION MICROPTALMIE FRANCE
ASSOCIATION NAEVUS₂₀₀₀ FRANCE EUROPE
ASSOCIATION NATIONALE DES CARDIAQUES CONGÉNITAUX
ASSOCIATION NEUROFIBROMATOSES & RECKLINGHAUSEN
ASSOCIATION OLLIER MAFFUCCI EUROPE
ASSOCIATION PEMPHIGUS – PEMPHIGOÏDES FRANCE
ASSOCIATION SOS DESMOÏDE
ASSOCIATION SANS DIAGNOSTIC ET UNIQUE
ASSOCIATION SCLÉROSE TUBÉREUSE DE BOURNEVILLE
ASSOCIATION SPINA BIFIDA ET HANDICAPS ASSOCIÉS
ASSOCIATION STRÜMPPELL-LORRAIN
ASSOCIATION SURRÉNALES
ASSOCIATION SYNDROME DE MOEBIUS FRANCE
ASSOCIATION TANGUY MOYA MOYA
ASSOCIATION DE SOUTIEN ET D'INFORMATION A LA LEUCÉMIE LYMPHOÏDE CHRONIQUE ET LA MALADIE DE WALDENSTRÖM
ASSOCIATION DES GROUPES AMITIÉ TURNER
ASSOCIATION DES MALADES SOUFFRANT D'ANGIO OEDÈME PAR DÉFICIT EN C₁ INHIBITEUR
ASSOCIATION DES MALADES DU SYNDROME DE MCCUNE-ALBRIGHT
ASSOCIATION DES PANCRÉATITES CHRONIQUES HÉRÉDITAIRES
ASSOCIATION DES PATIENTS DE LA MALADIE DE FABRY
ASSOCIATION DES PERSONNES CONCERNÉES PAR LE TREMBLEMENT ESSENTIEL
ASSOCIATION DU NAEVUS GÉANT CONGÉNITAL
ASSOCIATION POUR AIDER ET INFORMER LES SYRINGOMYÉLIQUES EUROPÉENS RÉUNIS
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 POUR LA LUTTE CONTRE LES MALADIES INFLAMMATOIRES DU FOIE ET DES VOIES BILIAIRES
ASSOCIATION POUR LA SENSIBILISATION AUX MALADIES RARES, ORPHELINES ET AUTO-IMMUNES À SAINT-PIERRE-ET-MIQUELON
ASSOCIATION POUR LA LUTTE CONTRE L'ALCAPTONURIE
ASSOCIATION POUR LA RECHERCHE SUR LA SCLÉROSE LATÉRALE AMYOTROPHIQUE
ASSOCIATION SYNDROME DE KLEINE-LEVIN
CHARCOT-MARIE-TOOTH FRANCE
CUTIS LAXA INTERNATIONALE
DEBRA FRANCE
DRAVET SYNDROME EUROPEAN FEDERATION
ENFANTS DE LA LUNE ASSOCIATION POUR LE XERODERMA PIGMENTOSUM
EURO-DYMA
EUROPEAN FEDERATION LESCH-NYHAN DISEASE
EUROPEAN FEDERATION FOR HEREDITARY SPASTIC PARAPLEGIA
EUROPEAN FEDERATION OF ASSOCIATIONS OF PATIENTS WITH HAEMOCHROMATOSIS
EUROPEAN PATIENT ORGANISATION FOR DYSIMMUNE AND INFLAMMATORY NEUROPATHIES
FOP FRANCE
FITIMA EUROPE - FONDATION INTERNATIONAL TIerno ET MARIAM
FRANCE LYMPHANGIOLEIOMYOMATOSE
FRANCE LYMPHOME ESPOIR
FRUCTOS'AMIS POUR LA VIE
FÉDÉRATION DES MALADES DRÉPANOCYTAIRES ET THALASSÉMIQUES SOS GLOBI
GENESPOIR: ASSOCIATION FRANÇAISE DES ALBINISMES
GENIRIS
GÉNÉRATION 22
HTAPFRANCE
HYPOPHOSPHATASIE EUROPE
INCONTINENTIA PIGMENTI FRANCE
INFLAM'ŒIL

LES AMIS DE REMI
 LES ENFANTS DU JARDIN
 LES PETITS BOURDONS
 LIGUE NATIONALE CONTRE LE CANCER
 LIGUE CONTRE LA CARDIOMYOPATHIE
 LUPUS FRANCE
 MED₁₃L SYNDROME ASSOCIATION
 MOSAÏQUES - ASSOCIATION DES «X FRAGILE»
 NAEVUS GLOBAL
 OUVRIER LES YEUX
 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
 VHL FRANCE
 VAINCRE LA MUCOVISCIDOSE
 VAINCRE LA PAPILLOMATOSE RESPIRATOIRE RÉCURRENTE
 VAINCRE LES MALADIES LYSOSOMALES
 VALENTIN APAC - ASSOCIATION DE PORTEURS D'ANOMALIES
 CHROMOSOMIQUES
 VIVRE SANS THYROÏDE
 WHITE SUTTON FRANCE
 WILLIAMS-FRANCE
 X FRAGILE FRANCE
 XTRAORDINAIRE

GEORGIA

GEORGIAN FOUNDATION FOR GENETIC AND RARE DISEASES

GERMANY

AHC-DEUTSCHLAND E.V.
 ARVC-SELBSTHILFE E.V.
 ALLIANZ CHRONISCHER SELTENER ERKRANKUNGEN E.V.
 ANGELMAN E.V.
 BSHV FÜR KINDER, JUGENDLICHE UND ERWACHSENE MIT
 SELTENEN, CHRONISCHEN SKELETTERKRANKUNGEN E.V.
 BUNDESVERBAND KLEINWÜSCHSIGE MENSCHEN UND IHRE FAMILIEN
 E.V.
 BUNDESVERBAND POLIOMYELITIS E.V.
 BUNDESVERBAND SCHILDDRÜSENKREBS - OHNE SCHILDDRÜSE LEBEN
 E.V.
 BUNDESVERBAND DER CLUSTERKOPFSCHMERZEN-SELBSTHILFE-
 GRUPPEN
 BUNDESVERBAND DER EHLERS-DANLOS-SELBSTHILFE E.V.
 BUNDESVERBAND FÜR ANGEBORENE GEFÄSSFEHLBILDUNGEN E.V.
 CRPS BUNDESVERBAND DEUTSCHLAND E.V.
 CHARGE SYNDROM E.V.
 CHOLESTERIN & CO E.V.
 CYSTINOSE SELBSTHILFE E.V.
 DEUTSCHE DUCHENNE STIFTUNG
 DEUTSCHE EHLERS-DANLOS INITIATIVE E.V.
 DEUTSCHE GBS CIDP SELBSTHILFE E.V.
 DEUTSCHE INTERESSENGEMEINSCHAFT PKU
 DEUTSCHE KLINEFELTER-SYNDROM VEREINIGUNG E.V.
 DEUTSCHE NARKOLEPSIE GESELLSCHAFT E.V.
 DEUTSCHE SARKOIDOSE VEREINIGUNG 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.
 EUROPEAN ASSOCIATION OF PATIENT ORGANISATIONS OF SARCOIDOSIS
 EUROPEAN CONGENITAL HEART DISEASE ORGANISATION
 EUROPEAN MEN ALLIANCE
 EUROPEAN NETWORK FOR ICHTHYOSIS
 FOP GERMANY (FÖRDERVEREIN FÜR AN FIBRODYSPLASIA OSSIFICANS
 PROGRESSIVA ERKRANKTE)
 FAUN STIFTUNG
 GAUCHER GESELLSCHAFT DEUTSCHLAND E.V.
 GESELLSCHAFT FÜR MUKOPOLYSACCHARIDosen E.V.
 HAE VEREINIGUNG E.V. (HEREDITARY ANGIOEDEMA)
 HSP-SELBSTHILFEGRUPPE DEUTSCHLAND E.V.
 HAND IN HAND GEGEN TAY-SACHS UND SANDHOFF E.V.
 HOFFNUNGSBAUM E.V.
 ICA-DEUTSCHLAND E.V. FÖRDERVEREIN INTERSTITIELLE CYSTITIS
 IEB E.V. 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 BEHCET IN DEUTSCHLAND

LEONA E.V.
 MPN-NETZWERK E.V.
 MPS EUROPE
 MARFAN EUROPE NETWORK
 MARFAN HILFE DEUTSCHLAND E.V.
 MORBUS-OSLER SELBSTHILFE E.V.
 MULTINATIONAL INTERSTITIAL CYSTITIS ASSOCIATION
 MYELITIS E.V.
 NCL-GRUPPE DEUTSCHLAND E.V.
 NEPHIE
 NETZWERK HYPOPHYSEN- UND NEBENNIERENERKRANKUNGEN E. V.
 OIFE - OSTEOGENEIS IMPERFECTA FEDERATION EUROPE
 PCH-FAMILIE E.V.
 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.
 SMA EUROPE
 SSADH-DEFIZIT 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.
 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

ASSOCIATION OF GREEK FRIENDS FOR PAEDIATRIC IMMUNOLOGY PRIMARY
 IMMUNODEFICIENCIES «HARMONY»
 CHILD'S HEART
 GREEK ALLIANCE FOR RARE DISEASES
 HELLENIC CYSTIC FIBROSIS ASSOCIATION
 HELLENIC FRIEDREICH'S ATAXIA ASSOCIATION
 HELLENIC LEAGUE AGAINST RHEUMATISM
 HELLENIC MYASTHENIA GRAVIS ASSOCIATION
 IMMUNE DEFICIENCY ASSOCIATION GALINOS
 KRIKOS ZOIS SOCIETY FOR PATIENTS AND FRIENDS OF PATIENTS WITH
 INHERITED METABOLIC DISEASE
 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
 TO MELLON- ASSOCIATION OF PEOPLE WITH GENETIC DISORDERS
 TOGETHER FOR LIFE
 VHLFA ALLIANCE IN GREECE
 "95" - RARE ALLIANCE 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 MITOCHONDRIÁLIS BETEGEK ALAPÍTVÁNYA
 PRIMER IMMUNHIÁNYOS 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

INDIA

INDIAN ORGANIZATION FOR RARE DISEASES
 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

DEBRA IRELAND

EUROPEAN SICKLE CELL FEDERATION

FIGHTING BLINDNESS

FRIEDREICH'S ATAXIA RESEARCH ALLIANCE

HUNTINGTON'S DISEASE ASSOCIATION OF IRELAND

IRISH CANCER SOCIETY

NEUROFIBROMATOSIS ASSOCIATION OF IRELAND

POPSYCLE FOUNDATION

RARE DISEASES IRELAND

SICKLE CELL AND THALASSAEMIE IRELAND

THE CAVAN TOMMY HOEY TRUST

THE IRISH FRAGILE X SOCIETY

THE IRISH SOCIETY FOR MUCOPOLYSACCHARIDE AND RELATED DISEASES

VASCULITIS IRELAND AWARENESS

ISRAEL

CDLS ISRAELI FOUNDATION

GRIN DISORDERS RESEARCH FOUNDATION

ISRAEL ADULT POLYGLUCOSAN BODY DISEASE

LITTLE STEPS ASSOCIATION

ITALY

ABC ASSOCIAZIONE BAMBINI CRI DU CHAT

ACMT- RETE PER LA MALATTIA DI CHARCOT-MARIE-TOOTH ODV

AIBWS ODV

ACONDROPLASIA - INSIEME PER CRESCERE - ONLUS

AMICI DELLA PORFIRIA - SAN PIO DA PIETRELCINA ONLUS

ANGELI NOONAN

ASSOCIAZIONE PERSONE CON MALATTIE REUMATICHE

ASSOCIAZIONE NAZIONALE PEMFIGO/PEMFIGOIDE

ASSOCIAZIONE FAMIGLIE DI SOGGETTI CON DEFICIT DELL'ORMONE DELLA CRESCITA ED ALTRE PATOLOGIE

ASSOCIAZIONE FONDAZIONE ITALIANA HHT "ONILDE CARINI"

ASSOCIAZIONE ITALIANA ADRENOLEUCODISTROFIA ONLUS

ASSOCIAZIONE ITALIANA CISTITE INTERSTIZIALE

ASSOCIAZIONE ITALIANA DEI CARDIOPATICI CONGENITI ADULTI - ITALIAN GUCH ASSOCIATION

ASSOCIAZIONE ITALIANA ESTROFIA VESCICALE-EPISPADIA ODV

ASSOCIAZIONE ITALIANA GAUCHER ONLUS

ASSOCIAZIONE ITALIANA GLICOGENOSI

ASSOCIAZIONE ITALIANA LARYNGECTOMIZZATI

ASSOCIAZIONE ITALIANA LEUCODISTROFIE UNITE

ASSOCIAZIONE ITALIANA LINFOSTIOTICITOSI EMOFAGOCITICA

ASSOCIAZIONE ITALIANA MALATI DI ALCAPTONURIA

ASSOCIAZIONE ITALIANA MALFORMAZIONE DI ARNOLD-CHIARI CHILD

ASSOCIAZIONE ITALIANA MIASTENIA ONLUS

ASSOCIAZIONE ITALIANA MIASTENIA E MALATTIE IMMUNODEGENERATIVE - AMICI DEL BESTA ONLUS

ASSOCIAZIONE ITALIANA MORBO DI HIRSCHSPRUNG

ASSOCIAZIONE ITALIANA MUCOPOLISACCARIDOSI E MALATTIE AFFINI

ASSOCIAZIONE ITALIANA NIEMANN PICK E MALATTIE AFFINI - ONLUS

ASSOCIAZIONE ITALIANA SCLEROSI LATERALE AMIOTROFICA (SEZ. LOMBARDIA)

ASSOCIAZIONE ITALIANA SINDROME X FRAGILE ONLUS

ASSOCIAZIONE ITALIANA SINDROME DI EHLERS-DANLOS

ASSOCIAZIONE ITALIANA SINDROME DI POLAND

ASSOCIAZIONE ITALIANA SINDROME E MALATTIA DI BEHÇET

ASSOCIAZIONE ITALIANA SIRINGOMIELIA E ARNOLD CHIARI

ASSOCIAZIONE ITALIANA SOSTEGNO MALATTIE METABOLICHE EREDITARIE ONLUS

ASSOCIAZIONE ITALIANA PER LA LOTTA ALLE PHTS

ASSOCIAZIONE ITALIANA PER LE MALFORMAZIONI ANORETTALI

ASSOCIAZIONE LAM ITALIA ONLUS

ASSOCIAZIONE LIGURE THALASSEMICI ONLUS

ASSOCIAZIONE MALATI DI HAILEY HAILEY DISEASE

ASSOCIAZIONE MALATTIE RARE DELL'ALTA MURGIA ONLUS

ASSOCIAZIONE NAZIONALE ANGIOMA CAVERNOSO CEREBRALE

ASSOCIAZIONE PER L'INFORMAZIONE E LO STUDIO DELLA ACONDROPLASIA

ASSOCIAZIONE PERSONE WILLIAMS ITALIA ONLUS

ASSOCIAZIONE S.P.R.I.N.T.

ASSOCIAZIONE SCLEROSI TUBEROSA

ASSOCIAZIONE SINDROME NEFROSICA ITALIA

ASSOCIAZIONE SINDROME DI ALSTRÖM / ITALIAN ASSOCIATION FOR ALSTROM SYNDROME

ASSOCIAZIONE STUDIO MALATTIE METABOLICHE EREDITARIE ONLUS

ASSOCIAZIONE VENETA PER LA LOTTA ALLA TALASSEMIA

ASSOCIAZIONE CONTO ALLA ROVESCIA

ASSOCIAZIONE PER LE IMMUNODEFICIENZE PRIMITIVE ONLUS

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

FOP ITALIA ONLUS

FEDERAZIONE SINDROME DI PRADER WILLI ITALIA

FONDAZIONE ALESSANDRA BISCEGLIA W ALE ONLUS

FONDAZIONE LEGA ITALIANA RICERCA HUNTINGTON

GLI AMICI DI DANIELA

GLI EQUILIBRISTI HIBM

GRUPPO «ITALIA - GLIOBLASTOMA MULTIFORME - CANCRO AL CERVELLO»

GRUPPO FAMIGLIE DRAVET ASSOCIAZIONE ONLUS

GRUPPO FAMILIARI BETA-SARCOGLICANOPATIE

GRUPPO ITALIANO PER LA LOTTA ALLA SCLERODERMIA ONLUS

HHT EUROPE

HHT ONLUS

INCONTINENTIA PIGMENTI ASSOCIAZIONE ITALIANA ONLUS

LND FAMIGLIE ITALIANE ONLUS

LA STRADA PER L'ARCOBALENO

LINF4 ODV

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

PKS KIDS ITALIA ONLUS

PARENT PROJECT APS

RETINA ITALIA ONLUS

RING 44 INTERNATIONAL

SOD ITALIA - ASSOCIAZIONE ITALIANA DISPLASIA SETTO OTTICA E

IPOPLASIA DEL NERVO OTTICO

UNIAMO - RARE DISEASES ITALY

UN FILO PER LA VITA ONLUS

UNIONE ITALIANA ITTIOSI

UNIONE ITALIANA LOTTA ALLA DISTROFIA MUSCOLARE

UNIONE TRAPIANTATI POLMONE - PADOVA ODV

UNITED ONLUS

WORLD FEDERATION OF INCONTINENCE PATIENTS

XLPRD INTERNATIONAL ASSOCIATION

JAPAN

JAPAN PATIENT ASSOCIATION

KAZAKHSTAN

PATIENTS WITH CANCER AND RARE DISEASES SUPPORT ASSOCIATION

KOSOVO

RARE DISEASES KOSOVO (SHOQATA E SEMUNDJE TE RRALLA KOSOVE)

LATVIA

ASSOCIATION OF PEOPLE WITH SPECIAL NEEDS «MOTUS VITA»

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

EEN HÄERZ FIR KRIIBSKRANK KANNER ASBL

FONDATIOUN KRIIBSKRANK KANNER

RETT SYNDROME EUROPE

SYNDROME DE MATFAN DEN-I-LU ASBL

MALAYSIA

MALAYSIA METABOLIC SOCIETY

MALAYSIAN RARE DISORDERS SOCIETY

MALTA

NATIONAL ALLIANCE FOR RARE DISEASES SUPPORT - MALTA

MEXICO

ASOCIACION DE GAUCHER DE 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
CMTC-OVM
CHILDHOOD CANCER INTERNATIONAL - EUROPE
CORNELIA DE LANGE SYNDROME WORLD FEDERATION
EUROPEAN CLEFT ORGANISATION
EUROPEAN SOCIETY FOR PHENYLKETONURIA
EUROPEAN VHL (VON HIPPEL-LINDAU) FEDERATION
EUROPEAN WALDENSTRÖM MACROGLOBULINEMIA NETWORK
FSHD EUROPE
FABRY SUPPORT & INFORMATIE GROEP NEDERLAND
FIBRODYSPLASIA OSSIFICANS PROGRESSIVA STICHTING NEDERLAND
ITP PATIENTENVERENIGING
INTERNATIONAL MITO PATIENTS
INTERNATIONAL PAINFUL BLADDER FOUNDATION
INTERNATIONAL PORPHYRIA PATIENT NETWORK
INTERSTITIËLE CYSTITIS PATIENTENVERENIGING
KAISZ - CHILDREN WITH A AUTOINMUUM OR AUTOINFLAMMATORY DISEASE
MSS (MARSHALL-SMITH SYNDROME) RESEARCH FOUNDATION
NATIONAL ASSOCIATION REUMAZORG NEDERLAND
NATIONALE VERENIGING L.E. PATIËNTEN
NEDERLANDS NETWERK VOOR LYMFODEEM EN LIPOEDEEM
NEDERLANDSE HYPOFYSE STICHTING (DUTCH PITUITARY FOUNDATION)
NEDERLANDSE LEVERPATIENTEN VERENIGING
NEDERLANDSE PHENYLKETONURIE VERENIGING / DUTCH PKU ASSOCIATION
NEDERLANDSE VERENIGING VAN HEMOFILIE-PATIËNTEN/NETHERLANDS HAEMOPHILIA SOCIETY
NEPHCEUROPE
NEUROFIBROMATOSE VERENIGING NEDERLAND
OSCAR NEDERLAND
PATIENTENORGANISATIE FIBREUZE DYSPLASIE
PATIËNTENVERENIGING VOOR BLAASEXTHROPHIE NEDERLAND
SARCOIDOSE.NL
SPIERZIEKTEN NEDERLAND - DUTCH ASSOCIATION FOR NEUROMUSCULAR DISEASE
STICHTING AA & PNH CONTACTGROEP
STICHTING AMYLOIDOSE NEDERLAND SAN
STICHTING CHRISTIANSON SYNDROME EUROPE
STICHTING HART4ONDERZOEK / HEART4RESEARCH
STICHTING HISTIOCYTOSE NEDERLAND
STICHTING IJZERSTERK
STICHTING KANS VOOR PKAN KINDEREN
STICHTING NET-GROEP
STICHTING OVERDRUKSYNDROOM NL
STICHTING RPF NEDERLAND / DUTCH RPF FOUNDATION
STICHTING SHWACHMAN DIAMOND SYNDROME SUPPORT HOLLAND
STICHTING SPIERKRACHT
STICHTING STOFWISSELKRACHT
STICHTING TAPS SUPPORT/TAPS SUPPORT FOUNDATION
STICHTING TERRE - RETT SYNDROME FOUNDATION
STICHTING VOOR AFWEERSTOORNISSEN
STICHTING ZELDZAME BLOEDZIEKTEN
THYROID CANCER ALLIANCE
VSOP - VERENIGING SAMENWERKENDE OUDER EN PATIËNTENORGANISATIES
VASCULITIS STICHTING
VERENIGING VAN EHLERS DANLOS PATIENTEN
VERENIGING VOOR ICHTHYOSIS NETWERKEN
VOLWASSENEN, KINDEREN EN STOFWISSELINGSZIEKTEN
WORLD ALLIANCE OF PITUITARY ORGANIZATIONS
WORLD DUCHENNE ORGANIZATION

NEW ZEALAND

RARE DISORDERS NZ

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

ANIRIDIA EUROPE
EUROPEAN HUNTINGTON ASSOCIATION
FRAMBU - RESOURCE CENTRE FOR RARE DISORDERS
HYPOPARA NORGE
INTERNATIONAL HUNTINGTON ASSOCIATION
MPS-FORENINGEN I NORGE
MORBUS ADDISON ASSOCIATION NORWAY
NORSK FORENING FOR ARVELIG SPASTISK PARAPARESE / ATAKSI
NORSK FORENING FOR EHLERS-DANLOS SYNDROM
NORSK FORENING FOR OSTEOGENESIS IMPERFECTA/THE NORWEGIAN OSTEOGENESIS IMPERFECTA ASSOCIATION
NORSK FORENING FOR TUBEROS SKLEROSE
NORWEGIAN FEDERATION OF ORGANISATIONS OF DISABLED PEOPLE (FUNKSJONSHEMMEDES FELLESGORGANISASJON)
NORWEGIAN ORGANISATION FOR PRADER WILLIS SYNDROME
OSLERFORENINGEN NORGE

POLAND

DEBRA POLSKA
FOUNDATION OF BORYS THE HERO /FUNDACJA BOHATERA BORYSA
FUNDACJA SMA (SMA FOUNDATION POLAND)
FUNDACJA SANFILIPPO
FUNDACJA UMIEC POMAGAC (FOUNDATION FOR RD MPS)
MATIO-FUNDACJI POMOCY RODZINOM I CHORYM NA MUKOWISCYDOZE/
MATIO POLISH CF FOUNDATION
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 RADZIWILLOWA CHILD'S HEART FOUNDATION

PORTUGAL

ALIANÇA PORTUGUESA DE ASSOCIAÇÕES DAS DOENÇAS RARAS
ASSOCIAÇÃO PORTUGUESA CDG E OUTRAS DOENÇAS METABÓLICAS
ASSOCIAÇÃO PORTUGUESA DE DOENTES NEUROMUSCULARES
ASSOCIAÇÃO PORTUGUESA DE LEUCEMIAS E LINFOMAS
ASSOCIAÇÃO NACIONAL DE DISPLASIAS ÓSSEAS
ASSOCIAÇÃO NACIONAL PARA DIVULGAR E ORIENTAR PARA COMBATER E ENFRENTAR A TAY SACHS E OUTRAS GANGLIOSIDOSES
ASSOCIAÇÃO PORTUGUESA DE CHARCOT-MARIE-TOOTH
ASSOCIAÇÃO PORTUGUESA DE INSUFICIENTES RENAIS
ASSOCIAÇÃO PORTUGUESA DE OSTEOGÊNESE IMPERFEITA
ASSOCIAÇÃO SANFILIPPO PORTUGAL
EVITA
FEDRA - FEDERAÇÃO PORTUGUESA DE DOENÇAS RARAS
LIGA PORTUGUESA CONTRA AS DOENÇAS REUMÁTICAS
RARÍSSIMAS - ASSOCIAÇÃO NACIONAL DE DEFICIÊNCIAS MENTAIS E RARAS
ANDLINFJA ASSOCIAÇÃO NACIONAL DE DOENTES LINFÁTICOS / NATIONAL ASSOCIATION OF SUFFERERS OF LYMPHATIC DISORDERS

REPUBLIC OF MOLDOVA

COP III PLO II

ROMANIA

ASOCIATIA COPILOR CU BOLI MITOCONDRIALE/ASSOCIATION OF CHILDREN WITH MITOCHONDRIAL DISEASES
ASOCIATIA COPILUL MEU- INIMA MEA
ASOCIATIA NATIONALA MIASTENIA GRAVIS ROMANIA /ROMANIAN MYASTHENIA GRAVIS ASSOCIATION
ASOCIATIA PERSOANELOR CU GLICOGENOZA (APG ROMANIA)
ASOCIATIA PERSOANELOR CU TALASEMIE MAJORA
ASOCIATIA ROMANA SPINA BIFIDA SI HIDROCEFALIE
ASOCIATIA SMACARE
ASOCIATIA WERDNIG HOFFMAN AWH
ASOCIATIA ROMÂNĂ DE CANCERE RAR
AUTOIMMUNE DISEASES PATIENTS ASSOCIATION
CHARCOT MARIE TOOTH ROMANIA ASSOCIATION
DMD CARE
MASTOCYTOSIS SUPPORT ASSOCIATION ROMANIA
NEURO MOVE CMT ASSOCIATION
ROMANIAN NATIONAL ALLIANCE FOR RARE DISEASES
ROMANIAN PRADER WILLIS ASSOCIATION

RUSSIAN FEDERATION

FABRY RUSSIA
HELP TO CYSTIC FIBROSIS PATIENTS
INTER-REGIONAL SUPPORT CENTRE FOR PATIENTS WITH ANIRIDIA «IRIS»
INTERREGIONAL PUBLIC ORGANISATION FOR GAUCHER DISEASE
INTERREGIONAL PUBLIC ORGANISATION OF PATIENTS WITH FABRY DISEASE «THE ROAD TO LIFE»
LOOK TO SEE
MPS RUSSIA

NGO «FRAGILE CHILDREN»

NATIONAL ASSOCIATION OF PATIENTS WITH RARE DISEASES »GENETICA»
RUSSIAN ASSOCIATION OF RARE DISEASES
RUSSIAN PATIENT UNION
RUSSIAN RETT SYNDROME ASSOCIATION
SPIORZ UNION OF PATIENTS WITH RARE DISEASES AND RARE DISEASE PATIENTS ORGANISATIONS
THE ASSOCIATION OF PRIMARY IMMUNODEFICIENCY PATIENTS

SERBIA

CHILD RARE DISEASE SUPPORT AND RESEARCH ASSOCIATION LIFE
CHILDHOOD CANCER PARENT ORGANISATION «ZVONCICA»
LYMPHOMA PATIENTS' ASSOCIATION
NATIONAL ORGANIZATION FOR RARE DISEASES OF SERBIA

SINGAPORE

RARE DISORDERS SOCIETY (SINGAPORE)

SLOVAKIA

DEBRA SR
ORGANISATION OF MUSCULAR DYSTROPHY IN THE SLOVAK REPUBLIC
SLOVAK ALLIANCE OF RARE DISEASES
SLOVAK CYSTIC FIBROSIS ASSOCIATION
ZDRUZENIE OJEDLINELYCH GENETICKYCH OCHORENIE

SLOVENIA

ASSOCIATION OF PATIENTS WITH BLOOD DISEASES - DRUSTVO BOLNIKV S KRVNIMI BOLEZNIMI
DEBRA SLOVENIA - DRUSTVO DEBRA SLOVENIJA
EAMDA - EUROPEAN ALLIANCE OF NEUROMUSCULAR DISORDERS ASSOCIATIONS
FABRY PATIENTS ASSOCIATION SLOVENIA / DRUŠTVO BOLNIKOV S FABRYJEVO BOLEZNIJO SLOVENIJE
FOUNDATION OF CHILD NEUROLOGY
VILJEM JULIJAN ASSOCIATION FOR CHILDREN WITH RARE DISEASES
ZAVOD BERNARDI VENTRELLA

SOUTH AFRICA

PRIMARY IMMUNODEFICIENCY NETWORK OF SOUTH AFRICA
RARE DISEASES SOUTH AFRICA NPC

SPAIN

AHUCE - ASOCIACIÓN NACIONAL HUESOS DE CRISTAL OI ESPAÑA
ACCIÓN Y CURA PARA TAY-SACHS
ALIANZA ESPAÑOLA DE FAMILIAS DE VON HIPPEL LINDAU
ASOCIACION ALBI ESPAÑA
ASOCIACION ANDALUZA DE FIBROSIS QUISTICA
ASOCIACION DE ENFERMEDADES RARAS DGENES
ASOCIACIÓ CATALANA DE LAS NEUROFIBROMATOSIS
ASOCIACIÓN ANDALUZA DE PACIENTES CON SÍNDROME DE TOURETTE Y TRASTORNOS ASOCIADOS
ASOCIACIÓN ARTROGRIPOSIS MÚLTIPLE CONGÉNITA
ASOCIACIÓN CHIARI Y SIRINGOMIELIA DEL PRINCIPADO DE ASTURIAS
ASOCIACIÓN CIUDADANA DE AFECTADOS DE CISTITIS INTERSTICIAL
ASOCIACIÓN DE AFECTADOS POR HIPERINSULINISMO CONGÉNITO
ASOCIACIÓN DE EPIDERMOLISIS BULLOSA DE ESPAÑA (DEBRA SPAIN)
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ASOCIACIÓN NACIONAL AMIGOS DE ARNOLD CHIARI
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CRANEOSINOSTOSIS SINDRÓMICAS

ASOCIACIÓN NACIONAL DE HIPERTENSIÓN PULMONAR
ASOCIACIÓN RETINA MURCIA
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ASOCIACIÓN DE AFECTADOS POR DISPLASIA ECTODÉRMICA
ASOCIACIÓN DE ATROFIA DE NERVO OPTICO DE LEBER
ASOCIACIÓN DE ESCLERODERMIA CASTELLON
ASOCIACIÓN DE FAMILIARES Y AFECTADOS POR LIPODISTROFAS
ASOCIACIÓN DE FAMILIARES Y ENFERMOS DE FPI Y FPF
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DUCHENNE PARENT PROJECT ESPAÑA
EUROPEAN NETWORK FOR RARE AND CONGENITAL ANAEMIAS
FEDER - FEDERACIÓN ESPAÑOLA DE ENFERMEDADES RARAS
FEDERACION ESPANOLA DE PADRES CON NIÑOS CON CANCER
FEDERACIÓ CATALANA DE MALALTIES MINORITÀRIES
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SENSE BARRERES DE PETRER

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THE CURE & ACTION FOR TAY-SACHS (CATS) FOUNDATION
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 THE OXALOSIS & HYPEROXALURIA FOUNDATION
 THE SNYDER-ROBINSON FOUNDATION
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VENEZUELA, BOLIVARIAN REPUBLIC OF

FUNDACIÓN FURA

ZIMBABWE

CHILD & YOUTH CARE, ZIMBABWE



PARTICIPATION OF EURORDIS' REPRESENTATIVES IN PUBLIC EUROPEAN / INTERNATIONAL **CONFERENCES** & Workshops 2020

8th International Conference on Rare and Undiagnosed Diseases, 7-8 February

Virginie Bros-Facer: Rare Barometer Voices (Survey)

RESTORE MOMENTUM conference, 17 February

Karolina Hanslik represented EURORDIS and presented RARE IMPACT initiative

A Rare Disease Day Policy Event at the European Parliament, 18 February

Yann Le Cam represented EURORDIS

Rare Disease Denmark – Rare Disease Day Workshop, 28 February

Matt Bolz-Johnson: Integration of ERNs into the NHS

Solve-RD Annual Meeting 2020, 5-6 March

Gulcin Gumus: Community Engagement Task Force; **Virginie Bros-Facer:** Winter School: capacity building programme for patients on scientific innovation and translational research

NORDIC Rare Disease Summit, 13 March

Yann Le Cam: European recommendations for improving overall access to medicines for patients with rare diseases across the EU; **Terkel Andersen:** The importance of empowerment for patients, relatives and society

DIA Europe 2020, 17-19 March

Simone Boselli and **Yann Le Cam:** From discovery to patient access: how can Europe remain at the forefront of innovation in the development of diagnostics and therapies in rare diseases, and ensure patients benefit from the next hundreds of treatments? **Virginie Bros-Facer:** Share and protect our health data: Results from Rare Barometer survey on Data Protection and Data Sharing

ACHSE/RARE IMPACT event on Access to Advanced therapies, 21 April

Karolina Hanslik represented EURORDIS/RARE IMPACT

The European Conference on Rare Diseases & Orphan Products (ECRD), 14-15 May

Yann Le Cam, Matt Bolz-Johnson, Valentina Bottarelli, Virginie Bros-Facer, Virginie Hivert, Anna Kole, Simone Boselli, Marta Campabadal, Maria Cavaller, Gulcin Gumus, Ines Hernando and **Clara Hervas** represented EURORDIS

Webinar “Making Sickle Cell Disease a Healthcare Priority in the EU”, 19 June

Simone Boselli represented EURORDIS

VSOP Workshop, 22 June

Matt Bolz-Johnson: Integration of ERNs into the NHS

RD-CODE Multi-stakeholder Workshop, 22-23 June

Gulcin Gumus: Patient journey through diagnosis

PARADIGM Patient Engagement Open Forum, 25 June

Elisa Ferrer and **Maria Cavaller** represented EURORDIS

PARADIGM Patient Engagement Open Forum, 9 July

Elisa Ferrer and **Maria Cavaller** represented EURORDIS

The European Commission online workshop on the Pharmaceutical Strategy for Europe, 14-15 July

Simone Boselli, Virginie Hivert, François Houÿez and **Maria Cavaller** represented EURORDIS

EPF Data Saves lives, 28 July

Virginie Bros-Facer: Share and protect our health data

Cancer League “What is a Fair Price” - Launch event of ECL’s paper on fair pricing of cancer medicines, 2 September

Simone Boselli: Reflection of Stakeholders on ECL’s Fair Price Definition

ESOF2020 – EuroScience Open Forum, 2-6 September

Gulcin Gumus: The role of rare disease patients and patient organizations in driving genomics research projects and medicine. How to include patients in an effective way?

European Joint-Programme for Rare Diseases (EJP-RD) second General Assembly and consortium meeting, 14-18 September

Virginie Bros-Facer and **Raquel Castro** represented EURORDIS

RWE4Decisions Stakeholder Meeting on “Real-World Evidence for Learning Healthcare Systems”, 22 September

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

Universités d’automne, Alliance Maladies Rares, 23-26 September

Anja Helm and **Konstantinos Aligiannis**: Connaître et comprendre les acteurs du paysage maladies rares (EURORDIS); **Céline Schwob** represented EURORDIS.

EUCOPE event “Addressing unmet medical needs in the EU”, 29 September

Simone Boselli represented EURORDIS

ARM Event “Cell & Gene Meeting on the Mesa”, 14 October

Karolina Hanslik represented EURORDIS and presented RARE IMPACT initiative

Friends of Europe Rethinking health systems / State of Europe week, 14 October

Yann Le Cam represented EURORDIS

NORBS Regional conference “Caring for Rare”, 23-24 October

Simone Boselli and **Gulcin Gumus** represented EURORDIS

EFPIA webinar “Orphan medicines: how to sustain innovation in an area of high unmet need?”, 30 October

Simone Boselli, featured speaker, represented EURORDIS

WODC Europe workshop on Orphan Medicinal Products (OMPs), 2 November

Simone Boselli represented EURORDIS

1er congrès de l’Alliance maladies rares, 3 November

Yann Le Cam, **Ariane Weinman**, **Virginie Hivert** represented EURORDIS

11th World Orphan Drug Congress, 3 November

Yann Le Cam represented EURORDIS

German Presidency Conference: “Health Innovation - the European Health Data Space and Real-World Evidence”, 10 November

Simone Boselli represented EURORDIS

Digital Health 2020 – EU on the Move, 11 November

Yann Le Cam represented EURORDIS

Virtual ISPOR Europe 2020, 17 November

Yann Le Cam, Patient and Public Involvement in Healthcare Decision Making: Are We Maximizing Opportunities?

I-Com “Designing the Future European Health Union? Scaling-up Ambitions, Powering Resilience”, 18 November

Yann Le Cam: The Pharmaceutical Strategy: A New Framework for Innovation and Affordability?

Presentation of the EJP RD Short Guide to Patient Partnerships, ICCBH virtual forum on bone fragility disorders in children, 18 November

Virginie Bros-Facer represented EURORDIS

RARE IMPACT Final meeting “From Possible to Accessible: Challenges and solutions for improving patient access to advanced therapies medicinal products at the European level” 23 November

Karolina Hanslik represented EURORDIS and presented RARE IMPACT initiative

Ass. p63 Syndrome E.E.C. International APS Onlus, 26-27 November

Anne-Laure Aslanian represented EURORDIS

Lupus Europe meeting, 28 November

Anja Helm represented EURORDIS

EUCOPE workshop on ATMPs, 30 November

Karolina Hanslik represented EURORDIS and presented RARE IMPACT initiative

Pharmaceutiques « Maladies rares : ne laisser personne de côté », 30 November

Yann Le Cam represented EURORDIS

European Huntington Association (EHA): Improving access to care and treatment for HD patients and families: what role for policymakers?, 3 December

Simone Boselli and **Ines Hernando** represented EURORDIS

ESCF: Capacity Building, Learning, and Networking Webinar, 4 December

Anja Helm: Patients & Organisation engagement/Representation

Presentation of the EJP RD Short Guide to Patient Partnerships, ITHACA meeting, 10 December

Virginie Bros-Facer represented EURORDIS



JOURNAL PUBLICATIONS 2020

Erice Call for Change: Utilising Patient Experiences to Enhance the Quality and Safety of Healthcare, Drug Safety, April

Contributing authors:
François Houÿez

Boosting delivery of rare disease therapies: the IRDiRC Orphan Drug Development Guidebook, Nature Reviews Drug Discovery, April

Contributing authors:
Virginie Hivert

The fourth edition of the European Network for Health Technology Assessment Forum, April, International Journal of Technology Assessment in Health Care, April

Contributing authors:
François Houÿez

Defining Patient Engagement in Research: Results of a Systematic Review and Analysis: Report of the ISPOR Patient-Centered Special Interest Group [Editor's Choice], Value in Health, June

Contributing authors:
Rob Camp

From Passive to Active: Patients as Contributors to Medicinal Product Risk Communication Research, Communicating about Risks and Safe Use of Medicines, Real Life and Applied Research, June

Contributing authors:
François Houÿez

A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome, Orphanet Journal of Rare Diseases, August

Contributing authors:
Virginie Bros-Facer, Gulcin Gumus

Our greatest untapped resource: our patients, Journal of Community Genetics, April 2021 (delayed due to COVID-19)

Contributing authors:
Matt Bolz-Johnson, Ines Hernando, Yann Le Cam

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



EUROPEAN COMMISSION

DG Health and Food Safety

- The Operating Grant for year 2020
- 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 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 2020. Health sector companies have supported EURORDIS through the EURORDIS Round Table of Companies¹, the European Conference for Rare Diseases and Orphan Products² (ECRD) online, the EURORDIS Black Pearl Awards³, as well as international initiatives such as Rare Disease Day⁴, Rare Barometer⁵, Rare Diseases International⁶, EURORDIS Open Academy⁷, NGO Committee for Rare Diseases⁸, 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



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5 SANOFI GENZYME



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

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

³ <https://blackpearl.eurordis.org>

⁴ <https://www.rarediseaseday.org>

⁵ <http://www.eurordis.org/voices>

⁶ <http://www.rarediseasesinternational.org>

⁷ <https://openacademy.eurordis.org>

⁸ <https://www.ngocommitteerarediseases.org>

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Special Mention

We also would like to sincerely thank all the generous individuals and organisations from across many countries who made a gift to EURORDIS in 2020.



EURORDIS
RARE DISEASES EUROPE

WORKPLAN

2021

ACTION PLAN 2021	58
TEAM CHART 2021	68
GOVERNANCE CHART 2021	69
EXTERNAL REPRESENTATION CHART 2021.....	70
REVENUE & EXPENSES 2021.....	72



ACTION PLAN 2021

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 & Newborn 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 European Health Union** in the areas of high added value and economic impact, such as rare

diseases, cancers and other health threats, built on the lessons of the COVID-19 crisis for more resilient and equitable healthcare and welfare systems

- + **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**
- + Lead to conclusion the **Rare 2030 Foresight Study on Rare Disease Policy**, to adopt policy recommendations for rare disease policy in 2030 and pave the way to a new EU policy framework for rare diseases. This built upon the following project's milestones and deliverables: development of Rare 2030-2040 Future Exploratory Scenarios with thorough contribution from the project's partners, 250-member Panel of Experts, Research Advisory Board; Presentation of Rare 2030 Scenarios at the 2020 European Conference on Rare Diseases and Orphan Drugs (ECRD) and voting on stakeholder preferences; Elaboration on meaningful policy options to integrate in the preferred policy scenario 2030, through discussions and deliberations at the ECRD, four national-regional workshops, European workshops with ERNs and stakeholders; Gathering patients and families perspective through a survey with the Rare BarometerVoices platform; a Young Citizen Conference followed by an online debate with the participation of 27 young patient advocates who brought the perspective of the next generation; Consolidation of all proposals and presenting them in the form of recommendations to key policymakers at the final event of Rare2030 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

- + **Launch of a two-year campaign in view of the revamping of the EU rare disease strategy and establishing a new policy framework for rare diseases**, comprising of:
 - Engaging with upcoming Trio of EU Presidencies 2022-2023 to support the EU policy strategic review and new policy framework for rare diseases
 - Re-launch and expand the European Parliamentary Advocates for Rare Diseases network, expanding it to national MPs, in coordination with the National Alliances
 - Organisation of the first Rare Disease Week in Brussels for rare disease advocates on the occasion of Rare Disease Day 2021
 - Designing and implementing a broad grassroots campaign to mobilise EURORDIS members at large, with greater integration of advocacy actions at the local, national, European level, as well as international as appropriate

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

- + Follow-up of agreement reached in December 2020 on the **EU Multiannual Financial Framework 2010-2027** that will fund EU policies and programmes, in order to secure that **specific proposals impacting on rare diseases**
- + Secure the measures in support of people living with rare diseases agreed upon in the new health programme 2021-2027 **"EU4Health"**, adopted in December 2020, are duly implemented and funded on a yearly basis, including the **sustainability of ERNs and Orphanet**, specific support to **rare cancers** within the EU Beating the Cancer Plan
- + Continue the collaboration with the [EU4Health Civil Society Alliance](#) to make sure that the EU4Health Programme is implemented with patients as meaningful stakeholders in decision-making on EU health programming and spending, while securing common objectives are transposed into annual work plans (improved access to quality healthcare services, medicines and medical products, reducing unfair and avoidable health inequalities and bringing innovation to our health systems beyond the COVID-19 crisis)
- + 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**
- + Continue promoting RDs within health, social and digital strategies across EU funding programmes 2021-2027, including **European Structural and Investment Funds**, notably the **European Social Fund**. 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 FOSTER THE INCLUSION OF RARE CANCERS ACROSS EURORDIS' ADVOCACY TRANSVERSAL ACTIVITIES:

- + Work together with all relevant actors on the inclusion of policies and measures for rare cancers in the Europe's Beating Cancer Plan, based on the **RARE CANCER AGENDA 2030**
- + Mainstream rare cancer specific measures in EURORDIS' contributions and advocacy in major EU policies, e.g. revision of the pharmaceutical strategy and in particular of the paediatric and orphan medicinal products legislation; the EU disability strategy and the Action Plan for the European Social Pillar; the European Health Data Space; etc.
- + Continue providing/reinforcing support to ePAG advocates in the four ERNs relevant to rare cancers (PaedCan, EURACAN, EuroBloodNet and GENTURIS)
- + Strengthen the partnership with various partners, including the four ERNs relevant to rare cancers, Rare Cancers Europe, patient organisations, ePAG advocates, on the implementation of the recommendations set out in the **RARE CANCER AGENDA 2030**
- + Together with the ePAG advocates, provide the patients' perspective on the development of registries in the frame of the project **STARTER** (EURACAN registry) and **ENROL** (EuroBloodNet registry)
- + Work together with the National Alliances for rare diseases for further liaising with rare cancer patient organisations and rare cancer patient advocates on issues that are relevant to the rarity of diseases (lack of information, development of and access to treatments, access to ERNs' expertise...)
- + Participate in the Rare Cancers Europe's awareness raising campaign
- + Includes rare cancers in the Rare Disease Week 2021 and informs key MEPs, notably in the Special Committee on Beating Cancer, of the work that EURORDIS has been carried out over the past years on rare cancers
- + Promote rare cancers in the programme of the EU Council French Presidency to help include rare cancers in national cancer control plans with synergies with rare disease plans, based on the French model. France has made a specific link between its NCCP and NRDP to implement national networks for rare adult solid cancers (where there was a gap), inspired by the model of national rare disease networks

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; supporting ePAG advocates and clinicians to strengthen their partnership in the

ERNs; building our institutional relationship with the group of hospitals managers; contributing to shape the development of clinical research networks through our involvement in ERICA and the development of clinical practice guidelines and other clinical decision-making support tools through our involvement in the ERN Clinical Advisory Body

- + **Promote integration of ERNs into national healthcare systems in collaboration with National Alliances (NAs) and ePAG advocates** by continuing to support them to play an active role on the integration of ERNs into national healthcare systems through face-to-face or online meetings tailored to their country needs and health systems specificities and through the development of tools to support national action at local level

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
- + Link the outcomes of the Rare 2030 foresight study with the three above-mentioned actions

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 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
- + Continue to **advocate for all aspects of holistic and social care within the European Commission's future Action Plan on the implementation of the European Pillar of Social Rights**, in line with the EURORDIS position paper on holistic care and EURORDIS' contribution to the EC consultation 'Have your say on reinforcing Social Europe' (2020)
- + **Advocate for the reasonable accommodation of people living with a rare disease in the workplace, by providing inputs to the European Parliament's Committee on Employment and to the European Parliament's Committee on Employment and Social Affairs Own-initiative Report on the 'Implementation of Council Directive 2000/78/EC establishing a general framework for equal treatment in employment and occupation in light of the UNCRPD'**
- + Continuing to **promote an integrated approach to medical and social care in discussions with ERNs**, ahead of the creation of the future cross-ERN working group on integrated care, to be launched in 2023

1.9 ADVOCATE TO IMPROVE ACCESS TO DISABILITY RIGHTS

- + Continue to **advocate for the European Disability Strategy Rights Agenda 2020-2030** to provide guidance to improve disability assessment and ensure flexible work arrangements and adequate leave of absence for people living with a rare disease. This advocacy will be based on, based on the EURORDIS position paper on Holistic Care and on EURORDIS contribution to the EC's public consultation on the roadmap for the future strategy (2020)
- + **Continuing to partner with the European Disability Forum (EDF)**, of which EURORDIS is a 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 POLICY 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
- + Continue the promotion of the **UHC4RareDiseases campaign**, which builds on the success of the UN Political Declaration on Universal Health Coverage (UHC) adopted in 2019 with the inclusion of rare diseases and counts with a series of advocacy tools to promote the implementation of UHC in Europe and other regions; continue also participating in UHC2030; and develop additional empowering tools and trainings to facilitate advocacy on UHC
- + 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

- + Support RDI and its members in strategising the advocacy with Member States for the adoption of a **UN General Assembly Resolution on persons living with a rare disease and their families**
- + Explore opportunities to convey the need to **address rare diseases within a human rights approach** at the Human Right Council (HRC) sessions in Geneva, establish closer relations with the Office of the High Commissioner for Human Rights (OHCHR), and study the possibility of advocating for the adoption of an HRC resolution and the nomination of a Special Rapporteur on the rights of persons living with a rare disease
- + Continue RDI's participation in **Rare Disease Day** working groups as well as integrating the new governance structure, in addition to the organisation of an online RDD event

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, disseminate the draft of the UN General Assembly Resolution on persons living with a rare disease, and continue 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 EURORDIS International Advocacy Volunteers based in NYC
- + Start preparations for the RDD policy event 2022 to be held in Dubai, UAE, as part of the World EXPO

1.11 EURORDIS RARE BAROMETER PROGRAMME: GENERATING DATA FROM PATIENT EXPERIENCE

- + Scale up the collection of patient experiences and perspectives to enrich a patient evidence-based policy development building on the 15,000+ patients and families who have joined Rare Barometer Voices and continue the growth internationally
- + Continue dissemination of reports of previous surveys, infographics or online dashboards presenting the results of surveys in 23 languages
- + Develop a survey on diagnosis for people living with a rare disease, to identify personal and external factors influencing the process of obtaining a timely and accurate diagnosis from a patient perspective
- + Analyse the survey on the future of rare diseases as part of the Rare 2030 project so that patients' needs and opinions are taken into account when exploring the potential impact of the scenarios on patients and identifying preferred scenarios
- + Explore the feasibility and plan a research project to develop validated scale(s) on the experience of patients and families in the healthcare system
- + Explore the feasibility to carry out a second COVID-19 survey, to measure the effects of the pandemics on the life of people living with a rare disease
- + A significant part of the year will be dedicated to **developing the sustainability of the Rare Barometer programme**, find funding to scale up the H-CARE pilot survey 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 organisation database for targeted outreach
- + Organise the EURORDIS Membership Meeting 2021 online

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 2021
- + 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; working with the Public Affairs Manager based in Brussels on EU & National Integrated Advocacy; empowering capabilities through regular bilateral meetings and webinars; organising 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 number of grants and 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 an 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. Explore possibilities to apply to an EJP Grant / SSA for the training of help lines on research for rare diseases and disease classification
- + 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 2021
- + Reflect on how genetic testing results are communicated to patients, the role of genetic counsellors, and how helplines can help patients informing family members. This is now part of IGPRare with Virginie Bros-Facer

2.1.3 RareConnect

- + Support the creation of new online communities for very rare diseases and undiagnosed disease patients
- + Conduct an assessment of EURORDIS' role within RareConnect taking into account other online communities

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; for a launch in early 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

- + Finalise the EURORDIS Open Academy sustainability plan, including the plan for the future Open Academy programme and for the transition phase until its implementation. **The new programme will be composed of three pillars: e-learning offer, a single annual training programme and an outreach component to all alumni**
- + Organise the fourth edition of the EURORDIS Winter School on Scientific Innovation and Translational Research, including one week of online training, in April 2021, preceded by 3 pre-training webinars and over 10 e-learning courses
- + Organise the fourteenth edition of the EURORDIS Summer School on Medicines Research & Development, including one week of online training, in June 2021, preceded by 3 pre-training webinars and over 15 e-learning courses
- + Organise the third edition of the EURORDIS Leadership School, for ePAG advocates and other rare disease patient advocates, including 5 webinars, from July to December, and three days of intensive training, in October 2021
- + Continue the EURORDIS Digital School 2020-2021 as an online open programme, with a total of 5 webinars and e-learning courses, from late Spring until October 2021
- + For each of the Open Academy schools, coordinate the contents of the courses 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, improving its information, its organisation and its technical aspects, to ensure better navigation and an improved monitoring of the progress of patient advocates who take the e-learning courses; **Increase the availability and outreach of the Open Academy's online training materials to more patient advocates, free of cost; improve the Summer and Winter School's e-learning courses and develop new courses on other topics**
 - Create a monthly newsletter for alumni, to inform them on training opportunities and on relevant patient engagement opportunities
 - 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 Rome or online, in autumn 2021

2.3 RAISING AWARENESS & INFORMING

2.3.1 Rare Disease Day 2021 & 2022

- + Coordinate the international Rare Disease Day 2021 & 2022 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 2021 campaign throughout the year with social media and stories from the global campaign
- + Design and launch a new site for RDD by preparing a call for tender and with backend investment to ensure effective, GDPR-compliant tracking of partner and user data
- + Evaluation and reposition if needed in strategic paid media approach and enlargement of successful new media sectors

- + Co-create and produce Rare Disease Day 2021 campaign materials (visuals, website update, video) building on strategic review recommendations and a call to action
- + Translations of the media assets and webinars in different languages Hindi, Arabic, French, Spanish, Portuguese, Russian and English in order to spread the RDD message in more and under-represented countries
- + Expand the movement of lighting up the building landmarks supporting the RDD message after the restoration of the Covid situation
- + Creation of important metrics for the performance of the RDD new site

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** and provide regular support to ePAGs in governance and operational aspects 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 relevant activities
- + **Enhance the communication capacities of the patient community to disseminate the work of the ERNs, raise awareness about their services and value** by setting a communications working group with ePAG advocates and RD national alliances that will develop approaches, tools and tips to increase the reach out and comms activities of the patient community around ERNs
- + **Build the evidence around the impact of patient engagement in the ERNs; finalise the ePAG impact assessment framework pilots** to refine the measures to evaluate the impact of the ePAGs engagement in the ERNs; **engage with the ERNs monitoring and evaluation WG** to align the framework with the overall ERN strategy on monitoring and evaluation; support the ePAG advocates on their **involvement in the refinement and definition of the ERN assessment, monitoring and evaluation system**
- + Contribute to the promotion of **common methods to capture feedback on patient experience across the ERNs**, building on the experience of the H-Care pilot survey and fund raising for the development of a validated scale to measure patient experience with healthcare for rare diseases
- + **Further build ERN team members capacities** on patient engagement methodologies

- + **Renew the commitment of the existing ePAG advocates** and support them in identifying gaps in terms of diseases and countries to recruit new advocates, in collaboration with the national alliances and the European Federations; **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 through the update of the common template for ePAG terms of reference**
- + **Promote and facilitate exchange of ePAG good practices and peer learning** through the organisation of quarterly webinars delivered by ePAG advocates, a **2-half day Steering Committee annual meeting on the 9th and 10th of June** and **all-ePAG annual workshop 4th and 5th of November (both meetings online)**
- + Deliver a comprehensive capacity-building programme **Leadership School for patient advocates** within EURORDIS Open Academy; **engage with the ERNs Coordinators Knowledge Management and Training WG** on the ERNs training strategy to ensure alignment with Open Academy training activities targeted at patients and families and on developing a **course on patient-clinician partnership**

3.2 PATIENT ENGAGEMENT IN THERAPEUTIC DEVELOPMENT

3.2.1 Support Patient involvement in EMA activities

- + Participate in the **EMA Committee for Orphan Medicinal Products (COMP)**, including in the activities aiming at giving guidance to the European Commission on the matter related to the revision of the orphan regulation
- + Participate in the **EMA Pharmacovigilance and Risk Assessment Committee (PRAC)**, including in the Impact Assessment – Stakeholder Engagement working group & Cross-Committee Taskforce on Registries

- + Participate in the **EMA Paediatric Committee (PDCO)**
- + Collaborate with EMA to identify and support the participation of patients in Protocol Assistance dossiers at the Scientific Advice Working Party (SAWP), and also in scientific committees' consultations
- + 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 Rare Diseases patient representatives which are not representing EURORDIS on these Committees and Working Parties, no matter whether they are EURORDIS members or not

3.2.2 Support patient involvement in European HTA Network, EUnetHTA JA 3, related HTA activities and through the 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 2021
- + Identify and mentor patients participating in HTA Joint Procedures
- + Take part in the HTx 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 online 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
- + Contribute to the Pharma Review 2021 onwards

3.2.4 Support patients' Community Advisory Boards (CABs) to engage with Industry (EURO-CAB programme)

- + Flesh out and plan the implementation of the sustainability strategy as discussed with the EURORDIS BoD with a view to the new operating model of the EuroCAB programme coming into effect in 2022
- + Continue our work to advise European Federations who are looking to set up a CAB and support the CAB meetings for those who have entered into mentorship agreements
- + Promote Community Advisory Boards to patient groups and health corporates
- + Provide metrics to support the evaluation of the impact of the CABs
- + Review training programme common to all CABs in light of the new operating model

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 contribute 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-centred
 - Develop a comprehensive map of resources and tools to support undiagnosed rare disease patients as well as provide a 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** – at the moment: Clinical Research Networks, Shared Molecular Aetiologies, Access to RD Treatment, Integrating New Technologies for the Diagnosis of Rare Diseases and

Sustainable Economic Models in Repurposing (as leader). The roadmap 2021 will be approved beginning of the year (see below)

- + Notably, in the past years, 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 navigating the therapeutic development ecosystem and describes the available tools and initiatives specific for rare disease development and how to best use them (the Orphan Drug Development Guidebook is available online). Broader dissemination is ongoing
- + Prepare the **IRDiRC conference** organised together with the RE(ACT)'s International Congress of Research on Rare and Orphan Diseases in Berlin, postponed to January 2021 (to be held online)
- + A new **road map** for 2021 IRDiRC activities shall be approved by the consortium assembly early next year. EURORDIS is co-applicant for 3 Taskforces/Working groups: one on unlocking the potential of currently disregarded rare diseases, one on a Drug repurposing guidebook and one on MedTech in rare diseases
- + EURORDIS will actively promote opportunities for **patient representatives** to get involved as Task Forces and Working groups that 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 standardised 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 organisations

+ 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 on holistic care, including social services, integrated care and disability rights
- + **Continue to support the work of the European Network of Resource Centres for Rare Diseases - RareResourceNet**, as part of its Board of Directors
- + **Seek opportunities** to develop key actions in holistic and integrated care within European projects and initiatives
- + **Carry on the 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 2021-2026

- + Finalise the Strategic Revision 2021-2026, 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, including a Theory of Change, for adoption at the AGA 2021
- + Build out an internal system to implement the strategy and oversee the effectiveness of its operation

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 Strategic Partnerships (MoUs)

- + 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

4.2 RESOURCE DEVELOPMENT & EURORDIS SUSTAINABILITY

- + 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 Director to take forward work with Donors and Foundations**
- + Finalise, adopt and implement sustainability plans for EURORDIS' major programmes
- + Invest in developing strategic projects proactively and seek opportunities to finance them
- + **Focus team efforts, build skills and allocate resources to innovative project development in 2021**

4.3 EURORDIS EVENTS

- + Organise the **EURORDIS Black Pearl Awards** in February 2021 as an online event
- + Organise **the first Rare Disease Week**
- + Organise the **EURORDIS membership meeting (EMM) 2021** fully online
- + Organise **two CNA workshops and one CEF workshop**
- + Organise **two major workshops of the ERTC** with relevant stakeholders

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

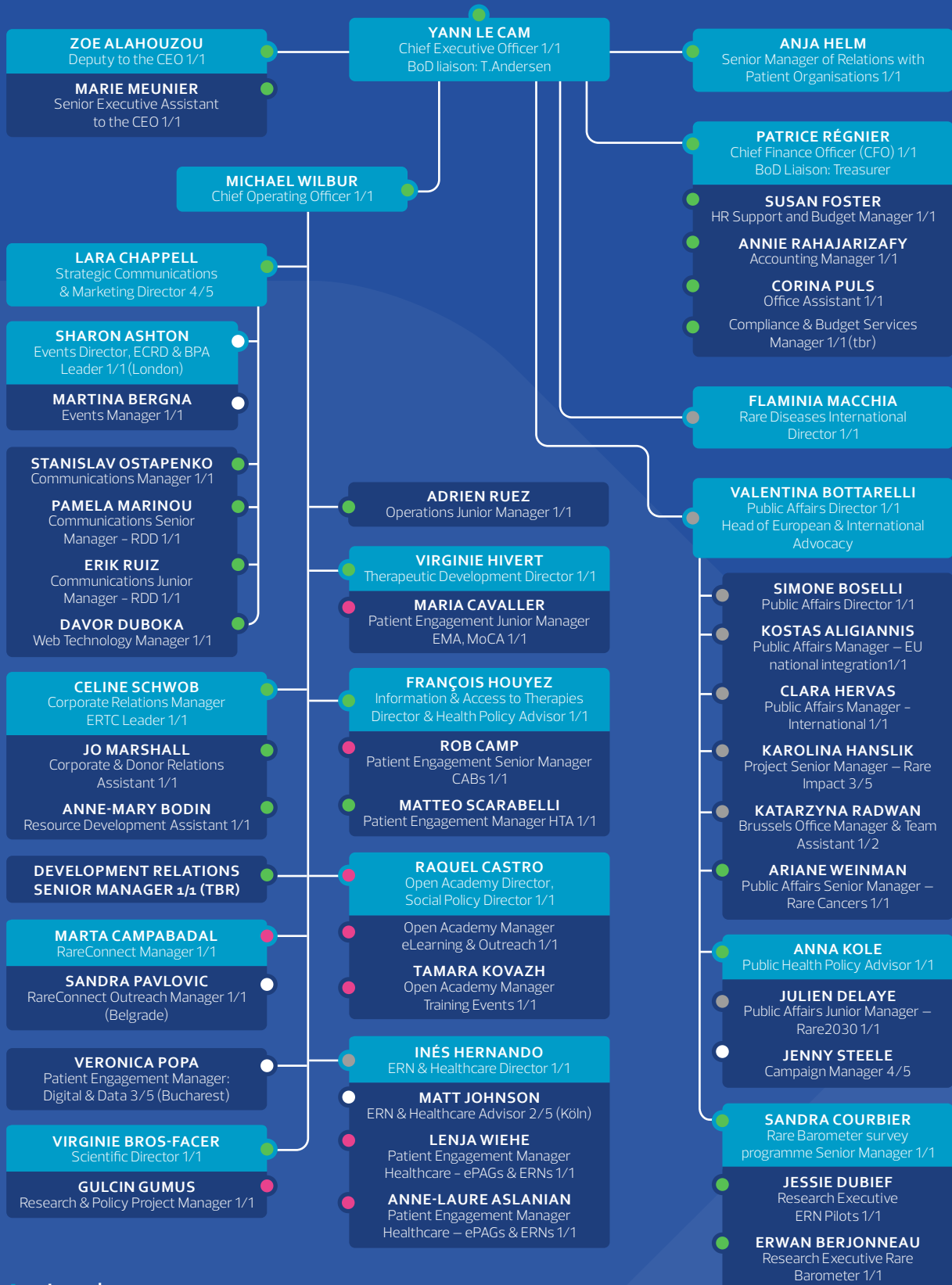
4.4.2 EURORDIS Volunteers

- + Improve **volunteers' visibility** in EURORDIS' communication and acknowledgements on the EURORDIS website
- + **Revise processes for effective and improved volunteer support and management**

4.4.3 EURORDIS Staff

- + **Appointment of following permanent positions:**
 - Strategic Project Design (based in Paris)
 - Fundraising Director (based in Paris)
 - Rare Barometer Research Executive (based in Paris)
 - Finance and Support Services Manager (based in Paris)
- + **Temporary positions in 2021 linked to a specific mission:**
 - Public Affairs Junior Manager, project Rare 2030 (Brussels, 1Q2021)
 - Rare Disease Day Senior Manager (Paris)
 - Patient Engagement Manager, Digital & Data
 - EU Rare Disease Strategy Campaign Manager

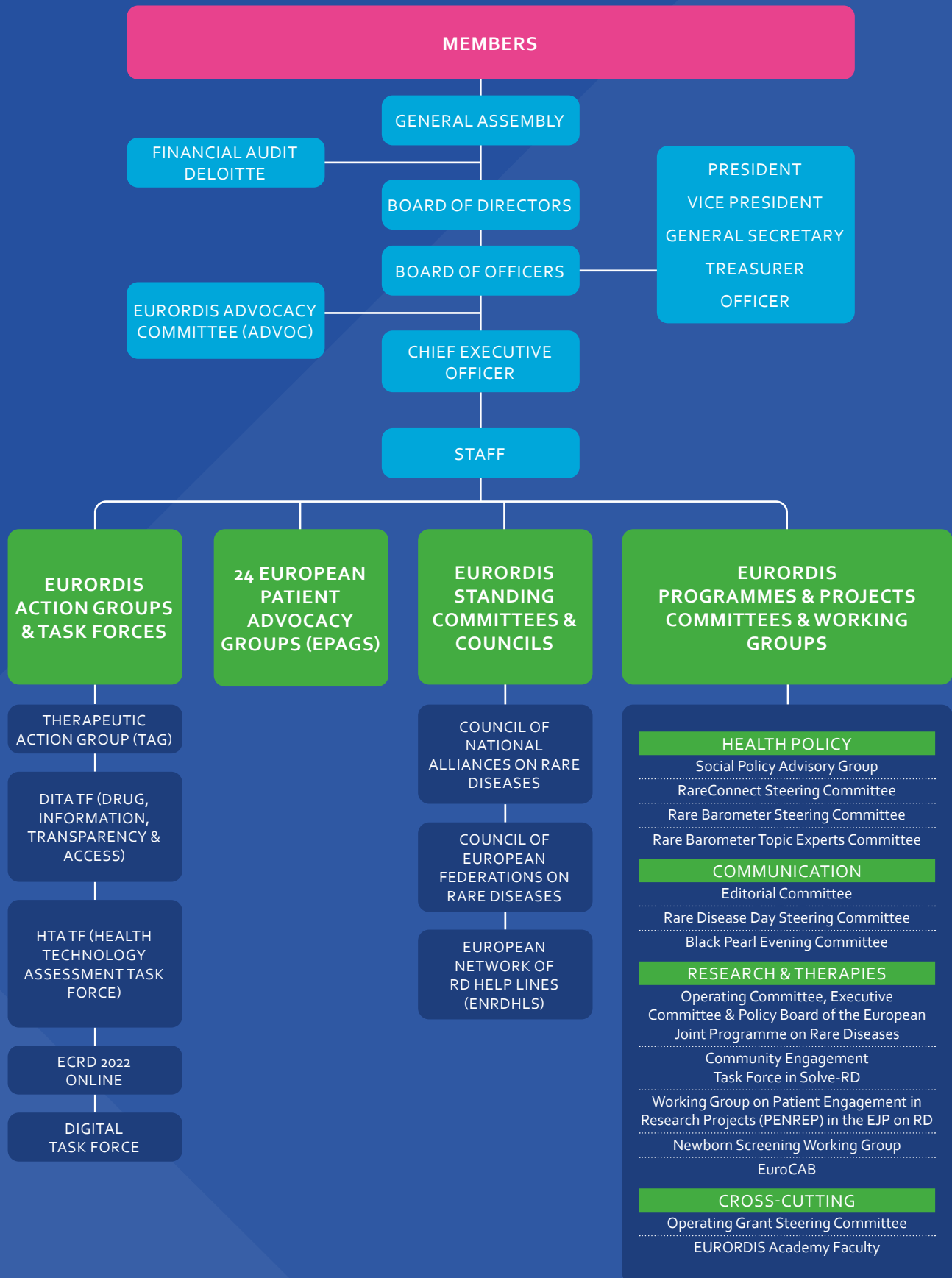
TEAM Chart 2021



Locations

- PARIS
- BARCELONA
- BRUSSELS
- OTHER

GOVERNANCE Chart 2021



EXTERNAL REPRESENTATION Chart 2021



EUROPEAN NOT-FOR-PROFIT ORGANISATIONS

EFPIA Think Tank: European Federation of Pharmaceutical Industries and Associations

EUPATI: Patient Engagement through Education

EUROPABIO Patients Advisory Group

EUCOPE: European Confederation of Pharmaceutical Entrepreneurs

EPF: European Patients' Forum

EDF: European Disability Forum

EFGCP: European Forum for Good Clinical Practice

FIPRA – International Policy Advisors

Friends of Europe

Orphanet

Rare Cancer Europe

Social Platform

Maladies Rares Info Service (French Helpline for RDs)

Rare Disease Platform in Paris

PFMD - Patient Focused Medicines Development Initiative

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

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

TOPRA Advisory Council: The Organisation for Professionals in Regulatory Affairs

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

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

HTx, Next Generation HTA

OpenMedicine

COST ACTIOPN 15105

(drug shortages)

C4C (Connect 4 Children)

CORBEL – MIUF

Solve – RD

reCOVID consortium IMI2

PARTNERSHIP LEARNED SOCIETIES

European Federation of Internal Medicine (EFIM)

European Hospital & Healthcare Federation (HOPE)

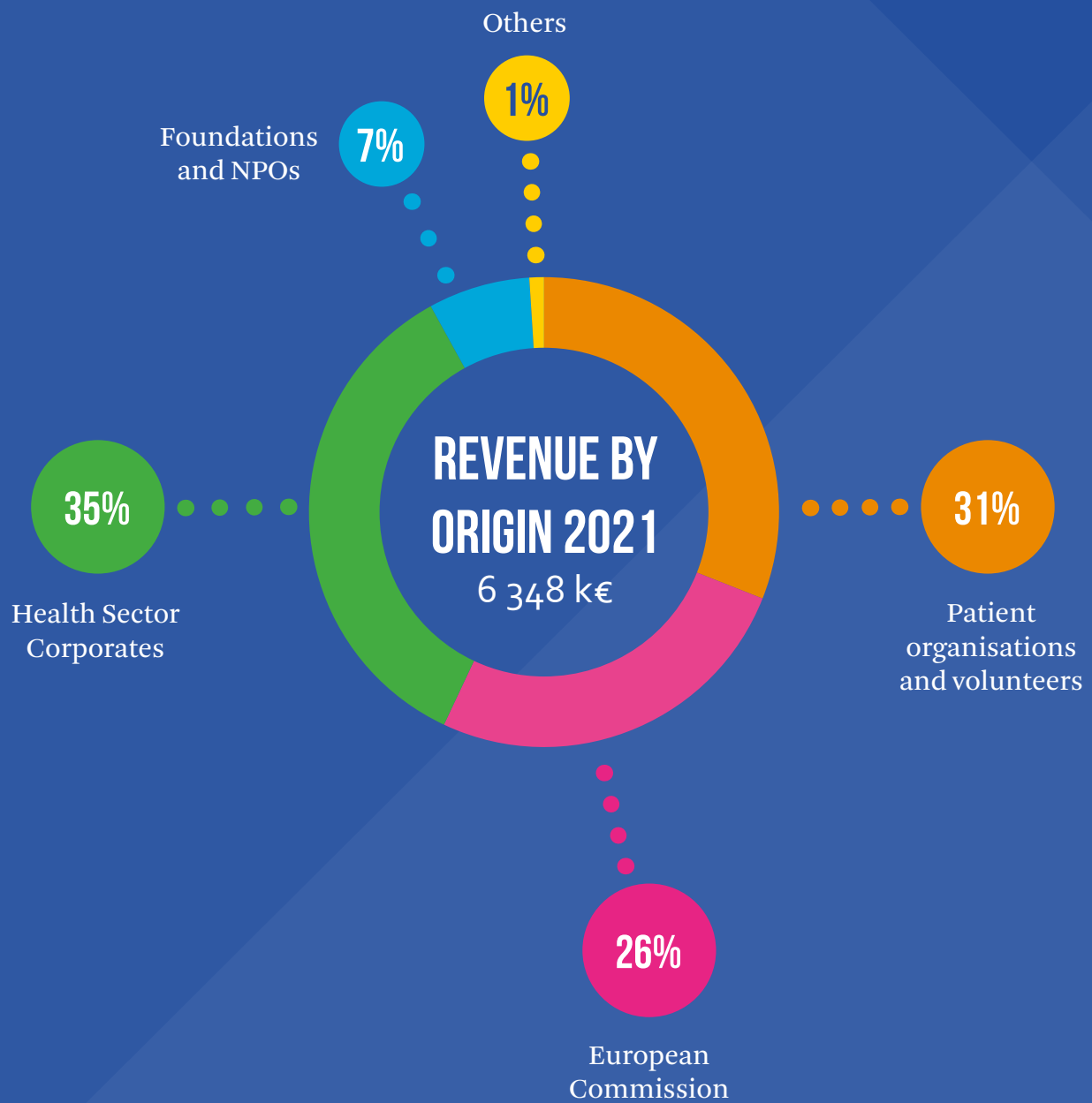
International Federation of Social Workers Europe (IFSW-Europe)

European Society of Human Genetics (ESHG)

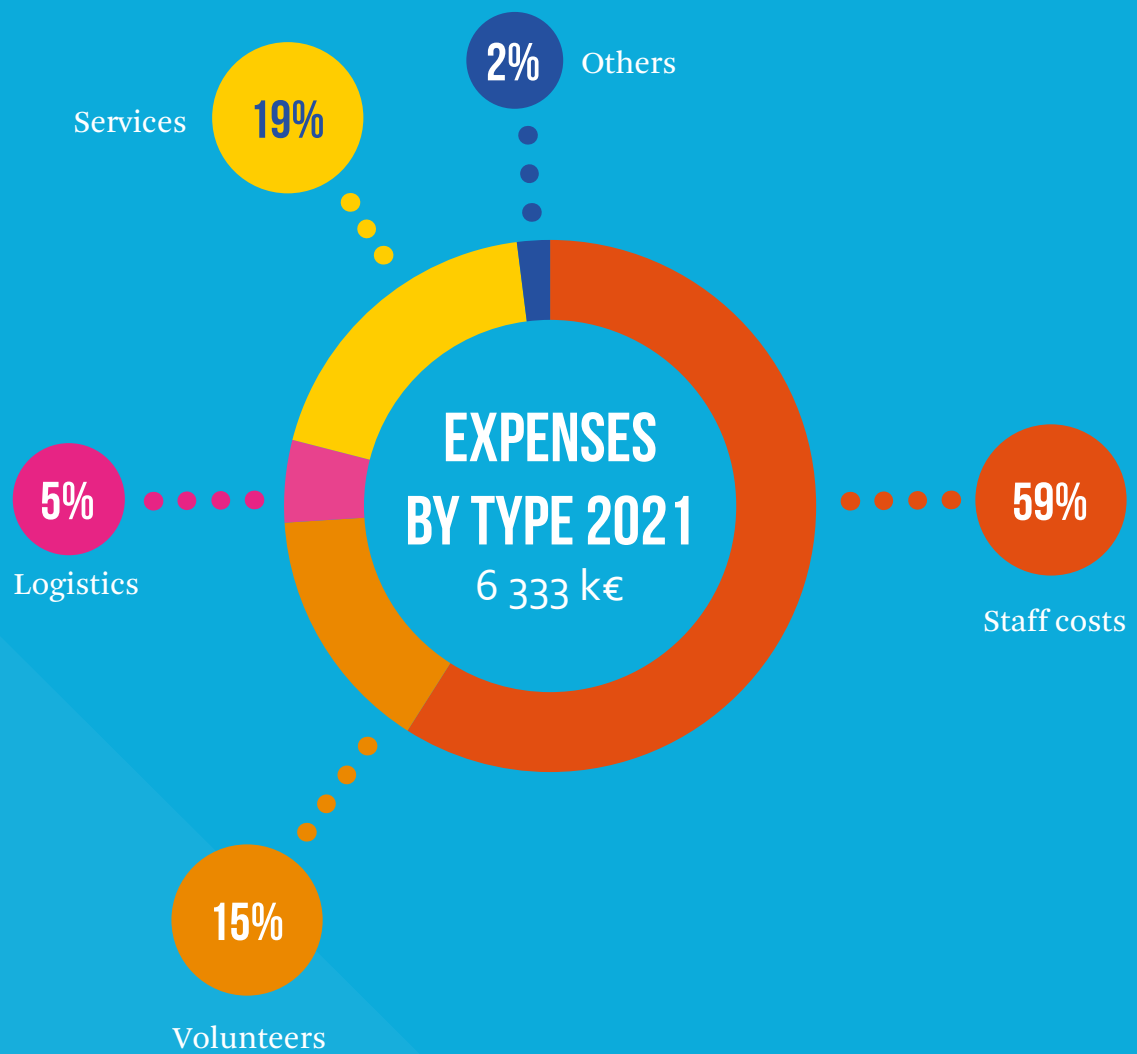
International Society for Pharmaco-economics and Outcomes Research (ISPOR)

European Connected Health Alliance - ECHalliance

REVENUE 2021



EXPENSES 2021



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)
DAG	Digital Action Group
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)
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

BBMRI Stakeholders' Forum	Biobanking and Biomolecular Resources Research Infrastructure
E-Rare	Network of ten partners – public bodies, ministries and research management organisations – from eight countries, responsible for the development and management of national/regional research programs on rare diseases
EunetHTA Forum	Support effective HTA collaboration in Europe that brings added value at the European, national and regional levels
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"
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
OA	EURORDIS Open Academy
Rare Barometer	EURORDIS survey programme
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

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

EUROPEAN COMMISSION

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

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
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
UHC	Universal Healthcare Coverage



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EURORDIS.ORG

