

EURORDIS

# ACTIVITY REPORT<sup>2021</sup>

& workplan 2022



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**YANN LE CAM**

Chief Executive Officer



**TERKEL ANDERSEN**

President of the  
Board of Officers



# FOREWORD

by the President and the Chief  
Executive Officer



Despite the increasing pressure on European and international National Healthcare Systems by the COVID-19 pandemic and the war in Ukraine in early 2022, which have reshaped political priorities,

EURORDIS and the rare disease community have successfully kept the challenges faced by people living with a rare disease (PLWRD) high on the political agenda.

In 2021, **EURORDIS continued several initiatives to prepare for the next decade of rare disease policy.** While we welcome more than twenty years of policy advances increasing support for PLWRD, many high unmet needs are still to be addressed and new challenges are emerging. A new impetus at the European level for further action is therefore vital.

In 2021, EURORDIS focused on **preparing for the next decade of rare disease legislation and public policy**, while **advocating for rare diseases to be recognised as a public health issue.** Following the conclusion of the Rare 2030 Foresight study, **our #30millionreasons campaign** - in view of the revamping of the EU rare disease strategy - **advocated for a goal-oriented European Action Plan for Rare Diseases.** Drawing on the Rare 2030 policy recommendations, our campaign explored and promoted EU Institutions' political options for updating, reviewing or replacing the EU's overall rare disease strategy. We have paved the way to a new EU policy framework for rare diseases, in line with the calls of the 2019 Court of Auditors' special report on cross-border healthcare.

Throughout the year, we have:

- Advocated for the adoption by the European Commission of a *European Action Plan for Rare Disease*, following the release of the Rare 2030 recommendations in 2021 calling for a new policy framework for rare diseases;
- Engaged with the **Trio of EU Presidencies 2022-2023 (France, Czech Republic, and Sweden)** and **Belgium and Spain in 2023-2024**, to support the EU policy strategic review and new policy framework for rare diseases;
- Consolidated the **Network of European Parliamentary Advocates for Rare Diseases**, and gained their support for a European Action Plan for Rare Diseases, leading to an Oral Question and a Plenary Debate

in the European Parliament on 24 November 2021;

- Run a broad **grassroots campaign**, consolidating advocacy actions at the local, national, and EU levels.

2021 was also an important turning point regarding **access to medicines and treatments with major innovations in the areas of gene and cell therapies**, with many Marketing Authorisation Applications (MAA) accepted and issued by the European Medicines Agency (EMA).

As with all of us, the year 2021 was marked for a second consecutive year by the COVID-19 pandemic. In this difficult context, EURORDIS decided to maintain all core activities and projects as planned, while moving them online and continuing to monitor the specific COVID-19-related issues faced by people living with a rare disease and their families.

In 2021, among the major advances and successes, we can note the **conclusion of the Rare 2030 Foresight study**, following two intensive years of multi-stakeholder consultation, which led to the publication of the Rare 2030 recommendations, "The Future of Rare Diseases Starts Today", and the **feedback conference at the European Parliament in February 2021** - a significant step in the advocacy for the European Action Plan for Rare Diseases.

The advocacy campaign **#30millionreasons**, encouraging the European Union to develop a European Action Plan for Rare Diseases, and the EURORDIS Action Plan proposal were also major steps forward and **became a matter of debate at the European Parliament in November 2021.**

In the lead up to Rare Disease Day 2021, **the first Rare Disease Week took place**, to train patients in European advocacy and let them meet, online, with key EU decision makers.

At the international level, EURORDIS' long-term work which started in 2009 has resulted in the **adoption by consensus of the first-ever UN resolution on "Addressing the needs of people living with rare diseases" in December 2021.** This political success was possible thanks to EURORDIS' close collaboration with Rare Diseases International and the UN NGO Committee on Rare Diseases.

2021 was also marked by progress in the different policy priority areas, including:

- 10 years of frontline **advocacy work that led to the adoption of the EU Health Technology Assessment (HTA) Regulation** in December 2021.
- The development of a strategic proposal for **the integration of ERNs into national health systems** as well as the efforts to strengthen the cooperation of patients and clinicians in European Patient Advocacy Groups (ePAGs).
- EURORDIS' response to the European Commission's **public consultation on the upcoming European Health Data Space (EHDS), with rare diseases as a pilot case.**
- The work on policy proposals for the **revision of the regulations on orphan and paediatric medicines and on general pharmaceutical legislation**, as well as the contribution to the Open Public Consultation on the revision of general pharmaceutical legislation.
- The joint work with the **European Expert Group on Financial Incentives for Orphan Drugs** to improve the framework for the development of therapies.

2022 will be a year of milestones: EURORDIS will **celebrate its 25th anniversary** and redefine its engagement strategy as it **reached over 1000 member organisations.**

In an exceptional conjuncture, EURORDIS is facing 4 major challenges in 2022: the uncertainty of funding from the European Commission's DG SANTE operating grant; the important turnover within the permanent team; the uncertainties related to the COVID-19 pandemic which continues to severely affect people living with a rare disease and which forces to organise activities with great flexibility; the war in Ukraine and its impact for people living with a rare disease across Europe.

EURORDIS will maintain its recurrent priorities:

- **The International Rare Disease Day 2022**, on February 28, with a 66% increase in downloads and more than 600 events organised in over 100 countries.
- The **European Conference on Rare Diseases and Orphan Products 2022 (ECRD)**, online.
- The **Council of National Alliances** for Rare Diseases (CNA) and the **Council of European Federations** (CEF).
- The engagement of patient advocates in the **committees of the European Medicines Agency (EMA), the Health Technology Assessment (HTA), and research projects.**

In 2022, we are committed to promoting the **European Action Plan for Rare Diseases**, based on the 8 final recommendations of the Rare 2030 recommendations.

In 2022, EURORDIS will:

- **Promote the European Action Plan on Rare Diseases and engage with the trio of EU Council Presidencies and the Parliamentary Advocates for Rare Diseases.**
- **Work towards removing barriers to timely and accurate diagnosis**, notably through:
  - A Rare Barometer survey on patient journey to diagnosis;
  - The SCREEN4CARE project, which focuses on newborn genetic screening and digital technologies;
  - A call to action to make newborn screening an area of cooperation at the European level to reduce inequalities in access.
- Contribute to the revision of the **Pharmaceutical Regulation**, where our priorities include product reuse, drug shortages and patient information. Regarding the **revision of the EU regulations on orphan drugs and paediatric medicines**, the priorities are to maintain incentives to attract R&D investments and to promote a product life cycle approach, recognising the continuum of data generation. Improving access to medicines, reducing development risks, and accelerating clinical research through earlier dialogue and regulatory support for potentially transformative therapies are also areas of great importance.
- Further increase **patient participation in the committees of the European Medicines Agency (EMA).**
- Work towards the **integration of ERNs, which will enter their new 5-year cycle, into National Healthcare Systems**, so as to establish patient healthcare pathways and cover progressively the full population, working together with the Joint Action on ERNs integration into NHS which will be starting in 2024 for 3 years.
- Contribute to the **elaboration of a European Health Data Space** and advocate for rare diseases as a use-case.
- Follow the development of the **Rare Diseases European Partnership** which will continue and expand the EJP-Co-Fund on Rare Diseases and will establish the European Clinical Research Networks for Rare Diseases, articulated with the ERNs, to enhance their clinical research readiness and attractiveness and scale up the European capacities across more rare diseases with basic common functions.
- Follow up on the **EURORDIS Strategic Review 2021-2030**. The priority in 2022 will be to refine the strategic objectives, guide the priorities area of work on which to focus and adapt EURORDIS' organisational capacities and architecture to deliver our strategy.

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# 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.

**984**

MEMBER PATIENT ORGANISATIONS



**73** COUNTRIES  
(27 EU COUNTRIES)

**53** NATIONAL ALLIANCES OF RD  
PATIENT ORGANISATIONS

**79** EUROPEAN FEDERATIONS OF  
SPECIFIC RARE DISEASES

 **FOUNDED IN**

**1997**

**6** MILLION €  
BUDGET

OUTREACH TO OVER

**2500**

PATIENT GROUPS



OVER

**460**

VOLUNTEERS

**82**  
PATIENT  
ADVOCATES

**380**  
MODERATORS

**50+** 

STAFF MEMBERS,  
WITH OFFICES IN PARIS,  
BRUSSELS, BARCELONA



# 2021-2030 STRATEGIC APPROACH

*In 2020 EURORDIS commissioned an external strategic review for the purpose of developing its strategy in 2021-2030. The strategic review collected input from EURORDIS members and stakeholders and incorporated the work done within the Rare2030 foresight project that developed recommendations for 2030 and beyond.*

*The resulting findings and proposed EURORDIS Strategy 2021-2030 were presented and discussed at the General Assembly 2021 and given the final validation by the EURORDIS Board of Directors.*

*The four Strategic Objectives are provisional and will be refined by the EURORDIS Board of Directors in 2022.*

## Vision

*All persons living with a rare disease can live longer and better lives reaching full potential and well-being included in a society that leaves no one behind*

## Mission

*EURORDIS works across borders and diseases to improve the lives of all persons living with a rare disease*

## FOUR STRATEGIC OBJECTIVES

### Strategic Objective 1

A new European policy framework to achieve measurable goals prolonging and improving the life of persons living with rare diseases. 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:

- ✦ By 2030, EURORDIS has promoted and facilitated the implementation of a **new European policy framework to achieve measurable goals prolonging and improving the life** of persons living with rare diseases and **their inclusion in society**, while designing a European **model of care** driven by **needs-led innovation** and collective responsibility prioritising **solidarity and equity**.

### Strategic Objective 2

An ecosystem of networks of member organisations, organisations at large and partners to achieve better and faster results

- ✦ By 2030, EURORDIS will have redefined and focused its **unique role in the rare disease ecosystem**; is leading **impactful and motivated networks** of member organisations and **advocates across diseases and borders**, which allows to effectively emphasise and highlight people's needs and integrate/mobilise national actions in key policy areas of the European with national **actions in key policy areas**; and as a **driver of change**, is creating **synergies across partners and key stakeholders** to achieve better and faster results.

### Strategic Objective 3

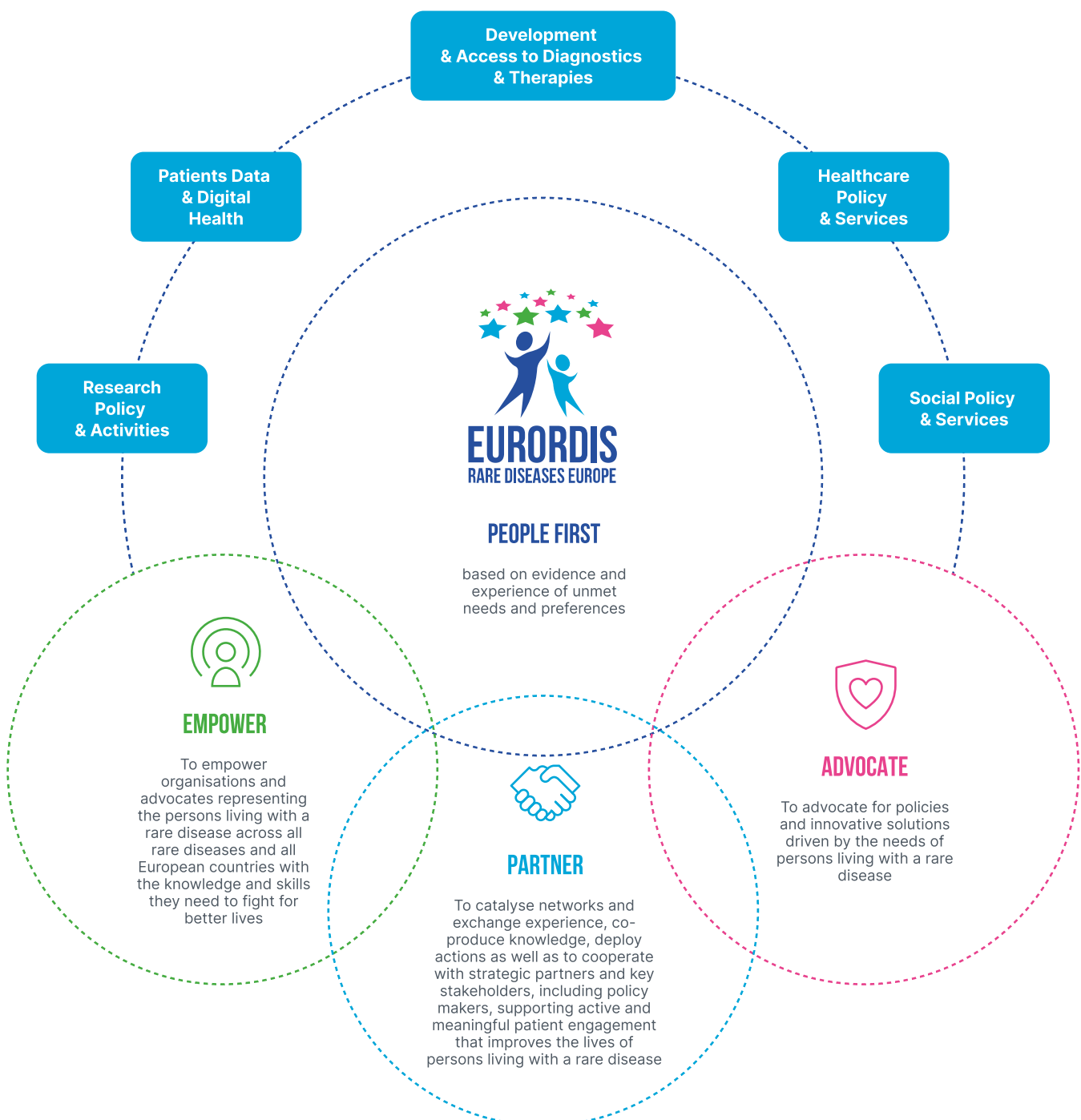
Focusing on priority areas where it matters the most during the next decade to achieve the goals

- By 2030, EURORDIS will have regularly assessed and consolidated its **priority areas of operations** to respond to the demands of both organisations and advocates while aligning with internal capacity, progressively focusing on the **new challenges of data and health digital technologies** as well as on **holistic life-long approach** and **inclusion in society**; as much as to strengthen its priority on changing the game in **research and knowledge**, **early diagnostics**, development of and access to **transformative or curative therapies**, integrated care, national and European **healthcare pathways** and **cross-border health care**.

### Strategic Objective 4

Inclusive of all rare diseases and all regions, to “leave no one behind”

- By 2030, EURORDIS will have achieved a “**leave no one behind**” strategy inclusive of **genetic and non-genetic rare diseases** – rare cancers, rare infections and rare poisonings – putting more focus on Eastern and Southern Europe and changing the game for **very rare diseases** and promoting the **next generation of young advocates**.







**EURORDIS**  
RARE DISEASES EUROPE

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# HIGHLIGHTS 2021

- 2021 was the second year marked by the **COVID-19 pandemic**. In this challenging context, EURORDIS' decision was to maintain all core activities and main projects as planned while moving all the activities online and to continue monitoring the specific Covid-19 related issues faced by the persons living with rare diseases (PLWRDs) and their families while disseminating information and taking appropriate supportive actions. The **EURORDIS Covid19 Resource Centre** created in 2020, was updated with information for our members, partners and stakeholders providing reliable sources of data, clinical guidelines, clinical trials and studies. Several statements were published to publicly convey the concerns of people living with rare diseases. The EURORDIS **COVID-19 Task Force** conducted bimonthly meetings to closely monitor the situation and activities.
- The Rare2030 **Foresight study**, a project co-funded by the EU and an academic consortium led by EURORDIS, ended in March 2021 with an outstanding achievement after two years of intensive work. **Rare2030** gathered the input of a large group of patients, practitioners and Key Opinion Leaders to propose policy recommendations that would lead to improved policy and a better future for people living with a rare disease (PLWRDs) in Europe through the next 10 years and beyond. The conclusions and recommendations were presented at the **final Rare2030** conference held at the European Parliament in February 2021. The Rare2030 recommendations "The Future of Rare Diseases Starts Today" on the most critical areas require sound policy were published at this occasion and widely spread throughout the year. They have been forming the basis of the advocacy work conducted by EURORDIS since then. An **Executive Summary** was also published.
- EURORDIS has launched the **#30million reasons advocacy grassroots campaign towards a European Action Plan for Rare Diseases**. Despite progress in recent decades, a high level of unmet needs remain as PLWRDs wait years for diagnosis, have disjointed care and a lack of treatment options. We need Europe to take action on rare diseases by 2030 in order to stop people losing their lives too young from rare diseases, improve the quality of life of PLWRDs, and ensure Europe is a global leader in rare disease innovation.
- The **Advocacy campaign towards a European Action Plan for Rare Diseases** was impactful: the European Parliament called for a European Action Plan on Rare Diseases asking a dedicated question to the **European Commission and the topic was discussed at the European Parliament plenary session in November 2021**. In addition, rare disease has been included as a topic on the common agenda of the **EU Trio Presidency – France, Czech Republic, Sweden**.
- In the framework of the Rare Disease Day 2021, the first **Rare Diseases Week** (RDW) was organised by EURORDIS. The RDW gathered 38 participants from 20 countries representing 20 different disease areas who met with their **European Parliamentarians** online. This was an important landmark to train patient advocates in European policy and introduce them to their elected parliamentarians to enable knowledge exchange.
- EURORDIS' longstanding work started in 2009 to promote rare diseases as an international public health priority culminated in the United Nation General Assembly adoption by consensus of the **first ever UN Resolution on "Addressing the needs of persons living with a Rare Disease"** in December 2021. Two years after the adoption of the UN Political Declaration on Universal Health Coverage (UHC), which explicitly included in its scope all PLWRDs, the UN Resolution is a landmark to recognise the 300 million PLWRDs in the world, to promote rare diseases across the UN programmes and agencies and to encourage all UN member states to address the specific challenges of PLWRDs. This political success was made possible thanks to the close work with Rare Diseases International and the NGO Committee for Rare Diseases.

- EURORDIS also participated in the working group that drafted the Friends of Europe report “**Building a path for rare diseases in the European Health Data Space**”, bringing the voice and needs of the rare disease patient community to help shape the different recommendations included in this report. EURORDIS responded to the public consultation on the EHDS, providing more specific feedback on the alternatives put forward by the Commission to enable the use of health data for healthcare provision, research and innovation as well as policy-making and regulatory decision; the development and use of digital health services and products and the development and use of Artificial Intelligence systems in health care.
- EURORDIS conducted important advocacy work for the re-introduction of the **Operating Grants for the functioning of health NGOs under the EU4H Work Programme 2022 together with the EU4Health Civil Society Alliance (EU4H CSA)**, gathering several health NGOs such as the European Patients Forum (EPF) and the European Public Health Alliance (EPHA).
- **EURORDIS’ membership** base continued to grow in 2021 reaching **984 members** by the end of the year, including **59 new members** approved throughout the year (a 4% increase). EURORDIS is the voice of all rare disease patients, including rare cancers as well as the most isolated rare diseases and including the undiagnosed. EURORDIS has members in a total of 73 countries and involves members and non-members in its activities.
- EURORDIS continued to coordinate, inform and empower the **Council of National Alliances (CNA – 39 members in 2021) and the Council of European Federations (CEF – 79 members in 2021)**. 2 CNA Workshops took place online (March and November), the later was again held partly in common with the Council of European Federations (CEF) in order to allow cross cutting discussions on common issues. The CNA has been particularly working on the preparation and coordination of the Rare Disease Day 2021 and 2022, the integration of ERN at national level, Newborn Screening and the EU Campaign for a new policy framework. Both the CNA and CEF have worked on the Covid-19.





- The **EURORDIS Open Academy** empowers patient advocates to have the confidence and knowledge required to bring their expertise to discussions on health care, research and medicines development. The Open Academy delivered **4 training programmes: Summer School** (Medicine Research and Development); **Winter School** (Scientific Innovation and Translational Research); the **Leadership School** and the **Digital School**. The programmes were provided in a blended format with online live sessions (due to the Covid19 pandemic) and e-learning courses. Across the 4 EURORDIS Open Academy programmes organised in 2021 there were **89 trainees** from more than 27 countries and 62 trainers. At the end of 2021, **EURORDIS Open Academy e-learning platform** had over **2,027 registered users/alumni** from more than **155 countries**. Over 61h of training were available on the platform at the end of 2021, across the topics of medicine research and development, research and digital media. 5 new e-learning courses were added to the platform.
- To improve public awareness globally and leave no one behind, the international campaign Rare Disease Day involved over 100 participating countries in 2021. The Rare Disease Day governance was strengthened with the National Alliances partnering. More than 1000 events were organised and the video was translated into 36 languages in addition to all the RDD materials broadly disseminated to increase our reach towards the general public and increase awareness.
- EURORDIS continued its support to the **24 European Patient Advocacy Groups (ePAGs)** aligned to the scope of the ERNs. An important effort has been done to facilitate the exchange of good practices on patient engagement among all patient advocates and to improve the overall collaboration between ERN clinicians and ePAG advocates, leading to the publication of a **preliminary set of measures to assess the collaboration between clinicians and patients in the ERNs** and the impact of the ePAG advocates engagement in the ERNs. One strategic proposal has been made with ERN Board of Member States following EURORDIS advocacy for the integration of ERNs into the national healthcare systems; as a result, a **joint action has been launched in the EU4Health Work Programme 2022**. In 2021, EURORDIS closely followed the uptake of its publication on the **Recommendations to achieve a mature ERN system in 2030** (published in December 2020), which makes part of the Rare2030 recommendations.

Major achievements were reached in 2021 to promote early diagnosing and reduce the diagnostic odyssey faced by **persons living with a Rare Disease**.

- EURORDIS, alongside its Council of National Alliances, Council of European Federations and its members, set out and published the 11 **Key Principles to support a harmonised European approach to Newborn Screening (NBS)** in January 2021. In order to implement these 11 key principles, a call for action to promote best practices on NBS was launched for the Steering Group on Health Promotion, Disease Prevention and Management of Non-Communicable Diseases (SGPP).
- The launch of the **new collaborative Innovative Medicines Initiative (IMI 2 JU) European project Screen4Care** in October 2021, under Horizon 2020. Screen4Care will run for a period of five years with a total budget of 25 million euros, provided by the IMI 2 JU, a joint undertaking of the European Union and the European Federation of Pharmaceutical Industries and Associations. Screen4Care offers an innovative research approach to accelerate rare disease diagnosis, which is based on two central pillars: **genetic newborn screening and digital technologies**. EURORDIS plays a major role and is involved in all **areas of focus**. EURORDIS leads the Patient Advisory Board and is directing and advising all partners on the priorities, needs and perspectives of persons living with rare diseases.
- **EURORDIS has played a frontline advocacy work resulting in the adoption of the EU on the Health Technology Assessment (HTA) regulation in December 2021**, with the scientific consultation and joint clinical assessment of all advanced therapies and all cancer medicines from 2025 and all other orphan products from 2028. A Consortium of 13 HTA bodies (EUnetHTA21) has been established by the European Commission through a call for tender, to bridge between the

end of Joint Action 3 and the approval/implementation of the Proposal for a Regulation on European Cooperation on **HTA** (now: Regulation 2282/2021). EURORDIS engaged with EUnetHTA21 to ensure the involvement of patients in the Consortium's activities and to contribute to the implementation – in particular policies and guidelines – of the future EU cooperation on HTA under the Regulation. EURORDIS continued to contribute to the implementation of the European Regulation on HTA through its dedicated Task Force.

- EURORDIS has continued to support and coordinate the **Therapeutic Action Group, a forum for discussion amongst patient representatives across Committees/Working Parties of the EMA**, and worked with the TAG members on policy proposals for the **revision of the Orphan and Paediatric regulations and the General Pharmaceutical legislation**. EURORDIS also contributed to the **Open Public Consultation on the revision of the general pharmaceutical legislation**.
- In 2021, the **European Expert Group on Orphan Drug Incentives (OD Expert Group)** launched 14 policy proposals to improve the incentives framework for rare disease therapy

development. The initiative, which kicked off in 2020, is led by EURORDIS and the European Confederation of Pharmaceutical Entrepreneurs (EUCOPE). It brings together a multi-stakeholder group of representatives from the broad rare disease community, including researchers, academia, patient representatives, members of the investor community, rare disease companies and trade associations, supported by a knowledge partner.

- An overall Strategic Review and Evaluation of the past 5 years (2015-2020) was conducted, leading to the revision of the overall Strategy. The new **EURORDIS Strategy 2021-2030** including the EURORDIS vision, mission, values, strategy and 4 overarching strategic objectives were presented and adopted at the Annual General Assembly in June 2021.
- 2021 was also marked by important staff turn-over (about 30%), mostly linked to the impact of the pandemic. The staff replacement and induction have been particularly difficult under the social distancing measures due to the Covid19 sanitary crisis. This turn-over has been used as an opportunity to reshuffle the team and re-allocate human resources on new priorities.





# 1. PATIENT ADVOCACY

## 1.1 OUR ADVOCACY GOALS WITHIN OUR STRATEGY PRIORITIES FOR 2021-2030

In the course of 2021, 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.
- ◆ 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 health care and making possible patient mobility.
- ◆ Promoting research and bridging patient's perspective and researcher's activities.
- ◆ Addressing the issues of genetic testing, genetic counselling & newborn screening.
- ◆ Voicing/expressing patient preferences and needs in sharing of health data in rare disease information systems and repositories and in contributing to a European Health Data Space that supports care and research for rare diseases.





## 1.2 ADVOCATES FOR RARE DISEASES AS A PRIORITY IN THE NEXT DECADE 2021-2030

In 2021, 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, 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. In 2021, therefore, advocacy focused on:

- ✦ **Preparing for the next decade of rare disease legislative & policy framework to take the necessary steps to requalify rare diseases as a public health issue.**
- ✦ **Following the conclusion of the Foresight Study Rare 2030, a #30millionreasons campaign in view of the revamping of the EU rare disease strategy and advocating for a goal-oriented European Action for Rare Diseases.**
- ✦ **Exploring and consolidating political options with EU Institutions - in the context of the above campaign - to update, review or replace the overall EU rare disease strategy on the basis of the Rare2030 policy recommendations for rare disease policy in 2030 (issued in February 2021), paving the way to**

Across the year, EURORDIS also contributed to the preparatory phases of policy and legislation with great impact on lives of people with rare diseases, by providing specific contributions to the definition of the roadmaps and to the stakeholders' consultations on the upcoming legislation for special populations (revision of the Regulations on orphan medicinal products and medicines for children) and the general pharmaceutical legislation and the future regulation on a European Health Data Space. Proposals for new legislation are expected from the European Commission in 2022. EURORDIS also collected inputs from members and ePAGs to submit an extensive contribution to the evaluation of the Cross-Border Healthcare Directive, which run in parallel to the ERN evaluation process. Last but not least, 2021 marked the culmination of the advocacy process that led to the adoption of the Regulation on Health Technology Assessment.

**the new EU policy framework for rare diseases**, and in line with the 2019 Court of Auditors special report on cross border health care. Actions included:

- ✦ **Advocating with the European Commission** for the transposition of Rare 2030 recommendations, with their specific call for a new policy framework for rare diseases, into a new policy initiative (Action Plan) with relevant Commissioners and cabinets and European Commission's services (DG SANTE, in particular).
- ✦ **Engaging with Trio of EU Presidencies 2022-2023, plus Spain**, to support the EU policy strategic review and new policy framework for rare diseases.
- ✦ **Consolidating the European Parliamentary Advocates for Rare Diseases network** and gaining their support for a European Action Plan for RDs, leading to an oral question and plenary debate.
- ✦ **Running a broad grassroots campaign** to mobilise EURORDIS members at large, with greater integration of advocacy actions at the local, national and European level.

EURORDIS followed up the discussions on the next budgetary period that led to the adoption of the new **EU Multiannual Financial Framework 2021-2027** and the revision of the **EU4Health programme** to respond to the pandemic crisis, with its ten-fold budgetary increase. In summer 2021, EURORDIS prepared the reflections on priority areas for funding that are relevant for the rare disease community and submitted them to the European Commission. In parallel, EURORDIS actively successfully contributed to the campaign of a coalition of health civil society organisations for the re-establishment of the Operating Grant for health NGOs, following the discontinuation of such grants in the first EU4Health Work Programme.

### 1.2.1 Parliamentary Advocates for Rare Diseases

*The network of Parliamentary Advocates for Rare Diseases brings together European and national members of parliament advocating to improve the lives of the 30 million people living with a rare disease in Europe. They 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.*

Following its relaunch in 2020, the **Network of Parliamentary Advocates for Rare Diseases** established a core group composed of a Chair (Frédérique Ries Renew Europe, Belgium) and four Co-Chairs from as many political groups.

MEP Ries, chair of the Network and one of the three sponsors MEPs of Rare 2030, hosted the final event of Rare2030, a foresight study that the European Parliament promoted and the European Commission co-funded. This landmark event marked the launch of the Rare2030 recommendations for a European policy for rare diseases and the beginning of the campaign #30millionreasons for the new EU policy framework on rare diseases and stronger EU-wide action in health, research, social affairs and other relevant policies.

Later in the year, due to the inaction of the European Commission, members of the Network, under the leadership of Mrs Ries, secured the passing of an

**Oral Question** to the European Commission calling for the prompt implementation of the Rare 2030 recommendations and the adoption of the EU action plan for rare diseases by 2023 in the form of a comprehensive plan with measures and targets, in order to leave no one living with a rare disease behind. This led to a European Parliament debate in November 2021 which the Commissioner for Health Stella Kyriakides took part in and nearly 20MEPs took the floor in support of a European Action Plan for rare diseases. Ahead of the plenary meeting, EURORDIS welcomed MEPs at a breakfast briefing to prepare the plenary debate. Following the debate, 43 MEPs addressed a letter to the Commissioner as, despite her commitment to implement some of the Rare 2030 recommendations, she stopped short of announcing a comprehensive policy framework to connect all policies and initiatives affecting people living with a rare disease at European and national levels.

Across 2021, EP members of the network for rare diseases and other MEPs joined forces to call on the European Commission to bring back the operating grants for health civil society organisations in the funding programme EU4Health, in support of the coalition that EURORDIS co-lead with the European Patient Forum, the European Public Health Alliance and Alzheimer Europe. An **open letter** addressed to the European Commission, co-signed by 22 health NGOs and 57 MEPs, emphasised how health and civil society organisations are vital partners to both European and national institutions to shape and implement public health strategies and policies. The MEPs support contributed to the positive conclusion of the campaign and the reinstatement of civil society operating grants in the EU4Health programme Work Programme 2022.

## 1.2.2 Foresight Study in Rare Diseases (Rare2030)

Over the past two years, EURORDIS has led the Rare 2030 Foresight Study with 7 institutional partners. With the participation of over 250 experts and almost 4000 patients and patient advocates, this study followed four steps of foresight:

- ✦ Establishing knowledge base
- ✦ Identifying trends and wild cards
- ✦ Preparing potential scenarios
- ✦ Developing policy recommendations

This participative and iterative multi-stakeholder study gave consensus for a future defined by equality – a future where the place of birth does not affect one's access to the best diagnosis, care and treatment; a future where innovations across the pathway are needs-led; a future, where every person living with a rare disease is valued in society.

Its conclusions were presented at The Rare 2030 Plenary Conference, "The future of rare diseases starts today", held in the lead up to the fourteenth edition of Rare Disease Day, bringing together over 1000 participants to discover the long-awaited recommendations from the Rare 2030 Foresight Study. Prominent EU policy makers, including Commissioner for Health and Food Safety Stella Kyriakides and French Health Minister Olivier Véran attended and spoke at the conference.

The report, "Recommendations from the Rare 2030 Foresight Study: The future of rare diseases starts today" sets out eight overarching recommendations across diagnosis, treatment, data, research and integrated care, each accompanied by measurable goals to monitor progress made across Europe.

The Recommendations offer a roadmap for a new rare disease framework by establishing measurable goals for the first time. In line with the UN Sustainable Development Goals, such goals set a common direction for all EU Member States to make tangible change for all their citizens living with a rare disease.

If a new framework for rare diseases is implemented by EU institutions and Member State governments, the Rare 2030 recommendations hope that by 2030:

- ✦ All people affected by a known rare disease will be diagnosed within six months of coming to medical attention
- ✦ 1000 new therapies will be available, and they will be 3 to 5 times more affordable
- ✦ The level of psychological, social and economic vulnerability of people with a rare disease and families will be reduced by one third

Complete recommendations and study outputs were formally submitted to the European Commission in May 2021.



### 1.2.3 Campaign for a European Action Plan for Rare Diseases

Following the submission of the results of the Rare 2030 Foresight Study, EURORDIS-Rare Diseases Europe in collaboration with its European Federations, National Alliances for Rare Diseases and 984 Patient Organisation Members, as well as the wider rare disease community embarked on a campaign to advocate for a proposed European Action Plan for Rare Diseases.

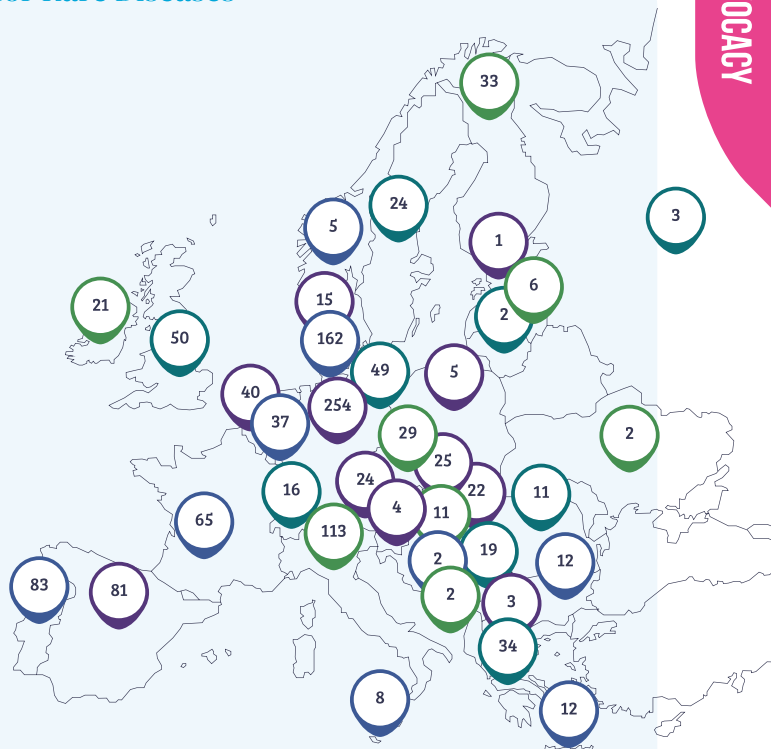
This campaign primarily addressed the European Commission. Other European institutions (European Parliament, European Council) and key opinion leaders were all part of this multi-layered approach.

One level of the campaign included a grassroots effort to hear from people living with rare diseases about their reason "Why does Europe need to take action on rare diseases now?"

Across Europe, over 2100 people – those living with a rare disease, parents, siblings, friends, advocates, healthcare professionals and researchers – shared their reasons why Europe must act. These are personal experiences, hopes and fears that are moving, motivating, sometimes devastating, impassioned and humbling. And all were made available in the form of a book, [website and video testimonials](#).

Other major milestones in the campaign included advocacy with Members of European Parliament in preparation of an Oral question with debate on the subject on 24 November 2021. Following the debate 41 member of European Parliament co-signed a letter to Commissioner for Health, Stella Kyriakides

A number of campaign materials helped address the range of key opinion leaders taking part in these advocacy efforts:



- ✦ A concept note describing the key needs and attributes of such a plan
- ✦ A confidential mock-up for campaigners to understand what a full plan could include
- ✦ Template letters to address the importance of a plan to health authorities at the European and national levels

## 1.3 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 legislation for "special populations" (the Regulation on Orphan Medicinal Products and the Regulation on Paediatric Medicines) has been under the microscope for quite some time.

In 2017, the European Commission launched the process for a joint evaluation of the legislation on medicines for children and rare diseases. A study was commissioned to understand the strengths and weaknesses of the Regulation on Orphan Medicinal Products. The purpose of the evaluation was two-fold:

- ✦ To focus on the output and results of the two Regulations: What respect patients' needs have been fulfilled in, what the societal consequences

have been experienced and what the synergy between two regulations has been achieved.

- ✦ 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 process included a stakeholder workshop on **"Medicines for Rare Diseases and Children: Learning from the Past, Looking to the Future"**, held in 2019, and the publication of the results in 2020. The evaluation report, while stressing the positive impact on the legislation on the medicine development, also highlighted that the drug development has not been efficiently supported in those rare and paediatric areas where the need for new medicines is the highest, while also indicated that European citizens do not have the same access to authorised treatments.



In 2021, in the context of the 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 and areas with no therapeutic options yet; 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.

In 2021, the European Expert Group on Orphan Drug Incentives (OD Expert Group) launched 14 policy proposals to improve the incentives framework for rare disease therapy development. The initiative, which kicked off in 2020, is led by EURORDIS and the European Confederation of Pharmaceutical Entrepreneurs (EUCOPE). It brings together a multi-stakeholder group of representatives from the broad rare diseases community, including researchers, academia, patient representatives, members of the investor community, rare disease companies and trade associations, supported by a knowledge partner.

EURORDIS also contributed to the WHO Europe Oslo Initiative (OMI) on pricing scheme and funding models to improve access to rare diseases therapies in Europe.

## 1.4 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 particularly, 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 alias, 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), as well as from shortages that can occur at any time. EURORDIS published the paper **“Breaking the Access Deadlock to Leave No One Behind”** in 2018 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.

The multistakeholder foresight study “Rare2030” (2019-2021) highlighted how science and technology offer an unprecedented chance to address the unmet medical needs of people living with a rare disease. This potential is currently not translated into actual health benefits for the large majority of people living with a rare disease due to issues concerning availability, accessibility and affordability of treatments. The ‘Rare 2030 recommendations’ were adopted in February 2021 and outlined the way forward, which is centred around patient-led innovation, solidarity and the added value of collaborative action at European level to drive change in the accessibility, affordability and sustainability of the life cycle of medicines for rare diseases.

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



*EURORDIS has been driving the RARE IMPACT initiative since 2018. RARE-IMPACT aimed to identify and validate the challenges to patients' access to advanced therapeutic medicinal products (ATMPs) 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.*

The first phase of 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, with seven solutions highlighted, alongside eleven country specific reports.

Given the remaining challenges highlighted in the report, and building upon the recommendations on the final report, participants to the RARE IMPACT Initiative initiated a second phase of the project, which is now concluded, focusing on three key areas with dedicated workstreams: price and the economics of ATMPs; evidence generation for ATMPs; processes and criteria of selection of Centres of Expertise for ATMPs.

In the first workstream, the project aimed to create a dialogue on the topic of pricing and economics of ATMPs, so as to improve the understanding around the ATMP innovation model, as well as the implications for sustainable health systems and innovations. The key objectives were: to help contextualise prices within the broader ATMP innovation model through collective exploration, to explore the economics of development of ATMPs in rare diseases, and to enable an open dialogue across stakeholders and advance mutual understanding of the pricing issues of ATMPs.

In the second workstream, the initiative engaged in a dialogue with stakeholders to better understand these challenges and to discuss the potential solutions to improving the evidence base for ATMPs, particularly through the collection of complementary, real-world data. Recommendations were proposed to address these issues, in order to support efficient decision-making and to facilitate timely patient access to ATMPs.

The ambition of the third workstream was to gather knowledge on the process for the selection of the Centres of Expertise that deliver commercially available Advanced Therapy Medicinal Products (ATMPs). The objectives were to better understand what criteria are used for the selection of Centres of Expertise at hospital, company, but also national and EU levels – European Reference Networks (ERNs) and others. The work has helped to identify the common and divergent approaches in the criteria that apply for Centres of Expertise selection.

## 1.4.2 Preparing the reimbursement decision: the HTA momentum

### Advocate for the EU HTA Regulation

EURORDIS played a frontline advocacy work resulting in the adoption of the EU on the Health Technology Assessment (HTA) regulation in December 2021, with the scientific consultation and joint clinical assessment of all advanced therapies and all cancer medicines from 2025 and all other orphan products from 2028.

### EUnetHTA JA3 – European Network for HTA, Joint Action 3 (2016-2020/2021)

The European Network for HTA (EUnetHTA) ended its mandate in 2020. An extension to fulfil Joint Action 3 last deliverables was granted by the European Commission, until May 2021.

The scope has been limited to Covid-19, reporting, and a White Paper. Through the extension period, EURORDIS remained committed to participating in consultations,

collaborating in EUnetHTA Events for stakeholders (contributing to the agenda and providing speakers), and informing its members on the work done.

### HTA Network and HTA Network Stakeholder Pool

The HTA Network (chaired by DG SANTE and composed of national HTA authorities and selected Stakeholders) is the body that advises the European Commission on the European Cooperation on HTA.

It is also supported by the HTA Network Stakeholder Pool, composed, among other categories of stakeholders, by 9 Patients and Consumers Organisations.

EURORDIS assumed the secretariat of the Patient and Consumers organisations of the Pool and represented them at the HTA Network (together with the Bureau Européen des Consommateurs).

## EUnetHTA 21

A Consortium of 13 HTA bodies (EUnetHTA21) has been established by the European Commission through a call for tender, to bridge the end of Joint Action 3 and the approval/implementation of the Proposal for a Regulation on European Cooperation on HTA (now: Regulation 2282/2021). EURORDIS engaged with EUnetHTA21 to ensure the involvement of patients in the Consortium's activities, and to contribute to the implementation of the future EU cooperation on HTA under the Regulation.

## Other activities

EURORDIS continued to engage with the HTA stakeholder community in public conferences and at national level to inform them about the European Commission initiatives on HTA and the work done by EUnetHTA, and to discuss the involvement of patients in HTA (especially with respect to the European Commission's Proposal for a Regulation on European Cooperation on HTA).

### 1.4.3 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 causes and identify possible solutions.

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



## 1.5 ADVOCATE FOR PROGRESS IN PATIENTS' RIGHTS TO CROSS-BORDER HEALTHCARE

In 2021, the European Commission began the evaluation of Directive 2011/24/EU on patient rights in cross-border health care ("Cross-border Healthcare Directive"), ten years after its entry into force and eight years after its transposition deadline the evaluation of the Directive. It is the ambition of the Directive on cross-border health care to ensure the right of every person living in Europe, including people living with a rare disease, to access the best possible diagnosis, care and treatment without unnecessary delay or physical, social or financial burden, when this cannot be met within the country of residence. The evaluation assessed how the Directive's objective to facilitate access to safe and high-quality cross-border healthcare in another Member State has been met and to what extent the Directive has promoted patient rights and cross-border cooperation between Member States for the benefit of EU citizens. It also looked into the approaches implemented by Member States in practice, how effectively they were working, what barriers patients still face seeking health care across borders and how the treatment and diagnosis of patients with rare and complex diseases have benefitted from the support of the European Reference Networks.

In order to contribute to the Stakeholder Consultation in July 2021, EURORDIS-Rare Disease Europe carried out an extensive consultation with people living with a rare disease and patient advocates, including surveys through EURORDIS' survey programme "Rare Barometer", dedicated webinars and targeted consultations with specific member groups (for example Rare Disease National Alliances and European Federations representing a disease area across Europe) and patient advocates in European Reference Networks. From this broad consultation, it unequivocally emerged that, despite its ambition, the Cross-border Health Care Directive was currently failing people living with a rare disease in Europe. This was submitted within the consultation framework and was developed into a response, **An empty promise: accessing cross-border healthcare for people living with a rare disease**, drawing on the summary of responses received with recommendations for action. This was shared with the European Union Institutions, membership and wider stakeholders to set out our strong position.



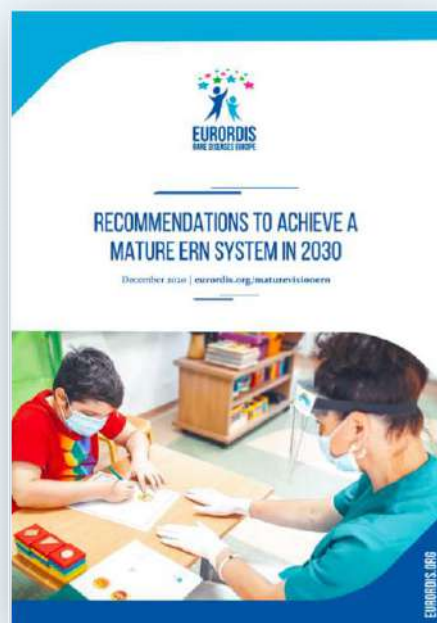
## 1.5.1 Advocate for the development of a mature ERN system

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 ePAG advocates. The paper, and the accompanying Policy Brief, reviews progress achieved so far and presents the rare disease patient community vision of a mature ERN system as well as specific recommendations on how to achieve this vision.

These recommendations were largely endorsed by the Rare2030 expert panel and used as a basis for the discussions on highly specialised healthcare and ERNs. Likewise, these recommendations have informed EURORDIS advocacy actions to ensure adequate funding for the ERNs under the EU4Health programme, as well as our proposal for a coherent strategy to expand the ERNs model (jointly submitted with the ERN Coordinators).

EURORDIS involvement to help shape the ERICA CSA proposal, and later in the future Rare Disease Partnership, was also largely driven by the proposals on clinical research networks included in the Mature ERN system paper.

We also used the vision and recommendations outlined in the paper to provide feedback and contribute to developing the proposal of the new Assessment, Monitoring and Evaluation and Quality Improvement System for the ERNs. Both directly and also via the support provided to the ePAG advocates who have been contributing to the different consultations organised by the EC contractor.



Finally, on an operational level, EURORDIS is implementing the vision around patient partnership established in the Mature ERNs paper, as well as the Rare 2030 Recommendations, by working with the ERN Coordinators, project managers and ePAG advocates to formalise the involvement of patient representatives in the ERNs governance structure and foster a partnership culture between clinicians and patient representatives involved in the Networks.

# 1.6 ADVOCATE TO IMPROVE ACCESS TO QUALITY RARE DISEASE DIAGNOSIS

Throughout 2021, EURORDIS continued to advocate for improved access to and quality of rare disease diagnosis.

## 1.6.1 Rare Barometer new Survey: the journey of rare disease patients to diagnosis

The last quarter of the year for the Rare Barometer Team has been largely dedicated to collecting insights on the journey of rare disease patients to diagnosis. A literature review has contributed to highlighting the gaps on this issue. A first qualitative study gathering around 60 patient representatives through an online dedicated platform was organised to form the basis of the quantitative survey questionnaire. A Topic Expert Committee comprised of sociologists, patient organisation representatives, geneticists, policy experts and industry partners was established to provide input on the development of the quantitative survey. The questionnaire has been finalised and sent for translation.



## 1.6.2 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 the most rare diseases (RD) by 2020 and fully integrates with the formation of ERNs.***

To date, the Solve-RD Project has analysed 11,763 datasets from 4457 families. The project has already solved 444 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. The infographic has been translated in 24 languages.

## 1.6.3 Undiagnosed Community

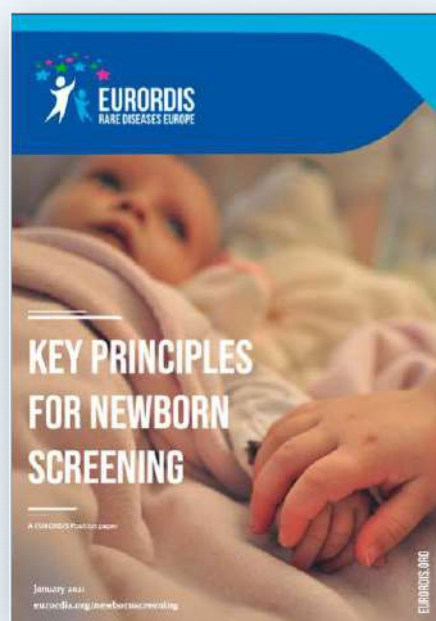
EURORDIS participates in Undiagnosed Diseases Network International (UDNI), an international network of clinical centres that was initiated in 2014 to address unmet needs of undiagnosed patients at a global level. The UDNI brings clinicians, researchers, genetic counsellors, and other medical professionals 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, clinical, 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, it has 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.

## 1.6.4 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 have been published in January 2021 and have been translated into 12 languages.***

In 2021, EURORDIS started to implement some elements of the call to action associated to the 11 Principles, also in recognition of the fact that early diagnosis, and indeed newborn screening, would be one of the building blocks of Europe's Action Plan for rare diseases EURORDIS is campaigning for. Given its ability to improve the lives of people living with a rare disease and their families. An overarching framework would encourage best practice and ensure its integration across EU Member States to ultimately reduce inequalities.

EURORDIS also has been one of the main organisers of the high-level technical meeting organised under the aegis of the Slovenian Presidency of the EU Council. The meeting, "Achieving Equity and Innovation in Newborn Screening and in Familial Hypercholesterolemia Paediatric Screening across Europe" took place on 11 October 2021 where EURORDIS presented the perspectives of people living with a rare disease. EURORDIS is also a co-author

of the publication "Towards Achieving Equity and Innovation Newborn Screening across Europe" which has been submitted to the International Journal of Neonatal Screening following the results of the Slovenian meeting. The publication will help passing the baton to the upcoming Czech Presidency of the EU Council who are planning to organise a meeting on Early Diagnosis in 2022.

At the end of 2021, EURORDIS also joined forces with UNIAMO, the Italian National Alliance, to promote the Italian extended newborn screening model as a European best practice. **The campaign**, in line with the 11 Principles and its call to action, supported a harmonised approach to newborn screening as the only way to ensure children across Europe undergo the most comprehensive screening available. With Italy having shown great leadership in their newborn screening programme, EURORDIS and UNIAMO invited the wide European community of rare diseases and the public at large to sign a petition asking the Italian Government to promote its model across Europe to guarantee every child's right to health.

## 1.7 ADVOCATE TO IMPROVE ACCESS TO DISABILITY RIGHTS

On 24 March 2021, the European Commission adopted the **new European Strategy for the Rights of Persons with Disabilities 2021-2030**. The Strategy is a part of the European Union's commitment to promoting, protecting and ensuring the full enjoyment of human rights by individuals with disabilities and complex needs. As a result of EURORDIS' advocacy over the past years, and of the support of the European Disability Forum who collaborated with EURORDIS in this effort, the Strategy takes into account rare diseases and disability related recommendations that EURORDIS promoted in its advocacy.

In line with EURORDIS' and members' **position paper on Holistic Care** (2019), with EURORDIS' **contribution to the EC consultation** (2020) and with the **Rare2030 recommendations** (2021), the strategy calls for:

- ✦ More awareness and support strategies for patients with disabilities related to rare diseases - including two text references to 'rare diseases', which is unprecedented in disability-policy documents;
- ✦ Reforms of social protection focusing on persons with disabilities and disability assessment frameworks;
- ✦ Improved labour market outcomes of persons with disabilities, including reasonable accommodation at work.

This is a great step forward for the rare disease community and a direct consequence of the collective decade-long plea to gather evidence on disabilities faced by people living with rare diseases.



This high-level European Strategy needs now to be duly implemented at the national and local levels so that its recommendations are fully transposed in daily lives of those affected. EURORDIS continued in 2021 and will continue in the years to come to follow the process of implementation of the Strategy to enable the rare disease community to seize the opportunities to provide inputs and to support advocacy at national/disease level. The Europe-wide campaign for Europe's Action Plan for Rare Diseases, described above, identified the reforms of the disability assessment frameworks as one of the 'flagships' of the European Plan, which should encompass all EU policy areas that are relevant for rare diseases and include them in a cohesive way.

## 1.8 GATHERING PATIENT EXPERIENCE AND PERSPECTIVE FOR EVIDENCE-BASED ADVOCACY

The Rare Barometer programme consists of surveys aiming to collect qualitative & quantitative data on the experiences, needs & expectations of RD patients & their families and carers. Rare Barometer secured a panel of rare disease patients who agreed to take part on a regular basis to EURORDIS' quantitative surveys. Figures and facts from the studies are used to develop EURORDIS' Advocacy positions. All studies are translated in 23 languages and made available to rare disease patient organisations.

After the end of the survey fieldwork, the results of the Rare 2030 survey on the future of rare disease were analysed through an in-depth report and summarised through a factsheet. Targeted results have been communicated to patient organisations from our network. Those results, together with past surveys conducted through the programme, have significantly contributed to the design of the Rare 2030 project recommendations. The webinar organised on the 18th of October 2021 gathered more than 80 participants and presented the results of the survey as well as the evidence-based process leading to the Rare 2030 recommendations, in order to provide advocacy tools for the rare disease community at large.

The team has worked on project proposals submitted to the 3rd EJP RD Joint Transnational Call focusing on Social sciences and Humanities research:

- ✦ One of them was coordinated by Rare Barometer, building on the H-Care survey pilot aiming to measure rare disease patients experience of their healthcare. Although not funded, the project proposal had been recommended by the scientific executive committee of the call, which highlighted the relevance, quality and scientific value of the proposal.

- ✦ The LIVES proposal has been accepted for funding. Rare Barometer will contribute to the development of a validated and open source scale aiming to measure the impact of rare diseases on young adult employment, education and inclusion in society.

The results of the H-care survey were presented on several occasions, including the EURORDIS Black Pearl Awards. Targeted results were also presented to hospitals who participated in the pilot.

The results of the past surveys, in particular "Share and protect our health data" and "the Impact of COVID19 on people living with a rare disease" were presented on several occasions and are continuously used by staff members to feed advocacy positions and intervention in conferences and events.

The Rare Barometer programme worked on 2 IMI projects that have been accepted for funding this year:

- ✦ Within Screen4Care, the programme will conduct a quantitative survey to contribute to a conceptual framework defining the notion of "actionable disease". It will also bring in the voice of patients for the co-creation of a platform aiming to facilitate the diagnosis of rare diseases.
- ✦ Within FACILITATE, the programme will voice the needs and concerns of rare disease patients regarding how they would like clinical trial data to be returned to them.

The work on Diagnosis is indicated in the above section on diagnosis.

## 1.9. EUROPEAN HEALTH DATA SPACE (EHDS)

Sharing health data to advance scientific research and improve clinical practice is of particular importance to the rare disease community, where knowledge and expertise are limited, patient populations are geographically dispersed, and their health data is scattered.

Over the last years, EURORDIS has actively identified and promoted the specific challenges and needs of people with rare diseases with regard to health data sharing at European and international levels. EURORDIS has produced **quantitative evidence** of rare disease patient perspectives on health data sharing and, based on these findings, we have participated in multi-stakeholder dialogue in the run up to the upcoming Regulation on the EHDS and have promoted rare diseases as a pioneering user-case and to formulate concrete proposals.

In 2021 EURORDIS responded to the consultation on the EHDS Roadmap proposing to use ERNs as a testing environment to implement and assess new health data governance models, by leveraging on their experience with health data sharing for primary purposes (experts' advice) and secondary uses (clinical practice guidelines, clinical decision support tools, clinical research). We also highlighted the need to have an institutional architecture that would bring under the same umbrella the technical and policy aspects of cross-border health data sharing, centred around an EU level agency, connected to the national health data agencies. Additionally, we stressed the need to clarify the impact of the Data Governance Act in the health sector and control of patients over their health data.



Later in the year, EURORDIS responded to the public consultation on the EHDS, providing more specific feedback on the alternatives put forward by the Commission to enable the use of health data for healthcare provision, research and innovation as well as policy-making and regulatory decision; the development and use of digital health services and products and the development and use of Artificial Intelligence systems in healthcare. EURORDIS also participated in the working group that drafted the Friends of Europe report "Building a path for rare diseases in the European

Health Data Space", bringing the voice and needs of the rare disease patient community to help shape the different recommendations included in this report.

We have also engaged with TEHDAS (Towards a European health data space) Joint Action, participating in two different work packages (WP5 and 8) and also as part of the general stakeholder forum. The project's goal is that in the future European citizens, communities and companies will benefit from secure and seamless access to health data regardless of where it is stored.

## 1.10 PROMOTE RARE DISEASES AS AN INTERNATIONAL PUBLIC HEALTH PRIORITY WITH RARE DISEASES INTERNATIONAL

*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 is the global alliance of people living with a rare disease of all nationalities across all rare diseases. The mission of RDI 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 80 member organisations from over 30 countries, that in turn represent rare disease patient groups in more than 100 countries worldwide and on 6 continents. The relationship between EURORDIS and Rare Diseases International is regulated by an MoU signed by both organisations (2018-2023), which includes financial, staff and services support. EURORDIS is seating on the Board and the RDI Advocacy Committee (Yann Le Cam is RDI's Treasurer and is Chairing the RDI Advocacy Committee). EURORDIS therefore contributes to many RDI activities. These activities are reported comprehensively in the RDI Activity Reports.*

### 1.10.1 Rare Disease Day Policy Event in the framework of RDD



On 4 March, to mark Rare Disease Day 2021, RDI hosted a policy event in partnership with the NGO Committee for Rare Diseases and EURORDIS. The event entitled '**Call for a UN Resolution on Persons Living with a Rare Disease – Why a UN Resolution & Why Now**' launched the international campaign for a UNGA Resolution.

261 participants from 83 countries attended the event, which brought together civil society organisations, PLWRD and families, UN Representatives, UN Member States, and policymakers.

The overarching goal of the event was to position PLWRD as a priority population requiring international action and propose that addressing the needs of this vulnerable population would contribute to achieving the UN 2030 Agenda.

At this event, the civil society partners presented the resolution proposed by the rare disease community with its "Five Key Ask", five key demands expressed by the draft text.

PLWRD and their families, represented by patient advocates from New Zealand, Brazil, Malta, and Kenya, appealed to the world's leaders, describing what it means to live with a rare disease and highlighting the potential impact of the resolution.

Nata Menabde, Executive Director at World Health Organisation, affirmed that health systems are not adapted for rare diseases and the needs of PLWRD. Todd Howland from the UN Office of the United Nations High Commissioner for Human Rights said that a lack of awareness and visibility fostered the discrimination and exclusion of PLWRD.

This was the first opportunity for the Member States to show support for the proposed resolution.

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## 1.10.2 Adoption of the UN Resolution on “Addressing the needs of persons living with a Rare Disease

EURORDIS' longstanding work started in 2009 to promote rare diseases as an international public health priority culminated in the United Nation General Assembly adoption by consensus of the **first ever UN Resolution on “Addressing the needs of persons living with a Rare Disease”** in December 2021. Two years after the adoption of the UN Political Declaration on Universal Health Coverage (UHC) which explicitly included in its scope all PLWRDs, the UN Resolution is a landmark to recognise the 300 million PLWRDs in the world, to promote rare diseases across the UN programmes and agencies, and to encourage all UN member states to address the specific challenges of PLWRDs. This political success was made possible thanks to the close work with Rare Diseases International and the UN NGO Committee for Rare Diseases.

EURORDIS spearheaded the work in the RDI Advocacy Committee to define the key asks, performance of benchmark analysis of best relevant provisions in other resolutions, and drafted the text of the resolution. When a core group of UN Member states was formed – Spain, Brazil, Qatar – after the NGO Committee for Rare Disease event in March 2021, EURORDIS and RDI provided both the draft as a base of work and a secretariat support to the core group. EURORDIS engaged the national alliances across Europe to reach out to their ministries of foreign affairs, health and social affairs, which allowed to secure the support of key EU member states, thereafter of the EU and of the several non-EU European countries, until the adoption by consensus.

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## 1.10.3 Universal Health Coverage for Rare Diseases (#UHC4RareDiseases) Campaign



Universal Health Coverage (UHC) ensures all people have equitable access to diagnosis, treatment, and care without facing financial hardship. For UHC to indeed be universal and cover all vulnerable communities, UHC policies must address the needs of PLWRD.

RDI is a member of the Civil Society Engagement Mechanism (CSEM) of UHC2030, a body founded by the World Bank and the World Health Organisation to advance equitable UHC policies and action.

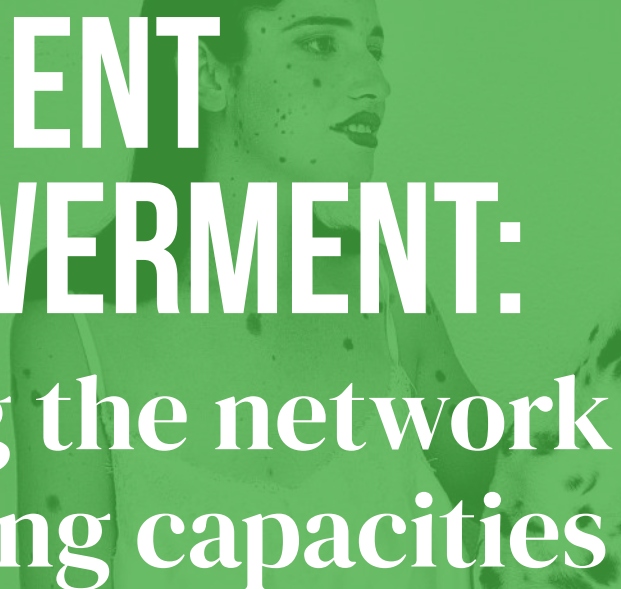
In September 2021 the CSEM developed the Health for All Advocacy Toolkit to strengthen capacity and mobilise civil society in support of UHC. RDI's #UHC4Rare Toolkit appears as an essential resource in the CSEM's toolkit, reflecting the growing visibility of the rare disease community in the international UHC movement.

To mark UHC Day (December 12), RDI partnered with the CSEM to host an in-depth discussion on health equity for PLWRD, entitled "Strengthening Care Systems to Achieve Equity." The event was an opportunity to urge policymakers to keep the promise to "leave no one behind" and achieve UHC by giving greater attention to equity and strengthening care for vulnerable groups.

The #UHC4RareDiseases Toolkit provides practical tools for patient advocates to raise awareness and ask policymakers to safeguard equity and consider the needs of PLWRD in national UHC strategies.

Overall, the UN Agenda 2030 and its Sustainable Development Goals are providing an important framework shaping EU objectives. They are also the framing the Rare 2030 recommendations and new European Action for Rare Diseases. The inclusion of rare diseases in UHC and the UN Resolution are revitalising the mandate of the European Union and to WHO-Europe to take action to address the unresolved challenges of persons living with rare diseases in Europe.





# 2. PATIENT EMPOWERMENT:

## Building the network & building capacities

### 2.1 COMMUNITY-BUILDING, NETWORKING & CAPACITY-BUILDING OF PATIENT ADVOCATES

#### 2.1.1 Membership

59 new members joined EURORDIS in 2021. At the end of 2021, EURORDIS had 984 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, 39 of which constitute the CNA.

The CNA's main activities in 2021 were:

- ✦ the preparation and coordination of the Rare Disease Day 2021 and 2022
- ✦ Integration of ERN at National level
- ✦ Newborn Screening
- ✦ EU Campaign for a new policy framework

In 2021, 2 CNA Workshops took place online. The first, in April, was a one-day workshop for CNA members only. The second, in November, was organised over 2 days, the first dedicated to the CNA and the second gathering CNA and CEF representatives.

The first workshop gathered 48 participants and focused on the following topics: - EU Campaign for a new policy framework - Rare Disease Day Global Campaign 2022 - Debrief Rare Disease Week 2021 and plans for the future - Covid 19.

The second CNA meeting took place in November over 2 days, the first dedicated to CNA only and the second in conjunction with the Council of European Federations.



### 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 2021 over 2 days, including one day jointly with the CNA:

- ✦ Revision of the general pharmaceutical legislation
- ✦ UN Resolution – What does it mean? What is next?
- ✦ Including rare cancers in Europe's Beating Cancer Plan
- ✦ Covid-19
- ✦ New CABs programme
- ✦ A new training by EURORDIS on how patients can take part in HTA
- ✦ How can federations prepare themselves for a new pilot at the EMA, when the evaluation of a new medicine starts?

- ✦ Access to orphan medicinal products: where do we stand?
- ✦ Update on newborn screening

Over the year, two Informal CEF meetings were organised in collaboration with European Huntington Federation and OIFE. These meetings are peer-to-peer meetings where federations exchange experiences, ideas and best practices on issues all federations encounter in their management: communication, fundraising, people raising and keeping volunteers engaged.

EURORDIS re-started the support program in 2021, to adapt to the situation created by the pandemic. The purpose of the programme is to provide small, quick, and flexible financial support for the organisation of online meetings. All European Rare Disease Federations and National Alliances that are members of EURORDIS and the CEF or CNA respectively and have an annual budget of less than 50.000 € could apply for this support. 18 500 euros were granted to 11 European Federations/National Alliances.

### 2.1.4 European Network of Helplines 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 the members of the European Network.**

In 2021, **19 helplines 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). One new helpline attended the meeting: Gene People, UK.

The network conducted its 14th Annual Caller Profile Analysis in October 2021 and participated in discussions on digital platforms that exist to record calls / emails, and to follow-up with enquirers. The network also participated in a training on the classification of rare diseases.

### 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 2021 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 to 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 to ensure the smooth running of webinars.*

Webinars focus on providing policy updates, involving patients in consultations and providing capacity-building trainings for patient advocates. Webinars comprised an important communication tool during 2021 when face to face meetings were not possible. More than 10 webinars were organised on a number of topics mentioned in this report (e.g. Covid, CNA webinars, CEF Webinars, webinars for ePAG advocates).

## 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 2021, 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 2021. The programmes were provided online instead, in a blended format with online live sessions and e-learning courses.

Across the 4<sup>th</sup> EURORDIS Open Academy programmes organised in 2021, there were 89 trainees, from more than 27 countries, and 62 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 over 2000 registered users from more than 155 countries while providing over 60h of training.**

### EURORDIS Winter School on Scientific Innovation & Translational Research

*Launched in 2018, the EURORDIS Winter School consists of a one 4,5-day face-to-face training (online since 2020), preceded by an online pre-training. 21 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 4<sup>th</sup> EURORDIS Winter School on Scientific Innovation & Translational Research was held from 12-16 April 2021. The programme was planned to fit an online format due to the pandemic.

23 patient advocates from 15 countries and 26 trainers participated in the 2021 edition of the EURORDIS Winter School, which covered important topics to support patient engagement in research. The pre-training, composed of e-learning courses and 3 webinars, took place from December 2020 to March 2021.

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.

Fascinating  
**Informative** fun lengthy  
 levelup motivating  
 Helping long **Interesting**  
 Intresting intense **comprehensive**  
 Educative  
 relevant **inspiring**  
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## EURORDIS Summer School on medicine Research & Development

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

34 participants, including 1 staff attendee, 26 patient advocates and 7 researchers, from 19 countries, representing over 19 rare diseases took part in this training.

The pre-training of the EURORDIS Summer School 2021 took place from March to June 2021. The 5 half-day trainings took place online, due to the COVID-19 pandemic, from 21-25 of June 2021.

This year, great effort was into optimising the programme for a suitable online delivery, while maintaining the interactivity and hands-on exercises. A session on "Patient engagement", including an intervention from Summer School alumni, was added to the intensive training week. The "Patient engagement" session aimed at discussing participants' particular experiences and plans to engage with regulators, companies and researchers, based the survey that had been sent to the participants ahead. Building on feedback from the previous edition, an additional networking session with participants and organising staff was included in the programme to enhance online exchange and networking opportunities.

## 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 for 2021 include learning how to:

- ✦ define objectives for social media activity
- ✦ segment audiences by creating personas
- ✦ create a channel strategy and content plan
- ✦ initiate and sustain discussions in the rare disease online community.
- ✦ create and curate content for the community of persons living with rare diseases.

These objectives were met through a free e-learning course and a webinar led by social and digital media experts.





## EURORDIS 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. Expert trainers from across Europe deliver the training webinars and sessions which normally consist of a series of webinars, from June to November, and an intensive 3-day training in October.*

This third edition of the EURORDIS Leadership School took place online, due to the COVID-19 pandemic, from June to November 2021. After two online pre-training courses in June, 5 webinars took place between July and November. Additionally, 3 half-day intensive training days took place from 6-8 October 2021, also online.

The topics of the Leadership School included self-awareness, emotional leadership, communicating with powerful, effective and influential communication, problem solving and negotiation.

## EURORDIS First Rare Diseases Week (RDW)

*In the framework of the Rare Disease Day 2021, the first Rare Diseases Week (RDW) was organised by EURORDIS during the week 22 of February 2021. The RDW, gathering 38 participants from 20 countries and 20 different disease areas met with their European Parliamentarians online (due to Covid-19). This was an important landmark to train patient advocates in European policy and introduce them to their elected parliamentarians to enable knowledge exchange.*

RDW is a week-long series of events organised by EURORDIS in Brussels, including a series of preparatory webinars. It targets rare disease patient advocates in a view to empower them with knowledge and skills to effectively participate in advocacy activities at the European level and influence the EU decisions that have a direct impact on the lives of persons living with a rare disease.

The RDW aims to:

- ✦ Enable patient advocates to understand the EU legislative and non-legislative processes; to establish more effective interaction between the advocates, their national competent authorities and policy makers on European legislations and policies development or implementation, to engage more with their Members of the European Parliament. The participants could also use this knowledge at local and national levels.

- ✦ Raise awareness on rare diseases and present a strong and united message to Members of the European Parliament (MEPs) and other policy-makers in Brussels on behalf of the rare disease community.
- ✦ Engage the members of the network of Parliamentary advocates for Rare Diseases, expanding their opportunities.
- ✦ Establish an EU/national network of people who can readily participate in advocacy campaigns in Brussels

**The preparation for Rare Disease Week includes a series of preparatory events (webinars) about EU institutions, the European Union legal framework, and enhancing your skills as a rare disease patient advocate.** A total of seven webinars have been planned for the first-ever Brussels Rare Disease Week on subjects such as introduction on RDW, EU affairs (3 webinars) (meet the EU institutions, legislative procedures, how to navigate your way in Brussels; skills webinar; Rare2030. All webinars are recorded and available for all RDW participants.

This was followed by 5 days of school online including Meeting with European Commission, Meeting with Permanent Representation, Meeting with MEP, Model European Union. In this framework the participants could also attend 2 events open to public – the Black Pearl Awards ceremony 2021 and the Rare2030 Final Policy conference.



## 2.2 RAISING AWARENESS & INFORMING

### 2.2.1 Rare Disease Day 2021



*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.*

For the 14th edition of Rare Disease Day in 2021 the events took place in over 100 countries and regions on every corner of the globe despite the global COVID-19 pandemic. In areas with restrictions, many events took place online in order to continue educating and informing people about the over 300 million people living with a rare disease globally and their families. The global campaign used 10 individual stories from around the world of people living with a rare disease and their families to represent the international community and its diversity.

Thousands of events took place in over 100 countries. We welcomed 3 new countries to the campaign: Mali, Nicaragua, Uzbekistan. In addition to holding events, people around the world were inspired by the campaign visuals and shared information, photos and Rare Disease Day branded materials online. Over 550 buildings and

monuments were lit up around the world in the colours of Rare Disease Day in a Global Chain of Lights to show solidarity for people living with a rare disease and their families.

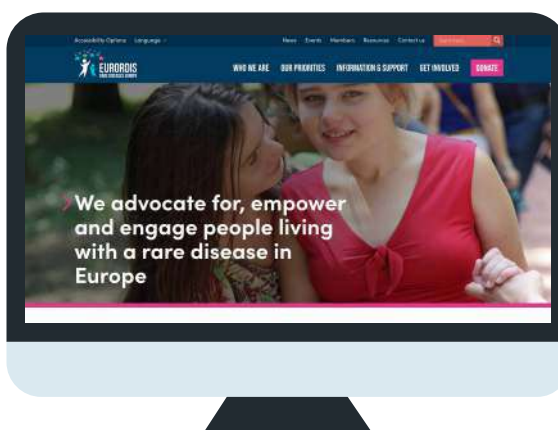
Media from all over the world covered the day, in which politicians, researchers, medical professionals and policymakers in Europe, the US and many more countries and regions participated.



### 2.2.2 EURORDIS Website

*The EURORDIS website outlines the events and activities of EURORDIS and provides information related 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 2021 we continued the backend development of our new website. We began translations and uploading the content. Having a website which is translated into 7 languages is technically challenging and requires several technical and development specifics. The expected launch is in the first semester 2022.





## 2.2.3 EURORDIS Newsletters

### eNews

The EURORDIS eNews is a monthly news report in English 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 provided via EURORDIS Facebook and Twitter.

Throughout 2021, 12 eNews issues were distributed to almost 11000 subscribers, available in 7 languages until August 2021, and subsequently in English only. This publication is year-round at a monthly frequency. This e-news publication, free of charge, gives stakeholders updates on the latest EURORDIS activities, 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 story topics in 2020 included: "UN Resolution is adopted: what does it mean for Europe?", "Lessons learnt: Building back better from COVID-19", "Revision of the EU pharmaceutical framework to ensure equitable access to rare disease therapies".



### 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 about 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 2400 contacts.

In 2021, our Member News was distributed twice a month to 2400 contacts at member organisations. The member news has separate sections such as Get Involved, Tools for you, and Policy Update. It also has a calendar of events to be sure our Members don't miss what is happening and we have noted high registration rates for webinars publicised through the Member News.

### Council of National Alliance Newsletter

A Council of National Alliances specific e-newsletter "CNA Update" was launched in 2020 to better target these important patient organisation members, who are heavily active in advocacy activities nationally. The CNA Update includes sections such as updates on the CNA meetings, the latest news such as Rare Barometer ongoing Surveys and results, updates on Advocacy activities. 11 NA newsletters were sent in 2021.





## 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 2021 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 and surveys, 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 EURORDIS Black Pearl Awards 2021



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 the unique ceremony 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 made by and for people living with rare diseases.

The 10th anniversary of the EURORDIS Black Pearl Awards was celebrated on 24 February 2021 fully online and marked the occasion of Rare Disease Day 2021. The event gathered 460 attendees representing all stakeholder groups of the rare disease community, connecting people from 55 countries.

### AWARDS OPEN to a public vote:



#### Young Patient Advocate Award

#### **Rachele Somaschini (Italy)**

For her impressive achievements in raising awareness about Cystic Fibrosis, as well as 200 000€ in funds for the Italian Cystic Fibrosis Foundation through her #CorrerePerUnRespiro project.



#### Visual and Audio Media Award

#### **Noémie Desquiotz-Sunnen (Luxembourg)**

For her outstanding achievements throughout her career as a musician, including the organisation of charity concerts.



#### Written Media Award

#### **Ana Ilić (Serbia)**

For her unique and raw poetry which has allowed greater and meaningful public to understand the true realities experienced by the rare disease community.



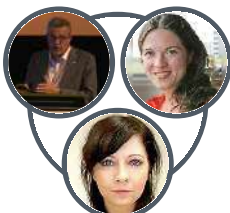
## Winners of the EURORDIS AWARDS 2021



### Lifetime Achievement Award

#### Prof. Milan Macek (Czech Republic)

For his lifelong dedication shown in addressing the needs of people living with a rare disease in the Czech Republic and in Europe, with EUCERD, the European Society of Human Genetics, the CF Network, Orphanet, and many more.



### European Rare Disease Leadership Award

#### Dr Enrique Terol (Spain), Dr Birute Tumiene (Lithuania) and Victoria Hedley (UK)

For their outstanding work in the development of the European Reference Networks, which has greatly contributed to a stronger European cooperation in the rare disease field.



### Policy Maker Award

#### MEP Kateřina Konečná (Czech Republic)

For her incredible leadership in championing the rare disease causes and her impressive commitment to advancing rare disease policies as a Member of the European Parliament.



### Scientific Award

#### Prof. Alain Hovnanian (France)

For his scientific excellence, comprehensive reach, dedication and groundbreaking advances in rare skin diseases (particularly Epidermolysis Bullosa and Pachyonychia Congenita).



### EURORDIS Volunteer Award

#### Ingunn Westerheim and Rebecca Tvedt Skarberg (Norway)

For the outstanding dedication they have shown to the rare disease community, to EURORDIS as volunteers, to OIFE, the European Joint Programme, Rare Disease Day, Rare Disease Week, Bond ERN and many others.



### EURORDIS Members Award

#### AKU Society (UK)

For their 17 years of advocacy and research in the rare disease field, which led to groundbreaking achievements in the rare disease community, particularly the DevelopAKUre and Patient-Centric leadership projects



### Company Award for Innovation

#### Orchard Therapeutics (UK)

For their leadership in harnessing the potential of hematopoietic stem cell gene therapy to change the course of severe inherited disorders, particularly for developing gene therapy for early onset metachromatic leukodystrophy (MLD).



### Company Award for Patient Engagement

#### Takeda (Global)

For their commitment to constructive collaborations with the rare disease patient community, from advocating for timely and accurate diagnosis and equitable patient access to therapies, through the multi-stakeholder dialogue on real-world evidence.



### Company Award for Health Technology

#### Epihunter (Belgium)

For creating an affordable and accessible digital solution that helps people with absence epilepsy to live more confident, fuller lives and providing clinicians and researchers with complete and accurate data to improve their care.

# 3. PATIENT ENGAGEMENT: Roles in decision-making

## 3.1 PATIENT ENGAGEMENT IN HEALTH CARE

### 3.1.1 European Reference Networks



EURORDIS supports patient organisations and patient representatives to ensure a meaningful involvement in the ERNs. There are 24 European Patient Advocacy Groups (ePAGs), one per ERN, bringing together patient representatives from European patient organisations. The ePAGs were created to ensure the patient voice sits at the core of the ERNs. ePAG advocates are supported by EURORDIS, they represent their wider patient communities through active engagement in the activities of their respective ERNs. Today, there are 314 ePAG patient advocates across the 24 ERNs.

EURORDIS supports ePAG advocates through dedicated individual ePAG calls (more than 90 calls in 2021), attendance to ERN annual meetings (9) and transversal working groups (17 calls). Specifically, in 2021 EURORDIS has increased its support through the ePAG horizontal working groups (17 calls), to facilitate cross-ePAG knowledge sharing on transversal ERN themes, such as research, clinical practice guidelines, monitoring and evaluation, governance, training and education and awareness raising. In addition, EURORDIS organised

5 ePAG Steering Committee calls to discuss strategic topics of relevance to the 24 groups. There has been also an important effort to facilitate the exchange of good practices on patient engagement among all patient advocates (6 webinars including on PROMs, joint patient-clinician ERN reflection sessions, clinical consensus statement, governance, as well as on scientific communication and registries).

In 2021, EURORDIS worked with the ePAG advocates to develop governance templates; it supported ePAG advocates to engage in the AMEQUIS workshops; worked closely with the ERN project managers, coordinators and advocates in several ERNs to organise joint reflection sessions targeted at improving the overall collaboration between ERN clinicians and ePAG advocates. Finally, EURORDIS, in collaboration with the members of the ePAG Steering Committee, developed a preliminary set of measures to assess the collaboration between clinicians and patients in the ERNs and the impact of the ePAG advocates engagement in the ERNs.

In terms of governance, EURORDIS and the members of the Steering Committee updated the ePAG Terms of Reference Template and the ePAG Constitution – the set of principles that govern patient engagement in the ERNs. EURORDIS also worked with CRANIO and ERN EYE ePAG advocates and ERN project managers to update the ePAG Terms of Reference and develop additional templates and a standard application form for ePAG advocates, to improve the governance structure around patient involvement in the ERNs.



EURORDIS supported the ongoing recruitment of new ePAG advocates and facilitated their induction organising 3 induction calls in 2021 to provide them with an overview of the ERNs system and the role of ePAG advocates.

Another priority area in 2021 was the exchange of good practices on topics related to the integration of ERNs into national health systems. EURORDIS held a workshop to discuss the relevant actions with National Alliances that was followed up by 2 webinars with sessions focussing on accreditation of centres of expertise; referral pathways, national networks of rare disease centres of expertise and care pathways. EURORDIS is preparing an integration toolkit on integration to summarise the best practices, tools and processes that were presented in these webinars as well as others identified through desk research.

In 2021 EURORDIS has continued to support ePAG advocates to develop Patient Journeys as a tool to collect the needs and expectations of the patient community regarding their care. These journeys allow them to share relevant information with the ERN clinicians and engage them in discussing the needs for refining or developing new standards of care. In 2021 we supported the development of diseases patient journeys for 25 different rare diseases.

In terms of annual meetings targeted at ePAG advocates, EURORDIS organised 2 gatherings in 2021 (i) ePAG online Steering Committee annual meeting (ii) an all-ePAG online meeting.

✦ The ePAG Steering Committee meeting took place on 9 June 2021. The AMEQUIS Consortium leads and EC officials were invited to attend this meeting to have an exchange of views on assessment, monitoring and evaluation of ERNs. The ePAG Steering Committee members also discussed other relevant topics, such as the new ePAG Terms of Reference template, the conceptual framework for patient engagement in the ERNs and the material compensation for ePAG advocates.

✦ The all-ePAG annual online meeting took place on 4-5 November 2021. The meeting was organised around the theme of making shared patient-clinician partnership, more like "business as usual" in the ERNs. It was the occasion for ePAG advocates and ERN clinicians and project managers to exchange experience on patient-clinician partnerships, learn about inspiring examples of collaboration and explore the development of common practices, behaviours and processes to champion patient-clinician shared leadership partnership.

## 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. The SPAG is composed of 8 volunteers who provide EURORDIS with their expertise to support EURORDIS' work in advocating for holistic and integrated care for people living with a rare disease and their families.

Across 2021, the Action Group continued to be active in advocating for holistic care for people living with rare diseases. In January 2021, EURORDIS-Rare Diseases Europe and the NoRo Resource Centre for Rare Diseases, with the support of the SPAG, contributed to the European Commission's consultation on the EU Employment and Social Innovation (EaSI) Programme, providing inputs on:

- ✦ The important measures to support the design, implementation and sustainability of EaSI funded projects;
- ✦ Unmet long term care needs of people living with a rare disease and how the pandemic has aggravated these needs;
- ✦ Summary of the follow-up actions of the INNOVCare project, which has been co-funded under the EaSI programme.

These contributions will shape the future calls of the EaSI programme and EURORDIS and NoRo have been invited to a number of follow up meetings to define the priorities of the programme and the upcoming calls for projects.

The SPAG worked with EURORDIS across the year to identify policy areas under the European Pillar for Social Rights and its Action Plan, as well as under the EU Disability Strategy 2021-2023, where it focused its advocacy work in the coming months. This also helped identify key actions and flagships of the upcoming European Action Plan for Rare diseases that EURORDIS is campaigning for, which should encompass holistic care and inclusion of people with rare diseases in society. During the last two meetings of the Group, its members demanded a renewal of the mandate of the Group in 2022 to enable it to accomplish its mission and the envisaged actions.



## 3.3 PATIENT ENGAGEMENT IN DIAGNOSIS

### 3.3.1 EURORDIS Newborn Screening Working Group

***Newborn screening is the process of systematically testing newborns just after birth for certain diseases. Ideally, this practice is the part of the 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 helps to 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 have been published in January 2021 and have been translated into 12 languages.***

### 3.3.2 SCREEN4CARE

The new collaborative Innovative Medicines Initiative (IMI 2 JU) – European project **Screen4Care** – was officially launched on 1 October 2021 with a kick off meeting (KOM) which took place online over 2 days on 26–27 October 2021 with around 100 participants including project partners as well as confirmed members of the Scientific Advisory Board and the Patient Advisory Board (PAB). Several key meetings for EURORDIS-led tasks have taken place as well, along with others where EURORDIS is a partner. Screen4Care will run for a period of five years with a total budget of 25 million euros, provided by the IMI 2 JU, a joint undertaking of the European Union and the European Federation of Pharmaceutical Industries and Associations.

Screen4Care offers an innovative approach to accelerate rare disease diagnosis, which is based on two central pillars: genetic newborn screening and digital technologies. EURORDIS plays a major role, located at the heart of Screen4Care and involved in all **areas of focus**. EURORDIS leads the Patient Advisory Board and is directing and advising all partners on the priorities, needs and perspectives of persons living with rare diseases. A Senior Project Manager was hired in September 2021 to manage this major project. Work on essential tasks has begun and planning for the accomplishment of central goals of the project has been laid out. The outputs of the Screen4Care project support EURORDIS's advocacy work on Newborn Screening (see chapter 1 on patient advocacy).



## 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 with involvement of several staff members.

In 2021, EURORDIS remained a member of the Consortium Assembly, Patient Advocacy Constituent Committee and Therapies Scientific Committee (member + former vice-chair) and continued involvement in the Task Forces (TF) and working groups (WG). Throughout the year, EURORDIS continued the dissemination of the **Orphan Drug Development Guidebook** co-created within IRDiRC's Therapies Scientific Committee with a multi-stakeholder group of experts. A sustainability plan has now been established in the context of an MoU signed between EURORDIS, EATRIS, Fondazione Telethon, CVBF and INSERM as EJP-RD lead partner); participated in the consortium assembly meetings and actively promoted opportunities for patient representatives to get involved in TF and WG of the 2021 road map for IRDiRC activities, to which EURORDIS significantly contributed. 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.

Current Task Forces and Working Groups where EURORDIS was involved (staff and/or volunteers) in 2021: Sustainable Economic Models in Repurposing (lead), IRDiRC-RDI Global Access WG (following on the work of the previous IRDiRC TF on Access to Treatment to which EURORDIS contributed) and TF on integration of new technologies for diagnostics. EURORDIS work in IRDiRC in 2021 led to peer-reviewed publications published in 2021 and January 2022.

### 3.4.2 European Joint Programme on Rare Diseases (EJP on RD) and the Rare Diseases Partnership preparation



*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 RD focuses 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 implementing 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 lead to the long-expected mind-set changes on patient-centred research.

EJP RD has two major objectives:

- ✦ To improve the integration, the efficacy, the production and the social impact of research on RD through the development, demonstration and promotion of Europe/world-wide sharing of research and clinical data, materials, processes, knowledge and know-how.
- ✦ 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 EJP RD proposal and as such is an active member of the Operating Group, which includes the leaders of the different pillars. EURORDIS co-leads Pillar 3 (capacity building and empowerment training courses for all relevant stakeholders including RD patient representatives). That Pillar gathers 45 partners across 5 work packages. EURORDIS has coordinated efforts to pull together a coherent programme of training courses and support activities that are delivered mostly on a pluriannual basis. EURORDIS is also involved in the transversal activities of Pillar 0 as well as Pillar 1 and Pillar 4.

Within Pillar 3, in 2021, EURORDIS provided training to 23 patient advocates (from 15 countries) on scientific innovation and translational research aspects on rare diseases through its EURORDIS Winter school that was held from 12 to 16 April 2021. The Expert Patients and Researchers EURORDIS Summer School took place from 21–25 June 2021. 33 trainees participated, including 26 patient advocates and 7 researchers from 19 countries. EURORDIS also oversaw all training activities from Pillar 3 partners.

In 2021, EURORDIS also participated in all project meetings, including the Policy Board meeting in early 2021 aimed to provide a comprehensive state of the art of existing or forthcoming initiatives and anticipate the

new Partnership on Rare Diseases. The RD Partnership is a co-funded partnership that will pull European Commission and EU Member States and Associated countries funding to optimise investments and research efforts in RDs by aligning them with coordinated strategies and programmes. The main goal of the RD partnership is to improve the life of patients living with a rare disease by developing diagnostics and treatments through multidisciplinary research and innovation programmes. The RD partnership will coordinate national, local and European research and innovation programmes. It will combine research funding and implementation of research supportive activities such as training, data access infrastructures, data standards, etc. In Spring 2021, the Commission services asked potential partners to further elaborate proposals for the candidate European Partnerships (49) planned to be launched in 2023 and 2024. The RD Partnership itself is planned to be launched in 2024. A Concept Paper has been developed by partners based on guidance and templates received from the EC. This has been done considering the initial concepts developed by the Commission and also thanks to the feedbacks received from Member States during early consultation. Together with 70 partners, EURORDIS contributed to the drafting of the concept paper of the EU Rare Diseases Partnership under Horizon Europe. The concept paper submitted on 21 December 2021 to the EC was published on the EC website in February 2022.

### 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. EURORDIS was particularly involved in the organisation of the c4c Multi-Stakeholder Meetings on Inflammatory Bowel Disease and Atopic Dermatitis by bringing relevant patient experts in the discussion. EURORDIS also led a workshop for patient organisations on pediatric drug development which is now available on the c4c website.





### 3.4.4 Collaborative Network for European Clinical Trials for Children (c4c)

The aim of the ERICA consortium is to build on the strength of the individual ERNs and create a platform that integrates all ERN's research and innovation capacity. The project started in March 2021. Through knowledge sharing, engagement with stakeholders in the rare disease domain and assembly of transdisciplinary research groups working across the global health spectrum, ERICA strives to reach the following goals:

- ✦ new intra- and inter-ERN rare disease competitive networks
- ✦ effective data collection strategies
- ✦ better patient involvement
- ✦ enhanced quality and impact of clinical trials
- ✦ increased awareness of ERN's innovation potential

Through the integration of ERN research activities, and outreach to European research infrastructures to synergistically increase impact and innovation, ERICA will strengthen the research and innovation capacity of the ERNs.

EURORDIS is involved in the following work packages:

- ✦ WP 2 on Data Collection, Integration and Sharing. Task 2.1 Coordination and Support of ERN registry activities (support). EURORDIS regularly engages and shares information with the members of ePAG Research and Registries Working Group to capture their feedback on specific WP2 deliverables.
- ✦ WP 3 on Patient Centred Research. Task 3.2 - Sharing information and engaging with ePAG advocates and our members to capture the patients' perspective on the disease-specific and common needs, so as to have a 360° vision of clinicians' and patients' views for PCOMs/PROMs development and implementation (support).
- ✦ WP 4 on Clinical Trial Support. Task 4.3. Drafting a framework for Patient Engagement in clinical trials (lead).

### 3.4.5 HTX - Next Generation HTA

HTx – Next Generation HTA is a Horizon 2020 funded project that aims at exploring new methods for complex decisions about pricing and reimbursement of treatments.

Which patients would benefit the most from a given high-cost new therapy? How to identify the best sequence or combination of treatments? What is the best therapeutic strategy for a given small populations or subgroup?

One of the goals of HTx is to explore to which extent real-world evidence can be used to solve such questions. Among the new methods to be applied, AI and machine learning play a special role in trying to make cost-effectiveness predictions.

This will be tested on 4 case studies, about 4 disease areas: Head and neck cancer, Diabetes 1 and 2, Multiple Sclerosis, Myelodysplastic Syndrome.

EURORDIS is part of the HTx consortium and is committed to inform the patient community about the progress and the potential of the project and to train the future patient advocates about HTx core concept and expected findings.

As EURORDIS covers the rare diseases scope of the HTx case studies (head and neck cancer, and myelodysplastic syndrome), we associated the European Multiple Sclerosis Platform to our communication and training effort to reach multiple sclerosis patient community.

On the training side, every year we include an HTx-dedicated sessions in EURORDIS Summer school, and in EMSP General Assembly from this year on. On the communication side, EURORDIS produced an introductory video for the public and interviewed several HTx experts to produce video materials to be disseminated. A toolbox for patients will be the final deliverable.



## 3.5 PATIENT ENGAGEMENT IN LIFECYCLE DEVELOPMENT

### 3.5.1 Scientific Committees and Working parties of the EMA (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 Medicinal 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

- ✦ supporting the **work of patient representatives** on the COMP, PDCO, CAT & PRAC (when applicable)
- ✦ supporting **participation of regular/ad hoc experts** when relevant
- ✦ promoting **ad hoc participation of patient experts** on the COMP particularly in the discussions on reassessment of the orphan status at the time of Marketing Authorisation
- ✦ identifying & selecting **patient representatives** to be appointed to EMA Scientific Committees. In 2021, EURORDIS informed its Members about the EC Call for Expression of Interest for CAT and PRAC. EURORDIS identified and endorsed one patient representative to be appointed to the CAT. In addition, EURORDIS put forward an application to be appointed to the EMA Management Board.

Patient 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), to the **Strategic and Learning Review meetings** held under the EU Presidency and to **workshops** organised by the Committees when relevant.

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

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

EURORDIS works closely with EMA, Public Engagement Team and SAWP Secretariat to analyse **Protocol Assistance dossiers**, identify and suggest patients from the EURORDIS network or beyond, in particular patients that have been trained to the R&D processes (e.g. EMA training day, EURORDIS Summer School, EUPATI).

After reviewing the data from 2021, it was estimated that the time spent per dossier ranges from 3 to 9 hours, depending on whether the patient representative was "experienced" or "new". As expected, with totally new patients, the Patient Engagement Manager is spending more time either on the phone or via emails to guide them through the process. In total, this represents 60 hours per month.

Last year 35 patients were identified. Finally, 34 were involved. The EMA has now 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 2021, 4 mentors were involved giving more representativeness and broader views. Only 1 drop-out happened. Nonetheless, we are still facing 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.

Procedures with EMA Public Engagement Team have been strengthened and optimised by holding a monthly conference call. Discrepancies between scientific officers' input on the need for patient input and EURORDIS views are discussed on a case-by-case basis, always reaching an agreed solution. Further registration of patients as experts has been improved by including the Patient Engagement Manager in all communications from EMA to patients and within the scientific advice team.

#### 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 Houÿez (member), and Russell Wheeler (volunteer, Leber Hereditary Optic Neuropath UK Society) were appointed as alternates by EURORDIS Board in September 2019.

*In 2021, all meetings were virtual. The EMA training for patient advocates took place online. The total number of days at PCWP over the year: 23.*



## Topics addressed

EURORDIS worked in particular on:

- ★ User testing of the Clinical Trial Web Portal for the public with two EURORDIS volunteers involved
- ★ Review of complex medicine names
- ★ International Conference for Harmonisation Guidelines on Clinical Development and Evaluation of Medicines – revision E6 and E8

- ★ COVID-19 treatments and vaccines
- ★ Shortages of Medicines: definition of critical medicines
- ★ Manufacturing sites and risk of shortages
- ★ EMA mandate expansion

## 3.5.2 Additional activities at the EMA (European Medicines Agency)

### 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 2021 EURORDIS staff and volunteers reviewed hundreds of documents for public information.

Documents that are destined for the general public include:

- ★ Public Summary of Opinion (PSO) of orphan drug designations explains 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

### CHMP oral explanations - Involvement of patients in the benefit/risk evaluation

Since September 2014, the CHMP has decided to invite patients to an oral explanation with the marketing authorisation applicant in some situations where their participation can add to the scientific discussions, first as a pilot, and since 2017 as a more systematic policy. These consultations are now detailed in the EMA Annual Report on Stakeholder Involvement.

Number of patients' representatives identified for CHMP oral explanations with the marketing authorisation applicant in 2020: 1

- ★ CHMP oral explanation with applicant: 1
- ★ Number of days as EURORDIS mentor: 1
- ★ Product to treat Progeria

### Scientific Committees' Questionnaires for patients

In 2019 the EMA initiated a new procedure to collect the opinion 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 retinitis pigmentosa (PDCO)) and 269 responses were obtained.

In 2021 EURORDIS was not contacted to take part in such surveys.

### NEW EMA pilot

Since 1 January 2021, the EMA started a new pilot procedure from the beginning of the evaluation of the marketing **authorisation application submission**.

For each product, EURORDIS is contacted to answer some important questions the CHMP rapporteurs have before they start analysing the benefit/risks: impact of the disease in the life of patients, treated or not, their unmet needs, the relative effects of their treatments, any aspect on the disease they do not understand well, groups of patients with different manifestation of the disease, or different responses to treatments, expectations from a new treatment, treatment constraints that are acceptable to patients, experiences from patients who took part in clinical trials.

To achieve this, EURORDIS explores websites of relevant patient organisations that might have published about the abovementioned questions, contacts its own members, and conducts interviews with 2 to 6 patients in average. In 2021:

Number of orphan products with a marketing authorisation submission: **18**

Number of patients contacted: **175** from 23 countries

Number of interviews: **41** from 11 countries

Number of reports sent: **14** (in 4 cases, no patients could be identified)

Average time given by EMA: **27** days

Number of reports sent on time: **6/14**

Number of reports sent with delay: **8/14** (average delay: 2.4 days [IC95% 1-4])

Number of workdays on pilot: **13.5** days

### 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 2021 patients were identified for the following SAG meetings:

- ✦ February: SAG for Neurofibromatosis type 1 (2 patients identified)
- ✦ 4 October: SAG for Familial Adenomatous Polyposis (1 patient identified)
- ✦ 26 October: SAG for Pompe Disease (3 patients identified)

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

*The EURORDIS Task Force on HTA (Health Technology Assessment) is a group of 10 EURORDIS volunteers from members organisations, 5 men and 5 women, from 8 EU countries, experienced and trained in HTA. 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.*

The Task Force members discuss the assessment, pricing, and reimbursement of treatments across Europe and advise EURORDIS on all aspects of HTA: from methodology to the engagement of patients. They help EURORDIS in its effort to contribute to the cooperation on HTA at the European level, mainly by sharing their knowledge of national HTA systems, discussing experts' opinions, and engaging with the scientific community.

Under the supervision of EURORDIS staff, the Task Force members meet regularly (**online only, from 2020**), raise

awareness among the patient community, participate to policy debates and to public consultations (from the European Commission, EUnetHTA, or learned societies).

In 2021, the HTA Task Force replied to the Responses to European Commission public consultations of EUnetHTA WP1, **White Paper of the Future-Model of Cooperation on HTA** (March –April 2021).

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

*The DITA Task Force represents a group of 16 volunteers who are trained (via the EURORDIS Summer School) and active in issues concerning therapeutic development and evaluation of medicines for rare diseases.*

*The Task Force follows the work plan of the Patients' and Consumers' Working Party at the EMA. It also supports and/or advises the EURORDIS representatives who participate in EMA Scientific Committees and Working Parties. It is consulted on papers prepared by EURORDIS.*

The General Terms of Reference were approved by the Board of Officers on April 20, 2009.

The Task Force participates in conference calls when necessary (if certain issues need to be discussed) and face-to-face meetings.

Due to COVID-19, the task force reorganised its work in 2020 and 2021 with intensive discussions on measures taken to protect people living with rare diseases, and possible consequences. A series of 11 online meetings replaced the two usual face-to-face meetings, where various subjects were discussed (e.g. International Conference for Harmonisation Guidelines on Clinical Development and Evaluation of Medicines – revision E6 and E8; COVID-19 Vaccination campaign report; Update on Covid-19 treatments / EMA; Structured Dialogue on Shortages of Medicines: definition of critical medicines; EURORDIS webinar on manufacturing sites and risk of shortages; Presentation of the EMA big data steering group; Reflections on the Pharmaceutical Committee – revision of the Orphan and Paediatric regulation; Open Public Consultation on the revision of EU rules on medicines for children and rare diseases; Revision of the EU directive on blood, tissues and cells; European Commission Dialogue on Shortages of Medicines).

Members of the task force contributed to the following consultations:

- ✦ Early dialogue with CHMP at submission phase
- ✦ Complex medicines names / Quality Review of Documents
- ✦ Discussions between advocates for Amyotrophic Lateral Sclerosis and the EMA





# 4. CROSS-CUTTING PRIORITIES

## 4.1 GOVERNANCE

### 4.1.1 Annual General Assembly

The EURORDIS Annual General Assembly was held on 10 June 2021. Due to the ongoing pandemic the meeting and elections were held online for the second time in a row. EURORDIS' full members voted on the Activity and Financial reports 2020, Action Plan and Budget 2021 and on the new EURORDIS Strategy 2021-2030. Members also voted on the five vacant positions for the Board of Directors. The following candidates were successfully re-elected: Avril Daly, Retina International (Ireland); Elizabeth Vroom, World Duchenne Organisation (the Netherlands); Simona Bellagambi, UNIAMO Rare Diseases Italy (Italy); Alexandre Mejat, AFM-Téléthon (France). The Board also extended a warm welcome to new member Anna

Arellanosova, Rare Diseases Czech Republic (Czech Republic). All Board members were elected for a full mandate of three years.

The Board of Officers (BoO), is elected annually by the Board of Directors following the General Assembly. This year, the Board of Directors decided to expand the Board of Officers from 5 members to 6, adding an additional officer. The BoO was thus elected as follows: President - Terkel Andersen, Denmark; Vice President - Avril Daly, Ireland; General Secretary - Geske Wehr, Germany; Treasurer - Alain Cornet, Belgium; Officer - Dorica Dan, Romania; Officer: Maria Montefusco, Sweden.

### 4.1.2 EURORDIS Strategy 2021-2030

In 2020 EURORDIS commissioned an external strategic review by Philanthropy Advisors for the purpose of developing its strategy in 2021-2030. The strategic review collected input from EURORDIS members and stakeholders and incorporated the work done within the Rare2030 foresight project that developed recommendations for 2030 & beyond and surveys to

patients, as well as to patient organisations across diseases and across Europe to collect their policy preferences. The resulting findings and proposed Strategy 2021-2030 were presented and discussed at the General Assembly 2021 and given final validation by the EURORDIS Board of Directors. The final document can be found in the opening pages of this report.



### 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 Organisation for Rare Disorders



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



#### JPA – The Japan Patients' Association



#### RDI - Rare Diseases International



#### RADOIR - Rare Diseases Foundation of Iran



#### RVA – Rare Voices Australia



EURORDIS also has partnerships with a number of learned societies:

#### European Federation of Internal Medicine (EFIM)



#### European Hospital & Healthcare Federation (HOPE)



#### International Federation of Social Workers Europe (IFSW-Europe)



#### International Society for Pharmacoeconomics and Outcomes Research (ISPOR)



#### European Society of Human Genetics (ESHG)



#### European Connected Health Alliance (ECHAAlliance)



## 4.2 HUMAN RESOURCES

### 4.2.1 EURORDIS Staff

As at the end of the year, the team was composed of 49 staff members representing 18 languages and 20 nationalities and dispatched in 8 countries.

The number of FTE was 48.0 in 2021 compared to 47.7 in 2020.

The main office is in France (31), followed by Belgium (6), Spain (6), Serbia (1), Germany (1), the United Kingdom of Great Britain and Northern Ireland (1), Romania (1) and Russia (1).

4 new positions were created in 2021 (in order of appearance):

- ✦ Rare Barometer Communications Junior Manager, Andrea Osvoll, Paris (limited contract)
- ✦ Compliance and Budget Manager, Imane Elgoumri, Paris (permanent contract)

- ✦ Health & Social Sciences Project Senior Manager, Edith Gross, Paris (permanent contract)

- ✦ Events Junior Manager, Anne L'Hévéder, Paris (permanent contract)

4 positions were stopped:

- ✦ Public Affairs Manager - EU & National Integrated Advocacy, Kostas Aliogiannis, Brussels (permanent contract)
- ✦ Senior Manager Public Affairs, Lead Rare Impact Consortium on Access to Gene & Cell Therapies, Karolina Hanslik, Brussels (limited contract)
- ✦ Director Social Policy, Raquel Castro, Barcelona (permanent contract)
- ✦ Senior Manager Patient Engagement in Therapy Development, Lead Community Advisory Boards (CAB), Rob Camp, Barcelona (permanent contract)

### 4.2.2 EURORDIS Volunteers

As stated in its mission, EURORDIS is the voice of rare disease patients at the EU level and is represented in EU institutions and in European and international conferences.

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

Each group of volunteers is coordinated by at least one EURORDIS staff member.

#### Volunteer RareConnect moderators:

RareConnect, created within EURORDIS, responds to rare disease patients' needs for information and connection by creating international online communities and discussion groups for specific diseases.

There are 380 volunteer RareConnect moderators, concerned/affected by a specific rare disease, moderating online communities of patients, parents and carers across countries for their disease or group of diseases, with respect to people's privacy in compliance with the EU rules on data protection.

#### Volunteer patient advocates:

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

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

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

Most of the EURORDIS volunteer patient advocates belong to different internal working groups and Task Forces. Some of them can belong to two task forces, and sometimes (but rarely) three.

#### EPAC: European Public Affairs Committee

This internal committee plays an active and key role in EURORDIS' advocacy activities. The EPAC members discuss all relevant advocacy issues for people living with rare diseases and their families. They can also provide their comments on EURORDIS' positions on some specific issues. The EPAC is governed by Rules of Procedure. As of end 2021, it was composed of 43 full members: **24 volunteers** as well as 19 staff members (CEO, Directors and managers) involved in advocacy. The EPAC members have a mandate to represent EURORDIS.

#### TAG: Therapeutic Action Group

The TAG includes **volunteers** who represent patients on the European Medicines Agency's scientific committees. These committees hold meetings every month over two to three days. The required expertise and involvement in terms of time are both significant.

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

In 2021, **16 volunteers** contributed to the DITA's work.

Selected volunteers are trained (via the EURORDIS Open Academy) and are 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).

### HTA (Health Technology Assessment) Task Force

The HTA is composed of **10 volunteers** and coordinated by two staff members, who also manage the DITA Task Force in order to ensure a good synergy between these two task forces. The HTA Task Force advises EURORDIS on all aspects regarding Health Technology Assessment policies and procedures. Its role is to inform EURORDIS on how health technologies are assessed at the national level, how patients are involved in these assessments and share views on the future European Cooperation on HTA.

### DAG: Digital and Data Advisory Group

The DAG includes 10 volunteers. The Advisory Group advises EURORDIS on all aspects regarding digital policies and procedures.

### SPAG: Social Policy Action Group

The SPAG is composed of 8 volunteers who provide EURORDIS with their expertise to support EURORDIS' work in advocating for holistic and integrated care for people living with a rare disease and their families.

### 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 2021, **28 members of this ePAGs Steering Committee have expressed their interest to become an EURORDIS volunteer.** 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 PRIVATE RESOURCE DEVELOPMENT

2021 saw steady growth in revenue across resource development channels, and successes in initiating and strengthening relationships with diversified supporters. This was particularly significant, considering the operational challenges posed by the ongoing COVID-19 pandemic and a prolonged key fundraising staff vacancy for much of the year.

- ✦ On top of our two annual EURORDIS Round Table of Companies meetings, additional webinars were held for ERTC companies throughout the year. In total, 77 different health sector corporations supported EURORDIS in 2021.
- ✦ After a slowdown in diversified fundraising in 2020 (partly due to the discontinuation of the position on diversification), we were encouraged to see an

uptick in support from individual donors and foundations in 2021 (Individual donor support up 707% over 2020; foundation support up 68% over 2020).

- ✦ There was a notable increase in engagement with foundations in 2021. In addition to receiving ongoing support from a major foundation partner, we were successful in receiving funding from a newly-engaged foundation, and we initiated conversations with several other foundations interested in the rare disease ecosystem.
- ✦ From November 2021, Brian Howard rejoined the EURORDIS staff as Head of Philanthropy and Partnerships.

## 4.4 FINANCE & SUPPORT SERVICES

Finance and support services' activities in 2021 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.
- ✦ 78 EURORDIS procedures were updated (accounting, HR, administrative and IT procedures). A Compliance and Budget Manager was recruited in order to update the procedures and create new ones in the next 2 years.



## 4.4.1 Contract Grants

### Renewed

Specific Grant Agreement (Operating Grant) for year 2021 (SGA 2021), single beneficiary, DG Sante, 12 months

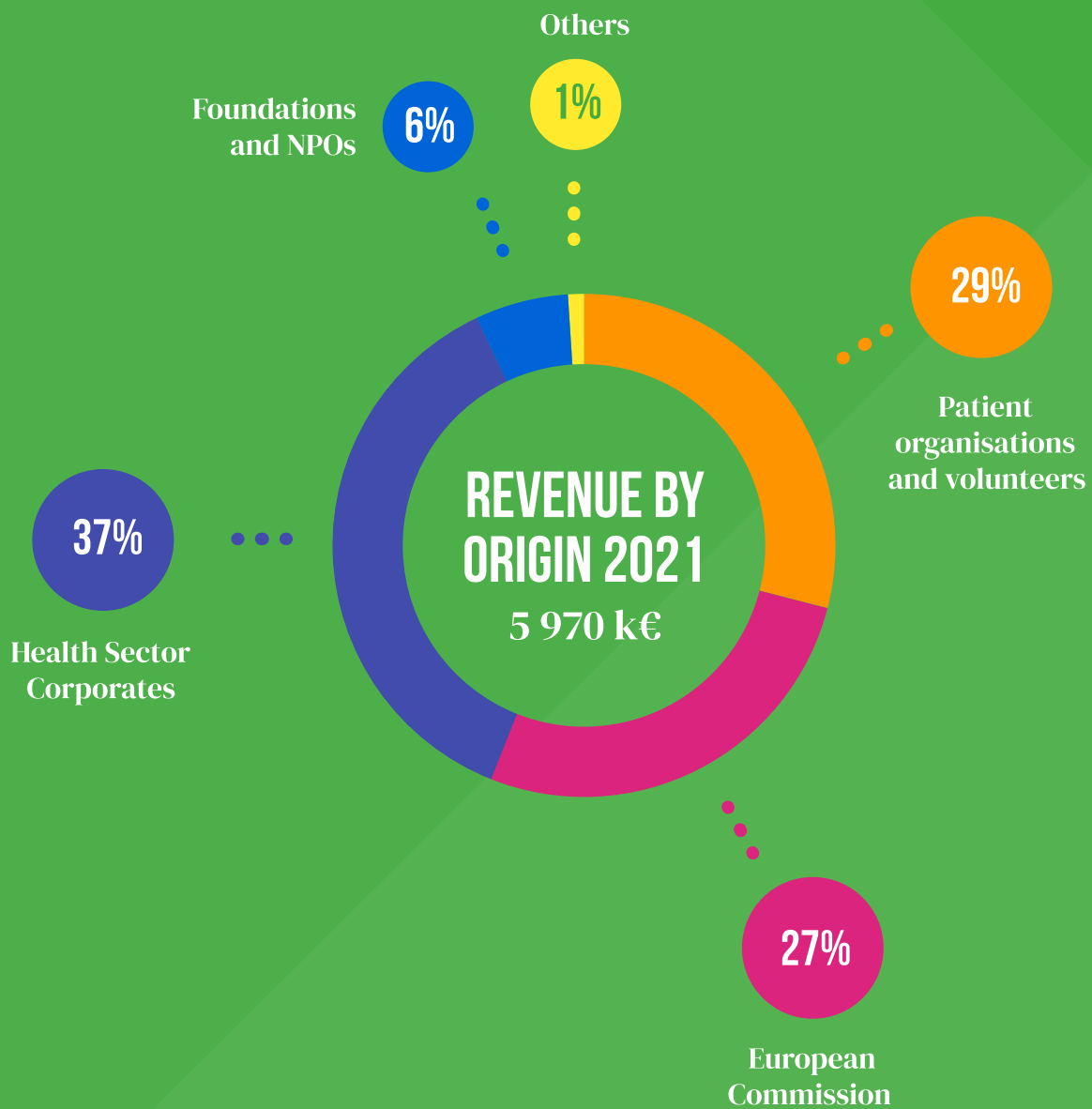
### Ongoing and new

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

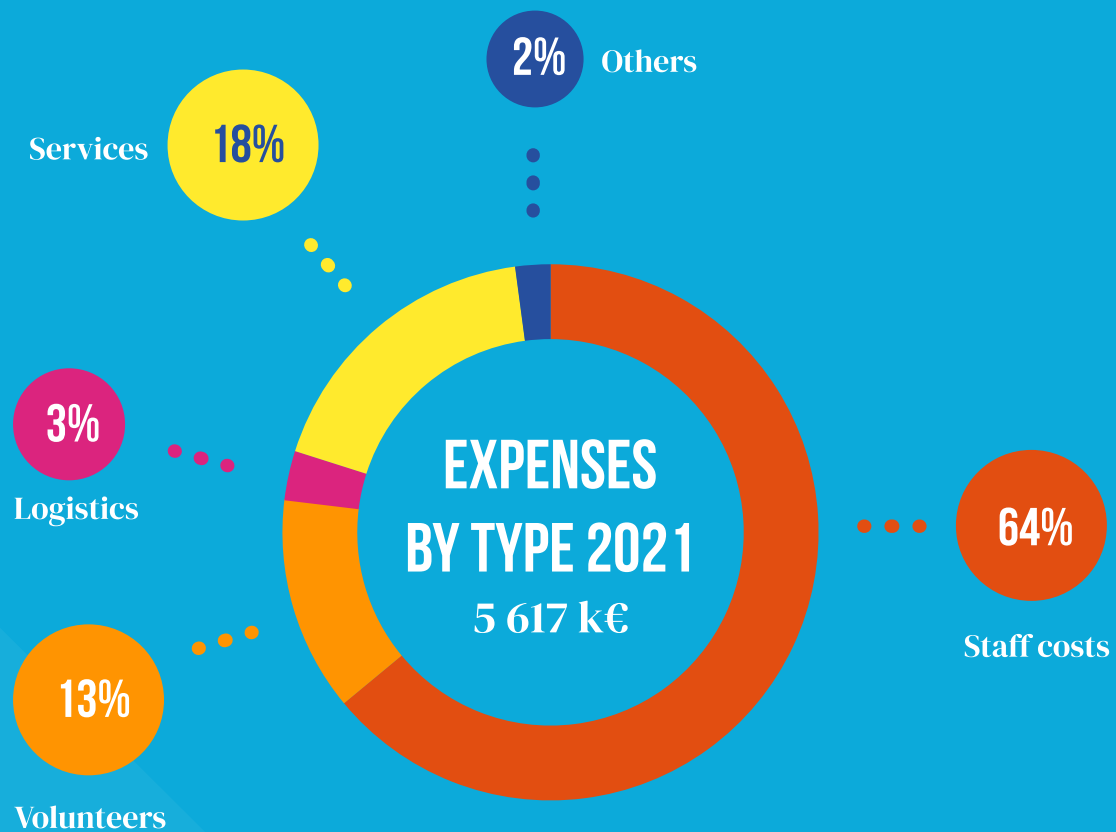
Framework Partnership Agreement 2018-2021 (Operating Grant), single beneficiary, DG Sante, 2018-2021

<u>ERICA</u> <u>Grant Agreement n° 964908 – ERICA</u> <u>Horizon 2020</u>	01/03/2021 – 28/02/2025	Partner, beneficiary	EURORDIS: 143,675€ Project: 2,313,808.75€
<u>European Joint Programme on Rare Diseases</u> <u>Grant Agreement n° 825575 – EJP RD</u> <u>Horizon 2020</u>	01/01/2019 – 31/12/2023	Partner, beneficiary	EURORDIS: 930,665€ Project: 100,362,308.32€
<u>Collaborative Network for European Clinical</u> <u>Trials For Children Grant Agreement n°</u> <u>777389 – conect4children</u> <u>IMI2</u>	01/05/2018 – 30/04/2024	Partner, beneficiary	EURORDIS 578,750€ Project: 67,000,000€
<u>Solve RD</u> <u>Grant Agreement n° 779257 – Solve-RD</u> <u>Horizon 2020</u>	01/01/2018 – 01/12/2022	Partner, beneficiary	EURORDIS: 375,000€ Project: 15,361,621€
<u>‘Next Generation Health Technology</u> <u>Assessment to support patient-centred,</u> <u>societally oriented, real time decision making</u> <u>on access and reimbursement for health</u> <u>technologies throughout Europe ’ – ‘HTx’</u> <u>Grant Agreement n° 825162 – HTX</u> <u>Horizon 2020</u>	01/01/2019 – 31/12/2023	Partner, beneficiary	EURORDIS: 625,097.50€ Project: 9,640,775€
<u>Rare 2030</u> <u>Grant Agreement n°PP-1-2-2018– Rare 2030</u> <u>IMI2</u>	01/01/2019 – 31/03/2021	Coordinator, Beneficiary	EURORDIS: 407,484€ Project: 1,300,000€
<u>RD CODE</u>	01/01/2019	Partner, beneficiary	EURORDIS: 29,952€
<u>Grant Agreement n° 826607 – RDCODE</u> <u>CHAFEA</u>	31/12/2021	Partner, beneficiary	Project: 749,885€
<u>SCREEN4CARE</u> <u>Grant Agreement n° 101034427 –</u> <u>SCREEN4CARE</u> <u>IMI2</u>	01/10/2021 – 30/09/2026	Partner, beneficiary	EURORDIS: 808,000€ Project: 11,938,568,75€

# REVENUE 2021



# EXPENSES 2021





# BOARD of Directors

June 2021 – May 2022

## BOARD OF OFFICERS



**Mr Terkel Andersen**  
PRESIDENT  
Danish Haemophilia Society



**Ms Avril Daly**  
VICE-PRESIDENT  
Rare Diseases Ireland



**Ms Geske Wehr**  
GENERAL SECRETARY  
European Network for Ichthyosis e.V



**Mr Alain Cornet**  
TREASURER  
Lupus Belgium



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



**Ms Maria Montefusco**  
OFFICER  
Rare Diseases Sweden

## DIRECTORS



**Mr Alexandre Mejat**  
AFM - Téléthon  
France



**Ms Anna Arellanesova**  
Rare Diseases Czech Republic  
Czechia



**Ms Elizabeth Vroom**  
World Duchenne Organisation  
Netherlands



**Ms Vlasta Zmazek**  
Rare Diseases Croatia  
Croatia



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



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



**Mr Lieven Bauwens\***  
IF for Spina Bifida  
and Hydrocephalus  
Belgium



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

# MEMBERS of EURORDIS

FULL MEMBER  
ASSOCIATE MEMBER

## ALBANIA

SHOQATA E SEMUNDJEVE TE RRALLA / RARE DISEASE ASSOCIATION ALBANIA

## ALGERIA

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

## ANDORRA

ASSOCIACIÓ DE MALALTIES MINORITÀRIES D'ANDORRA

## 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  
PHA AUSTRIA – INITIATIVE LUNGENHOCHDRUCK  
PHA EUROPE  
PRO RARE AUSTRIA, ALLIANZ FÜR SELTENEN ERKRANKUNGEN  
SMITH-MAGENIS-SYNDROM ÖSTERREICH  
USHER DEAFBLIND FORUM AUSTRIA

## BELARUS

BELARUSSIAN ORGANISATION OF PATIENTS WITH MPS AND OTHER RARE GENETIC DISORDERS

## BELGIUM

ASSOCIATION POUR L'INFORMATION ET LA RECHERCHE SUR LES MALADIES RÉNALES GÉNÉTIQUES  
BELGISCHE ORGANISATIE VOOR KINDEREN EN VOLWASSENEN MET EEN STOFWISSELINGSZIEKTE  
BELGISCHE VERENIGING VOOR LONGFIBROSE VZW  
BE-TSC VZW  
CF EUROPE  
CHILDREN'S TUMOR FOUNDATION EUROPE  
CONTACTGROEP MYELOOM EN WALDENSTRÖM PATIËNTEN  
DEBRA BELGIUM  
DYSNET  
EUROPEAN CHROMOSOME 11 NETWORK  
EUROPEAN CMT FEDERATION  
EUROPEAN FEDERATION OF WILLIAMS SYNDROME  
EUROPEAN HAEMOPHILIA CONSORTIUM  
EUROPEAN IDIOPATHIC PULMONARY FIBROSIS & RELATED DISORDERS FEDERATION  
EUROPEAN NETWORK FOR RESEARCH ON ALTERNATING HEMIPLEGIA  
EYE HOPE FOUNDATION  
FAMILIAL ADENOMATOUS POLYPOSIS ASSOCIATION  
FEDERG - FEDERATION OF EUROPEAN ASSOCIATIONS OF PATIENTS AFFECTED BY RENAL DISEASES  
HTAP BELGIQUE ASBL  
ICHTHYOSE BELGIQUE - ICHTHYOSIS BELGIË  
INTERNATIONAL FEDERATION FOR SPINA BIFIDA AND HYDROCEPHALUS  
INTERNATIONAL HUNTINGTON ASSOCIATION  
LIVER PATIENTS INTERNATIONAL  
LUPUS EUROPE  
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 BINDWEFSELAANDOENINGEN VZW.GEN  
ZEBRAPAD VZW

## BENIN

ALBINS SANS FRONTIÈRES

## BOSNIA AND HERZEGOVINA

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

## BRAZIL

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

## BULGARIA

ASSOCIATION OF PEOPLE SUFFERING BY ACROMEGALY IN BULGARIA  
ASSOCIATION OF TARLOV CYST PATIENTS IN 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 PATIENTS WITH GROWTH HORMONE DEFICIENCY  
NATIONAL ASSOCIATION OF SYRINGOMYELIA  
NATIONAL GAUCHER ORGANISATION  
NATIONAL PATIENTS' ORGANISATION  
PHA BULGARIA  
RETINA BULGARIA  
RETINA INTERNATIONAL  
THE BULGARIAN SOCIETY OF PATIENTS WITH PULMONARY HYPERTENSION

## BURKINA FASO

FONDATION INTERNATIONALE TIerno ET MARIAM

## CANADA

CANADIAN ORGANISATION FOR RARE DISORDERS  
PVNH SUPPORT & AWARENESS

## CHINA

CHINESE ORGANISATION 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 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 ASCOCIACE PRO VZACNA ONEMOCNENI)

## DENMARK

22Q11 DANMARK  
ADDISON FORENINGEN I DANMARK  
BLÆREEKSTROFIFORENINGENS  
CCHS DANMARK  
DANISH APERT SYNDROME ASSOCIATION /DANMARKS  
APERTFORENING  
DANMARKS BLODERFORENING / DANISH HAEMOPHILIA SOCIETY  
EHLERS-DANLOS FORENINGEN I DANMARK  
EUROPEAN POLIO UNION  
FORENINGEN FOR ATAKSI / HSP  
HAE SCANDINAVIA  
ICHTYOSIS ASSOCIATION IN DENMARK  
IMMUN DEFEKT FORENINGEN  
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 KVTI  
RINNEKOTI FOUNDATION, NORIO CENTRE  
SMA FINLAND RY  
SUOMEN VASKULITITIVHDISTYS RY / FINNISH VASCULITIS ASSOCIATION

## FRANCE

ACQ2 GENE ASSOCIATION  
AFA CROHN RCH FRANCE  
AFMKT-FRANCE  
AFM-TÉLÉTHON  
AGIR POUR LA MALFORMATION LYMPHATIQUE EN ALLIANCE  
ALLIANCE MALADIES RARES  
ALLIANCE SYNDROME DE DRAVET  
AMIS DE ADNP FRANCE  
AMYLOIDOSIS ALLIANCE THE VOICE OF PATIENTS  
ASL HSP-FRANCE  
ASS. INTERNATIONALE DE DYSTROPHIE NEURO AXONALE INFANTILE  
ASSEDEA  
ASSOCIATION ISIS  
ASSOCIATION "TURNER ET VOUS"  
ASSOCIATION AMS-ARAMISE  
ASSOCIATION ANÉMIES DYSERYTHROPOIÉTIQUES CONGÉNITALES  
ASSOCIATION BERNARD PÉPIN POUR LA MALADIE DE WILSON  
ASSOCIATION BPAN FRANCE  
ASSOCIATION CONTRE LES MALADIES MITOCHONDRIALES  
ASSOCIATION CRANIOPHARYNGIOME SOLIDARITÉ  
ASSOCIATION DES GROUPES AMITIÉ TURNER  
ASSOCIATION DES MALADES DU SYNDROME DE MCCUNE-ALBRIGHT  
ASSOCIATION DES MALADES SOUFFRANT D'ANGIO OEDEME PAR DÉFICIT EN C1 INHIBITEUR  
ASSOCIATION DES PANCRÉATITES CHRONIQUES HÉRÉDITAIRES  
ASSOCIATION DES PATIENTS DE LA MALADIE DE FABRY  
ASSOCIATION DES PERSONNES CONCERNÉES PAR LE TREMBLEMENT ESSENTIEL  
ASSOCIATION DU NAEVUS GÉANT CONGÉNITAL  
ASSOCIATION FRANÇAISE CONTRE L'AMYLOSE  
ASSOCIATION FRANÇAISE DE LA MALADIE DE FANCONI  
ASSOCIATION FRANÇAISE DE L'ATAXIE DE FRIEDREICH  
ASSOCIATION FRANÇAISE DE L'OSTEODYSTROPHIE HERÉDITAIRE D'ALBRIGHT  
ASSOCIATION FRANÇAISE DE L'OSTÉOGENÈSE IMPARFAITE  
ASSOCIATION FRANÇAISE DE NARCOLEPSIE-CATAPLEXIE ET HYPERSOMNIE

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 SYNDROMES D'EHLERS-DANLOS  
ASSOCIATION FRANÇAISE DU GOUGEROT-SJÖGREN  
ASSOCIATION FRANÇAISE DU LUPUS ET AUTRES MALADIES AUTO-IMMUNES  
ASSOCIATION FRANÇAISE DU SYNDROME DE CORNELIA DE LANGE  
ASSOCIATION FRANÇAISE DU SYNDROME DE KLIPPEL-FEIL  
ASSOCIATION FRANÇAISE DU SYNDROME DE LOWE  
ASSOCIATION FRANÇAISE DU SYNDROME DE RETT  
ASSOCIATION FRANÇAISE DU SYNDROME D'ONDINE  
ASSOCIATION FRANÇAISE DU SYNDROME PHELAN-MCDERMID  
ASSOCIATION FRANÇAISE LESCH-NYHAN ACTION  
ASSOCIATION FRANÇAISE NIEMANN PICK  
ASSOCIATION FRANÇAISE POUR LE SYNDROME DE GILLES DE LA TOURETTE  
ASSOCIATION FRANÇAISE STURGE WEBER "VANILLE-FRAISE"  
ASSOCIATION FRANCE VASCULARITES  
ASSOCIATION FRANCOPHONE CONTRE LA POLYCHONDRITE CHRONIQUE ATROPHIANTE  
ASSOCIATION FRANCOPHONE DES GLYCOGÉNOSES  
ASSOCIATION FRANCOPHONE DU SYNDROME D'ANGELMAN  
ASSOCIATION GRANDIR  
ASSOCIATION HEMOCHROMATOSE FRANCE  
ASSOCIATION HISTIOCYTOSE FRANCE  
ASSOCIATION HUNTINGTON FRANCE  
ASSOCIATION HYPOPARATHYROIDISME FRANCE  
ASSOCIATION ICHTYOSE FRANCE  
ASSOCIATION INTERNATIONALE MALADIES KYSTES TARLOV  
ASSOCIATION KOURIR  
ASSOCIATION MALADIES FOIE ENFANTS  
ASSOCIATION MARFANS  
ASSOCIATION MICROPHALMIE FRANCE  
ASSOCIATION NAEVUS 2000  
ASSOCIATION NATIONALE DES CARDIAQUES CONGÉNITAUX  
ASSOCIATION NEUROFIBROMATOSES & RECKLINGHAUSEN  
ASSOCIATION OLLIER MAFFUCCI EUROPE  
ASSOCIATION PEMPHIGUS – PEMPHIGOÏDES FRANCE  
ASSOCIATION POUR AIDER ET INFORMER LES SYRINGOMYÉLIQUES EUROPEENS RÉUNIS  
ASSOCIATION POUR LA LUTTE CONTRE L'ALCAPTONURIE  
ASSOCIATION POUR LA LUTTE CONTRE LES MALADIES INFLAMMATOIRES DU FOIE ET DES VOIES BILIAIRES  
ASSOCIATION POUR LA RECHERCHE SUR LA SCLÉROSE LATÉRALE AMYOTROPHIQUE  
ASSOCIATION POUR LA SENSIBILISATION AUX MALADIES RARES, ORPHELINES ET AUTO-IMMUNES À SAINT-PIERRE-ET-MIQUELON  
ASSOCIATION POUR L'AIDE AUX PERSONNES CONCERNÉES PAR LES MALADIES RARES MUCKLE WELLS SYNDROME ET CINCA  
ASSOCIATION POUR L'INFORMATION ET LA PRÉVENTION DE LA DRÉPANOCYTOSE  
ASSOCIATION POUR L'INFORMATION ET LA RECHERCHE SUR LES MALADIES RÉNALES GÉNÉTIQUES  
ASSOCIATION SANS DIAGNOSTIC ET UNIQUE  
ASSOCIATION SCLÉROSE TUBÉREUSE DE BOURNEVILLE  
ASSOCIATION SED1+  
ASSOCIATION SOS DESMOÏDE  
ASSOCIATION SPINA BIFIDA ET HANDICAPS ASSOCIÉS  
ASSOCIATION SURRÉNALES  
ASSOCIATION SYNDROME DE KLEINE-LEVIN  
ASSOCIATION SYNDROME DE MOEBIUS FRANCE  
ASSOCIATION TANGUY MOYA MOYA  
AVML VIVRE MIEUX LE LYMPHÈDÈME  
CHARCOT-MARIE-TOOTH FRANCE  
CUTIS LAXA INTERNATIONALE  
DEBRA FRANCE  
DIABETE INSIPIDE FRANCE  
DRAVET SYNDROME EUROPEAN FEDERATION  
ENFANTS DE LA LUNE ASSOCIATION POUR LE XERODERMA PIGMENTOSUM  
ENSEMBLE LEUCEMIE LYMPHOMES ESPOIR  
EURO-DYMA  
EUROPEAN FEDERATION FOR HEREDITARY SPASTIC PARAPLEGIA  
EUROPEAN FEDERATION LESCH-NYHAN DISEASE  
EUROPEAN FEDERATION OF ASSOCIATIONS OF PATIENTS WITH HAEMOCHROMATOSIS  
EUROPEAN PATIENT ORGANISATION FOR DYSIMMUNE AND INFLAMMATORY NEUROPATHIES  
EWENLIFE  
FÉDÉRATION SOS GLOBI  
FITIMA EUROPE - FONDATION INTERNATIONAL TIERNO ET MARIAM  
FOP FRANCE  
FRAGILE X FRANCE  
FRANCE LYMPHANGIOLEIOMYOMATOSE



FRUCTOS'AMIS POUR LA VIE  
GÉNÉRATION 22  
GENESPOIR: ASSOCIATION FRANÇAISE DES ALBINISMES  
GENIRIS  
HTAPFRANCE  
HYPOPHOSPHATASIE EUROPE  
INCONTINENTIA PIGMENTI FRANCE  
INFLAM'ŒIL  
LA MAISON 8P  
LES AMIS DE REMI  
LES PETITS BOURDONS  
LIGUE CONTRE LA CARDIOMYOPATHIE  
LIGUE NATIONALE CONTRE LE CANCER  
LUPUS FRANCE  
MED13L SYNDROME ASSOCIATION  
MOSAÏQUES - ASSOCIATION DES "X FRAGILE"  
NAEVUS GLOBAL  
OUVRIR LES YEUX  
PETIT CŒUR DE BEURRE  
POLYPOSES FAMILIALES - APTPEF  
PRADER WILLI FRANCE  
RETINA FRANCE  
SANFILIPPO SUD  
SOLIDARITÉ HANDICAP AUTOUR DES MALADIES RARES  
SYNDROME PACS1 - SCHUURS-HOEJUMAKERS  
TINTAMARRE  
UNION HUNTINGTON ESPOIR  
UNION NATIONALE DES ASSOCIATIONS PARENTS ET AMIS PERSONNES HANDICAPÉES MENTALES  
UNION NATIONALE DES SYNDROMES D'EHRLERS-DANLOS  
VAINCRE LA MUCOVISCIDOSE  
VAINCRE LA PAPILLOMATOSE RESPIRATOIRE RÉCURRENTE  
VAINCRE LES MALADIES LYSOSOMALES  
VALENTIN APAC - ASSOCIATION DE PORTEURS D'ANOMALIES CHROMOSOMIQUES  
VHL FRANCE  
VIVRE SANS THYROÏDE  
WHITE SUTTON FRANCE  
WILLIAMS-FRANCE  
XTRAORDINAIRE

## GEORGIA

GEORGIAN FOUNDATION FOR GENETIC AND RARE DISEASES  
SCN2A GEORGIA

## GERMANY

AHC-DEUTSCHLAND E.V.  
ALLIANZ CHRONISCHER SELTENER ERKRANKUNGEN E.V.  
ANGELMAN E.V.  
ARVC-SELBSTHILFE E.V.  
ASOCIATIA ROMANA SPINA BIFIDA SI HIDROCEFALIE  
BSHV FÜR KINDER, JUGENDLICHE UND ERWACHSENE MIT SELTENEN, CHRONISCHEN SKELETTERKRANKUNGEN E.V.  
BUNDESVERBAND ANGEBORENE GEFÄSFEHLBILDUNGEN E.V.  
BUNDESVERBAND DER CLUSTERKOPFSCHMERZEN-SELBSTHILFE-GRUPPEN  
BUNDESVERBAND FÜR PFFD, FFU, FIBULA- UND TIBIADEFEKTE E.V.  
BUNDESVERBAND KLEINWÜCHSIGE MENSCHEN UND IHRE FAMILIEN E.V.  
BUNDESVERBAND POLIOMYELITIS E.V.  
BUNDESVERBAND SCHILDDRÜSENKREBS - OHNE SCHILDDRÜSE LEBEN E.V.  
CHARGE SYNDROM E.V.  
CHOLESTERIN & CO E.V.  
CRPS BUNDESVERBAND DEUTSCHLAND 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 SARKOIDOSE VEREINIGUNG E.V.  
DEUTSCHE SYRINGOMYELIE UND CHIARI MALFORMATION DSCM E.V.  
DEUTSCHE UVEITIS-ARBEITSGEMEINSCHAFT E.V.  
DEUTSCHSPRACHIGE SELBSTHILFEGRUPPE FÜR ALKAPTONURIE E.V.  
DSAI E.V. - PATIENTENORGANISATION FÜR ANGEBORENE IMMUNDEFEKTE DUP15Q E.V.  
ESOPHAGEAL ATRESIA GLOBAL SUPPORT GROUPS  
EUROPEAN ASSOCIATION OF PATIENT ORGANISATIONS OF SARCOIDOSIS  
EUROPEAN MEN ALLIANCE  
EUROPEAN NETWORK FOR ICHTHYOSIS  
FAUN STIFTUNG  
FOP GERMANY (FÖRDERVEREIN FÜR AN FIBRODYSPLASIA OSSIFICANS PROGRESSIVA ERKRANKTE)  
FSHD EUROPE  
FUNDACJA SANFILIPPO  
GAUCHER GESELLSCHAFT DEUTSCHLAND E.V.  
GESELLSCHAFT FÜR MUKOPOLYSACCHARIDOSEN E.V.  
HAE VEREINIGUNG E.V. (HEREDITARY ANGIOEDEMA)  
HAND IN HAND GEGEN TAY-SACHS UND SANDHOFF E.V.

HOFFNUNGSBAUM E.V. - VEREIN ZUR FÖRDERUNG DER ERFORSCHUNG UND BEHANDLUNG VON NBIA-ERKRANKUNGEN  
HSP-SELBSTHILFEGRUPPE DEUTSCHLAND E.V.  
IEB E.V. DEBRA DEUTSCHLAND  
IMBS ALLIANCE  
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.  
MARFAN HILFE DEUTSCHLAND E.V.  
MORBUS-OSLER SELBSTHILFE E.V.  
MPN-NETZWERK E.V.  
MPS EUROPE  
MULTINATIONAL INTERSTITIAL CYSTITIS ASSOCIATION  
MYELITIS E.V.  
NCL-GRUPPE DEUTSCHLAND E.V.  
NEPHIE E.V. - SELBSTHILFE NEPHROTISCHES SYNDROM  
NETZWERK HYPOPHYSEN- UND NEBENNIERENERKRANKUNGEN E.V. - NETWORK PITUITARY AND ADRENAL DISORDERS  
OIFE - OSTEOGENEIS IMPERFECTA FEDERATION EUROPE  
PATIENTEN- UND SELBSTHILFEORGANISATION FÜR KINDER UND ERWACHSENE MIT KRANKER SPEISERÖHRE MIT KRANKER SPEISERÖHRE  
PATIENTENVERBAND FAMILIÄRE AMYLOID POLYNEUROPATHIE  
PCH-FAMILIE E.V.  
PEUTZ-JEGHERS-GERMANY E.V.  
PKD FAMILIÄRE ZYSTENNIERE E.V.  
PRO RETINA DEUTSCHLAND E.V.  
PULMONALE HYPERTONIE E.V.  
SANFILIPPO INITIATIVE E.V.  
SELBSTHILFE EPP E.V.  
SELBSTHILFE ICHTHYOSE E.V.  
SELBSTHILFEGRUPPE EKTODERMALE DYSPLASIE E.V.  
SELBSTHILFEGRUPPE FÜR PXE-ERKRANKTE DEUTSCHLANDS E.V.  
SELBSTHILFEGRUPPE GLYKOGENOSE DEUTSCHLAND E.V.  
SELBSTHILFEORGANISATION FÜR MENSCHEN MIT ANOREKTALFEHLBILDUNGEN  
SKLERODERMIE SELBSTHILFE E.V.  
SMA EUROPE  
SSADH-DEFIZIT E.V.  
TOM WAHLIG STIFTUNG  
VEREIN AHC18+  
VEREIN VHL (VON HIPPEL - LINDAU) BETROFFENER FAMILIEN E.V.  
XP - FREU(N)DE MONDSCHENKINDER  
ZNM - ZUSAMMEN STARK! E. V.

## GREECE

"95" - RARE ALLIANCE GREECE  
ANGELMAN SYNDROME GREECE  
ASSOCIATION OF GREEK FRIENDS FOR PAEDIATRIC IMMUNOLOGY  
PRIMARY IMMUNODEFICIENCIES «HARMONY»  
CHILD'S HEART  
HELLENIC CYSTIC FIBROSIS ASSOCIATION  
HELLENIC FRIEDREICH'S ATAXIA ASSOCIATION  
HELLENIC LEAGUE AGAINST RHEUMATISM  
HELLENIC MYASTHENIA GRAVIS ASSOCIATION  
IMMUNE DEFICIENCY ASSOCIATION GALINOS  
KARKINAKI AWARENESS FOR CHILDHOOD AND ADOLESCENT CANCER  
KRKOS ZOIS SOCIETY FOR PATIENTS AND FRIENDS OF PATIENTS WITH INHERITED METABOLIC DISEASE  
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  
PESPA (GREEK ALLIANCE FOR RARE DISEASES)  
PRADER WILLI SYNDROME ASSOCIATION HELLAS  
PULMONARY HYPERTENSION OF GREECE - HELLENIC PULMONARY HYPERTENSION  
TO MELLON- ASSOCIATION OF PEOPLE WITH GENETIC DISORDERS  
TOGETHER FOR LIFE  
VHLFA ALLIANCE IN GREECE

## GUATEMALA

ASOCIACIÓN NACIONAL GUATEMALTECA PARA LAS ENFERMEDADES DE DEPÓSITO LISOSOMAL  
PROCRECE

## HONG KONG, SAR OF CHINA

JOSHUA HELLMANN FOUNDATION FOR ORPHAN DISEASE

## HUNGARY

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

## ICELAND

AHC FEDERATION OF EUROPE

ALTERNATING HEMIPLEGIA OF CHILDHOOD ASSOCIATION OF ICELAND  
EINSTÖK BÖRN - SUPPORT GROUP FOR CHILDREN WITH RARE  
DISORDERS

GUÐRUN'S RETT SYNDROME RESEARCH TRUST

## INDIA

INDIAN ORGANISATION FOR RARE DISEASES

ORGANISATION 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

THE ULTRA RARE DISEASES, DISORDERS & DISABILITIES FOUNDATION

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

ACONDROPLASIA - INSIEME PER CRESCERE - ONLUS

AIBWS ODV

AMICI DELLA PORFIRIA - SAN PIO DA PIETRELcina ONLUS

ANGELI NOONAN

ANIRIDIA ITALIANA APS

ASSI GULLIVER - ASSOCIAZIONE SINDROME DI SOTOS ITALIA

ASSOCIAZIONE NAZIONALE PEMFIGO/PEMFIGOIDE

ASSOCIAZIONE CONTO ALLA ROVESCIA

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 LINFOISTIOCITOSI EMOFAGOCITICA

ASSOCIAZIONE ITALIANA MALATI DI ALCAPTONURIA

ASSOCIAZIONE ITALIANA MALFORMAZIONE DI ARNOLD-CHIARI CHILD

ASSOCIAZIONE ITALIANA MIASTENIA E MALATTIE

IMMUNODEGENERATIVE - AMICI DEL BESTA ONLUS

ASSOCIAZIONE ITALIANA MIASTENIA ONLUS

ASSOCIAZIONE ITALIANA MORBO DI HIRSCHSPRUNG

ASSOCIAZIONE ITALIANA MUCOPOLISACCARIDOSI E MALATTIE AFFINI

ASSOCIAZIONE ITALIANA NIEMANN PICK ONLUS

ASSOCIAZIONE ITALIANA PER LA LOTTA ALLE PHTS

ASSOCIAZIONE ITALIANA PER LE MALFORMAZIONI ANORETTALI

ASSOCIAZIONE ITALIANA RENE POLICISTICO ONLUS

ASSOCIAZIONE ITALIANA SINDROME DI EHLERS-DANLOS

ASSOCIAZIONE ITALIANA SINDROME DI POLAND

ASSOCIAZIONE ITALIANA SINDROME E MALATTIA DI BEHÇET

ASSOCIAZIONE ITALIANA SINDROME X FRAGILE

ASSOCIAZIONE ITALIANA SIRINGOMELIA E ARNOLD CHIARI

ASSOCIAZIONE ITALIANA SOSTEGNO MALATTIE METABOLICHE

EREDITARIE ONLUS

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 NAZIONALE PERSONE CON MALATTIE REUMATOLOGICHE

ASSOCIAZIONE PER LE IMMUNODEFICIENZE PRIMITIVE ONLUS

ASSOCIAZIONE PER L'INFORMAZIONE E LO STUDIO DELLA

ACONDROPLASIA

ASSOCIAZIONE PERSONE WILLIAMS ITALIA ONLUS

ASSOCIAZIONE POIC E DINTORNI ONLUS

ASSOCIAZIONE S.P.R.IN.T.

ASSOCIAZIONE SCLEROSI TUBEROSA

ASSOCIAZIONE SINDROME DI ALSTRÖM / ITALIAN ASSOCIATION FOR

ALSTROM SYNDROME

ASSOCIAZIONE SINDROME NEFROSICA ITALIA

ASSOCIAZIONE SPINA BIFIDA ITALIA

ASSOCIAZIONE STUDIO MALATTIE METABOLICHE EREDITARIE ONLUS

ASSOCIAZIONE VENETA PER LA LOTTA ALLA TALASSEMIA

CIDP ITALIA ONLUS

COLLAGENE VI ITALIA ONLUS - COL6

COSTELLO.CFC - ASSOCIAZIONE ITALIANA SINDROME DI COSTELLO -

CARDIOFACIOCUTANEA - RASOPATIE - ONLUS

DEBRA ITALIA ONLUS

DRAVET ITALIA ONLUS

EUROPEAN CONGENITAL HEART DISEASE ORGANISATION

FEDERAZIONE SINDROME DI PRADER WILLI ITALIA

FONDAZIONE ALESSANDRA BISCEGLIA VIVA ALE ONLUS

FONDAZIONE LEGA ITALIANA RICERCA HUNTINGTON

FOP ITALIA ONLUS

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

LA STRADA PER L'ARCOBALENO

LINFA ODV

LND FAMIGLIE ITALIANE ONLUS

MITOCON- INSIEME PER LO STUDIO E LA CURA DELLE MALATTIE

MITOCONDRIALI - ONLUS

P63 EEC SYNDROME INTERNATIONAL NET WORK WORD COMMUNICATION

- MALATTIE RARE ONLUS

PANDAS ITALIA

PARENT PROJECT APS

PKS KIDS ITALIA ONLUS

RETINA ITALIA ONLUS

RING 14 INTERNATIONAL

SOD ITALIA - ASSOCIAZIONE SEPTO OPTIC DYSPLASIA AND OTHER

NEUROENDOCRINE DISORDERS

UN FILO PER LA VITA ANAD IICB

UNIAMO - RARE DISEASES ITALY

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

ASSOCIATION OF SUPPORT TO PATIENTS WITH ORPHAN DISEASES IN THE

REPUBLIC OF KAZAKHSTAN

PATIENTS WITH CANCER AND RARE DISEASES SUPPORT ASSOCIATION

## KOSOVO

RARE DISEASES KOSOVO (SHOQATA E SEMUNDJE TE RRALLA KOSQVE)

## 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  
ATAxie VERENIGING NEDERLAND  
BIJNIEVERENIGING (DUTCH ADRENAL PATIENT SOCIETY)  
CHILDHOOD CANCER INTERNATIONAL -EUROPE  
CMTC-OVM  
CORNELIA DE LANGE SYNDROME WORLD FEDERATION  
EUROPEAN CLEFT ORGANISATION  
EUROPEAN SARCOIDOSIS FOUNDATION  
EUROPEAN SOCIETY FOR PHENYLKETONURIA  
EUROPEAN VHL (VON HIPPEL-LINDAU) FEDERATION  
EUROPEAN WALDENSTRÖM MACROGLOBULINEMIA NETWORK  
FABRY SUPPORT & INFORMATIE GROEP NEDERLAND  
FIBRODYSPLASIA OSSIFICANS PROGRESSIVA STICHTING NEDERLAND  
INTERNATIONAL MITO PATIENTS  
INTERNATIONAL PAINFUL BLADDER FOUNDATION  
INTERNATIONAL PORPHYRIA PATIENT NETWORK  
INTERSTITIELE CYSTITIS PATIENTENVERENIGING  
ITP PATIENTENVERENIGING  
KAISZ - CHILDREN WITH A AUTOINMUUM OR AUTOINFLAMMATORY DISEASE  
MSS (MARSHALL-SMITH SYNDROME) RESEARCH FOUNDATION  
NATIONAL ASSOCIATION REUMAZORG NEDERLAND  
NATIONALE VERENIGING L.E. PATIENTEN  
NEDERLANDS NETWERK VOOR LYMFODEEM EN LIPODEEM  
NEDERLANDSE HYPOFYSE STICHTING (DUTCH PITUITARY FOUNDATION)  
NEDERLANDSE LEVERPATIENTEN VERENIGING  
NEDERLANDSE PHENYLKETONURIE VERENIGING / DUTCH PKU ASSOCIATION  
NEDERLANDSE VERENIGING VAN HEMOFILIE-PATIENTEN/NETHERLANDS HAEMOPHILIA SOCIETY  
NEPHCEUROPE  
NEUROFIBROMATOSE VERENIGING NEDERLAND  
OSCAR NEDERLAND  
PATIENTENORGANISATIE FIBREUZE DYSPLASIE  
PATIENTENVERENIGING VOOR BLAASEXSTROPHIE 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 PIERRE ROBIN EUROPE - PIERRE ROBIN EUROPE FOUNDATION  
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  
VASCULITIS STICHTING  
VERENIGING VAN EHLERS DANLOS PATIENTEN  
VERENIGING VOOR ICHTHYOSIS NETWERKEN  
VOLWASSENEN, KINDEREN EN STOFWISSELINGSZIEKTEN  
VSOP - VERENIGING SAMENWERKENDE OUDER EN PATIENTENORGANISATIES  
WORLD ALLIANCE OF PITUITARY ORGANISATIONS  
WORLD DUCHENNE ORGANISATION

## 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

## NORTH MACEDONIA

ANIRIDIA EUROPE

EUROPEAN HUNTINGTON ASSOCIATION

FRAMBU - RESOURCE CENTRE FOR RARE DISORDERS

HJERNESVULSTFORENINGEN (NORWEGIAN BRAIN TUMOUR ASSOCIATION)

HYPOPARA NORGE

MORBUS ADDISON ASSOCIATION NORWAY

MPS-FORENINGEN I NORGE

NORSK FORENING FOR ARVELIG SPASTISK PARAPARESE / ATAKSI

NORSK FORENING FOR EHLERS-DANLOS SYNDROM

NORSK FORENING FOR OSTEOGENESIS IMPERFECTA/THE NORWEGIAN

OSTEOGENESIS IMPERFECTA ASSOCIATION

NORSK FORENING FOR TUBEROS SKLEROSE

NORWEGIAN FEDERATION OF ORGANISATIONS OF DISABLED PEOPLE

(FUNKSJONSHEMMEDES FELLESORGANISASJON)

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 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 RADZIWIŁLOWA CHILD'S HEART FOUNDATION

## PORTUGAL

ANDLINF|ASSOCIAÇÃO NACIONAL DE DOENTES LINFÁTICOS /  
NATIONAL ASSOCIATION OF SUFFERERS OF LYMPHATIC DISORDERS

ASSOCIAÇÃO NACIONAL DE DISPLASIAS ÓSSEAS

ASSOCIAÇÃO NACIONAL PARA DIVULGAR E ORIENTAR PARA COMBATER E ENFRENTAR A TAY SACHS E OUTRAS GANGLIOSIDOSES

ASSOCIAÇÃO PORTUGUESA CDG E OUTRAS DOENÇAS METABÓLICAS

ASSOCIAÇÃO PORTUGUESA DE CHARCOT-MARIE-TOOTH

ASSOCIAÇÃO PORTUGUESA DE INSUFICIENTES RENAI

ASSOCIAÇÃO PORTUGUESA DE LEUCEMIAS E LINFOMAS

ASSOCIAÇÃO PORTUGUESA DE NEUROMUSCULARES

ASSOCIAÇÃO PORTUGUESA DE OSTEOGÉNESE IMPERFEITA

ASSOCIAÇÃO SANFILIPPO PORTUGAL

EVITA

FEDRA - FEDERAÇÃO PORTUGUESA DE DOENÇAS RARAS

LIGA PORTUGUESA CONTRA AS DOENÇAS REUMÁTICAS

RARISSIMAS - ASSOCIACAO NACIONAL DE DEFICIENCIAS MENTAIS E RARAS

RD-PORTUGAL - UNIÃO DE ASSOCIAÇÕES DAS DOENÇAS RARAS DE PORTUGAL

## REPUBLIC OF MOLDOVA

COPILII PLOII

## 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 DE CANCERE RAR

ASOCIATIA ROMANA PENTRU BOLI NEUROLOGICE PERIFERICE/

ROMANIAN ASSOCIATION OF PERYPHERAL NEUROLOGICAL DISEASES

ASOCIATIA SMACARE

ASOCIATIA WERNIG HOFFMAN AWH

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 WILLI ASSOCIATION



## RUSSIAN FEDERATION

HELP TO CYSTIC FIBROSIS PATIENTS  
INTERREGIONAL PUBLIC ORGANISATION FOR GAUCHER DISEASE  
INTERREGIONAL PUBLIC ORGANISATION OF PATIENTS WITH FABRY DISEASE "THE ROAD TO LIFE"  
INTER-REGIONAL SUPPORT CENTRE FOR PATIENTS WITH ANIRIDIA "IRIS"  
LOOK TO SEE  
MPS RUSSIA  
NATIONAL ASSOCIATION OF ORGANISATION OF PATIENT WITH RARE DISEASES "GENETICS"  
NGO "FRAGILE CHILDREN"  
RUSSIAN ASSOCIATION OF RARE DISEASES  
RUSSIAN PATIENT ASSOCIATION  
RUSSIAN RETT SYNDROME ASSOCIATION  
SPIPORZ UNION OF PATIENTS WITH RARE DISEASES AND RARE DISEASE PATIENTS ORGANISATIONS  
THE ASSOCIATION OF PRIMARY IMMUNODEFICIENCY PATIENTS

## SERBIA

CHILD RARE DISEASE SUPPORT AND RESEARCH ASSOCIATION LIFE  
CHILDHOOD CANCER PARENT ORGANISATION «ZVONCICA»  
CITIZENS ASSOCIATION "BROMOLOGOS"  
DMD SERBIA  
LYMPHOMA PATIENTS' ASSOCIATION  
NATIONAL ORGANISATION 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  
EVROPSKA FUNDACIJA SATB2, USTANOVA ZA SATB2 SINDROM (SATB2 EUROPE)  
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

ACCIÓN Y CURA PARA TAY-SACHS  
AHUCE - ASOCIACIÓN NACIONAL HUESOS DE CRISTAL OI ESPAÑA  
ALIANZA ESPAÑOLA DE FAMILIAS DE VON HIPPEL LINDAU  
ASOCIACIÓ CATALANA DE LAS NEUROFIBROMATOSIS  
ASOCIACION ALBI ESPAÑA  
ASOCIACION ANDALUZA DE FIBROSIS QUISTICA  
ASOCIACIÓN ANDALUZA DE PACIENTES CON SÍNDROME DE TOURETTE Y TRASTORNOS ASOCIADOS  
ASOCIACIÓN ARTRORIPPOSIIS MÚLTIPLE CONGÉNITA  
ASOCIACIÓN CHIARI Y SIRINGOMIELIA DEL PRINCIPADO DE ASTURIAS  
ASOCIACIÓN CIUDADANA DE AFECTADOS DE CISTITIS INTERSTICIAL  
ASOCIACIÓN DE AFECTADOS DE NEUROFIBROMATOSIS  
ASOCIACIÓN DE AFECTADOS POR DISPLASIA ECTODÉRMICA  
ASOCIACIÓN DE AFECTADOS POR HIPERINSULINISMO CONGÉNITO  
ASOCIACIÓN DE ATROFIA DE NERVIÓ OPTICO DE LEBER  
ASOCIACION DE ENFERMEDADES RARAS D'GENES  
ASOCIACIÓN DE EPIDERMOLISIS BULLOSA DE ESPAÑA (DEBRA SPAIN)  
ASOCIACIÓN DE ESCLERODERMIA CASTELLÓN  
ASOCIACIÓN DE FAMILIARES Y AFECTADOS POR LIPODISTROFIAS  
ASOCIACIÓN DE HEMOGLOBINURIA PAROXISTICA NOCTURNA  
ASOCIACIÓN DE NEVUS GIGANTE CONGÉNITO  
ASOCIACIÓN DE PACIENTES ASMD ESPAÑA  
ASOCIACIÓN DE PACIENTES DE UVEITIS  
ASOCIACIÓN ENFERMEDAD DE KAWASAKI  
ASOCIACIÓN ESPAÑOLA ANIRIDIA  
ASOCIACIÓN ESPAÑOLA DE AFECTADOS POR SARCOMA  
ASOCIACIÓN ESPAÑOLA DE AMILOIDOSIS  
ASOCIACIÓN ESPAÑOLA DE ANGIOEDEMA FAMILIAR  
ASOCIACIÓN ESPAÑOLA DE ENFERMOS DE GLUCOGENOSIS  
ASOCIACIÓN ESPAÑOLA DE ENFERMOS DE POMPE  
ASOCIACIÓN ESPAÑOLA DE ENFERMOS Y FAMILIARES DE LA ENFERMEDAD DE GAUCHER ESPAÑA  
ASOCIACIÓN ESPAÑOLA DE ESCLERODERMIA  
ASOCIACIÓN ESPAÑOLA DE FAMILIARES Y ENFERMOS DE WILSON  
ASOCIACIÓN ESPAÑOLA DE FIBRODISPLASIA OSIFICANTE PROGRESIVA  
ASOCIACIÓN ESPAÑOLA DE FIEBRE MEDITERRANEA FAMILIAR

ASOCIACIÓN ESPAÑOLA DE ICTIOSIS  
ASOCIACIÓN ESPAÑOLA DE MASTOCITOSIS Y ENFERMEDADES RELACIONADAS  
ASOCIACIÓN ESPAÑOLA DE PACS1  
ASOCIACIÓN ESPAÑOLA DE PARAPARESIA ESPÁSTICA FAMILIAR STRÜMPPELL-LORRAIN  
ASOCIACIÓN ESPAÑOLA DE PORFIRIA  
ASOCIACIÓN ESPAÑOLA DE RAQUITISMOS Y OSTEOMALACIA HEREDADOS  
ASOCIACIÓN ESPAÑOLA DE SÍNDROME DE POLAND  
ASOCIACIÓN ESPAÑOLA DEFICIT DE LIPASA ACIDA LISOSOMAL  
ASOCIACIÓN ESPAÑOLA DEL SÍNDROME CDG  
ASOCIACIÓN ESPAÑOLA DEL SÍNDROME DE JOUBERT  
ASOCIACIÓN ESPAÑOLA DEL SÍNDROME DE SCHAAF-YANG  
ASOCIACIÓN ESPAÑOLA SÍNDROME DE SJÖGREN  
ASOCIACIÓN HHT ESPAÑA  
ASOCIACIÓN MADRILEÑA DE PACIENTES CON SÍNDROME DE GILLE DE LA TOURETTE Y TRASTORNOS ASOCIADOS  
ASOCIACIÓN NACIONAL AMIGOS DE ARNOLD CHIARI  
ASOCIACION NACIONAL DE DERMATOMIOSITIS JUVENIL  
ASOCIACIÓN NACIONAL DE HIPERTENSIÓN PULMONAR  
ASOCIACIÓN NACIONAL SÍNDROME DE APERT Y OTRAS CRANEOSINOSTOSIS SINDRÓMICAS  
ASOCIACIÓN RETINA MURCIA  
ASOCIACIÓN SÍNDROME DE ANGELMAN  
ASOCIACIÓN SÍNDROME DE LOWE DE ESPAÑA  
ASOCIACIÓN SÍNDROME LESCH NYHAN ESPAÑA  
ASOCIACIÓN STOP SANFILIPPO  
ASOCIACION XERODERMA PIGMENTOSUM  
ASSOCIACIO CATALANA DE LA DELECCIO 22Q  
ASSOCIACIÓ D'APRÀXIA OCULAR I MALALTIES ASSOCIADES  
ASSOCIACIÓN CATALANA DE ENFERMEDADES NEUROMUSCULARES DE NEU - ASOCIACIÓN DE ENFERMEDADES DE LOS NEUROTRANSMISORES  
DUCHENNE PARENT PROJECT ESPAÑA  
EUROPEAN NETWORK FOR RARE AND CONGENITAL ANAEMIAS  
FEDER - FEDERACIÓN ESPAÑOLA DE ENFERMEDADES RARAS  
FEDERACIÓ CATALANA DE MALALTIES MINORITÁRIES  
FEDERACIÓN DE ASOCIACIONES DE RETINOSIS PIGMENTARIA DE ESPAÑA  
FEDERACIÓN DE ATAXIAS DE ESPAÑA  
FEDERACIÓN ESPAÑOLA DE ENFERMEDADES NEUROMUSCULARES  
FEDERACIÓN ESPAÑOLA DE FIBROSIS QUISTICA  
FEDERACIÓN ESPAÑOLA DE HEMOFILIA  
FEDERACION ESPAÑOLA DE PADRES CON NIÑOS CON CANCER  
FEDERACIÓN ESPAÑOLA DEL SÍNDROME X FRÁGIL  
FUNDACIÓ CATALANA D'ELA MIQUEL VALLS (MIQUEL VALLS FOUNDATION)  
FUNDACIÓN ALPE ACONDROPLASIA  
FUNDACIÓN ANDRÉS MARCIO, NIÑOS CONTRA LA LAMINOPATÍA  
FUNDACION LIBELLAS  
FUNDACIÓN MARI PAZ JIMÉNEZ CASADO  
FUNDACIÓN NIEMANN-PICK DE ESPAÑA  
FUNDACIÓN SÍNDROME 5P MENOS  
FUNDACIÓN SÍNDROME DE DRAVET - DRAVET SYNDROME FOUNDATION  
FUNDACION SINDROME DE WEST  
FUNDACIÓN SÍNDROME WOLF HIRSCHHORN 4P-  
HIPERTENSION PULMONAR ESPAÑA ORGANISACIÓ DE PACIENTES  
INSTITUTO DE INVESTIGACIÓN Y DESARROLLO SOCIAL DE ENFERMEDADES POCO FRECUENTES  
LYMPHOMA COALITION  
MPS LISOSOMALES  
PEQUEÑOS SUPERHEROES  
SAF ESPAÑA  
SAMS - SÍNDROMES ARRÍTMICOS RELACIONADOS CON LA MUERTE SUBITA  
SENSE BARRERES DE PETRER  
SIMA ASOCIACIÓN DE AFECTADOS SÍNDROME DE MARFAN

## SWEDEN

AGRENSKA  
AORTA DISSEKTION FÖRENINGEN SKANDINAVIEN  
EUROPEAN CAVERNOMA ALLIANCE  
MARFAN EUROPE NETWORK  
NEUROFÖRBUNDET / SWEDISH ASSOCIATION OF PERSONS WITH NEUROLOGICAL DISABILITIES  
PRADER WILLI SYNDROME ASSOCIATION IN SWEDEN  
PRIMÄR IMMUNBRIST ORGANISATIONEN  
RARE DISEASES SWEDEN (RIKSFÖRBUNDET SÄLLSYNTA DIAGNOSER)  
SVENSK FÖRENING FÖR HYPOPARATYREOIDISM  
SVENSKA MARFANFÖRENINGEN - SWEDISH MARFAN ASSCIATION  
SVENSKA ÖDEM FÖRBUNDET (SWEDISH ASSOCIATION OF CHRONIC OEDEMA)  
SWEDISH CYSTIC FIBROSIS ASSOCIATION  
SWEDISH EDS ASSOCIATION (EDS RIKSFÖRBUND)  
SWEDISH MPS SOCIETY  
THYROID FEDERATION INTERNATIONAL  
WILHELM FOUNDATION

## SWITZERLAND

ASSOCIATION ENFANCE ET MALADIES ORPHELINES  
ASSOCIATION SUISSE ROMANDE INTERVENANT CONTRE LES MALADIES NEURO-MUSCULAIRES  
BLACKSWAN FOUNDATION  
CML ADVOCATES NETWORK  
ESPERARE  
FMF & AID GLOBAL ASSOCIATION  
FONDATION SANFILIPPO SUISSE  
FOUNDATION ORPHANHEALTHCARE  
FRAXAS - ASSOCIATION X FRAGILE SUISSE

HAEI - HEREDITARY ANGIOEDEMA INTERNATIONAL ASSOCIATION  
 HHT-SWISS  
**MARAVAL - MALADIES RARES VALAIS - SELTENE KRANKHEITEN WALLIS**  
**MARFAN FOUNDATION SWITZERLAND (MARFAN STIFTUNG SCHWEIZ)**  
**PRADER WILLI SYNDROM VEREINIGUNG SCHWEIZ**  
**PRORARIS**  
**SCHWEIZERISCHE GESELLSCHAFT FÜR PORPHYRIE**  
 SELBSTHILFE ICHTHYOSE SCHWEIZ  
 SJÖGREN EUROPE  
 SMA SCHWEIZ  
 SWISS FH/SCHWEIZERISCHE GESELLSCHAFT FÜR FAMILIÄRE FORMEN  
 DER HYPERCHOLESTERINÄMIE

## TAIWAN, PROVINCE OF CHINA

TAIWAN FOUNDATION FOR RARE DISORDERS

## TURKEY

**DMD AILELERI DERNEGI**  
**DUCHENNE KAS HASTALIĞI İLE MÜCADELE DERNEGI**  
 KİFDER  
 MUKOPOLISAKKARİDOZ VE BENZERİ LİZÖZOMAL DEPO HASTALIKLARI  
 DERNEGI / MPS TURKEY  
**PULMONER HİPERTANSİYON VE SKLERODERMA HASTA DERNEĞİ / PHA**  
**TURKEY**  
 SİSTİNOZİS HASTALARI DERNEĞİ / CİSTİNOZİS PATIENTS ASSOCIATION  
**SMA HASTALIĞI İLE MÜCADELE DERNEĞİ**  
 YUZÜMLÜ MUTLULUK DERNEĞİ

## UKRAINE

**ASSOCIATION OF PATIENTS WITH PULMONARY HYPERTENSION**  
**KHARKIV'S CHARITABLE FOUNDATION - «CHILDREN WITH SPINAL**  
**MUSCULAR ATROPHY»**  
 LCCF "SISTER DALILA" PULMONARY HYPERTENSION UKRAINIAN RARE  
 DISEASE ASSOCIATION  
 NGO "RARE DISEASES OF UKRAINE"  
 UKRAINIAN ASSOCIATION CRYSTAL PEOPLE  
**UKRAINIAN ASSOCIATION OF HELP FOR PATIENTS WITH CF**  
 UKRAINIAN NATIONAL CHARITABLE FUND ZAPORUKA  
 UKRAINIAN UNION OF PATIENTS' ORGANISATIONS  
 UKRAINIAN PARENT PROJECT «MIO-LIFE»

## UNITED KINGDOM OF GREAT BRITAIN AND NORTHERN IRELAND

ACRODYSOSTOSIS SUPPORT AND RESEARCH  
**ACTION DUCHENNE**  
 ADVOCACY FOR NEUROACANTHOCYTOSIS PATIENTS  
**AKU SOCIETY**  
 ALEX, THE LEUKODYSTROPHY CHARITY  
 ALSTROM SYNDROME EUROPE  
**ALSTROM SYNDROME UK**  
**ANN EDGAR CHARITABLE TRUST - NEUROENDOCRINE TUMOUR SUPPORT**  
**ANNABELLE'S CHALLENGE**  
**ASSOCIATION FOR GLYCOGEN STORAGE DISEASE**  
**ASSOCIATION FOR MULTIPLE ENDOCRINE NEOPLASIA DISORDERS**  
**ATAXIA UK**  
**BATTEN DISEASE FAMILY ASSOCIATION**  
**BEHCET'S UK**  
 BRITTLE BONE SOCIETY  
 CAMBRIDGE RARE DISEASE NETWORK  
 CANCER 52  
**CAVERNOMA ALLIANCE UK**  
**CDH INTERNATIONAL**  
**CDH UK**  
**CHILD GROWTH FOUNDATION**  
 CHILD LUNG FOUNDATION  
**CHILDHOOD TUMOR TRUST**  
 CHILDREN'S LIVER DISEASE FOUNDATION  
**CILIOPATHY ALLIANCE**  
**CONTACT A FAMILY**  
 CURE CLCN4  
**CYSTINOSIS FOUNDATION OF THE UK**  
**DANCING EYE SYNDROME SUPPORT TRUST**  
 DEGOS DISEASE SUPPORT NETWORK  
**ECTODERMAL DYSPLASIA SOCIETY**  
**EURO ATAXIA - EUROPEAN FEDERATION OF HEREDITARY ATAXIAS**  
 EUROPEAN LUNG FOUNDATION  
 EUROPEAN MYASTHENIA GRAVIS ASSOCIATION  
**EUROPEAN TUBEROUS SCLEROSIS COMPLEX ASSOCIATION**  
 FABRY INTERNATIONAL NETWORK  
 FEDERATION OF EUROPEAN SCLERODERMA ASSOCIATIONS  
 FETAL ANTI CONVULSANT SYNDROME ASSOCIATION  
 FH EUROPE  
 FINDACURE: THE FUNDAMENTAL DISEASES PARTNERSHIP  
**FOP FRIENDS**  
**GAUCHERS ASSOCIATION UK**  
**GENETIC ALLIANCE UK**  
 GLUT1 DEFICIENCY UK  
 HOPE FOR HASTI  
**HOPE FOR HYPOTHALAMIC HAMARTOMAS UK**  
**HUNTINGTON'S DISEASE YOUTH ORGANISATION**  
 INTERNATIONAL BRAIN TUMOUR ALLIANCE  
**INTERNATIONAL GAUCHER ALLIANCE**  
**INTERNATIONAL NIEMANN-PICK DISEASE ALLIANCE**  
**INTERNATIONAL PATIENT ORGANISATION FOR PRIMARY**  
**IMMUNODEFICIENCIES**  
**INTERNATIONAL PRADER-WILLI SYNDROME ORGANISATION**  
**JOINING JACK**  
**KRABBE UK**  
**LEBER'S HEREDITARY OPTIC NEUROPATHY SOCIETY**

**LGD ALLIANCE EUROPE**  
**MAX APPEAL**  
 MEBO RESEARCH  
**METABOLIC SUPPORT UK**  
**MPS SOCIETY**  
**MYOTUBULAR TRUST**  
**NEWLIFE THE CHARITY FOR DISABLED CHILDREN**  
**NIEMANN-PICK DISEASE GROUP UK**  
**NORTHERN IRELAND RARE DISEASE PARTNERSHIP**  
**ORGANISATION FOR ANTI-CONVULSANT SYNDROME**  
**PITT HOPKINS UK**  
**POLYCYSTIC KIDNEY DISEASE CHARITY**  
 POMPE SUPPORT NETWORK  
**PSC SUPPORT**  
 PSEUDOMYXOMA SURVIVOR  
 PTEN RESEARCH FOUNDATION  
 RARE AUTOINFLAMMATORY CONDITIONS COMMUNITY- UK  
 RARE DISEASES UK  
 RING 20 RESEARCH AND SUPPORT UK CIO  
 SALIVARY GLAND CANCER UK  
 SCHINZEL-GIEDION SYNDROME FOUNDATION  
 SMILE WITH SHIV  
**SPINAL MUSCULAR ATROPHY SUPPORT UK**  
**STIFF PERSON SUPPORT GROUP**  
**STURGE-WEBER UK**  
**THE AACR RESEARCH TRUST CHILDREN'S CHARITY**  
 THE AARSKOG FOUNDATION  
 THE CHROMOSOME 18 REGISTRY AND RESEARCH SOCIETY (EUROPE)  
**THE CURE & ACTION FOR TAY-SACHS (CATS) FOUNDATION**  
**THE EHE RARE CANCER CHARITY**  
**THE EHLERS-DANLOS SOCIETY**  
**THE FRAGILE X SOCIETY**  
 THE MADDI FOUNDATION  
 THE PBC FOUNDATION (UK) LTD  
 TIMOTHY SYNDROME ALLIANCE  
**TUBEROUS SCLEROSIS ASSOCIATION**  
 UK ATTR AMYLOIDOSIS PATIENTS' ASSOCIATION  
**UK MASTOCYTOSIS SUPPORT GROUP**  
**UNIQUE - RARE CHROMOSOME DISORDER SUPPORT GROUP**  
**UNITED KINGDOM THALASSAEMIA SOCIETY**  
**VASCULITIS UK (THE VASCULITIS TRUST)**

## UNITED STATES OF AMERICA

MCT8-AHDS FOUNDATION INC.  
 ALAGILLE SYNDROME ALLIANCE  
 ALSTROM SYNDROME INTERNATIONAL  
 APS FOUNDATION OF AMERICA, INC  
 ASSOCIATION FOR CREATINE DEFICIENCIES  
 BCM FAMILIES FOUNDATION  
 CACNA1A FOUNDATION  
 CURE PSP  
 CYSTINOSIS FOUNDATION  
 DEFEAT MSA ALLIANCE  
 EMPTY NOSE SYNDROME INTERNATIONAL ASSOCIATION  
 FMD CHAT  
 GLOBAL DARE FOUNDATION  
 GOULD SYNDROME FOUNDATION  
 INTERNATIONAL FOXG1 FOUNDATION  
 INTERNATIONAL PEMPHIGUS & PEMPHIGOID FOUNDATION  
 INTERNATIONAL WAGR SYNDROME ASSOCIATION  
 INTERNATIONAL WALDENSTROM'S MACROGLOBULINEMIA FOUNDATION  
 MYHRE SYNDROME FOUNDATION  
 NEUROMUSCULAR DISEASE FOUNDATION  
 NORD NATIONAL ORGANISATION FOR RARE DISORDERS  
 NTM INFO & RESEARCH  
 PROJECT 8P  
 PTEN HAMARTOMA TUMOR SYNDROME FOUNDATION  
 PURA SYNDROME FOUNDATION  
 REMEMBER THE GIRLS  
 SATB2 GENE FOUNDATION  
 SUPERFICIAL SIDEROSIS RESEARCH ALLIANCE  
 SYNGAP1 FOUNDATION  
 THE CUSHING SUPPORT & RESEARCH FOUNDATION  
 THE CUTE SYNDROME FOUNDATION  
 THE OXALOSIS & HYPEROXALURIA FOUNDATION  
 THE SNYDER-ROBINSON FOUNDATION  
 USHER SYNDROME COALITION

## URUGUAY

ASOCIACION ACONDROPLASIA URUGUAY  
 FUNDACION URUGUAYA PARA LA INVESTIGACION DE LAS ENFERMEDADES  
 RARAS

## VENEZUELA, BOLIVARIAN REPUBLIC OF

FUNDACIÓN FURA

## ZIMBABWE

CHILD & YOUTH CARE, ZIMBABWE



# PARTICIPATION OF EURORDIS' REPRESENTATIVES IN PUBLIC EUROPEAN / INTERNATIONAL CONFERENCES & Workshops 2021

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## EMA Public stakeholder meeting on the approval and roll-out of COVID-19 vaccines in the EU, 8 January

**François Houÿez** contributed to the discussion

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## RE(ACT) Congress & IRDiRC Conference 2021, 13-15 January

**Virginie Hivert**: The Galaxy Guide & Hands-on as Speaker and Methodologies to assess the effect of diagnosis and therapies on rare diseases patients as Co-Chair; **François Houÿez**: Patients as drivers in drug development and clinical trials

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## Live Policy Event: Unlocking innovation and access for rare disease patients in Europe, 22 February

**Yann Le Cam**: Increase access for patients in Europe through sustainable innovation

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## International Conference on Rare Diseases, 1 March

**Yann Le Cam**: Getting our rights 'right': An international framework for rare diseases; **Gulcin Gumus**: Newborn Screening: Harmonising approaches to NBS in EU; **Elizabeth Vroom**: Patient Preferences in NBS: The Duchenne Example; **Ines Hernando**: European Reference Networks - Recommendations to achieve a mature ERN system in 2030; **Karolina Hanslik**: RARE IMPACT: Collaboration to increase access in Europe; and Simone Boselli as Moderator

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## Meeting on Data Sharing for Duchenne, 3 March

**Elizabeth Vroom** as Meeting Host and **Gulcin Gumus** as Presenter

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## DIA Europe 2021, 15-19 March

**Maria Cavaller**: Patient Perspective on PED In Regulatory Decision-Making; **François Houÿez**: Trans-Diagnostic Drug Development Beyond Oncology; **François Houÿez**: Effective Risk Communication for Safe Use Behaviors in Healthcare: Innovation Through Formative and Evaluative Collaborations; **Matteo Scarabelli**: After Three Joint Actions with EUnetHTA – Which are the Main Achievements and the Remaining Challenges? **François Houÿez**: Refocus on Unmet Public Health Challenges – Who should Lead the Way? **Simone Boselli** represented EURORDIS in the sessions 'Enabling Cell and Gene Therapies: Creating an Appropriate Framework' and 'Cross-border Access to ATMPs in the European Union'

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## EC Structured Dialogue for the security of medicines supply, 25 March

**François Houÿez** participated in a series of meetings until October 2021

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## EMA Public stakeholder meeting on the approval and roll-out of COVID-19 vaccines in the EU, 26 March

**François Houÿez** contributed to the discussion

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## ICRDOD Bulgaria, 27 March

**François Houÿez**: How patients see the future cooperation on HTA in Europe – the case of orphan medicinal products

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## 9th Annual UDNI Conference & Mayo Clinic Science Session 2021, 11 April

**Gulcin Gumus**: Patient Engagement Working Group

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### EUnetHTA Stakeholder Forum 15 April

**Matteo Scarabelli as Chair of the Session:** Engaging Stakeholder in EUnetHTA

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### Duke University Global Challenges and the COVID Vaccine: Manufacturing and Supply, 20 April

**François Houÿez** represented EURORDIS

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### Nordic Rare Disease Summit 2021, 12-13 April

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

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### 2021 EUnetHTA Forum, 15 April

**Yann Le Cam:** EUnetHTA: The past, the present and the future - Lessons learnt and recommendations

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### Towards a European Health Union: Lessons learned, future challenges, 21 April

**Yann Le Cam** represented EURORDIS

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### 1st meeting with the Member States Representatives on the proposal for a Horizon Europe Partnership on Rare Diseases Brussels, 22 April

**Yann Le Cam:** Rare 2030 – Strategic recommendations

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### Rare Conversations, 5 May

**Rob Camp:** Research in rare disease: the role of Community Advisory Boards - EuroCAB

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### AESGP 57th Annual Meeting, 27 May

**François Houÿez:** Communicating risk in times of uncertainty

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### Partnering for Patients 2021, 28 May

**Matteo Scarabelli:** Patient involvement in HTA: where it starts, where is it going?

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### Slovak conference on HTA and pricing, 2 June

**François Houÿez:** the case of orphan drugs

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### French National Alliance, 4 June

**François Houÿez:** The New Early Access and Compassionate Use Schemes which impact for people living with a rare disease?

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### International Pharmacovigilance Days, 18 June

**François Houÿez:** Patient Organisations' Role in Pharmacovigilance

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### DIA Global, 30 June

**François Houÿez:** Patient-Focused Benefit-Risk Assessment and Risk Management: Methodology for Engaging with Patients: What has been Learned?

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### Realising the Potential of Transformative Therapies, 30 June

**Simone Boselli** represented EURORDIS

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### 5es Rencontres sur les Maladies Rares, 6 July

**Yann Le Cam:** Faciliter l'accès aux thérapies innovantes : comment aller plus loin

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### HTA Network Stakeholder Pool, 6-7 July

**Matteo Scarabelli, François Houÿez** represented EURORDIS

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### RARE Conversations, 8 July

**François Houÿez:** HTA, From national to EU value assessment: which provisions for orphan drugs?

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### ICPE Plenary Session on COVID-19 Communication, 24 August

**François Houÿez:** Plenary 2: Communicating COVID-19 Pharmaco-epidemiological Research to Patients: Why is it important? How can we do it? What are the implications now and for the future?

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### World Orphan Drug Congress USA 2021, 26-27 August

**Yann Le Cam:** A global patient perspective on the future of medicines development and access

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### 22q11 Conference, 4 September

**Gulcin Gumus:** Harmonising approaches to NBS in EU

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### Webinar EU4Health Work Programme 2022, 10 September

**Yann Le Cam:** Health Systems

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### National Press Foundation's workshop on rare diseases, 14 September

**Yann Le Cam:** How Rare 2030 Plan Can Promote Evidence-Based New Policy Framework

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### EU Law in the Pharmaceutical sector, 15 September

**François Houÿez:** debate on the future of Europe: Access to medicines

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### The 2021 ESMO Patient Advocates Track Sessions, 16 September

**Ariane Weinman:** How the integration of a specific section on rare cancers into national cancer control plans can support the rare cancer patients' journey from diagnosis to treatment

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### TOPRA Annual Symposium, 23 September

**Virginie Hivert:** From 'knocking on the door' to co-creation: 25 years of patient engagement in R&D

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## European Sickle Cell Disease Summit – ESCF, 24-25 September

**Gulcin Gumus** represented EURORDIS

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## Oslo Medicines Initiative: A new vision for collaboration between the public and private sectors, 27 September

**Yann Le Cam** represented EURORDIS

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## Seminario Taller sobre el Estado de Situación de Enfermedades Raras en la Región SICA y Región Andina, 28 September

**Yann Le Cam** represented EURORDIS

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## European Health Forum Gastein (EHFG) “Rise like a phoenix”, 29 September

**Yann Le Cam** represented EURORDIS

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## Technical meeting on Achieving Equity and Innovation in Newborn Screening and in Familial Hypercholesterolemia Paediatric Screening across Europe – An accompanying event of the Slovenian Presidency of the Council of the European Union 2021, 11 October

**Yann Le Cam:** The views of people living with rare diseases and their families: 11 key principles; **Simone Bosselli, Valentina Bottarelli and Gulcin Gumus** represented EURORDIS

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## How can we make the impossible possible for rare and paediatric disease? by EFPIA, 12 October

**Simone Bosselli** represented EURORDIS

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## International Longevity Coalition, 12 October

**François Houyez:** Improving vaccination uptake among patients with chronic conditions across Europe

---

## 7ème Edition des Rencontres RARE, 14-15 October

**Yann Le Cam:** Présidence française de l'Union européenne; **Virginie Hivert:** Quel est le rôle des patients dans la recherche et dans le cycle de vie du médicament?

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## NORD Breakthrough Summit 2021, 18-19 October

**Yann Le Cam:** International Collaboration in the Rare Community

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## Office of Health Economics, 20 October

**François Houyez:** The role of unmet need in pharmaceutical innovation

---

## Regional conference Rare Diseases Database Novi Sad, Serbia, 22 October

**Gulcin Gumus:** NBS: Harmonising approaches to NBS in Europe

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## World Health Summit, 24-26 October

**Yann Le Cam** represented EURORDIS

---

## Kick-off meeting of the Screen4Care project, 26 October

**Virginie Bros-Facer, Gulcin Gumus, Sandra Courbier and Edith Gross:** Overview and integration of perspectives and needs of rare disease patients in NBS and early diagnosis

---

## Health-&Pharmacoeconomics Opportunities for German/African Cooperation, 30 October

**François Houyez:** the future European Cooperation on HTA

---

## How to Optimize Cross-border Cooperation to Support Equitable Access to Advanced Therapies? by TRANSFORM Interest Group, 10 November

**Simone Bosselli** represented EURORDIS

---

## 8th Tay-Sachs & Sandhoff Family Conference, 9 November

**Gulcin Gumus:** Newborn Screening: Harmonising approaches to NBS in EU

---

## ISPOR 2021, 16 November

**François Houyez:** No Time to Wait: Can Early Access Schemes Work for Gene Therapies?

---

## 1st International Conference on Rare Diseases and Paediatric Research, 18 November

**Virginie Hivert:** The Orphan Drug Development Guidebook

---

## EMA Big Data Stakeholder Forum, 7 December

**Virginie Hivert:** Regulatory processes for data

---

## Helplines conference, 6 December

**Gulcin Gumus:** EURORDIS' involvement in undiagnosed diseases

---

## CIRS Ensuring meaningful patient engagement, 15 December

**François Houyez:** How can companies align internal and external stakeholders' expectations during development to support the review and reimbursement of new medicines?

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# JOURNAL PUBLICATIONS 2021

## Patient Contribution to the Development and Safe Use of Medicines During the Covid-19 Pandemic

January, Therapeutic Innovation & Regulatory Science

Contributing authors:

**François Houÿez**

## Evaluation of patient engagement in medicine development: A multi-stakeholder framework with metrics

February, Health Expectations

Contributing authors:

**Elisa Ferrer on behalf of  
EURORDIS**

## Impact of Covid-19 on clinical care and lived experience of systemic sclerosis: An international survey from EURORDIS-Rare Diseases Europe

February, Journal of Scleroderma and Related Disorders

Mentions:

**The Rare Barometer team**

## Understanding multi-stakeholder needs, preferences and expectations to define effective practices and processes of patient engagement in medicine development: A mixed-methods study

February, Health Expectations

Contributing authors:

**Elisa Ferrer on behalf of  
EURORDIS**

## Learning from the Pandemic to Improve Care for Vulnerable Communities: The Perspectives and Recommendations from the Rare Disease Community (Editorial)

March, International Journal of Integrated Care

Contributing authors:

**Raquel Castro, Sandra  
Coubier, Erwan Berjonneau**



**Our greatest untapped resource: our patients**

April, Journal of Community Genetics

Contributing authors:

**Matt Bolz-Johnson, Yann Le Cam & Ines Hernando**

**Sustaining Meaningful Patient Engagement Across the Lifecycle of Medicines: A Roadmap for Action**

May, Therapeutic Innovation & Regulatory Science (TIRS)

Contributing authors:

**Maria Cavaller-Bellaubi PharmD**

**Shaping national plans and strategies for rare diseases in Europe: past, present, and future**

May, Journal of Community Genetics volume

Contributing authors:

**Valentina Bottarelli, Ariane Weinman**

**Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases**

June, Accepted for publication, European Journal of Human Genetics

Contributing authors:

**Gulcin Gumus**

**Rare Diseases in Handbook Integrated Care (pp. 763-782)**

July, Springer, Cham

Contributing authors:

**Raquel Castro and Sandra Courbier**

**Essential list of medicinal products for rare diseases: recommendations from the IRDiRC Rare Disease Treatment Access Working Group**

July, Orphanet Journal of Rare Diseases

Contributing authors:

**Virginie Hivert**

**Patient engagement in healthcare: a preliminary set of measures to evaluate patient engagement in the European Reference Networks**

August, Rare Disease and Orphan Drugs Journal

Contributing authors:

**Lenja Wiehe; Anne-Laure Aslanian; Ines Hernando**

**IRDiRC: 1000 new rare diseases treatments by 2027, identifying and bringing forward strategic actions**

November, Rare Disease and Orphan Drugs Journal

Contributing authors:

**Virginie Hivert**

**Orphan Medicine Incentives: How to Address the Unmet Needs of Rare Disease Patients by Optimizing the European Orphan Medicinal Product Landscape Guiding Principles and Policy Proposals by the European Expert Group for Orphan Drug Incentives (OD Expert Group)**

December, Frontiers in Pharmacology

Contributing authors:

**Yann Le Cam**

# ACKNOWLEDGEMENTS

EURORDIS would like to thank the following organisations and companies for their financial support in 2021:

## PATIENT ORGANISATIONS AND PUBLIC ENTITIES

### AFM - TÉLÉTHON

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



### EUROPEAN COMMISSION

DG Health and Food Safety

- ◆ The Operating Grant for year 2021
- ◆ 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
- ◆ European Rare disease research coordination and support action (ERICA)
- ◆ The Innovative Medicines Initiative-Joint Undertaking (IMIJU) projects:
  - ◆ conect4children, a Collaborative Network for European Clinical Trials For Children
  - ◆ 'Shortening the path to rare disease diagnosis by using newborn genetic screening and digital technologies' — 'SCREEN4CARE



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



## Health Sector Corporates

EURORDIS appreciates the contributions received from health sector companies. Ensuring a sustained variety of funding is key to minimise potential conflicts of interest. EURORDIS had 77 different health sector corporate donors in 2021. Health sector companies have supported EURORDIS through the EURORDIS Round Table of Companies, the European Membership Meeting, the EURORDIS Black Pearl Awards, as well as international initiatives such as Rare Disease Day, Rare Barometer, Rare Diseases International, EURORDIS Open Academy and multi-lingual communications, as well as through contributions supporting project development and the Rare Impact project. The breakdown of each company's contributions by project is detailed on the EURORDIS website on the "Corporate Revenue" tab of the "Financial Information" section.

### TOP FIVE DONORS

1	TAKEDA	
2	SANOFI GENZYME	
3	PFIZER	
4	ROCHE	
5	CSL BEHRING	

### FOOTNOTES

1	<u>EURORDIS</u>	
2	<u>BLACKPEARL</u>	
3	<u>RARE DISEASE DAY</u>	
4	<u>EURORDIS VOICES</u>	
5	<u>RARE DISEASES INTERNATIONAL</u>	
6	<u>OPEN ACADEMY</u>	



## OTHER PHARMACEUTICAL &amp; BIOTECHNOLOGY COMPANIES &amp; HEALTH SECTOR CORPORATES MAKING DONATIONS TO EURORDIS



## OTHER CORPORATES PROVIDING DIRECT AND IN-KIND SUPPORT

### DLA Piper



### Aparito



## FOUNDATIONS AND NON-FOR-PROFIT CONTRIBUTORS

### Association of International Pharmaceutical Manufacturers (AIPM)



### Fondation Ipsen, under the aegis of Fondation de France



### Bertarelli Foundation



### Everylife Foundation



### Chan Zuckerberg Initiative



### 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 2021.



**EURORDIS**  
RARE DISEASES EUROPE

# WORKPLAN

## 2022

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# ACTION PLAN 2022

*Following the overall Strategic Review 2015-2020 and the design of the new EURORDIS long-term Strategy for 2021-2030, adopted at the General Assembly June 2021, the EURORDIS Action Plan 2022 is structured along our long term strategic goals "ADVOCATE, EMPOWER and PARTNER, focusing on PEOPLE FIRST – based on evidence and experience of person's needs and preferences - across "research policy & initiatives, patients' data & digital health, development & access to diagnostics & therapies, healthcare policy & services, social policy & services".*

To achieve our mission "EURORDIS works across the borders and diseases to improve the lives of all persons

living with a rare disease", EURORDIS pursue four overarching strategic objectives:

- ✦ A new European policy framework to achieve measurable goals prolonging and improving the life of persons living with rare diseases
- ✦ An ecosystem of networks of member organisations, organisations at large; and partners to achieve better and faster results
- ✦ Focusing on priority areas where it matters the most during next decade to achieve the goals
- ✦ Inclusive of all Rare Diseases, in all regions, to "Leave no one behind"

## STRATEGIC OBJECTIVE 1: A NEW EUROPEAN POLICY FRAMEWORK TO ACHIEVE MEASURABLE GOALS PROLONGING AND IMPROVING THE LIFE OF PERSONS LIVING WITH RARE DISEASES

*By 2030, EURORDIS has promoted and facilitated the implementation of a new European policy framework to achieve measurable goals prolonging and improving the life of persons living with rare disease and their*

*inclusion in society, while designing a European model of care driven by a need's led innovation and collective responsibility prioritising solidarity and equity.*

### 1.1 ADVOCATE, EMPOWER, PARTNER

**Our advocacy goals within our strategic objectives for 2021-2030**

- ✦ Promoting rare diseases as a priority for achieving sustainable development 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 diagnostics and quality of medical and social care at local level
- ◆ Promoting access to cross-border healthcare and making patient mobility possible
- ◆ Promoting research and bridging perspectives of persons living with rare diseases and researcher activities
- ◆ Addressing the issues of genetic testing, genetic counselling and newborn screening
- ◆ Voicing/expressing preferences and needs expressed by persons living with rare diseases by sharing of health data in rare diseases information systems and repositories and in contributing to a European Health Data Space that supports care and research for rare diseases

### Advocate for rare diseases as a priority in the next decade 2021-2030

- ◆ **Promote a European Health Union** in areas of action of high European added value and high economic impact. Such areas are health threats, cancers, and rare diseases, learning the lessons of the COVID-19 crisis and the impact of the Ukrainian war building more resilient and equitable healthcare and welfare systems
- ◆ **Prepare for the next decade of rare disease legislative and policy framework to take the necessary steps to requalify rare diseases as a public health issue**
- ◆ Explore and consolidate political options with EU Institutions to **update, review or replace the overall EU rare disease strategy** on the basis of the **Rare2030 policy recommendations for rare disease policy** (issued in February 2021). This action **paves the way to a new EU policy framework for rare diseases by 2030** and is in line with the 2019 Court of Auditors special report on cross-border health care
- ◆ **Continue the two-year #30millionreasons campaign in view of the revamping of the EU rare disease strategy and advocating for a goal-oriented European Action for Rare Diseases, comprising of:**
  - ◆ Engaging with Trio of EU Presidencies 2022-2023, plus Spain or further EU Council Presidencies, as appropriate, to support the EU policy strategic review and new policy framework for rare diseases
  - ◆ Re-launching and expanding the European Parliamentary Advocates for Rare Diseases network, expanding it to national members of parliament, in coordination with the National Alliances
  - ◆ Finalising the broad grassroots campaign to mobilise EURORDIS members at large, with greater integration of advocacy actions at the local, national, European levels.

### Promote the sustainability of rare diseases as a policy and budget priority in the EU programmes for the period 2021-2027

- ◆ Secure **proposals impacting on rare diseases** in the context of the **EU Multiannual Financial Framework 2010-2027** that will fund EU policies and programmes. In particular
- ◆ Ensure that the measures in support of people living with rare diseases agreed upon in the new health programme 2021-2027 **"EU4Health"** are duly implemented and funded on a yearly basis, including the **sustainability of ERNs and Orphanet**, the specific support to children and adult **rare cancers** within the EU Beating the Cancer Plan. Include the support for a comprehensive strategy for rare diseases in the form of an Europe's Action Plan and its key actions and flagships
- ◆ 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 future **European Partnership for Rare Diseases** (succeeding to the **European Joint Programme Co-Fund for Research on Rare Diseases**), **Clinical Research Networks for Rare Diseases** embedded within ERNs and supported by the RD Partnership and rare cancer 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** and the **Cohesion Fund** and the new **Cohesion's Action for Refugees in Europe (CARE)**
- ◆ Promote the needs of people with rare diseases as a priority in the Innovative Health Initiative-IHI (IMI's successor) programme 2021-2027



## STRATEGIC OBJECTIVE 2: AN ECOSYSTEM OF NETWORKS OF MEMBER ORGANISATIONS, ORGANISATIONS AT LARGE AND PARTNERS TO ACHIEVE BETTER AND FASTER RESULTS

*By 2030, EURORDIS has redefined and focused its unique role in the rare disease ecosystem; we are leading impactful and motivated networks of member organisations and advocates across diseases and borders, enabling to relay people's needs and integrate European with national actions in key policy areas; and as a driver of change, is creating synergies across partners and key stakeholders to achieve better and faster results.*

To achieve this objective, in 2022, EURORDIS will continue to inform, support, reinforce and empower its networks of European patient organisations at large, Members, volunteers, European federations, National Alliances, European helplines, international actors, providing them also with capacity-building through training, e-learning, resources and recommendations. The activities are detailed in the next sections:

### 2.1 EMPOWER AND PARTNER

#### Build the community, inform, support, empower our networks

##### Membership and European Patient Organisation at large

- ✦ Build the community through improved outreach to the Rare Disease community: EURORDIS Membership and other organisations representing persons living with a rare disease, involve EURORDIS members and non-members in activities (attend conferences, workshops and trainings) in order to represent the broad spectrum of RDs
- ✦ Reach the number of **1000 members** at EURORDIS (Membership)
- ✦ Ensure regular two-ways interactions with our members through the bi-monthly Member News sent out in six languages, ad hoc consultations, webinars and training opportunities
- ✦ Prepare and examine membership application, maintain an efficient process of regular membership reassessment
- ✦ Prepare the EURORDIS Membership Meeting 2023
- ✦ Maintain and develop the EURORDIS contact database that gathers data of 9631 contacts in over 5666 organisations (**over 2965 organisations representing persons living with a rare disease**) including EURORDIS's Membership. The database integrates the Orphanet database representing 6207 disorders, 2177 group of disorders and 1007 subtype of disorder (a total of 9351 entries). This database is used and updated daily by all EURORDIS staff members
- ✦ Enrich the information in the organisations' contact database for targeted outreach, EURORDIS informs and empowers the networks of organisations representing persons living with a rare disease in Europe

##### Volunteers

- ✦ EURORDIS relies on a unique network of over 460 volunteers organised by groups of interest/expertise
- ✦ All volunteer groups have a dedicated staff member who supports their work, providing them with a

forum of exchange of information, and guiding their overall work process

- ✦ Keep coordinating the volunteers and involving them into all EURORDIS activities

***EURORDIS coordinates, manages and builds capacities of several European networks which are actively contributing to Rare Diseases: the Council of National Alliances (CNA), the Council of European Federations (CEF) and the European Network of Rare Disease Help Lines (ENRDHL).***

##### Council of National Alliances (CNA)

- ✦ Overall network coordination, organisation of "hot topics" monthly calls and two meetings of the Council of National Alliances (CNA) with approximately 40 participants: one was held online in April and another one – a 2-day face-to-face – was held in Paris in November and offered 5 fellowships for patient representatives.
- ✦ Work with the CNA on: Rare Disease Day 2022 Campaign feedback and plans for 2023, the EU action plan and the national plans for rare diseases, the European Partnership for Rare Disease, the integration of ERNs into National Healthcare Systems, the Parliamentary advocates for rare diseases involving national parliament members, activities in support of Ukrainian people leaving with rare diseases and refugees
- ✦ 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
- ✦ Send monthly CNA newsletters to enhance capacity-building
- ✦ Reinforce the collaboration and integrate more the advocacy actions between EURORDIS and National Alliances; working with a dedicated Public Affairs Manager based in Brussels on EU & National Integrated Advocacy; empowering capabilities through regular bilateral meetings, peer-to-peer training, best practice sharing and webinars



### Council of European Federations (CEF)

- ✦ Overall coordination of the CEF, including regular informal CEF online meetings and the organisation of one face-to-face 2-day meeting with approximately 25 participants in Paris, including 1 joint day with the CNA, including 5 fellowships for patient representatives
- ✦ Continue the EURORDIS programme “Support to European Rare Disease Federations” to assist the weakest/youngest RD Federations in financing their network meetings (20 federations supported). The purpose of the programme is to provide small, quick, and flexible financial support for the organisation of online meetings
- ✦ Work more closely with EF on key items, in particular on the Ukrainian humanitarian crisis, on access to medicines – Real World Evidence, EU Fund, European Procurement, European RD Partnership and Rare Diseases Clinical Research Networks, Evaluation of the EU Directive on Cross-Border Health Care

### European Network of Rare Diseases Help Lines (ENRDHL)

- ✦ Overall coordination of the ENRDHL and organisation of one hybrid meeting with approximately 40 participants
- ✦ Share complex calls cases between helplines
- ✦ 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
- ✦ To share best practices and build capacity: organise a helpline workers for approximately 20 people on the management of complex calls, with an external trainer and videoconferencing. This training will be open to the EURORDIS’ staff facing direct requests. EURORDIS staff (about 11 staff members) is answering to individual request, and also to other categories of enquirers. In 2021, about 290 direct individual patient requests were handled by the team
- ✦ Implement the Caller Profile Analysis in Fall 2022

### Rare Cancers

- ✦ Continue to fully engage in the EU’s Beating Cancer Plan and Cancer Mission, as well as in other programmes where relevant
- ✦ Dedicate an experienced Public Affairs Senior Manager to maintain a high level of engagement of rare cancer patient advocates in all public consultations, notably through the Working Group composed of 30 ePAGs of the 4 relevant ERNs covering rare cancers. The WG serves as advisory group on advocacy issues in the field of (rare) cancer. Create a stronger sense of shared interest and solidarity across the +70 Rare Cancer EURORDIS member organisations
- ✦ Ensure a close coordination and collaboration with patient organisations, the European federations for specific rare cancers – paediatric or adult, the network WECAN, and, in the USA, the NORD Rare Cancer Coalition; fully engage in our co-founded multi-stakeholder platform Rare Cancer Europe; continue to collaborate with academic partners such as ESMO and ECCO
- ✦ Develop a well-sourced dedicated web section on rare cancers with substantial information on both EURORDIS’ policy activities and projects and on information relevant to patients and patient organisations
- ✦ Support actions and projects to implement recommendations for paediatric cancers and rare adult cancers set out in the Rare Cancer Agenda 2030 and in the European Parliament’s report “strengthening Europe in the fight against cancer; towards a comprehensive and coordinated strategy”. Integrate a component on rare cancers in the International Rare Disease Day and in the Rare Disease Week

### RareConnect

- ✦ RareConnect host, the Children’s Hospital of Eastern Ontario, is no longer able to invest in the Rare Connect Platform. In 2022, EURORDIS is considering options to maintain a low maintenance level with hosting at CHEO and limited support to moderators, or, find a new host and re-invest, or, retire the service by conducting an assessment of EURORDIS’ role within RareConnect taking into account other online communities

## 2.2 SUPPORT AND EMPOWER

### Empower: build capacity for our networks

#### Support and capacity-building of the Rare Diseases community with Covid19 updated information, resources and recommendations

Rare Diseases patients, are one of the most vulnerable populations affected by health threats such as Covid19. Since the beginning of the pandemic in March 2020, EURORDIS has been very active in providing resources for people living with a rare disease to better manage their already fragile situations, including specific online information, links towards the ERNs specific Covid19 and vaccination recommendations, as well as providing capacity building webinars. In 2022:

- ✦ Maintain the EURORDIS Covid19 Task Force meetings, maintain and develop the online Covid19 Resource Center launched on the EURORDIS’ website in March 2020, updated as required, and keep supporting the resilience of the network of Patient Organisations as well as the adaptation of the Rare Disease community to this new healthcare challenge, also by re-enforcing the network preparedness to new healthcare threats

### EURORDIS Open Academy & other trainings

EURORDIS has been training patient advocates since 2008. The OA's primary goal is to empower patient advocates in the various fields where patient engagement and empowerment is needed. The programmes are based on a blended approach with online trainings (webinars and e-learning courses) and face-to-face components.

- ★ Organise the 5<sup>th</sup> edition of the **EURORDIS Winter School** on Scientific Innovation and Translational Research, **including one week of online training, in March 2022, preceded by 3 pre-training webinars and over 10 e-learning courses**
- ★ Organise the 15<sup>th</sup> edition of the **EURORDIS Summer School** on Medicines Research & Development, **including one week of online training, in June 2022, preceded by 3 pre-training webinars and over 15 e-learning courses**
- ★ Further develop the **Open Academy's e-learning courses and digital platform:**
  - ★ Continue the **EURORDIS Digital School 2021-2022** as an online open programme with new e-learning courses and webinars including creating great mobile videos
  - ★ 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
- ★ 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
- ★ Increase the availability and outreach of the Open Academy's online training materials to more patient advocates, free of cost; in order to empower our various networks of patient advocates, including our Members, the EU networks, the ePAGs, our network of OA alumni
- ★ Maintain the new Open Academy website, which was launched in 2021
- ★ Launch a **monthly newsletter for alumni**, to inform them on training opportunities and on relevant patient engagement opportunities. Discuss further community tools such as LinkedIn group
- ★ Organise the second edition of the **Rare Diseases Week**
- ★ **ePAG webinar series "Building Good Practices' for European Patient Advocate Groups (ePAGs)"** will comprise of four quarterly online meetings to inspire other ePAG advocates to replicate or adapt good practices to their own needs and context and to encourage ePAG advocates to deliver webinars which they have developed to guide others through their own experience, identifying success factors and lessons learned

## STRATEGIC OBJECTIVE 3: FOCUSING ON PRIORITY AREAS WHERE IT MATTERS MOST DURING NEXT DECADE TO ACHIEVE OUR GOALS

*By 2030, EURORDIS has regularly assessed and consolidated its priority areas of operations to respond to the demands of both organisations and advocates while aligning with internal capacity, progressively focusing on the new challenges of data and health digital technologies as well as on a holistic life-*

*long approach and inclusion in society; as much as to strengthen its priority on changing the game in research and knowledge, early diagnosis, development of and access to transformative or curative therapies, integrated care, national and European healthcare pathways and cross-border healthcare.*

### 3.1 ADVOCATE, EMPOWER, PARTNER

**The EURORDIS Rare Barometer Programme: Patient experience surveys enabling evidence-based contributions to policy making and research**

EURORDIS conducts qualitative and quantitative surveys to collect and analyse robust evidence regarding the experiences of persons living with rare diseases including carers/family for the purpose of using this information in policy and decision-making processes and establishing EURORDIS' positions. In 2022, the Programme will focus on:

- ★ Scaling up the collection of experiences and perspectives of persons living with a rare disease to enrich an evidence based policy development building on the 15,000+ individuals with own experience who have joined Rare Barometer Voices and continue the growth internationally
- ★ Launching the **Rare Disease patients' journey to diagnosis survey**: this survey will be launched by EURORDIS in the first semester, following an information webinar held in March. The survey aims to understand the journey people living with a rare disease go through when seeking a diagnosis for their rare disease, for example by measuring the time taken to obtain a diagnosis, or the consequences of being undiagnosed or misdiagnosed. A report on the survey's results will be produced, as well as factsheet summarising the findings, translated in 26 languages.

- Stepping towards the development of a scale to measure rare diseases healthcare experience. This scale aims to include all of the aspects faced by people living with a rare disease in the evaluation of the care they received. The main outputs include a literature review report on high quality healthcare experience for rare diseases, with a conceptual framework for the development of a scale to measure healthcare experience and a discussion guide of online focus groups highlighting the unmet needs of people living with rare diseases and complementing the conceptual framework (translated in 6 languages)
- Continue dissemination of reports of previous surveys, infographics or online dashboards presenting the results of surveys in 23 languages
- The programme will apply for foundation and EC grants as well as developing support from corporate partners

### **Advocate and empower patients to reduce diagnostic delays for rare diseases and to tackle undiagnosed diseases, as a major priority area**

**It takes on average five years for a patient to get a diagnosis, when a diagnosis is made at all.**

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

- EURORDIS, alongside its Council of National Alliances, Council of European Federations and its members, have set out and published the **11 Key Principles to support a harmonised European approach to Newborn Screening (NBS)** in January 2016. NBS emerges as an issue more important than ever due to new diagnostic capacities and new or future potentially transformative treatments
- EURORDIS will keep chairing the EURORDIS Newborn Screening Working Group (NBS-WG) which was set up to review current policy and practice in the field of NBS, in order to develop principles for the 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
- Following the call to action of the 11 Key Principles on NBS, with the aim to reduce inequalities across the EU, EURORDIS will continue to support the promotion of best practices in the field of NBS and, in particular, the uptake in the context of the Steering Group on Promotion and Prevention of NCDs (SGPP, an advisory body to the EC) of the Italian-extended NBS programme, following the campaign initiated by UNIAMO in 2021

#### **Shape position on early diagnosis**

EURORDIS will advocate to consolidate early diagnosis as a key area of policy action under the European Action Plan for Rare Diseases.

- Linking to the conclusions of the Technical Meeting on NBS held under the Slovenian residency of the EU Council in October 2021, the Czech Presidency committed to organise a technical meeting on Early Diagnosis for RDs in July 2022 as a part of the Bicentennial of Mendel celebration. Along with CAVO, EURORDIS will support and promote the meeting that will not only aim to harmonise practices across Europe, but also explore the State

of the Art of NBS, including genomic approaches to NBS and how to strengthen capacities of European countries

- The meeting will be the opportunity to present the results of the international RareBarometer Survey on Diagnosis for people living with rare diseases. These results will also feature at a launch event at the European Parliament in autumn, with the sponsorship of members of the network of Parliamentary Advocates for Rare Diseases
- EURORDIS will also advocate for a future European Joint Action across Member States on NBS

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

- Work with the Global Commission to broaden the outreach of the Rare Barometer Survey on Diagnosis in cooperation with Rare Disease International (RDI) (international pilot)
- Focus the Global Commission on policy – an updated analysis of diagnostic policy and services across countries in the world, an international foresight study on diagnostic trends and scenarios – and on innovative genomic, digital and organisational solutions
- Invite new corporate partners to join the Global Commission and renew the composition of the Commission

#### **Contribute and bring the patient voice in the new SCREEN4CARE project: an innovative research approach to accelerate rare disease diagnosis with genetic newborn screening and digital technologies**

- Screen4Care, a new collaborative Innovative Medicines Initiative (IMI 2 JU) European project, was launched in October 2021. It will run for a period of five years with a total budget of 25 million euros, provided by the IMI 2 JU – a joint undertaking of the European Union under Horizon 2020 and the European Federation of Pharmaceutical Industries and Associations
- Screen4Care offers an innovative research approach to accelerate rare disease diagnosis, which is based on two central pillars: genetic newborn screening and digital technologies. EURORDIS plays a major role, located at the heart of Screen4Care and is involved in all areas of focus. EURORDIS leads the Patient Advisory Board and is directing and advising all partners on the priorities, needs and perspectives of persons living with rare diseases. A Senior Project Manager was hired in September 2021 to manage this major project

#### **Participate and bring the patient voice in the EU-funded project Solve-RD, a research project aiming to solve the unsolved rare disease by sophisticated omics approaches, genetic knowledge web (genes, genomic variants and phenotypes)**

- 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 follow up of genetic counselling for patient organisations and healthcare professionals

#### **Participate in the Undiagnosed Diseases Network International**

- ✦ Continue representing the EU RD patients' perspectives and voices of 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

#### **Advocate for and contribute to the implementation of the patient rights to Cross-border healthcare Directive**

EURORDIS will continue to promote the rights of persons living with a rare disease in the framework of the implementation of the Directive 2011/24/EU ("Cross-border Healthcare Directive"). In 2022, the European Commission will finalise the evaluation of the 10-year implementation of the Directive. EURORDIS will build on its contribution to the Commission's public consultations held in 2021, which resulted from extensive consultations with EURORDIS members, to continue to stress the weaknesses of the legislation, especially of its application in Member States, and how it comes short of its ambition to improve cross-border care, in particular for people living with rare diseases. EURORDIS will also continue to propose solutions to address existing shortcomings and will promote advocacy actions with the support of its network of Parliamentary Advocates, once the Commission's evaluation will be issued and the European Parliament will respond with an Own Initiative Report.

European Reference Networks (ERNs) are a central component and output of the Cross-Border Health Care Directive. EURORDIS continues to promote the deployment of ERNs and supports the implementation of their key functions through enabling the involvement of patient representatives in the ERNs and working along ERN Coordinators, the EC and the ERN Board of Member States to ensure that ERNs respond to the needs of people living with a rare or complex condition. Though not legal ERN members, patient representatives have now been de facto accepted as members, sitting on the boards of ERNs, clinical committees and working groups. In 2017 EURORDIS set up the European Patient Advocacy Groups (ePAGs) to structure the involvement of the patient community in the ERNs. EURORDIS has worked with the ePAGs to develop the rules for patient engagement in the ERNs and to integrate the ePAGs within the ERNs governance structures. EURORDIS supports the ePAG voluntary work through training, peer-learning and exchange of good practices; providing advice and expertise to enable their direct involvement in key ERN activities including governance, the development of clinical decision support tools, research, registries, assessment, monitoring and evaluation. EURORDIS works with the wider rare disease patient community to raise awareness on ERNs at national level and advance the integration of ERNs into national health systems, liaising with experts to exchange best practices and facilitate the involvement of patient organisations in this process at local level.

As part of the contribution to shape a mature ERN system, EURORDIS contributes to the ERNs' five-year evaluation, engaging with the Joint Action on Integration of ERNs. The European Commission launched a Joint Action in 2022 to support Member States over the next 3 years in this area. Activities will include exchanges of best practice and concrete proposals, and guidelines for better integration of ERNs in the national healthcare systems, including well-defined patient pathways, referral procedures, development of national networks on rare diseases.

To be adopted in 2022, an EURORDIS Toolkit on the Integration of ERNs into National Healthcare Systems will contribute to the knowledge basis for the Joint Action by providing an overview of the relevant areas of intervention and proposing tools and processes to define concrete actions in each of them. It will thus help Member States establish, in collaboration with the national rare disease community, the legal instruments, operational processes and structures to support an effective integration of the ERNs into their national health systems.

The Toolkit will support meaningful patient engagement in the development and consolidation of the ERNs and as such, it will thus contribute to the implementation of the Cross-Border Healthcare Directive.

#### **Advocating for holistic care**

In 2022 the EC will adopt a European Care Initiative – a flagship initiative of the European Pillar for Social Rights. It will support Member States to ensure that long-term care is delivered in an effective manner, in coordination within and between health, social and community. EURORDIS will make sure that specificities of PLWRDs and complex conditions be integrated in the initiative, in collaboration with the Social Platforms of which EURORDIS is a member, and is in line with the recommendations of EURORDIS' 'Achieving Holistic and Person-Centred Care Position Paper'. Specifically, EURORDIS will contribute to the EC call for evidence launched in March 2022 and will engage with the European Parliament's Rapporteur of the Own Initiative Report at the EP, Sirpa Pietikainen, co-chair of the network of Parliamentary Advocates for RDs.

Advocating for holistic care is also a cross-cutting priority in relation with Rare2030 and the European Reference Networks on Rare Diseases.

#### **Advocate to improve access to disability rights**

With the European Commission expected to issue in 2022 – as a part of the European Disability Strategy – a guidance scheme for Member States to improve national disability assessment systems, EURORDIS will continue to engage with the European Disability forum to ensure persons with all types of disability, including persons with rare conditions, invisible disabilities or multiple impairments, are not overlooked and are provided with adequate levels of disability allowance, social protection schemes, community services and independent living arrangements.

Based on existing tools (notably, Orphanet's functional consequences tool and disability factsheets), EURORDIS will build on the explicit recognition of RD specificities in the EU Disability Strategy and advocate to ensure that the scheme will single out and share best practices across EU Member States, define common standards for Member States voluntary uptake to ensure that the disabilities experienced by PLWRD are duly assessed, recognised and supported with adequate compensation measures.

The work of RDI on Commonalities and Specificities between Living with a RD and Living with disabilities will also be used.

#### **Advocate for a European Health Data Space (EHDS) enabling the secure sharing and use of data at EU level**

After the adoption of the EC proposal for an EU Regulation on EHDS, EURORDIS will engage with its members to contribute to the legislative process around EHDS and voice preferences and needs of the RD community. EURORDIS will build on existing work to single out rare diseases as exemplary use cases in the EHDS. Actions will include:

- ✦ An information webinar addressed to all members to present and discuss: the specificities of RDs in the EHDS (why RDs is a use case); the status quo and the position of the RD community (Rare2030 recommendations; EURORDIS contribution to the EC EHDS public consultation; survey on patients' preference in data sharing and peer reviewed article, etc.); the EC proposal and its relevance for people with RDs.
- ✦ Gathering input and feedback on the EHDS Regulation draft proposal from members of EURORDIS, including EURORDIS Digital Advisory Group and selected ePAGs; drafting a position paper
- ✦ Targeted consultation of the broad membership on the draft EURORDIS position
- ✦ Dissemination and communication on the position to relevant Member of the European Parliament and Member States in the context of the legislative procedure

#### **Advocate for the implementation of EU pharma regulatory and policy framework**

- ✦ Support the implementation of the HTA newly adopted Regulation through responses to

methodological consultations and on patient and medical experts engagement throughout 2022

- ✦ Prepare the ground in view of the adoption of the proposals for legislation on medicines for children and rare diseases (medicines for special populations), as well as the revision of the General Pharmaceutical Legislation, expected by the end of 2022, within the framework of the Pharmaceutical Package. Building on the position developed by EURORDIS and members over the years and lately captured in the contributions to the 2021 EC public consultations, consolidate key messages, engage with members and partner with key stakeholders as appropriate (EFPIA, EUCOPE, EuropaBio etc.)
- ✦ With the support of the French Presidency of the EU Council, continue the advocacy for a structured collaboration between Member States to improve access and better use of public resources, along with the establishment of EU Fund for co-financing the generation of post-marketing authorisation evidence for very rare diseases during the years following approval
- ✦ Continue the structured dialogue with EFPIA with the potential adoption of a Joint Statement on proposals for improving equitable access, as well as developing a feasibility study on the EU Fund for Real World Evidence (RWE). On the latter, follow up closely the implementation of the RWE4Decisions initiative
- ✦ Continue engagement within the Expert Group on Orphan Drug Incentives, with development of cross-cutting proposals on incentives to evolve the potential solutions to included in the forthcoming legislative proposal on therapies for rare diseases
- ✦ Explore action toward common European procurement and purchase of very low prevalence diseases, highly complex and costly treatments

## 3.2 EMPOWER

### **Support Patient Engagement in European Reference Networks (ERNs) and European Patient Advocacy Groups (ePAGs)**

- ✦ **Support European Patient Advocacy Groups (ePAGs)** to enable effective and meaningful involvement of patients in the operational and strategic activities of the ERNs as well as continuous learning and improvement of methods and tools used to conduct patient engagement in the ERNs. EURORDIS will work on 3 areas:
  - ✦ **Good governance and representativeness.** The EURORDIS team will oversee and support ePAGs and ERNs to develop and implement common rules and policies for patient engagement and supporting the recruitment of ePAG advocates to ensure optimal patient representation in all ERNs
  - ✦ **Conducting patient engagement:** steer and manage the ePAG Steering Committee and other transversal ePAG working groups (on

research and registries, assessment, monitoring and evaluation, communication, clinical practice guidelines and education & training); regular calls and webinars to support ePAG advocates' effective involvement across the different areas

- ✦ **Developing tools to enable engagement of persons with own experience of living with a rare disease in the ERNs:** Building on the experience and best practice gathered during the first cycle of ERNs, EURORDIS will develop: a practical guide on how to involve patients and patient representatives in clinical practice guidelines development and a practical guide on how to capture individual needs through the journey of a person who lives with a disease. Both guides will be developed as chapters of a larger toolkit to facilitate patient engagement in the ERNs, that EURORDIS plans to further develop and expand in the next years with additional guidance on different topics

- Finalise and publish a **toolkit on good practices to facilitate the integration of ERNs into national health systems**, building on the 2021 exchange on good practices complemented by desk research
- Organisation of the Annual all-ePAG meeting (online)**. All 300 ePAG advocates will be invited to participate along with ERN clinicians and project managers
- Launch of a pilot programme on coaching to improve patient-clinician partnership (M5)** based on the need to further improve the collaboration of patient representatives and clinicians identified in 2021. The pilot will deliver online coaching for a minimum of approximately 20 ePAG advocates and clinicians in 2 ERNs. It will combine soft skills training and coaching sessions. This pilot aims to show the value of developing a mutual understanding of partnership and shared leadership in the context of the ERNs to enable the Networks to effectively incorporate the experience and needs of the patient community in their strategic planning and operations.

## Support patient engagement in therapeutic development

### Support actively the patient engagement in EMA activities

- Bring our own knowledge and expertise developed with the patient community to the regulatory community and continue to support patient advocates to participate in a broad range of regulatory activities
- Representation of EURORDIS at the EMA as member of the Patients' and Consumers' Working Party (PCWP)
  - Work with International Council for Harmonisation ICH on E6 and E8 guidelines: on clinical trials and patient engagement in clinical trials
- Identification of and support to RD patients participating in EMA activities and scientific committees (COMP, CAT, PDCO, PRAC, SAWP, HMPC, CHMP) by supporting the work of permanent patient representatives (staff or volunteers) and the participation of ad hoc experts in the COMP, PDCO, CAT, PRAC, HMPC and CHMP; identifying & selecting patient representatives to be appointed to EMA Committees; supporting participation of ad hoc patient representatives in Protocol Assistance/Scientific Advice (SAWP), EMA/HTA Parallel Scientific Advice and Scientific Advisory Group meetings; Participation to EMA transversal activities within several working groups of the Scientific Committees (e.g. Cross-Committee Taskforce on Registries), EMA/HMA task force on Availability and its Concept Paper on the Prevention of Shortages, and to the EU regulatory network Strategy 2020-2025. When applicable: COMP (3 days/month), PDCO (4 days/month), CAT (2 days/month) and PRAC (4 days/month)
- Participation in other EMA meetings, including the Strategic Review and Learning Meetings organised in the context of the EU presidency, stakeholder forums and also in scientific committees consultations/ workshops

- Participate in the EMA Committee for Orphan Medicinal Products (COMP), including the activities aiming at giving guidance to the European Commission on matters related to the revision of the orphan regulation
- Collaborate with EMA to identify and support the participation of patients in Protocol Assistance dossiers at the Scientific Advice Working Party (SAWP)
- Coordination and animation of the EURORDIS Therapeutic Action Group (TAG), which consists of EURORDIS' and the rare disease community representatives at EMA committees and Working Parties (WPs) 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
- Monthly therapeutic reports, disseminated to patient representatives participating at the EMA, the EURORDIS TAG, the EURORDIS BoD, the European Public Affairs Committee (EPAC), the EURORDIS Summer School Alumni, in addition to the EURORDIS members at large via the member news

### Actively support the patient engagement in dialogue with payers: MoCA

- Mechanism of Coordinated Access to Orphan Medicinal products: Identification of & support to RD patients participating in MoCA

### Support patient involvement in European HTA Network, EUnetHTA JA 3, related HTA activities and through the EURORDIS HTA Task Force (HTA TF) - Contributing to the implementation of EU HTA Regulation (2021/2282)

Patient representatives are increasingly involved as partners of scientific research, in recognition of them being 'expert by experience'. Patient engagement in regulatory and HTA process is becoming mainstream practice, with the growing recognition of the patient reported outcomes as decisive in clinical development and assessment processes. Yet, the growing complexity of this landscape requires that patients build solid capacities in the scientific, ethic and regulatory fields, in order for them to be fully fledged partners

- Representing patients in the stakeholder bodies of the European HTA Cooperation contributing to the development and implementation of patient engagement rules and procedures (by EUnetHTA 21 and European Commission), responding to public consultations (especially from EUnetHTA21, in charge of methodological guidelines and procedural guidance) participating in public conferences for the Regulation implementation and on HTA at large
- Inform and engage our Members about the HTA Commission's initiatives (consultations, studies launched, calls for projects/tenders, and/or events)
- Management, animation and coordination of the EURORDIS HTA Task Force (12 members)



### **Support patient involvement in quality information on medicines through the EURORDIS Drug Information Transparency & Access Task Force (DITA TF) - Contributing to the EU Pharma Access strategy, to the STAMP Working Group on Repurposing and DITA Task Force**

EURORDIS supports patient involvement in quality information on medicines and access issues through:

- ✦ Management, coordination and animation of the EURORDIS Drug Information Transparency & Access Task Force (DITA TF) "Consists of 18 persons including 16 volunteers and 2 EURORDIS staff"
- ✦ Organise one annual F2F meeting with regular online meetings throughout the year

- ✦ Regular consultations and virtual meetings will take place. The DITA TF work plan is aligned on the one of the PCWP at EMA
- ✦ The review of public information on medicines (Medicines Overviews, package leaflets, Urgent Communication, Public Summary of Opinions)
- ✦ Work on repurposing of medicines will continue as member of the Repurposing Observatory Group (RepOG) (established by STAMP) and will help informing and engaging patients in repurposing scientific advice at EMA level & at national level

## **3.3 PARTNER**

### **Organise and produce the ECRD 2022 online**

- ✦ The European Conference on Rare Disease (ECRD) will take place at the end of June and will gather over 1000 participants online across a week. All types of stakeholders from the Rare Disease community will be participating to discuss various important topics related to the Rare Disease Policy

- ✦ ECRD coordination, organisation, production and follow-up, working with the ECRD Programme Committee on the 3 main policy tracks for ECRD202 (Ensuring healthy lives and promoting well-being for all people living with a rare disease at all ages; Building resilient infrastructure, promoting inclusive and sustainable industry and fostering innovation for people living with a rare disease; Reduce inequality within and among countries by focusing on equity for people living with a rare disease)

# **STRATEGIC OBJECTIVE 4: INCLUSIVE OF ALL RARE DISEASES, ALL REGIONS, TO "LEAVE NO ONE BEHIND"**

## **4.1 ADVOCATE, EMPOWER, PARTNER**

### **Current humanitarian crisis in Ukraine**

#### **Uniting for the two million Ukrainians living with a rare disease in and out of Ukraine**

EURORDIS' response is two-fold: both immediately responding to the needs of people living with rare diseases and advocating on behalf of their specific needs with the support of our multistakeholder networks.

- ✦ Developing and curating information to develop Decision Support Tools that help Ukrainians living with rare diseases (LWRD) navigate the different health and social care policies available across EUROPE and how they can access care
- ✦ Facilitating (where and if possible) solutions for getting Medicines into Ukraine and vulnerable people out.
- ✦ Mobilising our network of patient organisations, companies, European Parliamentarians and external stakeholders by sharing information on the needs of vulnerable people and best practices for addressing key challenges.

- ✦ Identifying volunteers who can be deployed by EURORDIS and our partner organisations to help individuals to navigate care pathways outside of Ukraine or to translate medical records (among others)
- ✦ Managing individual requests from patients either by helping them find appropriate organisations or by problem solving as best as we can
- ✦ 'Rare BNB': where we identify families in Europe who understand rare diseases and are happy to host a patient family coming from Ukraine and match these with people looking to leave Ukraine or to leave border countries, especially Poland, where facilities to treat patients with complex needs are already under strain
- ✦ Advocating on behalf of Ukrainians living with a rare disease through the media, parliamentary networks, and direct outreach to governmental and non-governmental organisations, departments and institutions in Ukraine and in Europe. (For clarity, this position links with advocacy teams, provides them with evidence, but does not lead the advocacy work)



### **Raise Awareness at EU and international level: Rare Disease Day 2022 campaign and preparation of Rare Disease Day 2023 campaign**

Since its launch in 2008, RDD plays a critical part in building an international rare disease community that is multi-disease, global, and diverse – but united in purpose. In 2022, we aim to:

- ✦ Coordinate the international Rare Disease Day 2022 & plan RDD 2023 in over 100 countries; focus the campaign on reframing the basic key messages about rare diseases: "Rare is many. Rare is strong. Rare is proud". 5% of the population, rare is 6000 diseases
- ✦ Extend the 2022 campaign throughout the year with social media and stories from the global campaign
- ✦ Co-create and produce Rare Disease Day 2022 campaign materials (visuals, website update, video) building on strategic review recommendations and a call to action
- ✦ Translations of the media assets and webinars into 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

### **Foster the inclusion of rare cancers across EURORDIS' advocacy transversal activities**

- ✦ Persons living with a rare cancer are included in all our evidence based analysis and proposals in advocacy
- ✦ Strengthen our network of 70+ member patient organisations in rare cancers
- ✦ Support the 4 ERNs on rare cancers and the engagement of patient representatives through ePAGs
- ✦ Foster research projects for rare cancers
- ✦ Support drug development and access for rare cancers, which represents about 40% of all orphan designations, risk/benefit assessment, HTA assessments and early dialogue with payers, that EURORDIS supports
- ✦ Rare Barometer surveys
- ✦ International Rare Disease Day – promote the inclusion of rare cancers and encourage rare cancer

patient groups to use that day to shine a light on their disease, needs and what they have achieved

- ✦ Communication on EURORDIS' activities for rare cancers
- ✦ Links with the European Parliament Group on Rare Diseases (some of them were also members of the European Parliament's Committee on Beating Cancer (BECA))

### **Promote rare diseases as an international policy priority through:**

#### **Strategic Partnerships (MoUs) with international patient organisations and stakeholders**

- ✦ Maintain partnerships with international organisations and review the MoUs as needed, in addition to being open to new partnerships with other international patient organisations
- ✦ Work towards the implementation in Europe of the UN Resolution addressing the needs of PLWRD, its impact on Europe's Action Plan as well as on National Plans and Strategies for Rare Diseases
- ✦ Promote rare diseases as an international public health priority in partnership with Rare Diseases International. In 2022, EURORDIS will continue to promote Rare Diseases as a European/international integrated health priority through Rare Diseases International (RDI) (MoU signed in 2018) and with the support of other partner major international rare disease patient groups such as NORD (USA), CORD (Canada), JPA (Japan), Rare Voices Australia (RVA), the Rare Diseases Foundation of Iran (RADOIR)
- ✦ Promote Rare 2030 recommendations towards WHO Europe with a focus on non-EU countries, engage the dialogue on networking of healthcare providers outside the EU, contribute to the WHO Europe Oslo Initiative (OMI) on pricing scheme and funding models to improve access to rare diseases therapies in Europe
- ✦ WHO (Geneva) contributes as a knowledge-partner in RDI to the deliverables of the MoU WHO-RDI, in particular on: the definition framework of rare diseases, the collaborative global network for rare diseases, the essential lists of medicines, medicines for children and in-vitro diagnostic, report on these activities to relevant European Commission DGs, participate to the side events of the World Health Assembly, May 2022.

## CROSS-CUTTING PRIORITIES: RELEVANT TO ALL OUR STRATEGIC OBJECTIVES

### 5.1 Communication and dissemination

#### Design a new Strategy for Communication and Dissemination by networks

- Finalise the EURORDIS new Communication strategy and begin its implementation. The Communication strategy will elaborate on the target audiences, channels, tools and messages for each network to improve outreach

#### Design and implementation of Communication and Dissemination tools and activities

This task includes the design of C&D tools as well as to maintain, regularly evaluate, update and to implement C&D activities as defined in the C&D strategy.

It includes:

- Finalise the technical development of the new eurordis.org website providing a clearer and easier navigation for users to find the information they need in our wide range of information and make it more responsive for personal devices e.g. smartphones and tablets, for a launch within the first semester 2022
- Monthly eNews produced and disseminated to 11,000 subscribers (none in August)

- Twice-monthly Member News produced (designed), disseminated to the over 988 EURORDIS members (none in August). It consists of a news report tailored specifically to the needs and interests of EURORDIS' membership
- Monthly National Alliance newsletters will be produced, offering information specific to national policy and opportunities or activities which involve National Alliances
- Management of EURORDIS online communication tools including social media (Facebook, Twitter, LinkedIn, Flickr, YouTube, Instagram), streamlined with the EURORDIS website
- Production and dissemination of reference documents, which include the Activity Report 2021 & Action Plan 2022, Factsheets, infographics, Brochures
- Maintain and cultivate relations with media covering the policies of the European Union
- Translate activities into the 7 official EURORDIS languages (EN, FR, DE, IT, ES, PT, RU) on the website (all 7), Member News (6 languages, excluding Russian), and a number of other languages on an ad hoc basis (approximately 30 for RDD video)







## 5.2 Governance

### EURORDIS Strategy 2021-2030

- ✦ Disseminate the new EURORDIS long-term strategy 2021-2030
- ✦ Refine the EURORDIS Strategic Objectives 2021-2030, develop operational criteria to prioritise activities contributing to reach these objectives, within the EURORDIS long term strategic goals
- ✦ Define the Strategic Alignment Framework to adapt the capabilities and organisation of EURORDIS to deliver on its strategic objectives over the next years

### Manage the Governance bodies, maintain the EURORDIS By-laws

- ✦ Regularly review and update the EURORDIS internal governance documents that comprise the EURORDIS by-laws.
- ✦ Conduct at least three Board of Directors (BoD) Meetings (in March, July and November) and four Board of Officers Meetings
- ✦ Ensure two-ways communication between Board and staff.
- ✦ Organise the Annual General Assembly online, gathering all Members in May 2022

## 5.3 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
- ✦ Finalise, adopt and implement sustainability plans for EURORDIS' major programmes
- ✦ Invest in developing strategic projects proactively and seek opportunities to finance them

- ✦ Companies, EMA policy on the handling of competing interests of scientific committee members and experts and the EU4Health rules
- ✦ Pursue opportunities to deliver new sources of diversified income from foundations; developing the activities of the **Head of Philanthropy & Partnerships to take forward work with Donors and Foundations**
- ✦ **Focus team efforts, build skills and allocate resources to innovative project development in 2022**

## 5.4 EURORDIS events

### Organisation of the following events

- ✦ **EURORDIS Black Pearl Awards** in February 2022 as an online event
- ✦ **ECRD 2022** fully online
- ✦ **Two CNA workshops and one CEF workshop**
- ✦ **Two major workshops of the ERTC** with relevant stakeholders

## 5.5 Human resources

### EURORDIS Volunteers

- ◆ Improve **volunteers' visibility** in EURORDIS' communication and acknowledgements on the EURORDIS website
- ◆ **Revise processes for effective and improved volunteer support and management**
- ◆ Assessment of volunteers' activities and reorganisation of the support provided to volunteers after the covid19 long period of solely remote work

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

- ◆ Enhance the induction and the integration of new staff members following the covid19 social distancing resulting in a period of high turn over
- ◆ Enhance new management team structure with an improved internal coordination meetings structure (core leadership meetings, operations meetings, management team meetings, unit team meetings, advocacy and strategy meetings, programmes' steering committee 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
- ◆ Prepare actions toward an integrated information system across the staff and volunteers
- ◆ Develop a Business Continuity & Contingency Plan for the Finance & Support Function
- ◆ Develop Procedures and Tools to safeguard the organisation and enhance quality of processes

### EURORDIS Staff

EURORDIS's quality **team consists of 50+ staff** with a very high and diverse level of expertise, qualifications, know-how, skills, interdisciplinarity, multi-linguism (over 15) and international profiles. Many of the staff have been EURORDIS' team members for several years, with deep knowledge, experience, and professional networks of people in the community an essential asset in an organisation representing persons with complex needs bringing together such a wide range of stakeholders.

- ◆ Support the emergence of a group of team members, forming a middle management having a 360 degree view of the organisation in order to enhance transversal work strategically, streamline operations, optimise time and fund allocation
- ◆ Organisation of an All-staff Team Seminar with +55 staff members in Lisbon in May 2022 to enhance team building, cooperation and cohesion between the different units

In 2022, EURORDIS will continue to adapt its staff members, staff organisation and fulfil the necessary positions within the team according to the evolution of its strategy and operational needs:

### Appointment in 2022 of following permanent positions – succession or new

- ◆ Research and Policy Initiatives Director (based in Paris), succession to Scientific Director
- ◆ Research Senior Manager Drug Repositioning (based in Barcelona), research project
- ◆ Patient engagement and Training in Drug Repositioning (based Barcelona), research project
- ◆ Patient Data Director (based in Brussels), new position
- ◆ Governance Manager (succession to Deputy to the CEO) (based in Paris)
- ◆ Rare Disease Day Senior Manager (based in Paris), succession to RDD Senior Manager
- ◆ Communications Junior Manager (based in Paris), new position
- ◆ Communications Manager (based in Paris), succession to previous Communications Manager which has been promoted to Communications Senior Manager, in place of Communications Director
- ◆ European Public Affairs Senior Manager (based in Brussels), renewed position after disruption

### Temporary positions in 2022 linked to a specific mission

- ◆ Senior Project Manager to manage EURORDIS Ukraine response



# TEAM CHART 2022



PARIS

BRUSSELS

BARCELONA

OTHER

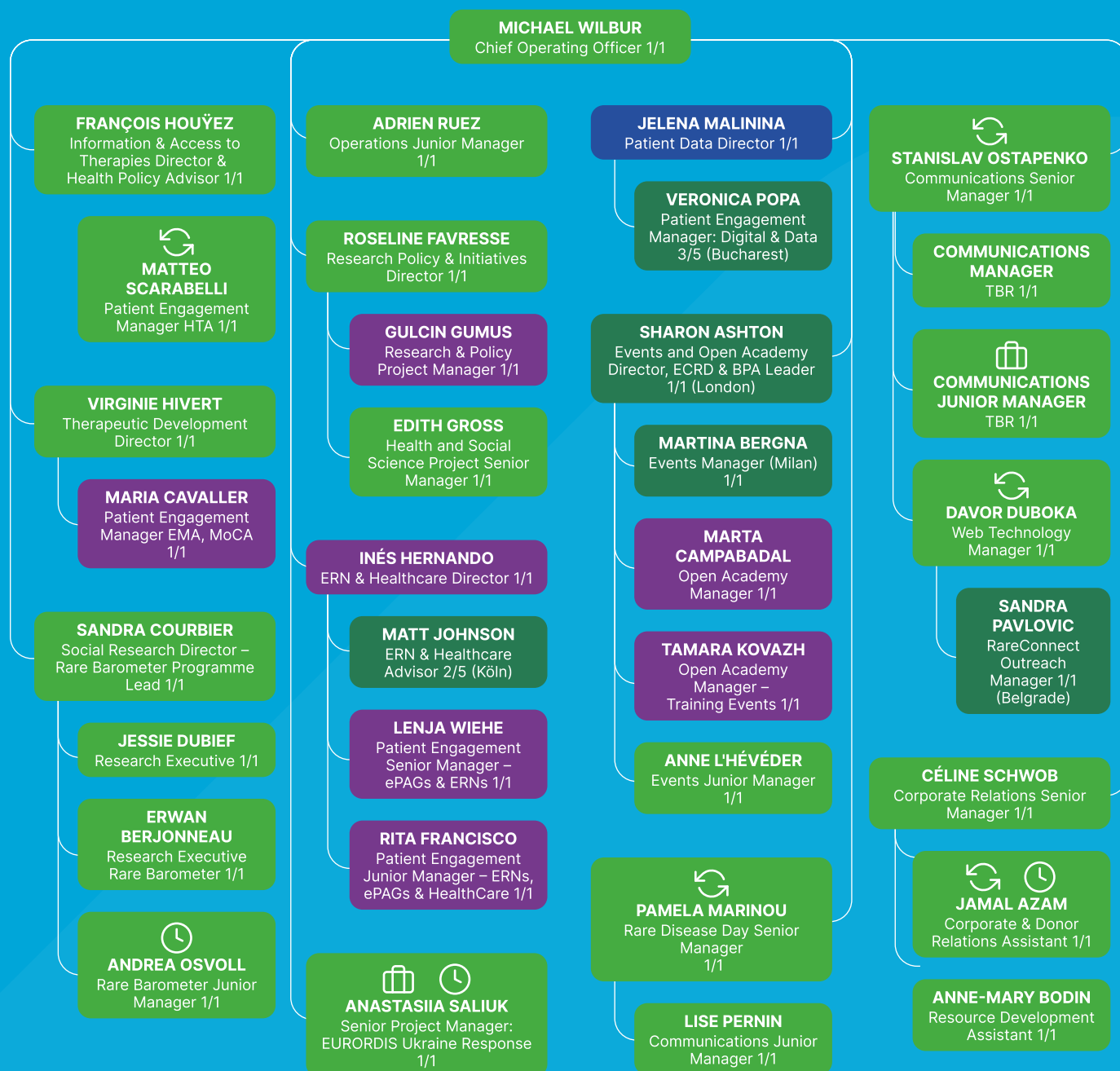
 Replacement

 Job creation

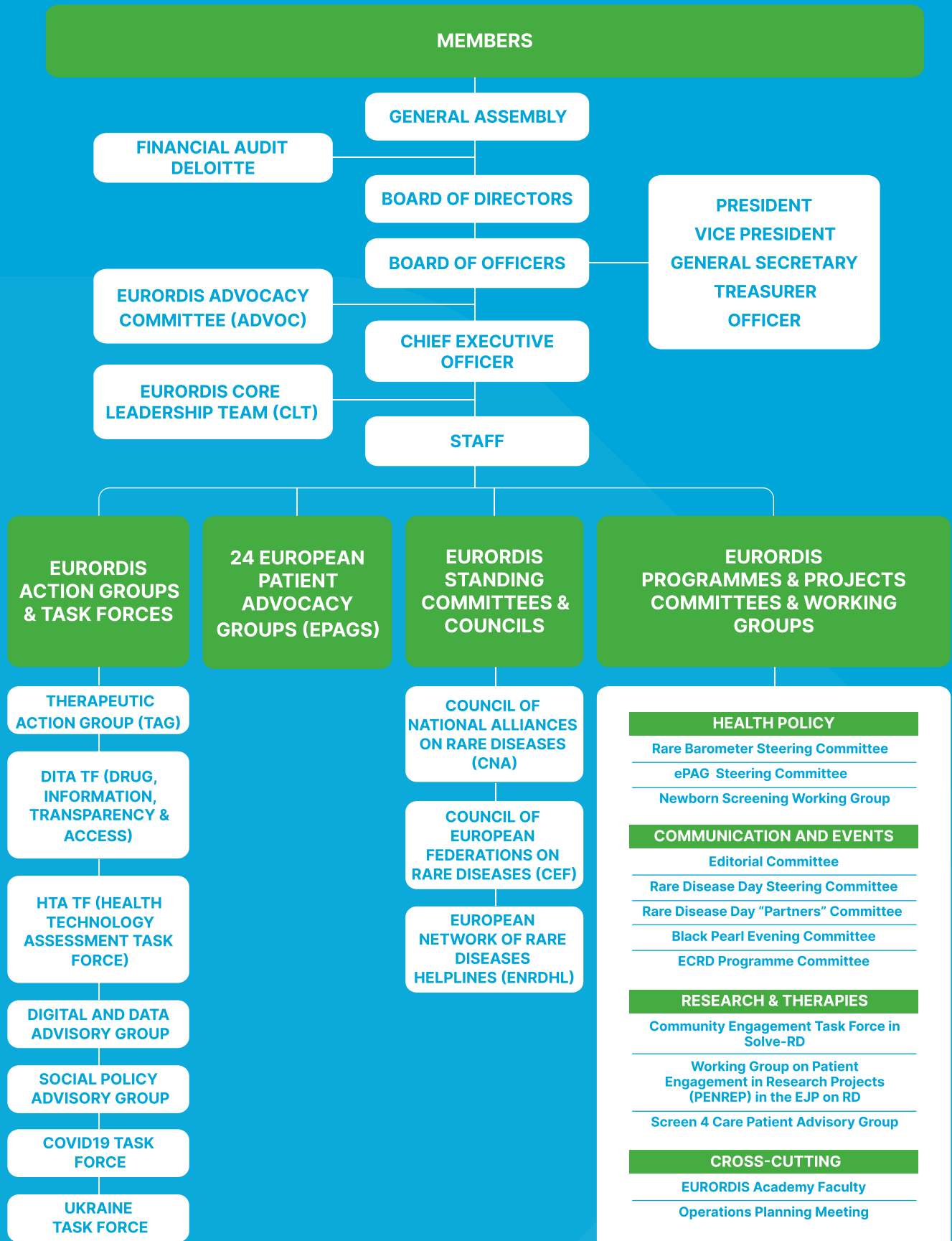
 Temporary contract

 Job evolution





# GOVERNANCE CHART 2022









# EXTERNAL REPRESENTATION CHART 2022



## EUROPEAN NOT-FOR-PROFIT ORGANISATIONS & INITIATIVES

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 Cancers Europe

WECAN: Workgroup of European Cancer Patient Advocacy Networks

European Cancer Organisation Patient Advisory Committee

Social Platform – The European Platform of European Social NGOs

Maladies Rares Info Service (French Helpline for RDs)

Rare Disease Platform in Paris

Get Real Institute

PFMD - Patient Focused Medicines Development Initiative

EJP RD – European Joint Programme on Rare Diseases

EU4Health Civil Society Alliance

## MEMBER OF EUROPEAN NETWORKS

BBMRI Stakeholders Forum

RD-Connect

HTx, Next Generation HTA

OpenMedicine

PARADIGM

C4C (Connect 4 Children)

CORBEL – MIUF

Solve – RD

reCOVID consortium IMI2

Rare 2030

## PARTNERSHIP WITH LEARNED SOCIETIES

European Federation of Internal Medicine (EFIM)

European Hospital & Healthcare Federation (HOPE)

International Federation of Social Workers Europe

European Society of Human Genetics (ESHG)

International Society for Pharmacoeconomics and Outcomes Research (ISPOR)

European Connected Health Alliance – ECHAlliance

European Union of Medical Specialists – EUMS

European Alliance for Personalised Medicine

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

RDI: Rare Diseases International

NGO Committee for Rare Diseases (United Nations, New York)

IRDiRC: International Rare Disease Research Consortium

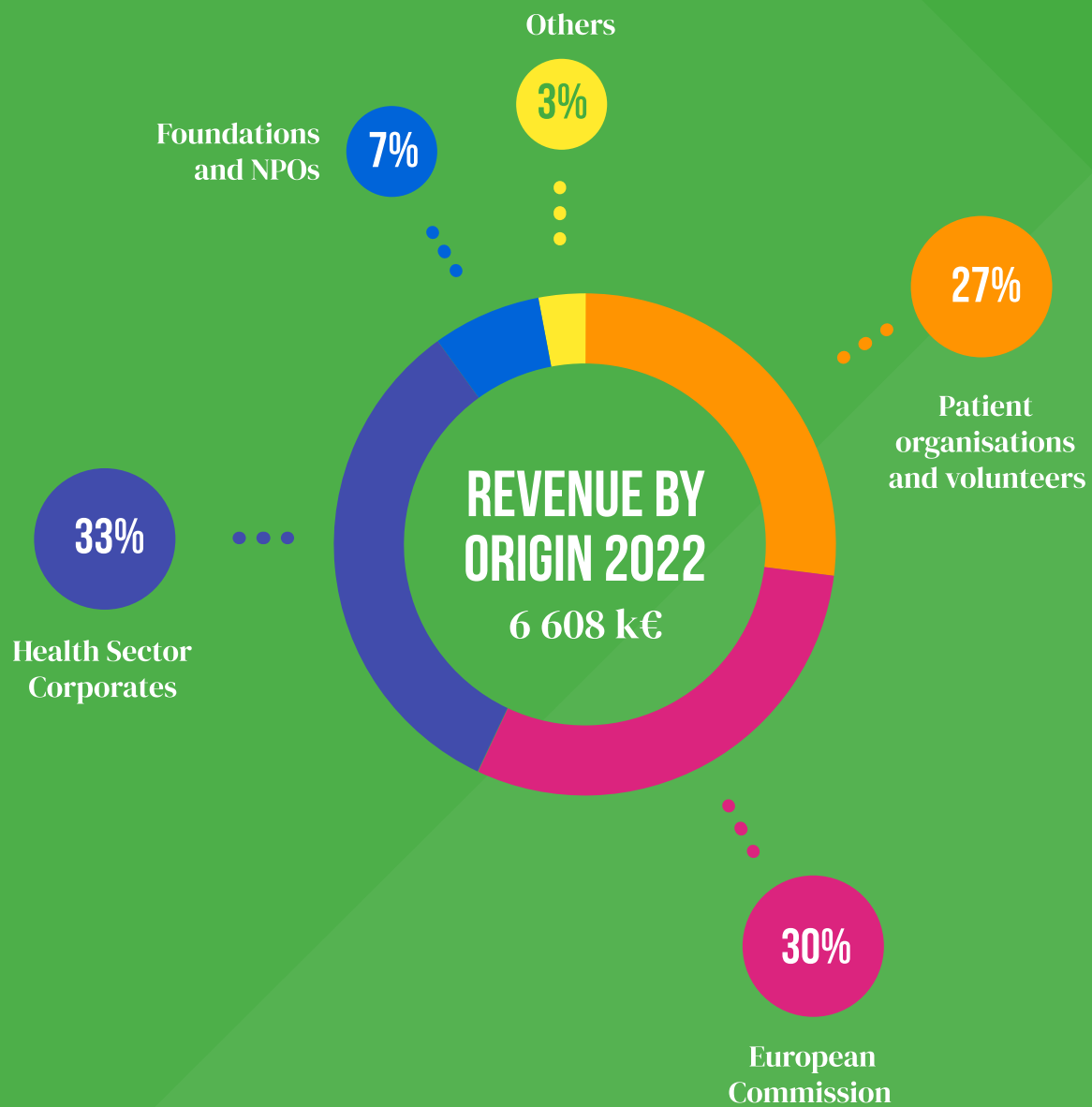
Global Commission to end the diagnostic odyssey for children

NEWDIGS: New Drug Development ParadIGMs

IAPO: International Alliance of Patients' Organisations

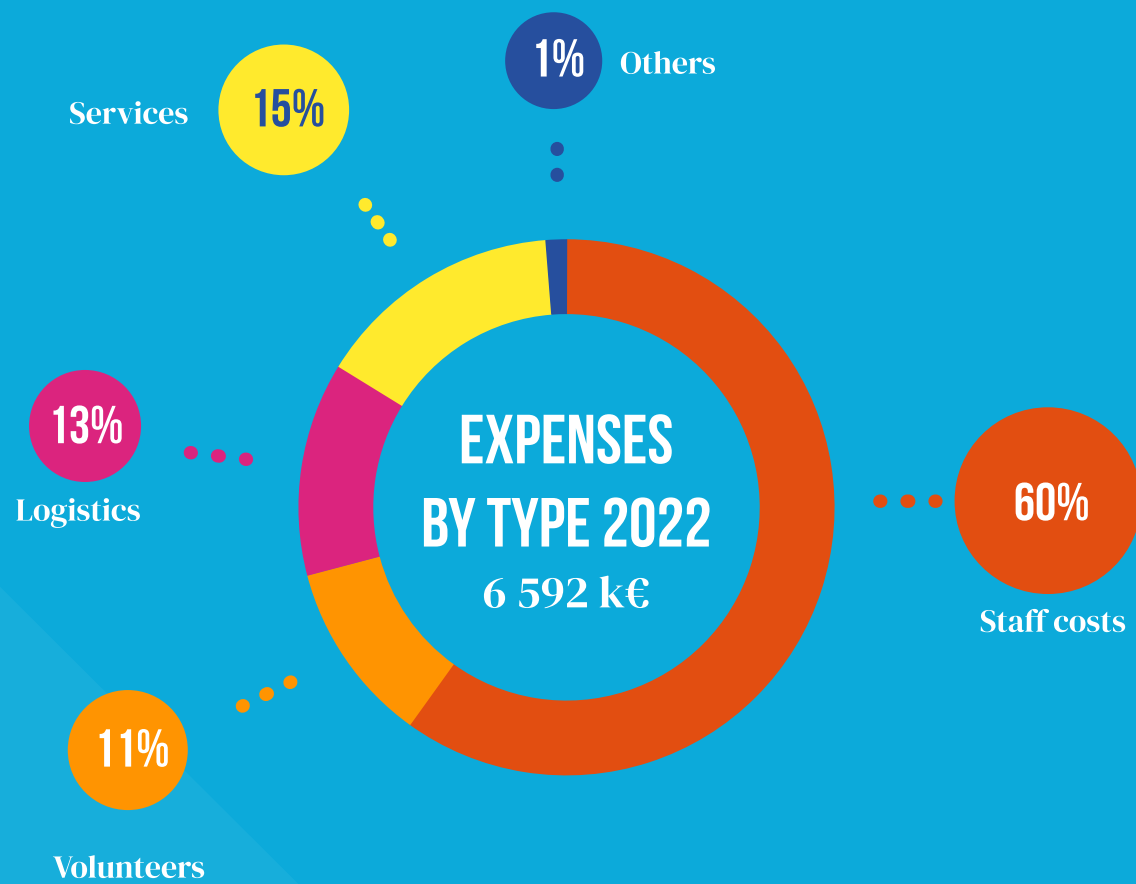
International partnerships (MoUs): NORD (USA), CORD (Canada), JPA (Japan), RVA (Australia), CORD (China), RADOIR (Iran)

# REVENUE 2022





# EXPENSES 2022



# ACRONYMS & DEFINITIONS

## EURORDIS INTERNAL COMMITTEES & TASK FORCES

<b>BoD</b>	Board of Directors (of EURORDIS)
<b>BoO</b>	Board of Officers (of EURORDIS)
<b>NA</b>	National Alliance (of Rare Disease Patient Associations)
<b>CEF</b>	Council of European Federations of Rare Diseases
<b>CNA</b>	Council of National Alliances (of Rare Disease Patient Associations)
<b>DITA</b>	Drug, Information, Transparency & Access (Task Force of EURORDIS)
<b>EPAC</b>	European Public Affairs Committee (includes current and some former Board members, and EURORDIS managers involved in advocacy)
<b>SPAG</b>	Social Policy Action Group (Task Force of EURORDIS)
<b>TAG</b>	Therapeutic Action Group (of EURORDIS) - Brings together EURORDIS' representatives (mainly volunteers) in EMA scientific committees
<b>DAG</b>	Digital and Data Advisory Group (of EURORDIS)

## EURORDIS PROJECTS/INITIATIVES OR IN WHICH EURORDIS IS INVOLVED

<b>ERTC</b>	EURORDIS Round Table of Companies (with pharma & biotech developing Orphan Medicinal Products)
<b>OA</b>	Open Academy (comprised of ESS, EWS, ELS, EDS)
<b>EURORDIS Summer School (ESS)</b>	4 day training on clinical trials for beginners. Since 2008, it has taken place each year in Barcelona, Spain.
<b>EURORDIS Winter School (EWS)</b>	4 day training on research
<b>EURORDIS Leadership School (ELS)</b>	3 day training on leadership
<b>EURORDIS Digital School (EDS)</b>	online training
<b>ePAG</b>	European Patient Advocacy Group
<b>ECRD</b>	European Conference on Rare Diseases and Orphan Products
<b>NGO Committee for Rare Diseases</b>	The NGO Committee for Rare Diseases engages at the UN level to elevate rare diseases to a priority within global public health
<b>RDD</b>	Rare Disease Day
<b>RDI</b>	Rare Diseases International
<b>CAB</b>	EURORDIS Community Advisory Board Programme
<b>EMM</b>	EURORDIS Membership Meeting

<b>AGA</b>	Annual General Assembly
<b>BPA</b>	Black Pearl Awards
<b>RDW</b>	Rare Disease Week
<b>RareConnect</b>	A safe, easy to use platform where rare disease patients, families and patient organisations can develop online communities and conversations across continents and languages
<b>Rare Barometer</b>	Rare Barometer is a community of people living with a rare disease who are willing to participate in EURORDIS-Rare Diseases Europe surveys and studies
<b>Rare 2030</b>	Rare 2030 is a foresight study that gathers 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.
<b>BBMRI Stakeholders' Forum</b>	Biobanking and Biomolecular Resources Research Infrastructure
<b>ECRIN</b>	European Clinical Research Infrastructures Network
<b>E-Rare</b>	Network of ten partners – public bodies, ministries and research management organisations – from eight countries, responsible for the development and management of national/ regional research programs on rare diseases
<b>EUnetHTA Forum</b>	Support effective HTA collaboration in Europe that brings added value at the European, national and regional levels
<b>EuroBioBank</b>	European Network of DNA, cell and tissue banks for rare diseases – EU project 2003 – 2006 for which EURORDIS was administrative coordinator. It is now the biobank network of RD-Connect
<b>EUROPLAN</b>	Fostering National Plans in Europe (project ended in 2018)
<b>EUPATI</b>	Innovative Medicines Initiatives Joint Undertaking "Fostering Patient Awareness on Pharmaceutical Innovation"
<b>EJA</b>	Joint Action on Rare Diseases of the EU Committee of Experts on Rare Diseases: Funded by EC and by Member States, divided in work packages corresponding to specific activities, e.g. continuity of Europlan (Work Package 4); developing guidelines for social services dedicated to RDs (Work Package 6)
<b>FACILITATE</b>	The project is focused on GDPR compliance in the context of clinical trials, and will look to find a way referencing previously completed trials to facilitate data sharing in a way that respects the needs and wishes of patients and adds value to all stakeholders
<b>Global Commission</b>	The Global Commission To End The Diagnostic Odyssey For Children With A Rare Disease is a group of multidisciplinary experts with the goal of achieving timely diagnosis for all rare diseases, especially those affecting children
<b>GCOF</b>	Genetic Clinics of the Future: To map the opportunities and challenges that surround the clinical implementation of next generation sequencing technologies, Horizon 2020, 2015-2017
<b>HTx Project</b>	HTx is a project funded by the European Union which aims to take this to the next level
<b>InnovCare</b>	Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions, DG Employment and Social Innovation (EaSI), 2015-2018
<b>IMI</b>	Innovative Medicines Initiative
<b>IRDIRC</b>	International Rare Disease Research Consortium
<b>PARADIGM</b>	PARADIGM's mission is to provide a unique framework that enables structured, effective, meaningful, ethical, innovative, and sustainable patient engagement (PE)
<b>SCOPE</b>	The Strengthening Collaboration for Operating Pharmacovigilance in Europe (SCOPE) Joint Action
<b>SCREEN4CARE</b>	Its goal is to shorten the time to diagnosis and treatment for patients with a rare disease using a dual strategy based on NBS and AI tools
<b>SOLVE-RD</b>	"Solve-RD - solving the unsolved rare diseases" is a research project funded by the European Commission for five years (2018-2022)



<b>TREAT-NMD</b>	Translational Research in Europe – Assessment and Treatment of Neuromuscular diseases
<b>Web-RADR</b>	Development of tools for patients and healthcare professionals to report suspected adverse drug reactions to national EU regulators, Innovative Medicines Initiative (IMI), 2014-2017
<b>RD-Action</b>	Joint Action to expand and consolidate the achievements of the former EUCERD JA, DG Sanco, 2015-2018
<b>EJP RD</b>	European Joint Programme for Rare Diseases

## EURORDIS & EUROPEAN REGULATORY NETWORK

<b>CAT</b>	Committee for Advanced Therapies
<b>CHMP</b>	Committee for Human Medicinal Products
<b>SAG</b>	Scientific Advisory Group at the Committee for Human Medicinal Products
<b>COMP</b>	Committee of Orphan Medicinal Products
<b>EMA</b>	European Medicines Agency
<b>HMA</b>	Heads of Medicines Agencies
<b>PCWP</b>	Patients and Consumers Working Party
<b>PDCO</b>	Paediatric Drugs Committee
<b>PRAC</b>	Pharmacovigilance and Risk Assessment Committee
<b>SAWP</b>	Scientific Advice Working Party
<b>EPAR</b>	European Public Assessment Report
<b>SPC / SmPC</b>	Summary of Product Characteristics
<b>MA</b>	Marketing authorisation

## EUROPEAN COMMISSION

<b>EP</b>	European Parliament
<b>EC</b>	European Commission
<b>MEP</b>	Member of European Parliament
<b>HaDEA</b>	European Health and Digital Executive Agency
<b>DG GROW</b>	Directorate General for Internal Market, Industry, Entrepreneurship and SMEs
<b>DG SANTE</b>	Directorate General for Health and Food Safety
<b>DG RTD</b>	Directorate General for Research and Innovation
<b>JRC</b>	Joint Research Centre of the European Commission (based in Ispra, Italy)
<b>CEGCC</b>	Commission Expert Group on Cancer Control
<b>CEGRD</b>	Commission Experts Group on Rare Diseases – 8 patients' representatives included 2 representatives of EURORDIS and 2 Observers
<b>EU HPF</b>	EU Health Policy Forum
<b>EU HPP</b>	EU Health Policy Platform

## NON GOVERNMENTAL PARTNERS

<b>AFM-Téléthon</b>	French Muscular Dystrophy Association
<b>DIA</b>	Drug Information Association
<b>CORD</b>	Canadian Organisation for Rare Disorders / Chinese Organisation for Rare Disorders
<b>EFPIA</b>	European Federation of Pharmaceutical Industries and Associations
<b>EPF</b>	European Patients' Forum
<b>EPPOSI</b>	European Platform for Patients' Organisations, Science and Industry
<b>EuropaBio</b>	The European Association for Bioindustries
<b>EUCOPE</b>	European Confederation of Pharmaceutical Entrepreneurs
<b>ESHG</b>	European Society of Human Genetics
<b>IAPO</b>	International Alliance of Patients' Organisations
<b>IFSW-Europe</b>	International Federation of Social Workers
<b>Inserm</b>	French National Institute for Health and Medical Research
<b>ISPOR</b>	International Society for Pharmacoeconomics and Outcomes Research
<b>MRIS</b>	Maladies Rares Info Services (French helpline for rare diseases)
<b>NORD</b>	National Organisation for Rare Disorders (USA) - EURORDIS' counterpart in the US
<b>RDI</b>	Rare Diseases International
<b>Orphanet</b>	The online portal for rare diseases and orphan drugs: orpha.net
<b>NORBS</b>	The National Organisation for Rare Diseases Of Serbia
<b>HUFERDIS</b>	Hungarian Federation of People with Rare and Congenital Diseases
<b>RADOIR</b>	Rare Disease Foundation of Iran
<b>San Pau</b>	Fundació de Gestió Sanitària de l'Hospital de la Santa Creu i Sant Pau (Health Management Foundation of the Hospital de la Santa Creu & Sant Pau)

## MISCELLANEOUS

<b>PLWRD</b>	Persons living with a rare disease
<b>CoE / CE</b>	Centre of Expertise / Excellence
<b>ERN</b>	European Reference Network
<b>EU MS</b>	Member State (of the European Union)
<b>EUNRDHL</b>	EU Network for Rare Diseases Helplines
<b>NP (RD)</b>	National Plan / Programme (for Rare Diseases)
<b>HTA</b>	Health Technology Assessment
<b>MAPPs</b>	Medicine Adaptive Pathways to Patients
<b>MEP</b>	Member of the European Parliament
<b>MoCA</b>	Mechanism of Coordinated Access to orphan medicinal products

<b>PACE-ERN</b>	Partnership for Assessment of Clinical Excellence in European Reference Network (PACE-ERN) Consortium
<b>PE</b>	Patient Engagement
<b>PLWRD</b>	Persons Living with a Rare Disease
<b>TRP</b>	Therapeutic Recreation Programme
<b>CAVOD</b>	Clinical Added Value of Orphan Drugs
<b>OMP</b>	Orphan Medical Product
<b>Orphan drug</b>	"Orphan drugs" are medicinal products intended for diagnosis, prevention or treatment of life-threatening or very serious diseases or disorders that are rare
<b>MA</b>	Marketing Authorisation (for a medical product)
<b>PV</b>	Pharmacovigilance
<b>EudraVigilance</b>	EudraVigilance is a system designed for collecting reports of suspected side effects
<b>ADR</b>	Adverse Drug Reaction
<b>CUP</b>	Compassionate Use Programme
<b>ATMP</b>	Advanced Therapy Medicinal Product
<b>NBS</b>	Newborn Screening
<b>NGS</b>	Next-Generation Sequencing
<b>UN</b>	United Nations
<b>CoNGO</b>	Conference of Non-Governmental Organisations in Consultative Relationship with the United Nations
<b>#Resolution4Rare</b>	#Resolution4Rare is a campaign to support the call for a UN Resolution on Addressing the Challenges of Persons Living with a Rare Disease (PLWRD) and their Families

## 32ND ERTC WORKSHOP

<b>BDSG</b>	Big Data Steering Group
<b>CTIS</b>	Clinical Trials Information System
<b>CTR</b>	Clinical Trials Regulation
<b>DARWIN</b>	Data Analysis and Real-World Interrogation Network
<b>DPA</b> s	Data Permit Authorities
<b>EHDS</b>	European Health Data Space
<b>EMA</b>	European Medical Association
<b>ESMA</b>	European Securities and Markets Authority
<b>NCA</b> s	National Component Authorities of ESMA
<b>PoC</b>	Poof of concepts
<b>RWE</b>	Real-World Evidence
<b>RWD</b>	Real-World Data





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