EURORDIS
RARE DISEASES EUROPE

ACTIVITY REPORT
2022

EURORDIS ACTIVITY REPORT 2022

Cover photo: “Unexpected and beautiful”
Nemaline Myopathy
Taken by Christina Da Silva, Spain

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EURORDIS IN BRIEF (AS OF DECEMBER 2022)

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 1,000 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.

1017 MEMBER PATIENT ORGANISATIONS

74 COUNTRIES (27 EU COUNTRIES)

57 NATIONAL ALLIANCES OF RD PATIENT ORGANISATIONS

80 EUROPEAN FEDERATIONS OF SPECIFIC RARE DISEASES

FOUNDED IN **1997**

6 MILLION € BUDGET

OUTREACH TO OVER **2500** PATIENT GROUPS

OVER **460** VOLUNTEERS

75 PATIENT ADVOCATES

380 MODERATORS

55+ STAFF MEMBERS, WITH OFFICES IN PARIS, BRUSSELS, BARCELONA

EURORDIS staff in Lisbon

FOREWORD
BY PRESIDENT & CHIEF EXECUTIVE OFFICER

AVRIL DALY
President of the Board of Officers

YANN LE CAM
Chief Executive Officer

We can be proud of how far we've all come over the past year – which was also our 25th anniversary – in spite of the political attention being devoted to COVID-19, health emergencies, and the war in Ukraine. Our community, which steadily grows every year, has reached the important landmark of 1,000 members.

Two EU Council Presidencies – held by France and the Czech Republic – committed to a European Action Plan for Rare Diseases, which is now supported by 22 EU Member States and the European Parliament, we celebrated Rare Disease Day in over 100 countries, and launched the largest-ever Rare Barometer survey on diagnosis. We also organised the 11th edition of the biannual European Conference on Rare Diseases and Orphan Products, proving once again the resilience and determination of our community to build a better future for all.

In 2022, EURORDIS launched its strategy for rare cancers. The year was also marked by an important evolution in EURORDIS leadership, with Terkel Andersen, who had served as EURORDIS President for the past 20 years, stepping down in June 2022 due to health reasons. Avril Daly, who had been EURORDIS Vice-President for the past 10 years, was elected by the Board of Directors as EURORDIS President in November 2022.

This year comes with new challenges, but also new opportunities. 2023 starts off with the second Rare Disease Week – empowering patient advocates in national, European, and international advocacy – Rare Disease Day, and the Black Pearl Awards all taking place in February.

In 2023, we will address the mental wellbeing of people with rare diseases, while focusing on a holistic approach to care during our EURORDIS Membership Meeting in Stockholm. We will foster the integration of ERNs into national healthcare systems and promote early diagnosis, particularly through newborn screening. We will also focus on the revision of the pharmaceutical legislation and implementing the EU Health Technology Assessment (HTA) legislation, and we will work to ensure that the European Health Data Space serves the best interests of people living with rare diseases. We will also continue our work on the important topic of Advanced Therapies and Medicinal Products (ATMPs).

EURORDIS will strive to empower its members and community through the Open Academy Schools and over 70 hours of eLearning, as well as new alumni programmes (Meetups and Masterclasses), with over 2,600 registered users participating in our eLearning courses from about 160 countries. These are only a few of the major activities to mention, so get ready for a new year of transformation, impact, and purpose in the service of our community.

HIGHLIGHTS

"What I do best",
Three Rare Brain malformations, Periventricular Nodular Heterotopia with overlying Polymicrogyria (posterior subtype) and Cerebellar Hypoplasia, Australia

- 2022 has marked the **25th anniversary of EURORDIS.** For this occasion, a communication has been launched including a special logo and a **series of videos** featuring the Board members looking back at our history, achievements and the way forward towards reaching our goals by 2030.
- The **EURORDIS membership** grows steadily every year, and this trend was confirmed in 2022 as EURORDIS has continued to develop and engage its network of members. 48 new members (patient organisations) (+3.5%) joined in 2022, reaching the important milestone of **1,000 Members in March 2022** for EURORDIS' 25th anniversary.
- In 2022, the strategic review initiated in 2021 has been completed. The General Assembly adopted a new vision, mission statement, values, strategy, strategic objectives and organisational model. The Board of Directors (BoD) furthered this review, refining the strategic goals and the organisational model, leading to the publication of a revised strategy in November 2022.
- The **EURORDIS leadership** has evolved in 2022. Due to health reasons, EURORDIS President for the past 20 years, Terkel Andersen, had to step down from his position in June 2022. Avril Daly, EURORDIS Vice-President for the past 10 years, became Acting President until the election of a new Board of Officers (BoO). During the BoD meeting of November 2022, a new structure of the BoO was decided. The BoO now consists of six officers, including two additional positions with specific responsibilities that have been added: one additional Vice-President to support the President in external representation; and one Deputy General Secretary to support the General Secretary, especially when it comes to the relationship with EURORDIS Members and, in particular, the National Rare Disease Alliances.
- In 2022, there were **75 EURORDIS volunteer patient advocates** who belonged to different groups or task forces that are managed by EURORDIS Patient Engagement Managers (e.g. the Therapeutic Action Group, the Drug Information, Transparency and Access Task Force, TGE Health Technology Assessment Task Force, the Digital and Data Advisory Group, the Social Policy Action Group, and the ePAG Steering Committee) and **380 volunteer RareConnect** moderators.
- After several years and the requirement of social distancing during COVID-19, the **EURORDIS team** (about 60 colleagues) gathered for **a team seminar in Lisbon,** where staff met with the Portuguese national alliance on rare diseases, and explored how to enhance team cooperation and efficiency. This was important as the year has also been marked by considerable staff turnover, with one third of the team being renewed and a significant reshuffle of the team to align with priorities and resources.
- In 2022, EURORDIS has continued working with experts, including clinicians, researchers, and **more than 300 partners** and stakeholders (organisations, institutes, private sector bodies, governments etc.) within co-funded European projects in which we are partner or leader, as well as in the framework of our partnerships and memberships to **over 70 European and international networks,** and within the EURORDIS Roundtable of Companies, which keep growing. EURORDIS acknowledges the support and collaboration with all our partners to achieve our mutual goals by 2030.
- The **campaign for a European Action Plan for Rare Diseases** has continued throughout 2022, with several key outputs helping to build momentum towards a European Action Plan for Rare Diseases – the new policy framework called for by the extensive review of the Rare 2030 foresight study. The European Action Plan for Rare Diseases was highly supported by the European Conference on Rare Diseases stakeholders in June 2022. In addition, two Ministerial expert conferences on Rare Diseases were held in under the French and Czech Presidencies of the Council of the EU, the latter of which concluded with a successful call for a European Action Plan led by the Czech Presidency of the Council of the European Union during the Expert Conference on Rare Diseases in Prague on 25–26 October 2022. More than 20 Member States are now supporting this call and 50 Members of the European Parliament have written a letter to the President of the European Commission, issued on Rare Diseases Day, calling for a European Action Plan for Rare Diseases.

- The **11th edition of the European Conference on Rare Diseases & Orphan Products (ECRD 2022)** was a virtual event for the second consecutive time. By maintaining this event online, it remained accessible to all, **reaching over 800 stakeholders** of the wider rare disease community from **61 countries.**

- In 2022, for **its 15th edition, Rare Disease Day** events took place in **over 110 countries** and regions on every corner of the globe, despite the continuation of the COVID-19 pandemic. As part of the Day's focus on equity, the global campaign promoted six 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, and we welcomed **five new countries** to the campaign.

- On 15 March 2022, in response to Russia's invasion of Ukraine, EURORDIS launched its **Ukraine Response programme,** aimed at supporting the Ukrainian rare disease community. EURORDIS' response has been to immediately respond to the needs of Ukrainians living with a rare disease, and to advocate on behalf of their specific needs with the support of our multi-stakeholder networks. Over the course of the year, a large cohort of Ukrainians living with a rare disease felt a direct positive impact from the programme on their lives, in Ukraine, in Poland and other neighbouring countries. In particular, thanks to a partnership with Airbnb.org, more than 300 families received 30 days of free housing in Europe as they were fleeing the war.

"Girlfriends for Life", MS, Austria

- The **Rare Barometer programme** launched the **journey of rare disease patients to diagnosis survey** in March 2022. A large communication effort has been conducted to disseminate the survey among the Rare Barometer panel and through social media. Rare disease patient organisations received a **communication toolkit, available in 27 languages** to invite their members to participate in the survey. The survey's participation was very high, reaching out a total of **13,307 respondents.** The results will be published in a peer-review publication in 2023.

- To reduce delays in diagnosis, EURORDIS focused on **newborn screening,** in particular in the context of the **Screen4Care project and of the Newborn Screening Working Group.** EURORDIS worked toward an "RDclopedia", a survey of rare disease-related initiatives in Europe, including registries, research projects, infrastructures and newborn screening programmes. The Newborn Screening Working Group was consulted to contribute to **the work on "actionability"** as a criterion for newborn screening.

- In 2022, EURORDIS contributed to the **International Rare Diseases Research Consortium (IRDiRC)** conference on **rare disease clinical research networks, a priority promoted by EURORDIS for a decade, paving the way for their inclusion in the future Rare Diseases Partnership.**

- **REMEDi4ALL,** an EU funded project in which EURORDIS participates together with 23 other organisations, launched in September 2022. The project aims to assemble a sustainable European Innovation Platform able to provide guidance and support to developers at all stages of the **drug repurposing pathway.** For this important project, a Senior Project Manager and a Patient Engagement & Training Manager have joined EURORDIS's team to work on the project under the supervision of the Therapeutic Development Director.

- 2022 was an important year as EURORDIS has taken a new step in strengthening its action for rare cancers, by adopting a **specific strategy for rare cancers – across all ages and types –** to imple-

"Walking on Water", AADC deficiency, United States

ment the strategic objective to leave no one behind. EURORDIS has always included rare cancers in its scope, and rare cancers benefit from our work across all priority areas. The overarching goal for the coming years is to implement the recommendations set out in the Rare Cancer Agenda 2030 and to ensure that rare cancers are always included in European cancer and rare disease policies, and in all national cancer plans, with distinct sections for paediatric and rare adult cancers.

- EURORDIS strengthened in 2022 the holistic approach to rare diseases, and accordingly decided that the EMM2023 will centre on the theme of a **"lifelong holistic approach to people's needs and their full inclusion in society",** in line with our Strategic Objective to deliver on the goal of social inclusion. The Board of Directors decided to create the position of **Social Policy and Initiatives Director** who started in October 2022 and is relaunching the Social Policy Action Group of volunteers.

Among the numerous and major achievements of 2022 that benefit people living with rare diseases, we can mention these few, and many more are detailed in this Activity Report:

- The successful outreach of Open Academy eLearning: At the end of 2022, the eLearning platform of the EURORDIS Open Academy had **over 2,654 registered users, from more than 157 countries. 70 hours of training** were available on the platform. Towards the end of 2022, a new way to engage and train the Open Academy alumni was implemented through the **Alumni Meetups.** This consists of online sessions dedicated to a single topic relevant to patient advocates. **Monthly newsletters** with news about the Open Academy and other training opportunities were sent to the alumni and the users of the Open Academy platform.
- EURORDIS continued to support patient partnership in the European Reference Networks (ERNs) on rare diseases. Among many outputs of this support, in 2022, EURORDIS developed three practical guides to support the work of ePAG advocates, including:
  - A guide on clinical practice guidelines development.
  - A guide on how patient organisations can listen to the community.
  - A guide on the ERN Evaluation (including a Q&A Factsheet on the Evaluation).
  - A toolkit was also published on good practices for facilitating the integration of ERNs into national health systems.
- 2022 was a successful year for our communications. With the team reshuffle and new team members joining, the team has reached several major achievements. EURORDIS launched a **new website.** We continued translations and uploaded all relevant content. The website is available in seven languages and is populated with relevant content regularly. The website saw an increase in the number of visits in 2022, reaching 518,581 annual visits. In November 2022, **EURORDIS launched its very first podcast,** entitled **"Rare on Air",** which explores the unique experiences, challenges and successes of people from our community, and investigates how a better world can be built for those who have a rare condition.
- EURORDIS had put forward one member of staff as a candidate for the selection procedure to appoint Civil Society representatives to the **Management Board of the EMA.** This candidate was selected and began her mandate on 15 June 2022. Two representatives of patient organisations are members of this Board, which is the EMA's integral governance body. The mandate of the Board lasts three years.

STRATEGY & IMPACT
2021-2030

"Emotions"
Facioscapulohumeral
Muscular Dystrophy
Taken by Sofia Kosinska,
Ukraine

In 2020, EURORDIS commissioned an external review for the purpose of developing its strategy for 2021-2030. The strategic review collected input from EURORDIS members and stakeholders, incorporating the work done within the Rare 2030 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 final validation by the EURORDIS Board of Directors.

The Board of Directors continued the strategic review throughout 2022 and adopted the following revised strategy in November.

VISION

EURORDIS' vision is a world where all people living with a rare disease can have longer and better lives and can achieve their full potential, within a society that values their well-being and leaves no one behind.

To achieve their full potential, people living with a rare disease need to be:

- Recognised as equal citizens with their rights fully respected
- Diagnosed timely and accurately
- Supported with state-of-the-art medical and social care, or cured
- Included in society in all aspects of life and enabled to live independently

MISSION

EURORDIS works across borders and diseases to improve the lives of all people living with rare diseases.

STRATEGIC OBJECTIVES

STRATEGIC OBJECTIVE 1: A NEW EUROPEAN POLICY FRAMEWORK TO ACHIEVE MEASURABLE GOALS PROLONGING AND IMPROVING THE LIVES OF PEOPLE LIVING WITH RARE DISEASES BY 2030

By 2030, EURORDIS will have promoted and facilitated the implementation of a new long-term European policy framework based on the Rare 2030 foresight study, guaranteeing that rare diseases are a sustainable public health priority and integrating European policies and national strategies across all countries in Europe, to achieve measurable goals improving the health and well-being of people living with rare diseases and their inclusion in society, as well as reducing inequalities across Europe.

By 2030, within the UN Sustainable Development Goals Agenda 2030, the One Sustainable Health Approach and the future European Health Union, we will have designed a European model of care, transformative and useful beyond rare diseases.

This innovative model will be driven by the needs of people living with rare diseases and a collective responsibility to prioritise solidarity and equity.

STRATEGIC OBJECTIVE 2: DELIVERING ON SIX PRIORITY AREAS

By 2030, EURORDIS will have made contributions to the goals of (based on the Rare 2030 foresight study):

- Earlier, faster and more accurate diagnosis – goal of diagnosis within six months.
- High quality national and European healthcare pathways, including cross-border healthcare – goal of improved survival average by three years over 10 years, 1/3 reduction of mortality under 5 years of age.
- Integrated medical and social care with a holistic lifelong approach and inclusion in society – goal of reducing the social, psychological and economic burden by one third.
- Research and knowledge development that are innovative and led by the needs of people living with a rare disease.
- Optimised data and health digital technologies for the benefit of people living with a rare disease and society at large.
- Expanded rare disease treatment development, availability, accessibility and affordability, particularly with regard to transformative or curative therapies – goal of 1,000 new therapies within 10 years.

STRATEGIC OBJECTIVE 3: BEING INCLUSIVE OF ALL RARE DISEASES, ALL REGIONS, "LEAVING NO ONE BEHIND"

By 2030, EURORDIS will have expanded its scope to "leave no one behind" in the rare disease community by covering:

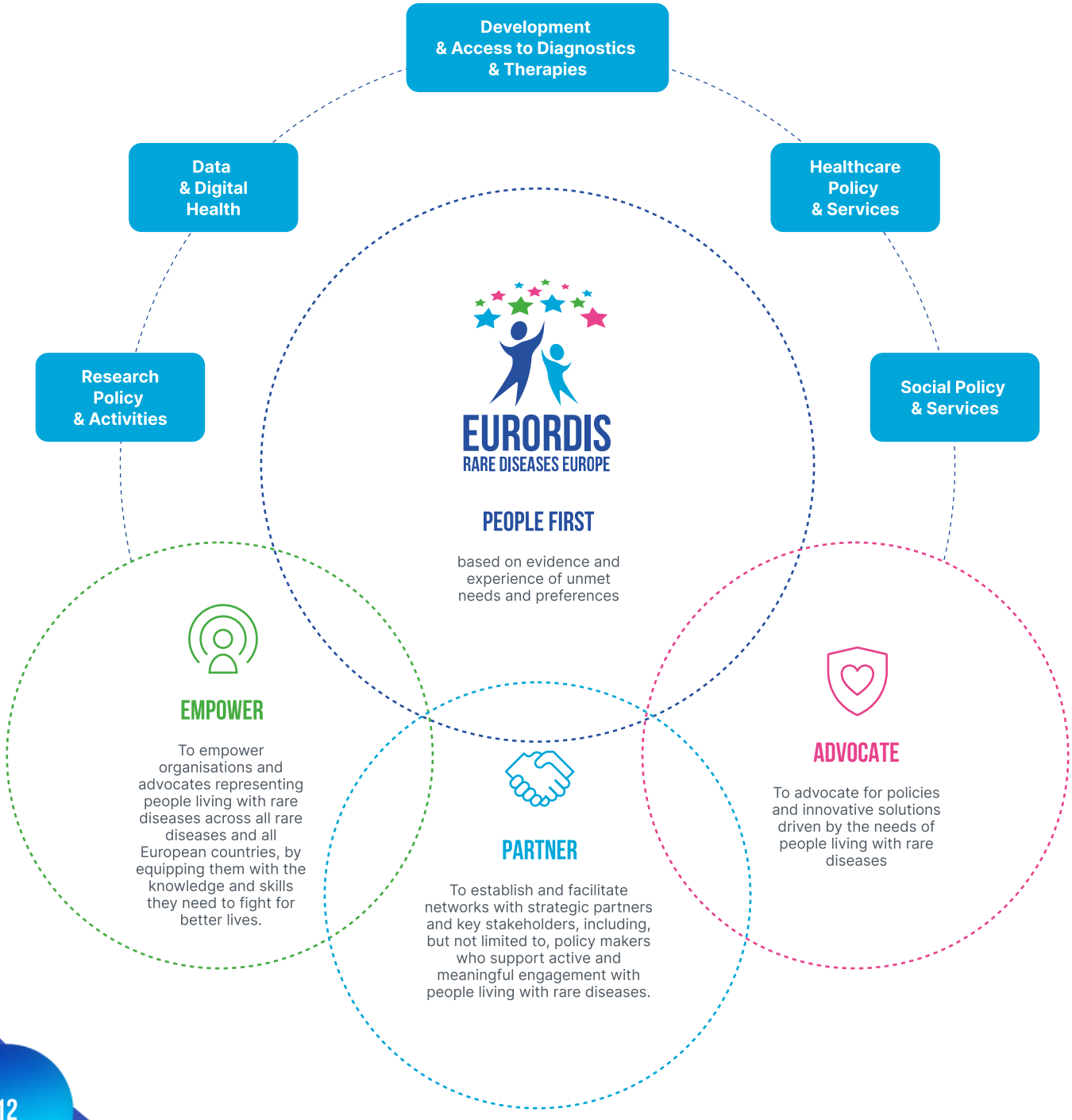
- All therapeutic areas including genetic and non-genetic rare diseases – rare cancers, rare infections and rare health hazards.
- All 48 countries in Geographical Europe prioritising Eastern and Southern Members of the EU, European Economic Area and EU accession countries.
- All rare disease prevalence and incidence levels, particularly the ones affecting fewer than one in 1,000,000.

ORGANISATIONAL MODEL TO FULFIL OUR STRATEGY AND REACH OUR STRATEGIC OBJECTIVES

EURORDIS is a Network Leverager within an ecosystem of networks of member organisations, advocates, partners and stakeholders. Through using our organisation's developed position as a Network Leverager, EURORDIS will achieve its strategic goals.

By 2030, EURORDIS will have catalysed and led impactful motivated networks of member organisations, and of advocates across disease areas, national borders, and age groups. EURORDIS will enable the relay to policymakers of the needs of people living with a rare disease and support the integration of European with national actions in key policy areas.

By the year 2030, EURORDIS will have established itself as a prominent driver of change, leveraging its unique role to initiate and cultivate networks of partners and stakeholders to facilitate the exchange of experiences, co-production of knowledge, and creation of synergies to effectively achieve our strategic objectives.



"No bad days!"
Morquio A syndrome - MPS IV a
Taken by Evgenia Tarakanova,
Russia

1. ADVOCATE

FOR POLICIES AND INNOVATIVE SOLUTIONS DRIVEN BY THE NEEDS OF PEOPLE LIVING WITH A RARE DISEASE

Milan Macek (National Coordination Centre for Rare Diseases, UH Motol and Charles University), Avril Daly (EURORDIS President) and Stella Kyriakides (EU Commissioner for Health and Food Safety) and Yann Le Cam (EURORDIS Chief Executive Officer).

Throughout 2022, EURORDIS has **supported its strategic objectives through advocacy efforts** and continued to promote rare diseases as a policy priority at national, European, and international levels.

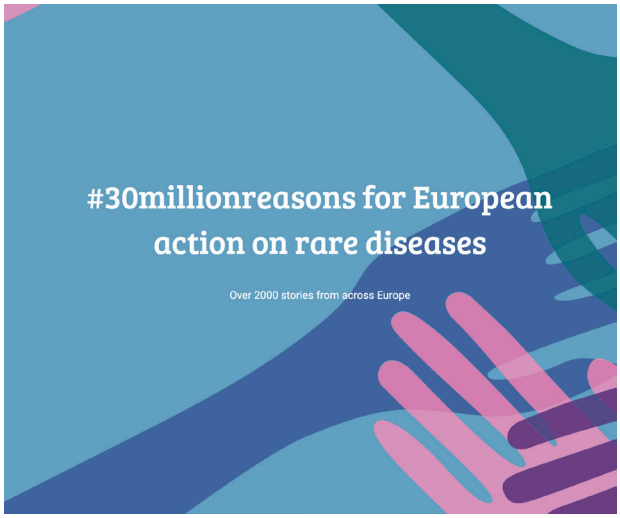
IN LINE WITH OUR STRATEGIC OBJECTIVE 1 TO REACH "A NEW EUROPEAN POLICY FRAMEWORK TO ACHIEVE MEASURABLE GOALS PROLONGING AND IMPROVING THE LIVES OF PEOPLE LIVING WITH RARE DISEASES BY 2030":

ADVOCATING FOR AN UPDATED POLICY FRAMEWORK FOR RARE DISEASES AND RARE CANCERS FOR 2021-2030

The campaign for a European Action Plan for Rare Diseases

This work has continued throughout 2022, with several key outputs helping to build momentum towards a **European Action Plan for Rare Diseases** – the new policy framework called for by the extensive review of the Rare 2030 foresight study.

EURORDIS launched and promoted a dedicated book and webpage sharing the more than 2,000 testimonies on why people living with rare diseases and their community want a European Action Plan for Rare Diseases. The book has been disseminated to over 50 Members of the European Parliament (MEPs), the European Commission, and policymakers at the level of EU Member States.



On 28 February 2022, the French Presidency of the EU Council hosted the "Ministerial Conference on Innovation and Care Pathways: For a European policy on rare diseases". This official event marked a significant milestone in the proposal of a European health union for rare diseases, as it provided the first opportunity for multi-stakeholder discussions. Representatives from 12 Member State Ministries of Health were either physically present or participated remotely. The conference was a major step towards the establishment of a comprehensive policy framework that addresses the needs of rare disease patients in Europe.

In late June 2022, the European Conference on Rare Diseases (ECRD 2022) – another event of the French EU Presidency – provided the opportunity for all stakeholders to consider how to transform the exhaustive review of the existing strategy on rare diseases, as emerged from Rare 2030, into proposals of concrete actions addressing unmet needs and inequalities persisting across Europe. As a result, EURORDIS coordinated a letter from all partners of the Conference, including European Reference Network (ERN) coordinators, pharmaceutical and non-pharmaceutical companies working in rare diseases, and national patient representatives, calling on the European Commission to introduce a European Action Plan for Rare Diseases.

EURORDIS also contributed to the programme and publication of a report on the Expert Conference on Rare Diseases in Prague on 25-26 October 2022 – an official event of the Czech Presidency of the EU Council. A major, conclusive output of the Czech EU Presidency was the Call to Action for rare diseases that was launched at the Expert Conference, which received the endorsement of 21 Member States (additional to the Czech Republic) at the EPSCO Council in December 2022. The Call to Action urged the adoption of a comprehensive European Action Plan for Rare Diseases, which would support and complement ongoing and future efforts at both the EU and Member State levels to address the unmet needs of individuals living with rare diseases. In addition, the Call to Action urged improvements in early diagnosis, maximising the potential of the revised EU legislation on orphan and paediatric medicines, enhancing access to treatments, and integrating the European Reference Networks (ERNs) into national healthcare systems to provide more comprehensive care.

These results were presented to the Commissioner for Health Stella Kyriakides at an in-person meeting on 6 December 2022, and a few days later to the European Commission unit (DG SANTE) responsible for rare diseases and ERNs.

Stakeholder Network for Rare Diseases

In 2022, EURORDIS continued preparing and launching the **Stakeholder Network on Rare Diseases** on the Health Policy Platform (HPP) of the

European Commission, co-leading the two kick-off meetings and the launch of a survey to identify priority areas of actions for the Network in the coming years. EURORDIS, along with Orphanet and the University of Newcastle, coordinates this dedicated forum for all rare disease stakeholders, identifying and implementing priority actions for the coming years. The Network has the ambition to reconstitute a multi-stakeholder forum for exchanges and debate on broad rare disease policy issues, as a follow-up of Rare 2030 and its Panel of Experts.

Securing proposals impacting on rare diseases in the context of the EU multi-annual financial framework 2021-2027

In 2022, EURORDIS supported proposals under the EU4Health Programme 2021-2027, including proposals relating to: the sustainability of European Reference Networks; Orphanet; support for children and adult rare cancers under Europe's Beating Cancer Plan; Horizon Europe 2021-2027 (including the European Partnership for Rare Diseases and the future flagship European Clinical Research Network for Rare Diseases); new flagship initiatives such as EU-level collaboration on newborn screening; European comprehensive health services for rare diseases; and an EU fund for the collection of real-world evidence.

Involving the European Network of National Alliances in advocacy activities

EURORDIS involved the European Network of National Alliances through its Council (CNA) in advocacy activities, including through in-person meetings and monthly "hot topics" calls. Throughout the year, EURORDIS provided ongoing support to its National Alliances by sharing medium-term advocacy plans and equipping them with tools and information to drive engagement by Member States in relevant EU initiatives and legislation. These efforts focused on strategic priority areas aligned with the EURORDIS-CNA Common Goals and Mutual Commitments. These efforts also included improving the access of National Alliances to our Rare Barometer surveys, allowing for more coordinated, evidenced-based advocacy.

CONVENING THE EUROPEAN NETWORK OF PARLIAMENTARY ADVOCATES FOR RARE DISEASES

In 2022, EURORDIS mobilised the Network around key advocacy priorities. In March, EURORDIS organised a briefing on the situation of people living with rare diseases in Ukraine for the co-chairs of the Network, which led to a parliamentary letter urging the European Commission to support Ukrainians living with a rare disease in its humanitarian response to the war. EURORDIS also engaged with various MEPs in the framework of the European Parliament resolution "Towards a common European action on care", including with the Rapporteur of the text. This led to the successful inclusion of specific provisions on rare diseases in the final text of the resolution (which are detailed later in this report). Throughout the year, EURORDIS also shared its recommendations with the Network on several key legislative files, including the European Health Data Space and the revision of pharmaceutical legislation. This ongoing work has provided the opportunity to recruit new members to the Network and to position EURORDIS as an expert on rare disease issues at the EU level.

PROMOTING 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 raise awareness of rare diseases directly among relevant international institutions; to equip patient groups with advocacy tools to engage their national authorities and drive local patient empowerment and; to foster greater international cooperation in the field of rare diseases.

Rare Diseases International

EURORDIS has worked with Rare Diseases International (RDI) since its inception on promoting rare diseases as an international public health initiative. EURORDIS provides the vision, strategy, leadership, and co-execution required to advance rare diseases within the international community through the United Nations (UN) system. All policy actions within the UN system are co-organised by the UN NGO Committee for Rare Diseases (of which EURORDIS is a founding member and Vice President), RDI, EURORDIS and Agrenska.

Three major RDI policy achievements so far co-led by EURORDIS include: a) the explicit integration of people living with a rare disease in the UN Political Declaration on Universal Health Coverage (UHC) adopted in September 2019; b) the unanimous adoption by the UN General Assembly of the UN Resolution on "Addressing the challenges of persons living with a rare disease and their families" in December 2021; c) the Memorandum of Understanding (MoU) between RDI and the WHO for the period 2019-2022.

Beyond the "Global Gathering for Rare Diseases" event with the WHO in May 2022 in Geneva, three events in 2022 shed lights on these adopted policies, promoted specific actions relating to their implementation. These three events were:

- The online event "Rare Diseases: A Global Priority for Equity" on Rare Disease Day (28 February 2022).
- The event "Addressing the challenges of persons with a rare disease as a Gender Equality, Human Rights and Equity Priority" at the UN High-Level Forum on Sustainable Development Goals (SDGs) on 6 July 2022. This hybrid event, sponsored by Spain and co-sponsored by Qatar and Brazil, with the participation of many other UN Member States, took place onsite at the UN Building in New York City and online.
- The online event "Universal Health Coverage for Rare Diseases: Developing pillars together", taking place on Universal Health Care Day (12 December 2022).

World Health Organization (Geneva)

EURORDIS' collaboration with the WHO is guided by the MoU RDI-WHO 2019-2022. As a co-leader in RDI governance, EURORDIS played a vital role in shaping the MoU, leading the Policy Committee, and contributing directly and indirectly to the three different areas of work within the MoU. In doing so, EURORDIS engaged relevant staff to ensure a comprehensive approach to the collaboration. Overall, the collaboration with the WHO is centred on delivering on UHC and the SDGs, based on the UN Resolution for people living with rare diseases. The deliverables for the year 2022 were submitted to the WHO at the end of March 2023. However, all developments were executed in close collaboration throughout the year, and the WHO review all draft deliverables ahead of their submission. In 2022, the three key relate to:

- The third and last phase of concept development for the WHO Global Network for Rare Diseases (WHO GNRD).
- The Operational Description of Rare Diseases. Based on the deliverable of 2021, in 2022 an international expert group finalised a consensus on a Core Definition of Rare Diseases and a Descriptive Framework for Rare Diseases. These were submitted for peer review. These pieces of work provided a robust base to define rare diseases within the UN system for WHO policy and actions, as well as for the implementation of UHC and the UN Resolution. This work has had a strong impact on the International Classification of Diseases (ICD) and its nomenclature – hence the close association of ORPHANET and the experts in the WHO ICD-11 Working on Rare Diseases.
- The WHO Essential Medicines List and the WHO Essential In Vitro Diagnostic List. The common goal with the WHO has been to analyse the current products listed for rare indications, to promote the opportunities to apply for rare diseases, and to build the capacities of applicants, so that there will be more applications and more products included in the essential lists. This project has been a collaborative effort between the International Rare Disease Research Consortium (IRDiRC), with the Working Group on Access, piloted and supported by RDI, and EURORDIS. The goal has been to identify essential medicines and in vitro diagnostic tools deserving of inclusion in the essential lists by leveraging the networks of both IRDiRC and ERNs within the EU. By working together, we have been aiming to create a comprehensive list of the most essential medicines and diagnostic tools. Analyses are submitted for peer review.

WHO Regional Committee for Europe

In the dynamic of RDI-EURORDIS collaboration with WHO Geneva, EURORDIS started a dialogue and engagement with WHO Europe. In 2022:

- EURORDIS had an online meeting with the WHO Director of the Division on National HealthCare Policies and Systems based on Rare 2030 to identify the areas of potential collaboration, including: early diagnosis and newborn screening, the identification of experts and their local and regional networking in synergy with the ERNs, and the response to the situation in Ukraine.
- The WHO Europe Director General Hans Kluge participated in the ECRD 2022 as

a keynote speaker, highlighting that WHO Europe – following the UN Resolution, our work with the WHO, and the Rare 2030 study – was ready to progressively integrate rare diseases in their scope of actions.

- EURORDIS continued to be highly engaged in the WHO Oslo Medicine Initiative (OMI), contributing speakers to the webinars on identifying solutions to address the challenges of highly priced innovative medicines, and the review of final reports.
- EURORDIS was invited to address the ministerial meeting lunch of the WHO Regional Committee Meeting in Tel Aviv, Israel, which saw Member States adopt a statement giving WHO Europe a new mandate on highly innovative, highly priced medicines (now called the Novel Medicines Platform).

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "EARLIER, FASTER AND MORE ACCURATE DIAGNOSIS: – GOAL OF DIAGNOSIS WITHIN SIX MONTHS":

ADVOCATING TO IMPROVE ACCESS TO QUALITY RARE DISEASE DIAGNOSIS

Newborn screening

EURORDIS remained dedicated to reducing diagnostic delays for rare diseases and addressing undiagnosed diseases. To achieve this goal, we advocated for a harmonised European approach to newborn screening based on the EURORDIS position paper on the 11 Key Principles for newborn screening. We also called for the creation of an EU-level expert working group to design and implement this approach, fostering collaboration and coordination among Member States.

In 2022, we continued to lead and moderate the EURORDIS Newborn Screening Working Group (NBS-WG) and disseminate the 11 Key Principles. EURORDIS chaired the session at the Czech EU Presidency technical meeting on "Early Diagnosis of Patients with Rare Disorders in the EU: Crucial Role of the Newborn Screening" in Brno on 23 July, and contributed to the "Early Diagnosis" session of the Czech EU Presidency Expert Conference on Rare Diseases in Prague on 25-26 October.

EURORDIS also presented the 11 Key Principles at international conferences and at the SSIEM Annual Symposium, and co-authored the article "Towards Achieving Equity and Innovation in Newborn Screening across Europe", which has been published in the International Journal of Nursing Studies (IJNS) as part of the special issue on "Neonatal Screening in Europe". A presentation was also made during the World Orphan Drug Congress Roundtable discussion on Newborn Screening Equity.

EURORDIS Rare Barometer Programme: Patient experience surveys enabling evidence-based contributions to policymaking and research

The Rare Barometer programme consists of surveys aiming to collect qualitative and quantitative data on the experiences, needs and expectations of people living with a rare disease. Rare Barometer secured a panel of 20,000 people living with a rare disease who agreed to take part on a regular basis in EURORDIS' quantitative surveys. Figures and facts from the studies are used to develop EURORDIS' advocacy positions. All studies are translated into 23 languages and made available to rare disease patient organisations.

The Journey to Diagnosis for People Living with a Rare Disease

The diagnosis survey was launched in March 2022 by a webinar presenting the content of the survey, how its results will be used, and how to disseminate it. The 14,525 people who were part of the Rare Barometer panel in March 2022 received an email inviting them to participate in the survey in their own language. 4,100 participants of the panel took the survey, of which 3,069 answered 80% of the questions, representing a 21% response rate (3,069/14,525), similar to other studies. 19,159 other respondents were reached out to through social media posts, Facebook advertisements, and patient organisations, of whom 10,238 completed 80% of the questionnaire. Rare disease patient organisations received a commu-

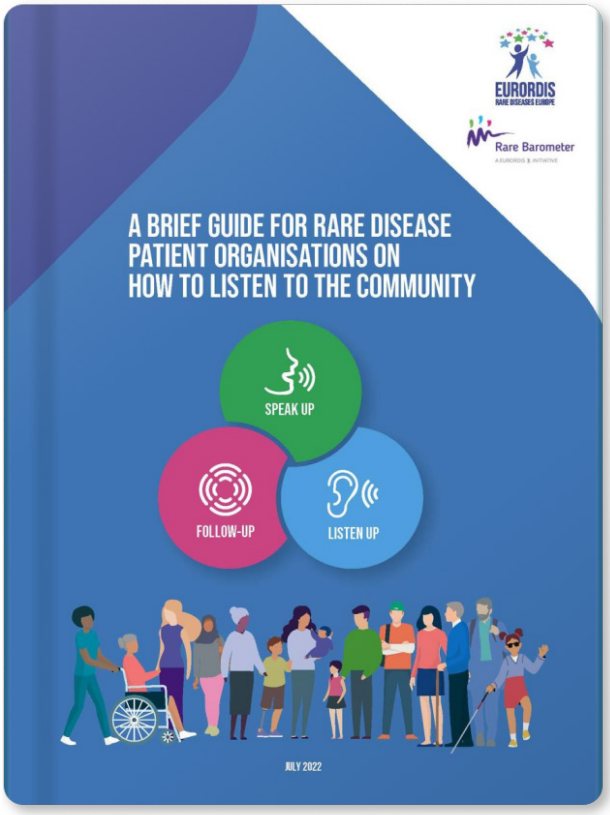
nication toolkit, available in 27 languages, with a template email and suggested social media posts and images to invite their members to participate in the survey. The second half of 2022 was largely dedicated to cleaning and analysing the data of the diagnosis survey, and EURORDIS members received 45 dashboards with their preliminary results, available in 27 languages. The final European results will be published in 2023.

Rare Barometer Survey: Needs assessment in Ukraine

A survey was conducted between 25 April and 15 June 2022 to assess the needs of rare disease patients and their families from Ukraine, whether they still lived in Ukraine or moved to another country. The answers from more than 400 respondents were used to shape EURORDIS' response and help people living with a rare disease impacted by the war in Ukraine. Survey results were communicated in the EURORDIS report on the challenges faced by people living in Ukraine affected by a rare disease.

Participation in the "Speak Up. Listen Up. Follow Up" guide

Rare Barometer participated in the development of the "Speak Up. Listen Up. Follow Up" guide, initiated and coordinated by the EURORDIS ERN and healthcare team to help patient advocates gather input from their community using methods from the social sciences, such as quantitative surveys, focus groups or interviews. The guide is structured into eight steps which range from defining objectives, to selecting the best approach to collect data, to analysing the feedback from rare disease communities. It is accompanied by complementary resources which include a Rare Barometer question repository with questions in 23 languages adapted to the rare disease population.



Disseminating results of past surveys

Results of past surveys, in particular the **"Share and protect our health data"** and **"Juggling care and everyday life: the balancing act of the rare disease community"** surveys, were presented on several occasions and were continuously used by staff members to feed advocacy positions and interventions at conferences and events.

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "OPTIMISED DATA AND HEALTH DIGITAL TECHNOLOGIES FOR THE BENEFIT OF PEOPLE LIVING WITH A RARE DISEASE AND SOCIETY AT LARGE":

ADVOCATING FOR A EUROPEAN HEALTH DATA SPACE TO ENABLE THE SECURE SHARING AND USE OF DATA AT THE EU LEVEL

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 are scattered. EURORDIS has actively identified and promoted the specific challenges and needs of people with rare diseases with regard to health data sharing at the European and international levels. On 3 May 2022, the European Commission published a **draft proposal on the European Health Data Space (EHDS).** The proposal is highly relevant for the rare disease community, since it includes a strategic vision on how people's health data are managed, shared and used across the EU for treatment and for research. EURORDIS has advocated for the construction of an EHDS that optimises electronic health records, ensures the ethical use of data for research, policymaking, and treatment development, increases digital health literacy in the rare disease community and beyond, and encourages the formation of patient and public partnerships.

To contribute to the development and implementation of the EHDS, the following activities were undertaken in 2022:

- Informational and consultative meetings were held within EURORDIS networks to present and explore the impact of draft EHDS proposals on the rare disease community and to agree on the advocacy strategy during the co-decision process on the EHDS.
- A **EURORDIS position paper on the EHDS proposal** was developed based on the gathered internal and membership feedback. We disseminated the position paper to various stakeholders, including representatives from the European Commission, the European Parliament, and the Council of the European Union, as well as healthcare professionals, payers, consumers, and representatives from the pharmaceutical, digital, and MedTech industries. In the framework of the legislative process, EURORDIS replied to the European Commission's public consultation on the EHDS and held meetings with key MEPs to present the main concerns and asks of the rare disease community as expressed in the position paper, and to ensure that they would be integrated into the final act.

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "INTEGRATED MEDICAL AND SOCIAL CARE WITH A HOLISTIC LIFELONG APPROACH AND INCLUSION IN SOCIETY – GOAL OF REDUCING THE SOCIAL, PSYCHOLOGICAL AND ECONOMIC BURDEN BY ONE THIRD":

ADVOCATING FOR INTEGRATED AND HOLISTIC CARE

Early in the year, EURORDIS identified the European Care Strategy, to be presented by the European Commission in the autumn of 2022, as an opportunity to address several long-term and holistic care needs of people living with rare diseases. As such, EURORDIS submitted a contribution to the Call for Evidence on a European Care Strategy in March 2022, which called on the European Commission to ensure that people living with a rare disease are included in the scope of the Strategy and that its approach to care is holistic. In parallel, EURORDIS engaged with the Rapporteur of the aforementioned European Parliament resolution "Towards a common European action on care", leading to the successful inclusion of specific provisions on rare diseases in the final text of the resolution. Specifically, MEPs recognised that long-term care needs extend to different groups, including people living with rare diseases. MEPs also called on the European Commission to undertake research to better understand the impact of inadequate care provision, and to secure funding for research projects on the social impact of rare diseases, as well as for the co-creation and transfer of good practices and innovative care models. Despite EURORDIS' advocacy efforts and the European Parliament's support, the final scope of the European Commission's proposals for the European Care Strategy, published in September, were not inclusive of people living with rare diseases and their holistic care needs.

ADVOCATING FOR QUALITY AND ADEQUATE SOCIAL RIGHTS

In cooperation with the Social Platform and its members, EURORDIS continued to follow the ongoing discussions and consultations on relevant EU flagship initiatives and legislative proposals deriving from the European Pillar of Social Rights Action Plan. In addition, the 11th European Conference on Rare Diseases & Orphan Products (ECRD 2022) provided the opportunity to advance advocacy on social rights within its second goal of "Reducing Inequalities". The three sessions on reducing inequalities, which involved over 20 speakers, focused on a range of social topics,

from disability assessments to social care, education, employment, and other aspects of independent living.

ADVOCATING TO IMPROVE ACCESS TO DISABILITY RIGHTS

In 2022, EURORDIS continued to follow the implementation of the European Strategy for the Rights of Persons with Disabilities 2021-2030, to ensure that the disability-related needs of the rare disease community were voiced and addressed. EURORDIS followed both the work of the European Commission on the flagship initiatives of the Strategy, as well as the European Parliament discussions on the Rights of Persons with Disabilities.

From November 2022, EURORDIS drafted a response to the European Commission's Call for Evidence on the European Disability Card, which was submitted in early January 2023. The EU Disability Card, to be proposed by the European Commission by the end of 2023, will make it easier for people with disabilities to access some services while travelling within the EU. Many people living with a rare disease and disability are at risk of not being able to access the Card, because they do not have disability status in their country. Besides providing input on the design of the proposal for the EU Disability Card, EURORDIS used its contribution to advocate for the improvement of national disability assessment frameworks. This is a key priority for the rare disease community and one of the "flagships" identified within the campaign for a European Action Plan for Rare Diseases.

EURORDIS attended the European Day of Persons with Disabilities annual conference, organised by the European Commission in partnership with the European Disability Forum.

"Mihajlo in sunset", Spinal Muscular Atrophy, Bosnia and Herzegovina

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "EXPANDED RARE DISEASE TREATMENT DEVELOPMENT, AVAILABILITY, ACCESSIBILITY AND AFFORDABILITY, PARTICULARLY WITH REGARD TO TRANSFORMATIVE OR CURATIVE THERAPIES – GOAL OF 1,000 NEW THERAPIES WITHIN 10 YEARS":

ADVOCATING FOR THE IMPLEMENTATION OF AN EU PHARMACEUTICAL REGULATORY AND POLICY FRAMEWORK THAT ADDRESSES THE CHALLENGES OF PEOPLE LIVING WITH RARE DISEASES: ON EU PHARMACEUTICAL LEGISLATION, ORPHAN MEDICINAL PRODUCTS AND PAEDIATRICS REGULATIONS

In 2022 we continued contributing to the overall EU debate on the revision of the General Pharmaceutical Legislation and the legislation for special populations (rare disease and paediatric patients). Highlights of the year include:

- The development of a position outlining the requests of the patient community in view of the revision of the Orphan Medicinal Products (OMPs) and Paediatrics Regulations.
- Continued dialogue with the European Commission to relay the key asks and concerns of people living with rare diseases ahead of their proposed legislative revisions.
- Evolving our advocacy actions relating to the General Pharmaceutical Legislation and the revisions of the OMP and Paediatrics Regulations over the two conferences of the successive French and Czech Presidencies of the EU Council. Presented at the Czech EU Presidency's Expert Conference on Rare Diseases in October 2022, key elements of our position were integrated into the aforementioned Call to Action signed by 22 EU Member States following the conference.
- Continued participation in the European Expert Group of Orphan Drug Incentives, a multidisciplinary and cross-functional expert group bringing together representatives of the broad rare disease community, including researchers, academia, patient representatives, members of the investor community, rare disease companies and trade associations. Development and presentation of recommendations on the revision of the Orphan Medicinal Products Regulation at the Rare Diseases Conversations Conference in November 2022.

ADVOCATING FOR THE STRENGTHENING OF EU CLINICAL RESEARCH CAPABILITIES: ACCELERATING CLINICAL TRIALS IN THE EU (ACT EU)

The Accelerating Clinical Trials in the EU (ACT EU) initiative aims to develop the European Union further as a competitive centre for innovative clinical research. It has been established by the European Commission, the Heads of Medicines Agencies (HMA) and the European Medicines Agency (EMA).

During the 35th EURORDIS Round Table of Companies Workshop, we explored the strategy of ACT EU to better appreciate which political actions are being taken towards the goal of positioning the EU as a competitive clinical research centre. At the Workshop, we learnt about several important public initiatives and public-private partnerships that are helping to advance the ACT EU goals of developing and implementing innovative approaches to clinical trials. In addition, the Workshop included acquiring insights into the Recommendations for Decentralised Clinical Trials, which represent one of the first tangible outcomes of this initiative.

ADVOCATING FOR THE IMPLEMENTATION OF THE REGULATION ON EU COOPERATION ON HEALTH TECHNOLOGY ASSESSMENT

EURORDIS continued its advocacy for the implementation of the EU Cooperation on Health Technology Assessment (HTA), through the activities of our HTA Task Force. Task Force members discussed the assessment, pricing, and reimbursement of treatments across Europe and advised EURORDIS on all aspects of HTA: from methodology to the engagement of patients. The Task Force supported 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. The Task Force replied to consultations from EUnetHTA21 in 2022.

Look the future in the eye, Leukemia, Italy

ADVOCATING TO IMPROVE ACCESS TO TREATMENT FOR RARE DISEASES

EURORDIS promoted the proposal of structured cooperation across European countries in pricing and reimbursement policies and the development of a European Fund to finance evidence generation post-marketing authorisation which places particular emphasis on the **rarest of diseases and advanced therapies.** In 2022, EURORDIS undertook this work through: continuing dialogue with multi-stakeholder platforms, such as the Expert Group on Orphan Drug Incentives, led by EURORDIS and EUCOPE; contributing and participating in multi-stakeholder initiatives such as RWE4Decisions; participating in European Commission-led conferences and EU Council Presidency high-level conferences; and continuing operations and support for early dialogue with payers through Mechanism of Coordinated Action (MoCA) meetings (as well as preparing a publication highlighting ten years of MoCA activities, which is foreseen to be published in 2023). Additionally, we advocated for improved access to rare disease treatments through the programme development of Track A, B and E of ECRD 2022.

ADVOCATING FOR THE HARMONISATION OF COMPASSIONATE USE PROGRAMMES (CUP) AND/OR A EUROPEAN FUND FOR CUP

CUPs have proven to be one of the most effective approaches in bringing innovative medicines to patients and ensuring access to those most in need.

Since the adoption of a position on compassionate use for medicines used in rare diseases in 2016, EURORDIS has continuously participated in efforts to improve CUPs in Europe.

In 2022, EURORDIS contributed to shaping the European Commission's proposal for the revision of the pharmaceutical legislation, highlighting the need to extend the scope of Article 83 or Reg. 726/2004 (either by adopting a European scheme, or by facilitating the adoption of an Early Access programme similar to the French programme which is the most effective in the EU). CUPs save lives as they are the most immediate

solution to improve access to life-saving medicines in the EU. This is even more needed in the context of emergency preparedness, where the Member States' regulatory route to implement EMA recommendations of use for medicines and vaccines addressing health threats can only be via compassionate use. In the absence of a scheme for a CUP, more than half of Member States are not ready.

Collaborative efforts on equity of access and sustainable approaches to the financing of innovative pharmaceuticals (RARE IMPACT)

RARE IMPACT aimed to identify and validate the challenges to patients' access to advanced therapeutic medicinal products (ATMPs), specifically 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 has been driving the RARE IMPACT initiative. The recommendations of Rare Impact have been used to develop project proposals which will help us to make use of the findings.

Monitoring actual access to medicines after the reimbursement decision

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 has been made to remedy part of the causes that explain medicine shortages, as patients with rare diseases are particularly affected.

However, the extent of the problem is difficult to quantify and the consequences for patients' 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 to 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 website's "shortages catalogue". For all shortages affecting medicines to treat rare diseases, the EMA consults EURORDIS.

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "HIGH QUALITY NATIONAL AND EUROPEAN HEALTHCARE PATHWAYS, INCLUDING CROSS-BORDER HEALTHCARE: GOAL OF IMPROVED SURVIVAL AVERAGE BY THREE YEARS OVER 10 YEARS, 1/3 REDUCTION OF MORTALITY UNDER 5 YEARS OF AGE":

ADVOCATING FOR PROGRESS IN PATIENTS' RIGHTS TO CROSS-BORDER HEALTHCARE

Following the survey and the response to the European Commission's consultations in 2021, EURORDIS continued in 2022 to follow up the implementation of actions relating to the evaluation of the EU cross-border legislation. EURORDIS organised a meeting with the European Commission (DG SANTE) to present the recommendations included within "An empty promise: accessing cross-border healthcare for people living with a rare disease". EURORDIS invited DG SANTE to the November meeting of the Council of National Alliances to present the follow-up actions from the evaluation and discuss best practices on prior authorisation.

ADVOCATING FOR THE MENTAL HEALTH OF PEOPLE LIVING WITH RARE DISEASES

The mental health of people living with rare diseases remains an unseen and neglected unmet need that is frequently overshadowed by the other medical complexities associated with rare diseases. The rare disease community identified as an absolute priority the need to look beyond the physiological symptoms of a rare disease (for example, in EURORDIS' 2019 position paper on "Achieving Holistic Person-Centred Care to Leave No One Behind". Included in the UN Resolution, the need to tackle the needs of people living with a rare disease and their families and to take concrete action to address the psychological impact associated with these complex conditions has recently emerged in a more distinctive manner, especially in the aftermath of the pandemic.

Mental health is now also increasingly recognised as a public health priority in the EU, which will be shrined in the new **Commission Communication on a comprehensive approach to mental health.**

In 2022, EURORDIS recognised the unmet mental health needs of the rare disease community through three key actions:

- Adopting mental health as a new strategic priority.

- Commencing the development of a new EURORDIS initiative on mental health.
- Undertaking a targeted, evidence-based review to demonstrate the increased vulnerability of people living with rare diseases and their families.

These actions constitute the basis for our efforts to see people living with rare diseases recognised as among the vulnerable population groups identified in the European Commission's comprehensive approach to mental health.

IN LINE WITH OUR STRATEGIC OBJECTIVE 3 OF BEING "INCLUSIVE OF ALL RARE DISEASES, ALL REGIONS, 'LEAVING NO ONE BEHIND'", EXPANDING THE SCOPE OF OUR MEMBERSHIP BY COVERING "ALL THERAPEUTIC AREAS INCLUDING GENETIC AND NON-GENETIC RARE DISEASES, RARE CANCERS, RARE INFECTIONS AND RARE HEALTH HAZARDS" AND "ALL RARE DISEASE PREVALENCE AND INCIDENCE LEVELS, PARTICULARLY THE ONES AFFECTING FEWER THAN ONE IN 1,000,000":

RARE CANCERS

2022 was an important year as EURORDIS adopted a specific strategy for rare cancers – across all ages and types – to implement the strategic objective to leave no one behind. The overarching goal for the coming years is to implement the recommendations set out in the Rare Cancer Agenda 2030 and ensure that rare cancers are always included in European cancer and rare disease policies, and in all national cancer plans, with distinct sections for paediatric and rare adult cancers.

EURORDIS has always included rare cancers in its scope, and rare cancers benefit from our work across all priority areas. This includes promoting research and drug development for rare and ultra-rare diseases, advocating for equal access to available therapies, implementing access to cross-border healthcare, and developing European Reference Networks.

However, while rare cancer patients and caregivers share many of the same burdens as other rare disease patients, they also face some unique challenges that require special attention. The Board has recognised the need to increase the visibility of EURORDIS' actions for rare cancer patients and to better integrate this group into its activities. This will allow the organisation to address rare cancer patients' specific needs, while also learning from the achievements of different rare cancer patient organisations.

As part of this strategy, EURORDIS has established the **Rare Cancer Advocates Network,** made up of the 33 European Patient Group Advocates (ePAGs) within the ERNs PaedCan (paediatric cancers), EURACAN (rare adult solid cancers), EuroBloodNet (rare adult blood cancers and rare haematological diseases), GENTURIS (genetic tumour risk syndromes), as well as ePAGs in other ERNs' sub-networks covering rare tumours, such as Endo-ERN (rare endocrine conditions).

The Network is represented in European institutions or in other European settings by four ePAGs together with EURORDIS. These four ePAGs represent each one of the four main groups of rare cancers: paediatric cancers, rare adult solid cancers, rare adult blood cancers, and genetic tumour risk syndromes.

One major achievement in 2022 resulting from the collective work of rare cancer ePAGs, and also Rare Cancers Europe, was the inclusion of a specific section for rare adult cancers in the European Parliament report on Europe's Beating Cancer Plan, as rare adult cancers are not specifically addressed in the Plan. The paediatric cancer community supported the amendments to show solidarity within the whole rare cancer community.

VERY RARE AND ULTRA-RARE DISEASES

EURORDIS has also been focusing on policy initiatives and solutions to address the challenges of **very rare and ultra-rare diseases.** This has been done through advocacy actions such as informing the revision of the EU Regulation on Orphan Medicinal Products and different projects such as: SOLVE-RD (undiagnosed diseases); Screen-4Care (early diagnosis and newborn screening); various activities undertaken within the EMA Committees; proposing access to very rare disease and advanced therapies; and proposing the provision of highly specialised services through the ERNs at the EU level.

"Stuck in the middle with you"
Kleefa Syndrome
Taken by Loredana Huljev, Croatia

2. EMPOWER

ORGANISATIONS AND ADVOCATES REPRESENTING PEOPLE LIVING WITH A RARE DISEASE ACROSS ALL RARE DISEASES AND ALL EUROPEAN COUNTRIES, BY EQUIPPING THEM WITH THE KNOWLEDGE AND SKILLS THEY NEED TO FIGHT FOR BETTER LIVES

"Konstantin Aniskin"
Syndrome Sanfilippo
Taken by Natalya
Shugaeva, Russia

In 2022, EURORDIS has **supported its strategic objectives through the empowerment of its networks.** This includes:

IN LINE WITH OUR STRATEGIC OBJECTIVE 1 TO SUPPORT "A NEW EUROPEAN POLICY FRAMEWORK TO ACHIEVE MEASURABLE GOALS PROLONGING AND IMPROVING THE LIVES OF PEOPLE LIVING WITH RARE DISEASES BY 2030" AND OUR STRATEGIC OBJECTIVE 3 TO BE "INCLUSIVE OF ALL RARE DISEASES, ALL REGIONS, 'LEAVING NO ONE BEHIND'":

BUILDING THE COMMUNITY AND INFORMING, SUPPORTING AND EMPOWERING OUR NETWORKS

Membership: Reaching the milestone of over 1,000 EURORDIS members

EURORDIS is a grassroots movement of patient organisations active in the field of rare diseases. EURORDIS membership grows steadily every year, and the trend was confirmed in 2022 as EURORDIS has continued to develop and engage its network of members. 48 new members (+3.5%) joined in 2022, reaching the milestone of 1,000 Members in March 2022 for EURORDIS' 25th anniversary.

The membership of EURORDIS is involved in the organisation's activities and in its decision-making processes through: the Member News (a bimonthly newsletter sent to all members in six languages); webinars; Rare Barometer surveys; direct mailings; and face-to-face meetings, in particular the EURORDIS Membership Meeting (EMM) and the General Assembly.

EURORDIS also reaches out to over 2,900 European patient organisations at large, while maintaining and developing the EURORDIS contact database.

European Network of Rare Disease National Alliances and the Council of National Alliances

The National Alliances bring together the many rare organisations within a particular country. The Council of National Rare Disease Alliances (CNA), established and coordinated by EURORDIS, allows national representatives of rare disease patients to work together on common European actions. The main achievements of the CNA so far include: the creation and implementation of Rare Disease Day; the active role of National Alliances in the promotion and development of national plans for rare diseases; the adoption of the "Common Goals and Mutual Commitments between EURORDIS & National Alliances in Europe"; the EU campaign for a new policy framework; advocacy for the integration of ERNs at the national level; and the development of EURORDIS' position on newborn screening.

In 2022, 25 EU countries participated in the CNA, as well as 11 non-EU countries (Albania, Bosnia, Georgia, Kosovo, North Macedonia, Montenegro, Norway, Ukraine, Russia, Serbia and Switzerland). Two CNA workshops took place. The first workshop, in April, was a one-day online workshop for only CNA members (which had 46 participants). The second workshop, in November, was organised over two days – the first day was dedicated to the CNA and the second day gathered CNA and Council of European Federations (CEF) representatives (which had 69 participants). CNA members also continued meeting once a month online for a CNA "hot topics" discussion and they received the monthly "CNA Update" newsletter.

European Network of Disease-Specific European Federations and the Council of European Federations

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.

In 2022, EURORDIS continued coordinating the European Network of Disease-Specific European Federations and the CEF, working more closely with them on key strategic items, in addition to improving access to disease-specific evidence through our Rare Barometer surveys, allowing for more coordinated evidence-based advocacy at the European level. EURORDIS also provided continuous support to 18 of the weakest or younger rare disease European Federations by financing their network meetings. Representatives of the European Federations gathered in November 2022 over two days, which included one day jointly hosted with the CNA and one day dedicated to a peer-to-peer session.

European Network of Rare Disease Helplines

The European Network of Rare Disease Helplines (ENRDHL) aims to better serve the needs of callers by sharing resources, best practices, common tools and knowledge with helplines. The ENRDHL was created in September 2006 and is coordinated by EURORDIS. The ENRDHL aims to increase awareness, efficiency, and best practice standards among its members.

EURORDIS has continued coordinating and developing the ENRDHL by identifying common interests, through analysing the profile of callers and the purpose of their inquiries (Caller Profile Analysis). In 2022, two helplines joined the network – ACHSE in Germany and the Huntington Association in Bulgaria – bringing the total number of network members to 22 helplines from 15 countries. An in-person training was organised on how to answer to complex cases, gathering 24 trainees and two trainers. The annual meeting of the ENRDHL took place (in a hybrid format) in July 2022, gathering 25 participants. The 15th Annual Caller Profile Analysis conducted in October 2022 analysed a total of 1,806 enquiries from 18 helplines, of which 1,086 related to specific rare diseases. This year, the work resulted in an analysis of the diseases in question, for each helpline and for the whole network, and a comparison with the Orphanet database.

RareConnect

RareConnect is a safe online tool that connect people living with rare diseases. The platform was initiated by EURORDIS and is now hosted by the Children's Hospital of Eastern Ontario (CHEO), with the participation of EURORDIS and other partners. RareConnect continued to provide a community for people living with a rare diseases, with nearly 3,000 new people joining the community in 2022. Whilst the platform continues to host a number of vibrant communities, a user survey conducted in 2022 shows that the overwhelming majority of users no longer depend solely on RareConnect to find their community, as the number of digital support communities for rare disease has increased greatly and become more accessible through mainstream social media. In 2022, EURORDIS, CHEO and the Steering Committee for RareConnect have been exploring options for the future of RareConnect.

IN LINE WITH OUR STRATEGIC OBJECTIVE 1 TO REACH "A NEW EUROPEAN POLICY FRAMEWORK TO ACHIEVE MEASURABLE GOALS PROLONGING AND IMPROVING THE LIVES OF PEOPLE LIVING WITH RARE DISEASES BY 2030":

BUILDING CAPACITY FOR OUR NETWORKS: TRAINING FOR PATIENT ADVOCATES

Brussels Rare Diseases Week 2023

In 2022, EURORDIS prepared the second (and first in-person) edition of Rare Disease Week (RDW). Besides organising RDW itself and the venue of the delegation in Brussels planned for February 2023, the team delivered the online pre-training programme in November and December 2022. Specifically, EURORDIS created two training modules for RDW participants, in collaboration with the Open Academy, and organised two webinars as a preparation for RDW (one on EU institutions and the other on advocacy and communication skills). The pre-training received a satisfaction rate of 89% from the participants, with the main result being their increased knowledge of EU decision-making processes and advocacy. This pre-training also familiarised RDW participants with EURORDIS's key policy asks, giving them the tools and skills for a successful advocacy work in Brussels.

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO CONTRIBUTE TO EXPANDING TREATMENT "DEVELOPMENT, AVAILABILITY, ACCESSIBILITY AND AFFORDABILITY, PARTICULARLY WITH REGARD TO TRANSFORMATIVE OR CURATIVE THERAPIES: GOAL OF 1,000 NEW THERAPIES WITHIN 10 YEARS", AND THE AMBITION OF ACHIEVING "RESEARCH AND KNOWLEDGE DEVELOPMENT THAT ARE INNOVATIVE AND LED BY THE NEEDS OF PEOPLE LIVING WITH A RARE DISEASE"

EURORDIS Open Academy

The EURORDIS Open Academy empowers patient advocates with the knowledge and skills to take part in patient engagement roles side-by-side with all stakeholders and to advocate for rare diseases on a European and national level.

Our Open Academy offer is composed of various schools for EURORDIS members, and open access e-learning courses on the Open Academy website. At the end of 2022, the training platform had reached over 2,650 registered users from more than 155 countries.

Trainings in 2022 included the EURORDIS Summer School and the EURORDIS Winter School, comprising e-learning modules, pre-training

webinars and online intensive days delivered in English. Due to the COVID-19 pandemic, there were no face-to-face sessions for any of these trainings in 2022.

The EURORDIS Summer School on Medicines Research & Development provided rare disease patient advocates with the knowledge and skills needed to become experts in medicines' research and development. The 15th edition of the Summer School took place online on 6-10 June 2022, with pre-training running from January to June 2022, including e-learning modules and three webinars.

22 trainees and four researchers, from 16 countries, participated in the training, covering topics including market access, regulatory frameworks, ethics and pharmacovigilance.

The EURORDIS Winter School on Scientific Innovation and Translational Research was launched with the aim of deepening patient representatives' understanding of how pre-clinical research translates into real benefits for rare disease patients. The training equipped participants with knowledge and skills so they could be empowered to effectively participate in discussions with researchers, policymakers and companies responsible for research or research infrastructures.

The 5th edition of the EURORDIS Winter School took place online on 21-25 March 2022, with pre-training running from October-March 2022, including e-learning modules and three webinars. 30 patient representatives from 19 countries and representing over 26 rare diseases participated in this training course. Specific topics covered included genetics and diagnosis, genome editing tools, translational research, pre-clinical models, drug repurposing, IRDiRC and European Joint Programme on Rare Diseases (EJP RD) activities, patient participation in research projects, and European Reference Networks.

The Open Academy also contributed to the organisation of the international course "Training for patient representatives and advocates on leadership and communication skills" – part of a series of training activities proposed by the EJP RD.

The two-day course took place online on 10-11 November 2022, and was preceded by webinars. The 2022 edition was hosted by the Istituto Superiore di Sanità (ISS). The course aimed to teach participants presentation, negotiation and leadership skills, through plenary presentations and role-playing sessions. The course endeavoured to improve participants' ability to communicate and represent needs in an assertive way, influence important decisions, and guide strategic decision-making when engaging with healthcare providers and other rare disease stakeholders.

In 2022, the Open Academy Digital School wrapped up with the final online course and webinar about video creation with smartphones. Participants of the webinar learned: basic tips on how to film videos with their phones; the importance of knowing what, where and when their audience consumes content; and how the social media platform that they choose to share a video on should inform the type of video that they produce.

Further to a review of our training offering, we have decided to implement the following changes in 2023:

- The two Schools will be run in parallel, from January to June, with post-module webinars in September.
- There will be a common application process for both Schools to maintain or increase the number of participants.
- A rebranding of the Schools will focus on topics rather than the "Winter" and "Summer" labels.
- There will be continuous learning through the introduction of post-School online meetups on relevant topics and extended online masterclasses.
- There will be increased alumni outreach and engagement through alumni newsletters, alumni online meetups, online masterclasses, and a EURORDIS-animated Facebook group.

PATIENT ENGAGEMENT IN LIFECYCLE DEVELOPMENT

Scientific Committees and Working Parties of the European Medicines Agency

EURORDIS is in the unique position of having, and having had, 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); the Pharmacovigilance and Risk Assessment Committee (PRAC); the Scientific Advice Working Party (SAWP); and the Committee on Herbal Medicinal Products (HMPC).

Identification of and support to rare disease patients' participating in EMA activities and EMA Scientific Committees:

- **Committee for Orphan Medicinal Products (COMP):** EURORDIS supported the three rare disease patient representatives appointed to the COMP, and the participation of one EURORDIS expert at the monthly plenary meetings and the EMA Strategic Learning Review Meetings (SLRMs) that took place online in March, and in Bonn September 2022.
- **Committee for Advanced Therapies (CAT):** EURORDIS supported one rare disease patient representative appointed to the CAT in July 2022, whose application to the CAT was also supported by EURORDIS. The patient representative participated in the monthly plenary meetings and the EMA Strategic Learning Review Meeting that took place in Paris December 2022.
- **Paediatric Committee (PDCO):** EURORDIS supported the two rare disease patient representatives (member and alternate) appointed to the PDCO who participated in the monthly plenary meetings and the EMA SLRMs that took place online in March, and in Prague in October 2022. EURORDIS supported the organisation of one of the SLRM sessions on newborn screening (NBS) with patient participation. The PDCO alternate became a member in November 2022. For the next PDCO mandate, EURORDIS proposed and issued a letter of support for two patient representatives, and endorsed and proposed a patient representative as a EURORDIS candidate, as an answer to the European Commission's call of expression of interest to appoint Civil Society representatives to the PDCO (deadline 3 November 2022).
- **Pharmacovigilance Risk Assessment Committee (PRAC):** EURORDIS supported the two rare disease patient representatives (member and alternate) appointed to the PRAC in June 2022 who participate in the monthly plenary meetings and the EMA SLRMs that took place online in March, and in Prague in October 2022. (Of note: the previous mandate ended in February 2022 and the two patient representatives did not reapply).
- **Scientific Advice Working Party (SAWP):** EURORDIS has worked closely with the EMA, the Public Engagement Team and the SAWP Secretariat to analyse Protocol Assistance dossiers, and to identify and suggest patients from the EURORDIS network or beyond, in particular patients that have been trained on research and development processes. For Protocol Assistance, EURORDIS reviewed 130 dossiers in 2022, of which 28 needed patient input. EURORDIS subsequently identified, mentored and involved 24 patients for input, and supported the EMA by identifying patients for scientific advice and Scientific Advice Groups (SAGs) as well as other activities.
- **Committee on Herbal Medicinal Products (HMPC):** EURORDIS contributed to the review of herbal monographs. However, there was no activity in this domain in 2022 as our Drug Information, Transparency and Access (DITA) Task Force began a new mandate under a new composition. In 2023, a call for volunteers to review the herbal monographs will be made.

Patient representatives at Patients' and Consumers' Working Party and other meetings

The Patients' and Consumers' Working Party (PCWP) – of which EURORDIS is a member – is a unique forum where all EMA Scientific Committees meet with patients and consumers. François Houÿez (member) and Russell Wheeler (volunteer, Leber Hereditary Optic Neuropath UK Society) were appointed as alternate members by the EURORDIS Board in September 2019.

A number of PCWP meetings – some of which combined with the Healthcare Professionals' Working Group – were held. These included:

- One meeting with all eligible organisations (patients, consumers and healthcare professionals).
- Other meetings on the launch of the Regulatory Science Research Network.
- An information session on advanced therapeutic medicinal products (ATMPs).
- A workshop on patient experience data in EU medicines development and regulatory decision-making.

EURORDIS, who contributed to the **Council for International Organizations of Medical Sciences (CIOMS) report on "Patient Involvement in the Development, Regulation and Safe Use of Medicines",** had several opportunities to present the published report at the EMA, including to all eligible patient organisations meeting on 15 November 2022.

Another important piece of work in 2022 was EURORDIS' contribution to the Decentralised Clinical Trials project, which published in EudraLex in December 2022.

Additional activities at the European Medicines Agency

Review of EMA documents for public information

The EMA is responsible for providing information about medicines authorised via the centralised procedure that includes information directed to people living with rare diseases and the wider public. During the preparation of this information, the EMA interacts with patients' and consumers' organisations to ensure that their information is appropriately worded and comprehensible to the target audience.

EURORDIS is extensively involved in these activities. In 2022, EURORDIS staff or volunteers reviewed **over 150 documents for public information,** including:

- **Public summaries of opinion (PSOs).** In 2022, 144 PSOs were reviewed by EURORDIS for orphan drug designations. PSOs explain in lay terms the disease, the number of individuals potentially affected in the EU, the medicinal product, and the stage of the product's development. A link to the product's sponsor, and to EURORDIS and Orphanet are provided in the PSOs to help patients obtain more information.
- **Medicine overviews.** In 2022, EURORDIS reviewed two medicine overviews. Medicine overviews are shorter documents based on the European Public Assessment Reports (EPARs) 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 leaflets (PLs) for Orphan Medicinal Products.** In 2022, EURORDIS reviewed eight PLs. PLs contains information on what the orphan 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. However, the number of PLs reviewed by EURORDIS has been decreasing over the years as the EMA Stakeholder Engagement unit establishes working relations directly with relevant rare diseases organisations.

Other documents disseminated by the EMA to the public and EURORDIS members include the EMA's monthly highlights newsletter, safety alerts, information on referrals, information on medicines with the black symbol, and risk management plan summaries.

Committee for Medicinal Products for Human Use (CHMP)

Since 1 January 2021, the EMA has been running a new pilot procedure from the beginning of the evaluation of a 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 and risks of a medicine. These questions relate to: the impact of the disease in the life of patients, treated or not; patients' unmet needs; the relative effects of patients' treatments; any aspect of the disease the CHMP does not understand well; groups of patients with different manifestations of the disease, or different responses to treatments; expectations from a new treatment; treatment constraints that are acceptable to patients; and the experiences of patients who took part in clinical trials.

To answer these questions, EURORDIS explores the websites of relevant patient organisations that might have published information relevant to the questions, contacts its own members, and conducts interviews with two to six patients on average.

EURORDIS contributes to the activities of the CHMP in different ways. These include:

- Taking part in early contacts between CHMP rapporteurs and patients. A report was published in 2022 on this activity, and the decision to move the procedure from a pilot phase to a permanent exercise was proven to be useful.
- Identifying patients for Scientific Advisory Group (SAG) meetings.
- Mentoring patients invited to oral explanations with the Marketing Authorisation Applicant.
- Reviewing documents intended for the public.

Regarding the early dialogue between CHMP rapporteurs and patients, EURORDIS published seven notes (for seven different products), representing 21 interviews with patients of one hour each. To interview 17 patients, it was necessary to contact 60 patients.

- **EMA task forces and working groups:** The EURORDIS Director of Treatment Information and Access was appointed as a full member of the EMA Emergency Task Force, in the scope of the extended mandate of the EMA. In 2022, this represented 36 meetings of two to three hours each. A EURORDIS volunteer member from our DITA Task Force is a member of the EMA Raw Data Advisory Group, which is guiding a new pilot project analysing the raw data from clinical trials.
- **EMA training:** One EURORDIS member of staff took part in the EMA's online training for patient advocates on 17-18 October 2022, which covered the topics of regulatory pathways for medicines, scientific advice, document reviews, and synergies between regulators and Health Technology Assessments (HTAs).
- **EMA Management Board:** EURORDIS had put forward one member of staff as a candidate for the selection procedure to appoint Civil Society representatives to the Management Board of the EMA. This candidate was selected and began her mandate on 15 June 2022. Two representatives of patient organisations are members of this Board, which is the EMA's integral governance body. The Management Board has a supervisory role with general responsibility for budgetary and planning matters, the appointment of the EMA's Executive Director, and the monitoring of the EMA performance. The mandate of the Board lasts three years.

Supporting patient involvement in Health Technology Assessment (HTA) activities: EURORDIS Task Force on HTA

The EURORDIS Task Force on Health Technology Assessment is a group of eight EURORDIS volunteers from member organisations from eight EU countries, experienced and trained in HTA. The Task Force aims to share experience and knowledge between EURORDIS members and staff about all HTA-related matters, and informs EURORDIS' positions. The objectives of the Task Force 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 Task Force are to raise awareness among patients and the HTA communities about the value of patient engagement, and to explore and discuss new methods of analysing cost-effectiveness.

The previous Task Force mandate came to an end in June 2022. This was followed by a call for expression of interest to join the Task Force and a renewal of the group.

Task Force members discuss the assessment, pricing, and reimbursement of treatments across Europe and advise EURORDIS on all aspects of HTA, from assessment 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 (several times per year online, and face-to-face twice per year), raise awareness among the patient community, participate in policy debates, and contribute to public consultations (led by the European Commission, EUnetHTA, or learned societies). The Task Force replied to consultations from EUnetHTA21 between April and November 2022, which have now been published.

The HTA and DITA task forces' joint meeting was held in May 2022. Both task forces reviewed the achievements from their respective mandates (2019-2022), considered the revision of the pharmaceutical legislation, and discussed: a proposal to the GetReal Institute; the analysis of raw data; and the engagement of patients in HTA. Feedback on the two task forces from

participants and final remarks, which included a point on the renewal of the task forces' mandates, closed the meeting.

After having been renewed through a call for interest and the selection of participants, the new **HTA Task Force met in-person** in Paris in December 2022. The Task Force members brainstormed on how they could contribute to EURORDIS' projects and initiatives, in addition to other pieces of work they could be involved in (such as consultations and conference attendances), in order to: support patient engagement in European HTA and related HTA activities; contribute to the implementation of the HTA Regulation (2021/2282); and advocate for improved access to treatment for rare diseases.

Supporting patient involvement in dialogue with payers through the Mechanism of Coordinated Access to Orphan Medicinal Products (MoCA)

In 2022, EURORDIS identified, supported and involved three rare disease patients in different MoCA meetings. EURORDIS contributed to the MoCA Steering Committee with the aim of raising awareness about the MoCA, and followed up on specific actions such as: updating the EURORDIS MoCA webpage; submitting the MoCA poster at the ECRD 2022; drafting a paper about the experience of the first 10 years of the MoCA (publication expected in 2023); and delivering a presentation about the MoCA at the Czech EU Presidency Expert Conference on Rare Diseases in October 2022.

Supporting patient involvement in developing quality information on medicines: Drug Information, Transparency and Access Task Force (DITA Task Force)

The DITA Task Force represents a group of volunteers who are trained (via the EURORDIS Open Academy Schools) and active in issues concerning therapeutic development and the evaluation of medicines for rare diseases. The Task Force follows the work plan of the Patients' and Consumers' Working Party (PCWP) at the EMA. The Task Force also supports or advises EURORDIS representatives who participate in EMA Scientific Committees and Working Parties. The Task Force is consulted on papers prepared by EURORDIS. The General Terms of Reference were approved by the Board of Officers in 2009.

The Task Force participates in conference calls when necessary (if certain issues require discussion) and holds face-to-face meetings. Following an online meeting in March 2022, a final face-to-face meeting of the 2019-2022 task force took place in May 2022. The call for candidates for a new Task Force was made in September 2022, when a new EURORDIS HTA Patient Engagement Manager was hired. After the appointment of DITA Task Force members by the EURORDIS Board of Directors, activities resumed, with a first meeting of the new Task Force in Paris in November 2022. The Task Force is now composed of 13 members, of whom five were already members from 2019 to 2022.

The two main topics addressed by the DITA Task Force in 2022 included:

- Contributions to the ICH Guidelines on General Principles for Clinical Trials, which were led by the departing Task Force.
- Contributions to patient registries and improved access to data, featuring a discussion with a Clinical Research Organisation (CRO). These topics were addressed by the new Task Force.

In 2022, the DITA Task Force contributed to EMA discussions or guidelines on the following topics:

- EMA/HMA guidelines on Decentralised Elements in Clinical Trials.
- ICH Principles for Clinical Trials.
- The revision of the pharmaceutical legislation.
- The Clinical Trial Information System and its interface with the public.
- The EMA pilot on raw data.
- The manufacturing of ATMPs.
- The role of the public sector.
- Involvement in reviewing and contributing to the regulatory science research topics, and patient involvement in regulatory science research.

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "OPTIMISED DATA AND HEALTH DIGITAL TECHNOLOGIES FOR THE BENEFIT OF PEOPLE LIVING WITH A RARE DISEASE AND SOCIETY AT LARGE":

EURORDIS Digital Advisory Group (DAG)

2022 marked the launch of FACILITATE, an Innovative Health Initiative (IHI) project aiming to return clinical trial data to trial participants, while creating a framework that would allow for the reuse of the data for further research.

EURORDIS continued to coordinate the Digital and Data Advisory Group (DAG), which was involved in the qualitative research on the importance of returning clinical trial data to trial participants, as well as what types of data – and in which contexts the data – would be relevant to them.

The DAG has also been engaged in reviewing the ethical framework of Screen4Care – a project aiming to accelerate rare disease diagnosis through genetic newborn screening and digital technologies.

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "HIGH QUALITY NATIONAL AND EUROPEAN HEALTHCARE PATHWAYS, INCLUDING CROSS-BORDER HEALTHCARE – GOAL OF IMPROVED SURVIVAL AVERAGE BY 3 YEARS OVER 10 YEARS, 1/3 REDUCTION OF MORTALITY UNDER 5 YEARS OF AGE":

Supporting patient partnership within the European Reference Networks (ERNs)

In 2016, EURORDIS, in collaboration with the European rare disease community, established 24 European Patient Advocacy Groups (ePAGs) to optimise the involvement of patient organisations and patient representatives in the strategic and operational delivery of the 24 ERNs. Each ePAG corresponds to the scope of one of the 24 individual ERNs, aligning patient organisations, clinicians, experts and researchers working on the same rare or complex disease or highly specialised intervention. These groups are composed of appointed patient representatives, some of whom were elected in 2016 and others who have been co-opted.

ePAG advocates play a fundamental role to connect the Networks with the wider rare disease patient community and, where relevant, to champion the diversity of views of the patient community relevant for each ERN. Today, there are 321 ePAG patient advocates across the 24 ERNs.

In 2022, EURORDIS supported a **patient partnership approach** in the development and consolidation of ERNs.

EURORDIS contributed to the **ERNs' Five-Year Evaluation,** and reviewed the evaluation framework that was presented at the final workshop organised by the European Commission and the AMEQUIS Consortium. EURORDIS organised two webinars to explain in simple terms the framework and process to ePAG advocates, with a focus on their role in the evaluation process. ERN project managers and clinicians were also invited to the Demystifying AMEQUIS webinar and the Evaluation webinar. EURORDIS developed a guide for ePAG advocates based on the ERN Evaluation Manual & Toolkit, specifically to support patient representatives who are active in the ERNs to understand their role and contribution in the evaluation process of the Networks. In addition, EURORDIS conducted 15 one-to-one calls with individual ePAGs. EURORDIS and the patient representatives that are part of the ePAG AMEQUIS Task Force met on several occasions throughout the year to reflect, exchange and discuss the topic of ERN evaluation.

Activities to ensure good governance and representativeness included building on the work done in 2021 by a small working group to streamline the rules on patient engagement in the ERNs. In March 2022, the group shared with all ERN Coordinators a new approach to structure the involvement of patient organisations and patient representatives in the ERNs.

This new approach featured two different levels of patient engagement, and to facilitate its implementation, the group developed four governance templates that need to be adapted by each ERN to reflect their specific structure. CRANIO, ERNICA, GUARD HEART and eUROGEN ERNs have used the templates to update their terms

of reference for patient engagement and by-laws. EURORDIS also updated and reviewed the ePAG Constitution that was then adopted by the EURORDIS Board of Directors to describe more clearly our support structure and methods.

To improve representativeness and ensure optimal patient representation in the ERNs, EURORDIS has been actively supporting ERNs to recruit new patient representatives from underrepresented countries and diseases. Particular focus was placed on the recruitment of patient organisations from Eastern and Nordic European countries, and from specific diseases. 81 expressions of interest for 21 ERNs were received from a total of 23 countries, including from Eastern European and Nordic countries. EURORDIS received expressions of interest from 52 candidates – the rest were sent directly to the ERNs management teams. Of these 52, six candidates have decided not to proceed with the full application and two have been officially appointed as ePAG advocates. The onboarding process for the rest will continue in 2023.

Activities to conduct patient engagement within the ERNs

EURORDIS has moved progressively from providing hands-on support to individual ePAG groups, to supporting patient representatives via cross-ePAG working groups (WGs) focusing on transversal topic areas. Through these WGs, EURORDIS supports and provides patient representatives involved in the ERNs with the information, knowledge and skills that they need to engage and partner effectively with clinicians in the Networks' collaborative activities.

In 2022, EURORDIS led and managed several ePAG WGs. These included:

- The **ePAG Steering Committee,** which convened four times to discuss strategic aspects related to structuring and enhancing patient involvement in the ERNs.
- The **Connecting Patients with ERNs Working Group,** which held five calls in 2022 to facilitate collaboration between ePAG advocates and Rare Disease National Alliances on ERNs. The group also developed communication resources to improve information sharing at the local and national levels.
- The **ePAG Clinical Practice Guidelines Working Group,** which hosted four webinars in 2022 on the topics of: the added value of patient involvement in guideline development; producing plain language summaries; the roadmap to guideline implementation; and the appraisal and adoption of an existing guideline. The group supported patient partnership in the development and implementation of clinical practice guidelines (CPG) and other clinical decision support tools (CDST).

"Rare Diseases are often invisible", Denys-Drash Syndrome, Belgium

- The **ePAG Research and Registries Working Group,** which held four calls in 2022 to disseminate information on research activities and support patient involvement in research.
- The **ePAG Training and Education Working Group,** which held four calls in 2022 to develop training and capacity-building materials to address the training needs of ePAG advocates.
- The **ePAG AMEQUIS Task Force,** which organised two calls and two webinars in 2022 to support patient involvement in the ERNs' monitoring, evaluation, and quality improvement system. Topics covered included Demystifying AMEQUIS and patient involvement in the Five-Year ERN Evaluation.
- The **Patient Partnership Working Group,** which held two calls in 2022 to enhance collaboration with ERN project managers and exchange information on tools and processes for patient involvement in the ERNs.

Through this working group structure, EURORDIS and the patient representatives involved in

the ERNs share information and updates, learn from each other, develop materials, and support ePAG advocates to engage on important topics. In 2022, the EURORDIS team has held in total 27 meetings with the patient representatives in these WGs.

Developing tools to enable patient engagement in the ERNs

In 2022, EURORDIS developed practical guides to support the work of ePAG advocates, including: a guide on clinical practice guidelines development; a guide on how patient organisations can listen to the community; and a guide on the ERN Evaluation (including a Q&A Factsheet on the Evaluation). A toolkit was also published on good practices for facilitating the integration of ERNs into national health systems.

Pilot coaching on patient-clinician partnership in the ERNs

Based on the need to further improve the collaboration of patient representatives and clinicians, EURORDIS organised three team coaching sessions with clinicians and patient representatives of ReCONNET (7.5 hours), ERN Lung (2 hours) and ITHACA ERN (6 hours). A total of 16 clinicians and ERN project managers and 25 patient representatives participated in these sessions.

The purpose of these sessions was to support a strong patient-clinician partnership culture by identifying common goals, listening to each other openly and actively, and learning how to work together more effectively.

Annual all-ePAG meeting

All 300 ePAG advocates were invited to participate in the annual all-ePAG meeting along with ERN clinicians and project managers. This year, the meeting comprised five independent online sessions around the theme: "Listening to your patient community."

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "INTEGRATED MEDICAL AND SOCIAL CARE WITH A HOLISTIC LIFELONG APPROACH AND INCLUSION IN SOCIETY: GOAL OF REDUCING THE SOCIAL, PSYCHOLOGICAL AND ECONOMIC BURDEN BY ONE THIRD":

EURORDIS Social Policy Action Group

The Social Policy Action Group (SPAG), is a group of volunteer rare disease advocates who disseminate and contribute to the positions of EURORDIS and its members, advocating for holistic and integrated care. The SPAG ended its latest three-year mandate in March 2022. At the time, eight representatives composed the SPAG. Following the end of the mandate, EURORDIS initiated a reflective process on the future of the SPAG. This included considering the possibility of expanding the group to include experts from other organisations involved in the social field, defining the scope necessary to ensure effective advocacy moving forward, and assessing the internal resources needed to coordinate and support the SPAG. Through this process, EURORDIS aimed to determine the best approach to sustain the effectiveness of the SPAG in its advocacy efforts.

In the summer, the EURORDIS Board of Directors decided to create the position of Social Policy and Initiatives Director. This position was filled in at the end of October, and one of this director's tasks is to relaunch the SPAG during 2023, with new Terms of Reference and a renewed membership.

ORGANISING THE EURORDIS MEMBERSHIP MEETING 2023 ON A HOLISTIC APPROACH TO PEOPLE'S NEEDS AND FULL INCLUSION IN SOCIETY

EURORDIS decided in 2022 – together with the EURORDIS Membership Meeting Programme Committee – to host the **EURORDIS Membership Meeting 2023 (EMM2023)** in Stockholm on 25-27 May 2023. EMM2023 will centre on the theme of a **"lifelong holistic approach to people's needs and their full inclusion in society",** in line with our Strategic Objective to deliver on the goal of social inclusion. EMM2023 will gather up to 250 participants from our members to network and participate in capacity-building workshops, including on digital health.

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "EARLIER, FASTER AND MORE ACCURATE DIAGNOSIS: GOAL OF DIAGNOSIS WITHIN SIX MONTHS":

EURORDIS Newborn Screening Working Group

The 11 Key Principles for Newborn Screening were published in January 2021 and have been translated into 13 languages. In 2022, the EURORDIS Newborn Screening Working Group continued discussions on harmonising newborn screening in the EU member states and how to take action accordingly. The Working Group members also joined several webinars and conferences.

IN LINE WITH OUR STRATEGIC OBJECTIVE 3 OF BEING "INCLUSIVE OF ALL RARE DISEASES AND REGIONS", AND OUR "LEAVING NO ONE BEHIND" GOAL OF EXPANDING OUR SCOPE TO COVER ALL 48 COUNTRIES IN GEOGRAPHICAL EUROPE, PRIORITISING EASTERN AND SOUTHERN MEMBERS OF THE EU, EUROPEAN ECONOMIC AREA, AND EU ACCESSION COUNTRIES, WE HAVE BEEN UNITING SINCE FEBRUARY 2022 FOR THE TWO MILLION UKRAINIANS LIVING WITH A RARE DISEASE WITHIN AND OUTSIDE OF UKRAINE AS A RESULT OF THE WAR:

On 15 March 2022, in response to Russia's invasion of Ukraine, EURORDIS launched its Ukraine Response programme, aimed at supporting the Ukrainian rare disease community. EURORDIS' response has been to immediately respond to the needs of Ukrainians living with a rare disease, and to advocate on behalf of their specific needs with the support of our multi-stakeholder networks.

Over the course of the year, a large cohort of Ukrainians living with a rare disease felt a direct impact of the programme on their lives. In particular, thanks to a partnership with Airbnb.org, more than 300 families received 30 days of free housing in Europe as they were fleeing the war. Between June and November 2022, over 500 Ukrainian families affected by rare diseases were supported in Poland through the "Razem z Ukraina" project. This initiative provided families with access to dedicated family assistants for consultations, essential medical equipment, psychological support, housing options, and reimbursement for travel costs to and from Europe. In addition to that, EURORDIS supported the work of a Rare Disease Hub for Ukraine – an entity tasked to ensure very close partnership with ERNs to support the medical management of families coming from Ukraine.

In the field of advocacy, EURORDIS worked on informing European, national and Ukrainian authorities of the specific challenges facing Ukrainians living with a rare disease and the ways to address them, in particular through a series of reports on the specific issues in neighbouring countries, or by voicing the concerns of our patient organisations as part of the Supporting Ukraine network of the European Commission. Where possible, we connected patient and humanitarian aid organisations in Ukraine and neighbouring countries to deliver frontline response to Ukrainians living with a rare disease and their families.

RAISING AWARENESS & INFORMING

Recurrent EURORDIS events that support EURORDIS strategic objectives:

Rare Disease Day 2022

Rare Disease Day is an annual, awareness-raising event co-ordinated by EURORDIS at the international level and by national alliances and patient organisations at 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 people's lives.

In 2022, for its 15th edition, Rare Disease Day events took place in **over 110 countries** and regions on every corner of the globe, despite the continuation of the COVID-19 pandemic. As part of the Day's focus on equity, the global campaign promoted six 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, and we welcomed **five new countries** to the campaign: the Ivory Coast, Kenya, Greece, Kazakhstan and Portugal. A variety of awareness events were held, including political conferences online, medical symposia, family days, and marches.

In addition to holding events, people around the world were inspired by the visuals, campaign in-

formation, photos, and Rare Disease Day-branded materials available online. These materials enabled people to participate in the Day's interactive "Share your colours" call to action.

More than 650 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.

The Day received widespread media coverage from various parts of the world, with participation from politicians, researchers, medical professionals, and policymakers across Europe, the United States, and many other countries and regions.

Website

The Rare Disease Day website continued to be a central point for people living with a rare disease to download usable campaign materials. They could also visit the site to gain information on the worldwide movement and all of the events taking place around the world for Rare Disease Day.

Webinar series: Capacity building

Two webinars were held during the months leading up to Rare Disease Day for the global community to share their experiences and help prepare for the event. The two webinars were held on the following subjects:

- How to speak to young children about rare diseases.
- How to use Rare Disease Day to advance your advocacy objectives.

EURORDIS Black Pearl Awards 2022

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, policymakers, scientists, companies and media at a 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 **11th edition of the EURORDIS Black Pearl Awards was celebrated online on 8 February 2022.** The event gathered 377 attendees representing all stakeholder groups of the rare disease community and connecting people from 38 countries.

Winners of the EURORDIS Black Pearl Awards 2022

- **Lifetime Achievement Award: Dr William Gahl (USA).** For his long-standing dedication to the rare disease community (with a particular focus on cystinosis), and for establishing the NIH Undiagnosed Diseases Program (which made more than 300 rare disease diagnoses and discovered 30 new genetic diseases), later expanding the programme to a national Undiagnosed Diseases Network and a worldwide Undiagnosed Diseases Network International.
- **European Rare Disease Leadership Award: Dr Anne-Sophie Lapointe (France).** For her extraordinary journey from mother of two children with a rare disease, to volunteer in a patient organisation, to public servant. Dr Lapointe has been demonstrating incredible dedication to the cause as President of Vaincre les Maladies Lysosomales (VML), at the National Council of Rare Diseases France, at the Ethical Committee of INSERM, and then leading the French National Plan on Rare Diseases and the National Plan on Genomics at the Ministry of Health.
- **Policy Maker Award: Professor Hans-Georg Eichler (Austria).** For his strong advocacy for drugs licensing in his capacity as Senior Medical Officer of the European Medicines Agency, as well as his role at the forefront of the campaign for clinical

research to be more innovative to reduce the time and money it takes for a drug to come to the market. The award also recognises Prof. Eichler's active engagement with the Clinical Trials Transformative Initiative (CTTI) and with several EURORDIS projects.

- **Scientific Award: Prof. Franz Schaefer (Germany).** For his positive impact in rare disease research and patient communities on an international level, particularly through initiatives such as the eRare-funded PodoNet Project for Research in hereditary and immune mediated steroid resistant nephrotic syndrome, the European Joint Programme for Rare Diseases, ERICA (the ERNs' research coordination programme), the ERN Coordinators Group, and several of the cross-ERN Working Groups.
- **Young Patient Advocate Award: Danielle Drachmann (Denmark).** For founding the Ketotic Hypoglycemia International (KHI) – the world's largest patient organisation for patients with idiopathic (unexplained) ketotic hypoglycemia, for her role of Patient Representative at the European Health Parliament and the European Medicines Agency, and her work at the Center for Research with Patients and Relatives and at the Research Committee at H.C Andersen's Children's Hospital (Odense University Hospital).
- **EURORDIS Volunteer Award: Graham Slater (UK).** For his outstanding contributions to several organisations and projects on rare diseases, such as the TOFS (UK EA support group), the Esophageal Atresia Global Support Group (EAT), the UK National Health Service's Clinical Reference Group for specialised surgery in children, the European Reference Network for rare Inherited and Congenital (digestive and gastrointestinal) Anomalies (ERN ERNICA), and several of EURORDIS' working groups.
- **EURORDIS Members Award: Childhood Cancer International Europe (Netherlands).** For their outstanding advocacy of childhood cancers on a national and international level, for establishing an "EU network of youth cancer survivors", and for their role as patient advocates in the European Reference Network for Paediatric Cancer and several other EU projects such as Harmony, PanCare and Accelerate.

- **EURORDIS Media Award: Ewenflix (France).** For offering all people living with a rare disease and their families free and accessible online tools where patients, parents and caregivers talk about how rare diseases affect their everyday life, and share personal tips or advice.
- **Company Award for Innovation: Lysogene.** For their work as a pioneering gene therapy company which has established itself among major players driving European-led innovation, in just over ten years. The award particularly recognises Lysogene's promising advances towards delivering novel treatments in neurodegenerative and neurodevelopmental disease areas with high unmet medical needs.
- **Company Award for Patient Engagement: Spinal Muscular Atrophy (SMA)-NBS Alliance.** For the collaborative effort to ensure that newborn screening programmes in all European countries diagnose all newborn children with SMA. EURORDIS welcomed the companies' mutual engagement with patient advocates and multiple stakeholders around a single cause, setting a model example which has the potential to encourage similar initiatives across other rare diseases.
- **Company Award for Health Technology: Aparito.** For creating innovative technologies that report patient experiences accurately. By placing patient needs at the heart of the solution, Aparito's expertise facilitates medicines development and contributes to addressing complex regulatory challenges.
- **Social Media Award (open to public voting): Milica with @my.baby.noa (Serbia).** For educating and bringing people closer to the life of a child with special needs via her social media platforms, providing hope and positivity to patients living a similar experience.

European Conference on Rare Diseases and Orphan Products 2022

The European Conference on Rare Diseases and Orphan Products is recognised globally as the largest, patient-led rare disease event in which collaborative dialogue, learning, and conversation take place, forming the groundwork to shape future rare disease policies.

The 11th edition of the European Conference on Rare Diseases & Orphan Products (ECRD 2022) was a virtual event for the second consecutive time. By maintaining this event online, it remained accessible to all, reaching over **800 stakeholders of the wider rare disease community from 61 countries.**

Over the five days of the conference, participants discussed how rare disease policy should be put into action in Europe to work towards meaningful goals, aligned with the conclusions from the Rare 2030 foresight study and the UN Sustainable Development Goals (SDGs) – to improve health and well-being, to reduce inequalities, and to foster innovation. All this helped to build momentum for post-ECRD 2022 follow-up actions and their implementation, to secure a better future for all people living with a rare disease.

Through a custom-built platform, registrants were able to participate in **19 sessions with over 120 expert speakers and chairs** and meet with fellow attendees in both facilitated and open networking sessions. Over 220 high quality e-posters were showcased on the platform.

For the first time ever, four online satellite meetings were hosted by National Rare Disease Alliances, gathering key opinion leaders and decision-makers on a Member State level to consider how EU policies and national strategies will align in the coming years, with a particular focus on how a new policy framework for rare diseases could drive national efforts.

INFORMING AND EMPOWERING OUR NETWORKS: COMMUNICATION AND DISSEMINATION

Communication strategy review

Throughout 2022, EURORDIS conducted a review of its Communication and Dissemination Strategy – aligned with the new EURORDIS strategy for 2022-2023 – to leverage our networks. A one-page strategic plan was developed to communicate EURORDIS' vision and action-oriented strategic plan to align every employee in the organisation. The communication strategy is expected to be finalised in 2023.

EURORDIS website

In 2022, we launched a new website. We continued translations and uploaded all relevant content. The website is available in seven languages and is populated with relevant content regularly. The website saw an increase in the number of visits in 2022, reaching 518,581 annual visits.

EURORDIS newsletters

eNews

The EURORDIS eNews is a monthly English-language newsletter that communicates breaking news of interest to patient advocates, people living with a rare disease and their families, and policymakers.

Throughout 2022, **12 eNews issues** were written, produced and distributed via email. This eNews publication, available free of charge, features a lead story relating to the work of EURORDIS, and provides stakeholders with news on the latest EURORDIS activities and other developments in the rare disease community.

Following the lead story are short summaries of news, which link to the specific activity or section of our website. Each eNews lead story ap-

pears on the website (eurordis.org) in full, regularly appearing on the homepage.

In 2022, typical headlines which followed the lead story included the following:

- RareConnect
- Patients, organisations, services
- International Events (featuring a link to the event's website)
- Members' Corner (featuring a link to the EURORDIS member's website)
- Living with a Rare Disease
- New Orphan Drug designations and Market Authorisations
- European Conference on Rare Disease and Orphan Products
- What's new at EURORDIS
- Get involved
- EURORDIS Black Pearl Awards

In 2022, lead story topics included:

- European Health Data Space: Unlocking the potential of health data for rare diseases.
- Demystifying HTA: patients' role in new legislation.
- Involving those who matter most: Improving patient engagement in the development of medicines.

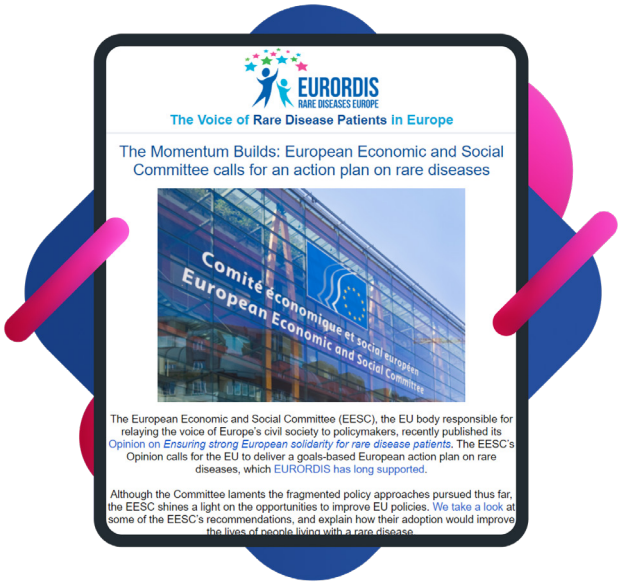
Member News

The EURORDIS Member News provides updates relevant to the rare disease patient community, as well as offering a space for us to remind our members of EURORDIS activities that they can participate in (such as events and webinars) and publications on EURORDIS policy positions. This newsletter is translated into six languages and disseminated to over 2500 contacts.

2022 saw the very successful **EURORDIS Member News enter its seventh year,** and it was sent twice a month (except in August), so a total of 22 editions were sent to recipients over the course of the year.

Council of National Alliance Newsletter

A Council of National Alliances (CNA) specific newsletter, "CNA Update", was launched in 2020 to better target these important patient organisation members, which are heavily active in advocacy activities at the national level.



The CNA Update includes sections for updates on the CNA meetings, the latest news (such as Rare Barometer surveys and results), and updates on advocacy activities. 11 CNA Updates were sent in 2022.

Social Media

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

We continued to successfully use our social media pages to publicise activities and interact with the rare disease community, particularly the patient community. Growth was seen on Instagram, as more engaging content was developed to interact with the users. Social media content in 2022 included:

- Scheduled content taken from the eNews.
- Spontaneous content received from staff.
- Information of interest from, and to, the rare disease community (including information received from members and projects).
- Posts encouraging the public to register for our events, take part in our capacity-building trainings, respond to our surveys, and submit photos to the EURORDIS Photo Award.
- Sharing important information with the rare disease community relating to the response to the war in Ukraine.

Launch of the new monthly EURORDIS Podcast "Rare on Air"

In **November 2022, EURORDIS launched its very first podcast, entitled "Rare on Air",** which explores the unique experiences, challenges and successes of people from our community, and investigates how a better world can be built for those who have a rare condition.

Two episodes went out in November-December 2022, focusing on "Breaking down barriers: Living with a rare disease and disability" and "Screening at birth: The key to longer, healthier and better lives". The podcast targets EURORDIS members, people living with a rare disease, and patient advocates as a primary audience, as well as rare disease clinical specialists, policymakers, and trade journalists as a secondary audience.

All episodes are recorded and disseminated in English.

Translations

All main EURORDIS information documents, such as brochures, and accompanying social media posts are translated.

- The Member News is translated in six languages (English, French, Germany, Italian, Portuguese and Spanish).
- The EURORDIS website is available in the seven official EURORDIS languages (English, French, Germany, Italian, Portuguese, Spanish and Russian).

Other printed materials

Other printed materials which were produced and disseminated included:

- A summary of the Activity Report 2021 & Workplan 2022.
- A summary of EURORDIS' reports on Ukraine.
- A summary of the EURORDIS position on the EHDS.

Other online materials

Other online materials which were published included:

- The Activity Report 2021 & Workplan 2022, and the Financial Report 2021.
- A EURORDIS report from the Czech EU Presidency Expert Conference on Rare Diseases (December 2022).
- The EURORDIS Proposal on the Revision of the Orphan Medicinal Products and Paediatric Regulation (November 2022).
- The EURORDIS Position on the European Health Data Space (October 2022).
- A EURORDIS report from the High-level Ministerial Conference on Rare Diseases (March 2022).

Other new online content

Other new online content which was published included:

- 21 press releases – including several press releases specific for the rare disease community about the war in Ukraine, which were also translated into Ukrainian.
- The regular updates of the Ukraine Resource Centre that was created in response to the Russian invasion of Ukraine in February 2022, with useful information and resources.
- A new landing page dedicated to the 25th anniversary of EURORDIS, alongside five professionally made videos with EURORDIS Board Members and senior management.
- A series of videos (nine videos) and articles (five articles) following major changes in the structure of the EURORDIS Board of Directors.
- 10 interview articles with Black Pearl Awardees.

"Happy Moments of Life"
Hereditary Hypophosphatemic
Taken by Pranab Basak, India

3. PARTNER

TO ESTABLISH AND FACILITATE NETWORKS WITH STRATEGIC PARTNERS AND KEY STAKEHOLDERS, INCLUDING, BUT NOT LIMITED TO, POLICYMAKERS WHO SUPPORT ACTIVE AND MEANINGFUL ENGAGEMENT WITH PEOPLE LIVING WITH A RARE DISEASE

"Friends for life - we are in this together!"
Retinoblastoma
Taken by Monika Koenig, Germany

EURORDIS has supported its strategic objectives through partnerships within five horizontal strategic priority areas. This includes:

RESEARCH POLICY AND ACTIVITIES

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "RESEARCH AND KNOWLEDGE DEVELOPMENT THAT ARE INNOVATIVE AND LED BY THE NEEDS OF PEOPLE LIVING WITH A RARE DISEASE":

TRANSLATIONAL RESEARCH

European Joint Programme on Rare Diseases (EJP RD, January 2019 – December 2023, Horizon 2020) and the Rare Disease Partnership (RD Partnership, Horizon Europe)

European Joint Programme on Rare Diseases

The European Joint Programme on Rare Diseases (EJP RD) brings over 130 institutions from 35 countries to create a comprehensive, sustainable ecosystem allowing a virtuous circle between research, care and medical innovation. The EJP RD focusses on maximising the potential of already funded tools and programmes by supporting them further, scaling them up, linking them up, and adapting them to the needs of end users through implementation tests in real settings.

In 2022, two **EURORDIS Schools were supported by EJP RD funding.** 23 patient advocates and 6 researchers from 18 countries took part in the 2022 Summer School (online, 6–10 June 2022) (see Open Academy Schools under Empower).

In 2022, EURORDIS took an active role in the 4th EJP RD General Assembly in Porto, Portugal, by chairing and presenting at several sessions, including one on "Facilitating patient partnerships". EURORDIS took part in a recorded video for the "ASHG TV Thought Leadership Film Series". It was broadcast during the ASHG meeting in all partnering hotels and in ASHG venues.

As part of the **PENREP (Patient Engagement in Research Projects),** EURORDIS organised and took part in different meetings, including a workshop to reflect on the main successes and remaining challenges around patient engagement in research. As a follow-up, a survey was defined and sent out to all patient groups who participated in projects funded by the EJP RD in 2019, which were evaluated at midterm in autumn 2022.

The EJP RD Joint Translational Call (JTC) 2023 launch in December 2022 attracted more than 100 participants. A panel session on patient partnerships in research was coordinated by EURORDIS at the EJP RD 2019 midterm evaluation meeting. The session welcomed testimonies from two patient experts involved in EJP RD JTC evaluation and one patient expert who was a partner in one of the JTC funded projects.

Two surveys were performed to assess patient engagement in research projects: one onsite amongst researchers attending the midterm evaluation meeting and an online survey amongst the patient partners involved in EJP RD 2019-funded projects. Those surveys should be extended to other funded projects to reach a critical number of answers and to identify trends and bottlenecks. Successful examples of the patient partnerships developed in the EJP RD-funded projects have also been embedded in the EJP RD Massive Open Online Course (MOOC) on Translational Research.

In late 2022, EURORDIS applied to the Nature Inclusive Health Research Award, highlighting the PENREP guidelines and, more generally, patient engagement in EJP RD research funding activities. The PFMD and EURORDIS also began discussions around patient engagement in early research.

In 2022, EURORDIS worked with ERICA to develop a **joint EJP RD/ERICA workshop on the topic: "Ethics and Regulatory considerations for ERN Data Access Committees (DAC)".** The workshop took place in June and encompassed subjects such as data access policy and data sharing for the ERNs.

In 2022, EURORDIS took **an active role in several EJP RD MOOCs:**

- The writing and development of the **second EJP RD MOOC,** "From Lab to Clinic: Translational Research for Rare Diseases", which was launched in October; totalling 705 enrolled learners so far. The MOOC was co-developed by EURORDIS, the Foundation for Rare Diseases, ERN EURO-NMD, Leiden University Medical Centre and EATRIS. The course is open, free and run on a continuous basis with defined facilitation windows (where mentors, including EURORDIS, are actively answering

comments and questions from learners).

- **The firstly developed MOOC,** "Diagnosing Rare Diseases: from the Clinic to Research and back", ran two facilitation windows in 2022. Taking all sessions together, since its start in April 2021, more than 4,800 learners from 140 countries have enrolled to follow the course. An active mentoring service is in place, including EURORDIS as an active mentor and educator.
- EURORDIS is part of a **third EJP RD MOOC** development, together with the ERN EpiCARE, Fondazione Gianni Benzi, and the Foundation for Rare Diseases on the topic "Rare Diseases Research Data: ethical and legal considerations". This MOOC will be delivered in 2023.

In 2022, EURORDIS continued to ensure the co-leadership of all EJP RD Pillar 3 training and capacity-building activities. EURORDIS also supported the organisation of the EJP RD paediatric training in Lyon, France, in early July 2022, as well as the EJP RD Leadership School held online, while providing expert reviews for the 2022 workshop and research mobility fellowship calls for ERN research training programmes.

Rare Disease Partnership

The Rare Disease Partnership concept paper developed by EJP RD members and additional experts was validated by the European Commission in February 2022, and published on the Commission's website. This draft was released to ensure transparency of information on the status of preparation of the Rare Disease Partnership. Following this release, the European Commission sent a letter to Member States asking them for their commitment to support the European Partnerships they wish to co-fund.

In March 2022, EURORDIS supported the calls organised by the EJP RD Coordination Team with EU Member States, relevant organisations, and agencies, to ensure that they: a) are fully aware about the ongoing developments of the Rare Disease Partnership; b) can exchange on the Rare Disease Partnership concept and identify needs and strengths in view of their country's participation; c) are best placed to decide and influence when their country will be consulted to commit to the Rare Disease Partnership.

In spring 2022, EURORDIS supported the process of the Rare Disease Partnership's development through an active communication with the National Rare Disease Alliances, to make sure that the Rare Disease Partnership was high on their agenda and that EU Member States' financial commitment was high. A presentation of the concept paper and funding decision mechanism was introduced to the Council of National Alliances (CNA) in early March 2022. Follow-up was ensured in March and April 2022 with National Alliances to accompany the Member State funding decision process. This involved multiple one-to-one interactions and support letters provided to ministries, as well as matchmaking National Alliances with relevant representatives from Ministries, Agencies, and Academia, in collaboration with the EJP RD Coordination Team. In November 2022, an updated presentation was given at the CNA meeting held in Paris. These efforts were highly effective, as the majority of EU Member States committed to participating in the Rare Disease Partnership.

The Strategic Research and Innovation Agenda (SRIA) – the strategy document – was drafted between December 2022 and January 2023 by the SRIA Task Force and a more restricted core group. EURORDIS took an active role in this group and coordinated the writing of one of the five strategic objectives ("All activities empower, as equal partners, people living with rare diseases") and was involved in drafting different sections of the document. The strategy document was shared for comments and inputs from the EU Board of Member States in February 2023, ahead of the launch of an open consultation phase.

European Rare Disease Research Coordination and Support Action (ERICA, March 2021 – February 2025, Horizon 2020)

The aim of the ERICA consortium is to build on the strength of the individual ERNs and create a platform that integrates all ERNs' research and innovation capacity. 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; and increased awareness of ERNs' 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.

In 2022, EURORDIS work focused mainly on WP2-ERN registries. EURORDIS contributed to shape the agenda of the final workshop organised in Heidelberg, Germany, as part of the workshop's scientific committee. EURORDIS led the design and delivery of the session on stakeholders' perspectives, where representatives from industry, clinicians, researchers, HTA agencies, the EMA

and EURORDIS shared their expectations and needs regarding collaboration with the ERN registries and the use and access to the data that is being (or will be) collected by the ERNs registries. Messages around the needs and expectations of the rare disease patient community regarding the ERN registries were discussed with members of the ePAG Research and Registries Working Group and delivered by EURORDIS Board Member, Elizabeth Vroom.

Rare Disease Moonshot (December 2022 – December 2027)

The Rare Disease Moonshot is a coalition of partners from industry, research and patient organisations, joining forces to accelerate scientific discovery and drug development in rare and paediatric diseases for which there is currently no therapeutic option. The coalition works to pool expertise, reduce fragmentation in research and foster greater collaboration between organisations. The seven organisations forming the Rare Disease Moonshot coalition are: the Critical Path Institute (C-Path); the European Infrastructure for Translational Medicine (EATRIS); the European Clinical Research Infrastructure Network (ECRIN); the European Federation of Pharmaceutical Industries and Associations (EFPIA); the European Confederation of Pharmaceutical Entrepreneurs (EUCOPE); EuropaBio; and EURORDIS-Rare Diseases Europe.

EURORDIS contributes to the **Rare Disease Moonshot** initiative that was launched in December 2022 to enhance public-private partnerships in research. Since 2022, EURORDIS has been active in the setting up and formal kick-off of the Rare Disease Moonshot through bimonthly calls. EURORDIS contributed to the organisation of a **workshop to boost public-private partnerships** to unlock uncharted territories in rare disease research and development. The aim of the workshop was to deliver recommendations that will inform the planning processes of funders and researchers when planning new initiatives or upscaling and deploying results from ongoing or past projects.

UNDIAGNOSED DISEASES

Collaborative project on diagnostic characterisation of rare diseases (Solve-RD, January 2018-June 2023, Horizon 2020)

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 yet known. Solve-RD echoes the ambitious goals set out by IRDiRC to deliver diagnostic tests for most rare diseases.

To date, the Solve-RD project has analysed 21,348 datasets (phenotype and exome/genome sequencing data) from 6,000 families. The project has already solved 511 rare disease cases (constituting an 8.5% diagnostic yield) for which a molecular cause was not previously known.

EURORDIS is a member of the Steering Committee for the work package that addresses disseminating 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 for ultra-rare diseases, and supporting the needs of the undiagnosed community.

The EURORDIS-led Community Engagement Task Force (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 into 25 languages.

In 2022, EURORDIS continued being actively involved in the project by chairing the session on diagnostic and clinical utility at the Solve-RD Annual Meeting. The Task Force members prepared the interview questions for Sonia Vallabh, who gave the keynote lecture at the Solve-RD Annual Meeting. The interview was published on the Solve-RD website and in the project's newsletter, and additionally communicated via EURORDIS' website, eNews newsletter and social media accounts. Moreover, the interactive version of the CETF Patient Journey diagram was launched on the new EURORDIS website on 13 March 2022, and subsequently the Solve-RD website. The CETF also designed and launched a survey on the needs and priorities of undiagnosed patients.

Undiagnosed Diseases Network International

The Undiagnosed Diseases Network International (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. Patients' and patient representatives' participation are instrumental in ensuring the long-term success of this initiative, as they can offer their expertise to the UDNI institutional, clinician and non-clinician members on how to ensure the effort is patient-focused, patient-friendly, and patient-driven.

EURORDIS participates in UDNI, an international network of clinical centres that was initiated in 2014 to address the unmet needs of undiagnosed patients at a global level. NORD, EURORDIS and the Wilhelm Foundation have collaboratively developed a patient engagement membership which was adopted by the UDNI's Board.

In 2022, EURORDIS – together with NORD and the Wilhelm Foundation – continued: chairing the UDNI Patient Engagement Group; created a workflow for reviewing incoming applications to the Group in order to facilitate the application process; created a welcome message for new members; and organised three teleconferences with member patient organisations. The efforts for outreach resulted in the UDNI Patient Engagement Group comprising 20 member patient organisations from all continents. EURORDIS also facilitated international collaboration for Undiagnosed Day. EURORDIS also gave a talk at the Undiagnosed Day webinar organised by the Wilhelm Foundation.

PAEDIATRIC CLINICAL TRIALS

Conect4Children (May 2018 – April 2024, IMI 2)

The **Conect4Children (c4c)** research network brings together pharmaceutical companies and national paediatric networks, as well as EU multinational subspecialty networks, large patient advocacy groups, children's hospitals, and other public research organisations from across Europe. It 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 aims to facilitate the development of new drugs and other therapies for the entire paediatric population.

In 2022, EURORDIS was particularly involved in the organisation of the c4c Multi-Stakeholder Meetings on Inflammatory Bowel Disease, Atopic Dermatitis and Paediatric Diabetes Type 1, bringing relevant patient experts (young patients and parents) into the meetings. EURORDIS also led a workshop for patient organisations on paediatric drug development and a follow-up webinar for patient experts about c4c activities, which is now available on the c4c website.

EURORDIS is currently involved in the drafting of a white paper on paediatric patient engagement. To support the drafting of this white paper, EURORDIS, with other project partners, is organising a workshop involving experts in patient involvement, ethics, methodologies of involvement, diversity, health literacy, policy, and patients' rights. EURORDIS is also contributing to c4c work on data quality standards and on an education and training programme, including through being a member of the related Education Board.

DATA AND DIGITAL HEALTH

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "OPTIMISED DATA AND HEALTH DIGITAL TECHNOLOGIES FOR THE BENEFIT OF PEOPLE LIVING WITH A RARE DISEASE AND SOCIETY AT LARGE":

FACILITATE (January 2022 – December 2025, IMI2)

FACILITATE is a four-year Innovative Health Initiative (IHI) project that aims to return clinical trial data to trial participants, as well as to develop a framework that would allow the re-use of clinical trial data for secondary purposes.

FACILITATE

EURORDIS is tasked with bringing the voice of patients to the centre of the process by conducting consultations with its patient expert group in the digital area – the DAG (Digital and Data Advisory Group – as well as by conducting a research overview into existing studies on the views of patients.

The kick-off meeting took place online in January 2022, and the project started working on its first deliverable, the Consortium Handbook.

In 2022, EURORDIS was, and still is, heavily involved in the two deliverables of: The Report

on Case Studies for the Technical Development of FACILITATE; and the Review of the Ethical Frameworks, which is still ongoing. EURORDIS is also involved in communication and stakeholder engagement relating to the project, and the Report on Legal Requirements. In November, the first General Assembly took place face-to-face in Modena, Italy.

Toward a European Health Data Space (TEHDAS, February 2021 – August 2023, CHAFEA Third EU Health Programme)

TEHDAS (Joint Action Towards the European Health Data Space) is an initiative aimed at developing joint European principles for the secondary use of health data. The work involves 25 countries, and it is based on the European Commission's Health Programme 2020.

EURORDIS was involved in Work Package 8 (WP8). This Work Package was tasked with obtaining a better understanding of citizens' attitudes towards sharing their health data. The two main objectives of this work package were to determine citizens' opinion on: health data re-use and data altruism. We participated and gave our opinion on both aspects. As WP8 is coming to an end, recommendations for both explored directions have been published.

Together4RD

Together4RD is a multi-stakeholder initiative aimed at supporting collaboration between European Reference Networks (ERNs) and industry in areas that will address the unmet medical needs resulting from the 95% of rare diseases that currently do not have a dedicated treatment.

In 2022, EURORDIS contributed to **Together4RD,** an initiative aiming to collect and leverage data to support research in rare diseases, generate new knowledge, and support the development of new treatments. EURORDIS is an active member of the **Together4RD Steering Group** and has: co-developed the **Together4RD Recommendations** to unlock ERN and industry collaboration for the benefit of people living with rare diseases; co-authored the White Paper on ERN-Industry Collaboration; sent **speakers to Together4RD events,** workshops and webinars (including an event in the European Parliament); led a Thought Leader Session on the initiative at ECRD 2022; led a Plenary Session on the initiative at the World Orphan Drug Congress Europe; and led multi-stakeholder online workshops on the initiative and other webinars.

DEVELOPMENT AND ACCESS TO DIAGNOSTICS AND THERAPIES

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO EXPANDING TREATMENT "DEVELOPMENT, AVAILABILITY, ACCESSIBILITY AND AFFORDABILITY, PARTICULARLY WITH REGARD TO TRANSFORMATIVE OR CURATIVE THERAPIES – GOAL OF 1,000 NEW THERAPIES WITHIN 10 YEARS":

Next Generation Health Technology Assessment" (HTx, January 2019-December 2023, Horizon 2020)

HTx aims to create a framework for the Next Generation Health Technology Assessment to support patient-centred, societally-oriented and real-time decision-making on access to, and reimbursement for, health technologies throughout Europe. This framework is to address questions such as: "Which patients would benefit the most from a given high-cost new therapy?"; "How can the best sequence or combination of treatments be identified?"; and "What is the best therapeutic strategy for a given small population or subgroup?".

One of the goals of HTx is to explore the extent to which 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. New methods will be tested on four case studies, about four disease areas: head and neck cancer, Diabetes 1 and 2, Multiple Sclerosis, and Myelodysplastic Syndrome.

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

In 2022, the HTx project undertook multiple activities and initiatives within the project. The first was the hosting of a panel discussion, "Building Bridges Between Patient and Treatment – Perspectives on Harmonizing Health Technology Assessment with Up and Down-Stream Stakeholders", at the Health Technology Assessment International (HTAi) Conference. Multiple workshops were also conducted, including: the HTx shared decision-making workshop that took place virtually; the HTx methods workshops in York; and the HTx workshop about novel payment models for gene therapy. In addition to these activities, two research publications were published. EURORDIS also led a study on the qualitative evaluation of acceptability, and barriers to introducing the cost dimension in shared decision-making within the clinical context.

GetReal Institute

The **GetReal Institute** builds on the success of two IMI projects – GetReal and The GetReal Initiative – and brings together a wide variety of stakeholders to drive the sustainable development and adoption of tools, methods and best practices in the generation and use of real-world evidence (RWE) for better healthcare decision-making.

In 2022, EURORDIS became a member of Get Real Institute, as a co-founder and Board member working on reducing barriers to the use of secondary data, bridging the gap between Randomised Control Trial (RCT) and RWE, and addressing the evidence needs of downstream healthcare decision-makers. As a member of the GetReal Board of Directors, EURORDIS contributed to the Initiative's activities held in 2022, including involvement in: defining the GetReal Institute's objectives, strategy and values; supporting the survey to GetReal Institute members on the Institute's objectives and strategic partnerships; and planning and organising the first GetReal Institute Conference in March 2023.

EURORDIS also supported the launch and continuation of three projects: the development of recommendations or guidelines on the use of external comparators for regulatory, HTA and payer decision-making; establishing criteria for when results from RWE are sufficiently compelling to be used as pivotal evidence; and a multi-stakeholder shared learning platform for RWE as a component of a broader "test and learn" environment in Europe.

VACCELERATE (January 2021-January 2024, Horizon 2020)

VACCELERATE is a clinical research network for the coordination and conduct of COVID-19 vaccine trials. The network is comprised of academic institutions from all over Europe. The consortium is led by the University Hospital Cologne, Germany, and currently includes 29 national partners in 18 EU-member states and five countries associated to the EU Horizon 2020 research programme.

In 2022, EURORDIS has contributed to VACCELERATE, a project that works on Clinical Trial Site Capacity Building, Public Health Needs, Volunteer Registries, Booster Vaccination Doses in the Elderly, Booster Vaccination Doses in Adults, and the vaccination regimen in children aged 12-16 years.

REMEDi4ALL (September 2022 – August 2027, Horizon Europe)

REMEDi4ALL aims to assemble a sustainable European Innovation Platform able to provide guidance and support to developers at all stages of the repurposing pathway. The multidisciplinary consortium of 24 leading institutions and organisations aims to create a vibrant European research and innovation ecosystem that facilitates cost-effective patient-centric development and the implementation of repurposed medicines in areas where there are high unmet medical needs, including the rare disease field.

The project started in September 2022, with a kick-off meeting in Amsterdam, the Netherlands. EURORDIS have hired a Senior Project Manager and a Patient Engagement & Training Manager. To ensure patient-centricity in all steps of the developmental process, EURORDIS has started to actively work with all related stakeholders and within all related work packages. EURORDIS has established fruitful relationships and collaboration with all demonstrator project leaders and started activities to support patient engagement, such as the identifying and onboarding of patient champions for the demonstrator projects, and establishing patient focus and patient advisory groups.

International Consortium for Rare Disease Research (IRDiRC)

The I**nternational 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 been a member of IRDiRC since its launch in 2011. Since then, EURORDIS has led, or contributed to, many activities of the Consortium aimed at reaching IRDiRC Goals. As a member of the Consortium Assembly (in the Patient Advocacy Constituent Committee), we have an oversight (and a voting right when relevant) on all the decisions taken by the Consortium: strategic choices, inclusion of new members, and the creation of task forces and working groups.

In addition to being a constituent Committee member, EURORDIS has always brought the expertise of its staff and members to the IRDiRC Scientific Committees, Task Forces and Working Groups, including: the Therapies Scientific Committee, the Sustainable Economic Models in Repurposing Taskforce (leading); Drug Repurposing Guidebook Task Force (which involved a workshop held in 2022); the group leading the Pluto Project on Disregarded Rare Diseases (which involved a workshop held in 2022); the IRDiRC-RDI Global Access Working Group; the Working Group on Medical Technology for Rare Diseases; the Task Force on the Integration of New Technologies for Diagnostics; and the group leading the Newborn Screening Initiative. We also applied to the newly created Regulatory Scientific Committee, but our patient representative candidate was not selected.

EURORDIS work in IRDiRC has led to peer-reviewed publications[1]. In 2022, we resubmitted our manuscript entitled "Sustainable Approaches for Drug Repurposing in Rare Diseases: Recommendations from the IRDiRC Task Force" to the Rare Disease and Orphan Drugs Journal, which shall be eventually published in 2023. EURORDIS remains committed to ensure sustainability of the Orphan Drug Development Guidebook, co-created within the IRDiRC Therapies Scientific Committee with a multi-stakeholder group of experts in the context of a Memorandum of Understanding signed between EURORDIS, EATRIS, Fondazione Telethon, CVBF and INSERM.

Screen4Care (S4C, October 2021-September 2026, IMI JU)

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 people living with rare diseases. EURORDIS promotes the patient voice by chairing the Screen4Care (S4C) Patient Advisory Board (PAB), which consists of 15 patient representatives involved in the EU-

1 Hechtelt Jonker A, Hivert V, Gabaldo M, Batista L, O'Connor D, Aartsma-Rus A, Day S, Sakushima K, Ardigo D. Boosting delivery of rare disease therapies: the IRDiRC Orphan Drug Development Guidebook. Nat Rev Drug Discov. 2020 Aug;19(8):495-496. doi: 10.1038/d41573-020-00060-w. PMID: 32313251.
Gahl WA, Wong-Rieger D, Hivert V, Yang R, Zanello G, Groft S. Essential list of medicinal products for rare diseases: recommendations from the IRDiRC Rare Disease Treatment Access Working Group. Orphanet J Rare Dis. 2021 Jul 13;16(1):308. doi: 10.1186/s13023-021-01923-0. PMID: 34256816; PMCID: PMC8278724.
Hivert V, Jonker AH, O'Connor D, Ardigo D. IRDiRC: 1000 new rare diseases treatments by 2027, identifying and bringing forward strategic actions. Rare Dis Orphan Drugs J 2022; 1:3. http://dx.doi.org/10.20517/rdodj.2021.02

Edith Sky Gross, EURORDIS Health and Social Science Senior Project Manager and Gulcin Gumus, EURORDIS Research and Policy Project Senior Manager together with the Newborn Screening Working Group of Screen4Care

RORDIS Newborn Screening Working Group, the Digital and Data Advisory Group, and ePAGs. The PAB provides strategic recommendations, guidance, and advice across all Screen4Care activities. This entails identifying patients' preferences on existing symptom checkers and co-designing the S4C virtual platform. The outputs of the S4C project support EURORDIS' advocacy work on Newborn Screening.

In 2022, EURORDIS worked toward an **"RDclopedia",** a survey of rare disease-related initiatives in Europe, including registries, research projects, infrastructures and newborn screening programmes. EURORDIS took part in forming the criteria for the choice of conditions to be screened as part of the "treatable" panel of conditions and advised on the co-creation transversal task force regarding the use of machine learning on electronic health records. We also participated in the Ethical, Legal and Safety Team (ELST) and provided the patient perspective on all outputs of the project. The Newborn Screening Working Group was consulted to contribute to the **work on "actionability"** as a criterion for newborn screening.

Global Commission to End the Diagnostic Odyssey for Children with Rare Diseases

EURORDIS continued to follow this initiative and to engage in the Patient Empowerment Education & Awareness Campaign. EURORDIS also supported the communication efforts of the Global Commission.

HEALTHCARE POLICY AND SERVICES

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND CONTRIBUTE TO THE GOAL OF "HIGH-QUALITY NATIONAL AND EUROPEAN HEALTHCARE PATHWAYS, INCLUDING CROSS-BORDER HEALTHCARE," TO IMPROVE SURVIVAL BY AN AVERAGE OF 3 YEARS OVER 10 YEARS AND REDUCE THE MORTALITY OF CHILDREN UNDER 5 YEARS OF AGE BY ONE THIRD, AS WELL AS THE GOAL OF "EARLIER, FASTER, AND MORE ACCURATE DIAGNOSIS," TO DIAGNOSE A RARE DISEASE WITHIN 6 MONTHS:

H-CARE

The **H-CARE** project aims to develop a feedback mechanism that regularly and robustly measures the healthcare experience of people living with rare diseases through the development and validation of Patient Reported Experience Measures (PREMs). Once validated, these PREMs could then be used as a measure to evaluate the patient-centred care provided across all 24 ERNs as part of their evaluation and monitoring system. Experience and satisfaction with care were originally discussed by the ERNs' Monitoring Working Group as being an important component of the 18 core monitoring indicators of ERNs. However, the implementation of the indicator was postponed as it was considered too complex to be implemented in a timely manner. The H-CARE project started in 2019 as an initiative of ERNs ERKNet, eUROGEN, GENTURIS and LUNG with the support of the EURORDIS Rare Barometer Survey Programme. A pilot survey conducted in 2020 showed that there is a need to develop and validate PREMs for rare disease patients and family members to robustly measure their healthcare experience within all 24 ERNs.

In 2022, EURORDIS worked towards the completion of a literature review, as a first step to develop PREMs. 4,767 titles and abstracts were screened and a data extraction form was drafted and shared with the Scientific Working Group of experts in PREM development and rare diseases. The H-CARE project was presented to ERNs and the European Commission in order to secure funding for the development and validation of scales to measure the healthcare experience of rare disease patients and family members by 2026.

Joint Action on the integration of European Reference Networks into national health systems (JA, October 2023-2026, DG SANTE)

The **Joint Action** on the Integration of ERNs aims at developing models and structures to enable Members States to connect their national health systems with the ERNs. It will include activities on different areas such as care pathways, data interoperability, dissemination at national level, quality assurance and designation of centres of expertise, national networks, referral systems, and the revision of national rare disease plans and strategies.

In 2022, EURORDIS engaged with representatives from the ERN Board of Member States, as well as the European Commission to convey concrete proposals on the scope and activities of the future Joint Action on the integration of ERNs into national health systems and into the EU cross-border healthcare system. It also duly informed the National Rare Disease Alliances about the possibility to participate as affiliated entities and encouraged them to reach out to the national competent authorities to learn whether their designated national Competent Authorities could include them as affiliated entities with a budget allocation. Finally, EURORDIS also contributed to this effort by publishing a toolkit outlining best practices for connecting ERNs and national health systems.

SOCIAL POLICY AND SERVICES

IN LINE WITH OUR STRATEGIC OBJECTIVE 2, WHICH AIMS TO DELIVER ON PRIORITY AREAS AND CONTRIBUTE TO THE GOAL OF "INTEGRATED MEDICAL AND SOCIAL CARE WITH A HOLISTIC LIFELONG APPROACH AND INCLUSION IN SOCIETY," WITH A GOAL OF REDUCING THE SOCIAL, PSYCHOLOGICAL, AND ECONOMIC BURDEN BY ONE THIRD:

EURORDIS continued to seek opportunities to develop project proposals aimed at addressing key holistic and social care priorities, notably within the Social Innovation (EaSI) strand of the European Social Fund Plus (ESF+).

In addition, EURORDIS and the Romanian National Alliance for Rare Diseases (RONARD) remained in contact with the European Commission as one of the implementors of previous successful projects funded by the EaSI programme – the INNOVCare project. In December, the INNOVCare project was added to the European Commission's publications catalogue, as an example of successful good practice.

Yann Le Cam (EURORDIS) speaks at a panel during the Expert Conference on Rare Diseases in Prague, October 2022

4. CROSS-CUTTING PRIORITIES

"Feeling the grass"
Chung Jansen Syndrome
Taken by Lisa Marrington,
United Kingdom

EURORDIS supported its strategic objectives through its cross-cutting priorities. These include:

GOVERNANCE

EURORDIS Governance Bodies: Annual General Assembly, Board of Directors and Board of Officers

The EURORDIS Annual General Assembly was held online on 18 May 2022. EURORDIS' full members voted on the Activity and Financial Reports for 2021, and the Work Programme and Budget for 2022.

Members also voted on the six vacant positions for the Board of Directors (BoD). The following candidates were successfully re-elected to the BoD: Terkel Andersen (Danish Haemophilia Society, Denmark); and Geske Wehr (Selbsthilfe Ichthyose E.V., Germany).

The Board also extended a warm welcome to new member Kirsten Johnson (Fragile X Society,

United Kingdom) and Rebecca Tvedt Skarberg (Osteogenesis Imperfecta Federation Europe, Norway).

All Board members were elected for a full mandate of three years.

The Board of Officers (BoO) is elected annually by the BoD following the General Assembly. In May, 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.

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Revised Board Terms of Reference

During the BoD meeting in July 2022, the roles and responsibilities of both the BoD and the BoO were discussed and refined in order to reinforce the distinction between both the BoD and the BoO, to give more time for strategic discussions to the BoD, and to give some clear delegation to the BoO when it comes to administrative, financial, legal and HR matters. A synthesis was produced and adopted by the BoD, which became the basis to revise the Terms of Reference of the BoD and the BoO. The revised Terms of Reference were adopted in November 2022.

Changes in the Board of Officers structure and the election of a new Board of Officers in November 2022

In 2022, the BoO structure was modified. The BoO previously consisted of six officers, namely: President; Vice-President; Treasurer; General Secretary; and two Officers. Due to health reasons, EURORDIS President for the last 20 years, Terkel Andersen, had to step out of his position in June 2022. Avril Daly, EURORDIS Vice-President for the past 10 years, became Acting President until the election of a new BoO during the BoD meeting of November 2022. During that Board meeting, a new structure of the BoO was also decided.

The BoO consists of six officers, including two additional positions with specific responsibilities that have been added: one additional Vice-President to support the President in external representation; and one Deputy General Secretary to support the General Secretary, especially when it comes to the relationship with EURORDIS Members and, in particular, the National Rare Disease Alliances.

In November, the BoO was thus elected as follows: President – Avril Daly, Ireland; Vice Presidents – Dorica Dan, Romania and Maria Montefusco, Sweden; General Secretary – Geske Wehr, Germany; Deputy General Secretary – Simona Bellagambi, Italy; Treasurer – Alain Cornet, Belgium.

Core Leadership Team

The EURORDIS Core Leadership Team (CLT) prepares and executes on the strategies, ensures day-to-day decision-making, and consists of the Chief Executive Officer (CEO), the Chief Financial Officer (CFO), the Chief Operating Officer (COO), the Public Affairs Director & Head of European and International Advocacy, and the Governance Senior Manager. The CLT meets every week to tackle the different strategic, management and operational issues that can be possibly solved at this governance level, in order to advance towards the achievement of EURORDIS' missions and activities.

EURORDIS Strategy 2021-2030

In 2020, EURORDIS commissioned an external strategic review by Philanthropy Advisors for the purpose of developing its strategy from 2021-2030. The strategic review involved gathering opinions from EURORDIS members and stakeholders. It also incorporated the recommendations from the Rare 2030 foresight study, and feedback collected via surveys completed by people living with rare diseases and patient organisations across Europe

The resulting findings and proposed Strategy for 2021-2030 was presented and discussed at the 2021 General Assembly.

Throughout 2022, the EURORDIS Board of Directors and the CLT continued the strategic review, leading to the revised Strategy for 2021-2030 that was adopted by the BoD in November 2022 and presented in this report.

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 by 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 - National Organization for Rare Disorders (USA)

CORD - Canadian Organization for Rare Disorders

JPA - Japan Patient Association

RVA – Rare Voices Australia

RADOIR - Rare Diseases Foundation of Iran

RDI - Rare Diseases International

EURORDIS also has partnerships with a number of learned societies, that are listed under the "External representation" webpage and detailed in our Work Programme 2023.

HUMAN RESOURCES (STAFF AND VOLUNTEERS)

EURORDIS Staff

In July 2022, the EURORDIS Board of Directors took the decision to create the position of Human Resources Director to lead EURORDIS Human Resources. The hiring was launched in September 2022 and completed in February 2023.

The number of FTE was 48.7 in 2022, compared to 48.0 in 2021.

As at the end of the year, the team was composed of 55 staff members across seven countries.

The main office is in France (33), followed by Spain (nine), Belgium (six), the United Kingdom (three), Italy (two), Germany (one) and Romania (one).

Six new positions were created in 2022 (in order of appearance):

- Patient Data Director, Jelena Malinina, Brussels
- Senior Project Manager: EURORDIS Ukraine Response, Anastasiia Saliuk, Paris
- Drug Repurposing Project Senior Manager, Claudia Fuchs, Bolzano
- Social Policy & Initiatives Director, Raquel Castro, Barcelona
- Patient Engagement & Training Manager, Judit Baijet, Barcelona
- Office Assistant, Aminata Doumbia, Paris

Four positions were stopped:

- Public Health Policy Director, Anna Kole, Paris
- Julien Delaye, Public Health Policy Junior Manager, Brussels
- Sandra Pavlovic, Rare Connect Manager, Belgrade
- Flaminia Macchia, Rare Diseases International Executive Director, Brussels

In May 2022, an All-Staff Team Seminar was organised in Lisbon over three days and gathered more than 55 staff members from our different

offices to enhance team cooperation and efficiency.

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 organisation's volunteers are governed by the **EURORDIS Charter of Volunteers, adopted by the EURORDIS General Assembly on 8 May 2014 in Berlin.** This Charter sets outs the values of EURORDIS, the volunteers' commitments, and EURORDIS' commitments to 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' need for information and connection by creating international online communities and discussion groups for specific diseases.

There are **380 volunteer** RareConnect moderators, concerned or 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:

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

They are selected via a Call for Expression of Interest and rigorous selection criteria to join a specific group of volunteers, and/or be a candidate to an EU high level committee. The volunteers must: a) be committed to the cause of rare diseases; b) master English; and 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 European Commission 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 (though rarely) three.

In 2022, there were **75 EURORDIS volunteer** patient advocates who belonged to different groups or task forces. One volunteer can belong to two or more groups or task forces.

- **EPAC: European Public Affairs Committee.** This internal committee plays an active and key role in EURORDIS' advocacy activities. The EPAC members discuss all relevant advocacy issues for people living with rare diseases and their families. They can also provide their comments on EURORDIS' positions on some specific issues. The EPAC is governed by Rules of Procedure. As of end 2021, it was composed of 43 full members: **20 volunteers** as well as 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 **5 volunteers** who represent patients on the European Medicines Agency's scientific committees. These committees hold meetings every month over two to three days. The expertise required and involvement in terms of time are both significant.
- **DITA (Drug Information, Transparency and Access) Task Force.** In 2022, **23 volunteers** contributed to the DITA Task Force's work (including members from mandate 2019-2022 and from mandate 2022-2025), as the Task Force was renewed in 2022. Selected volunteers are trained (via the EURORDIS Open Academy) and active on issues concerning therapeutic development of medicines for rare diseases as well as access. The Task Force supports and/or advises the EURORDIS representatives who participate in EMA Scientific Committees and Working Parties, or in the European Network of Health Technology Assessment (EUnetHTA) and the HTA Network (DG SANTE).
- **HTA (Health Technology Assessment) Task Force.** The HTA Task Force is composed of **8 volunteers** and coordinated by two staff members, who also manage the DITA Task Force in order to ensure a good synergy between the two groups. The HTA Task Force advises EURORDIS on all aspects regarding HTA policies and procedures. Its role is to inform EURORDIS on how health technologies are assessed at the national level and how patients are involved in these assessments, and to share views on the future European Cooperation on HTA.

- **DAG: Digital and Data Advisory Group.** The DAG included **11 volunteers** in 2022. The DAG 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 rare diseases 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 2022, **20 members of this ePAGs Steering Committee** have expressed their interest to be or remain a 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 of further strengthening patient advocates' involvement and raising awareness of ERNs amongst the wider rare disease community.

PRIVATE RESOURCE DEVELOPMENT

2022 saw continued growth in engagement and revenue across resource development channels.

- Our first of two annual EURORDIS Round Table of Companies Workshops was held online in April, following the successful virtual model developed for this event during the pandemic. Held over two afternoons, the meeting was attended by 165 participants, including 95 representatives from 46 companies.
- In October, we were thrilled to return to an in-person meeting in Barcelona. The meeting was attended by 146 participants, including 77 representatives from 46 companies.
- On top of these meetings, two webinars were held for corporate members on the topics of Rare Disease Day and the crisis in Ukraine.
- In total, 77 different health sector corporations supported EURORDIS in 2022, including five new corporate donors.
- Diversified fundraising continued to move in a positive direction, with individual donations growing by 20% over 2022.
- Foundation support decreased by 12% from 2021, notably due to a one-year gap in funding from a significant non-profit contributor. Nonetheless, we were successful in realising contributions from multiple first-time non-profit partners, laying the groundwork for continued engagement and support in subsequent years.
- Corporate, non-profit, and individual supporters alike responded with urgency and generosity to our Ukraine Emergency Response programme. In total, over €439,000 was raised to launch this new area of work.

FINANCE & SUPPORT SERVICES

Finance and support services' activities in 2022 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.
- 49 EURORDIS procedures were created or updated (accounting, HR, administrative and IT procedures).

Contract Grants

Ongoing

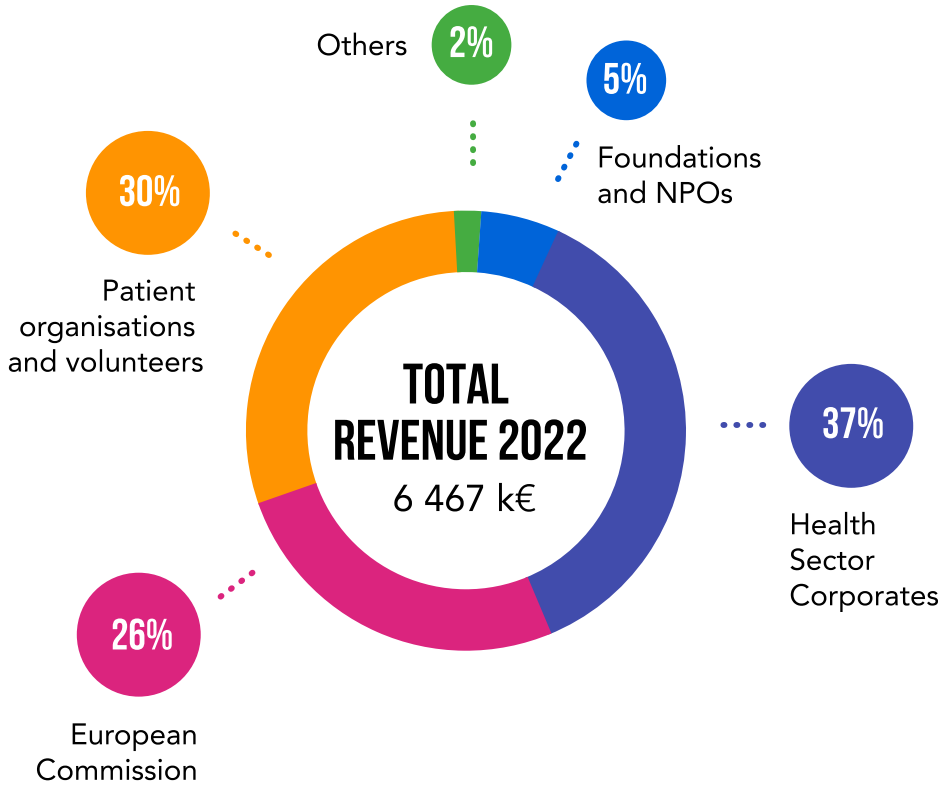
Advocacy and core activities, AFM-Téléthon, 2019-2022

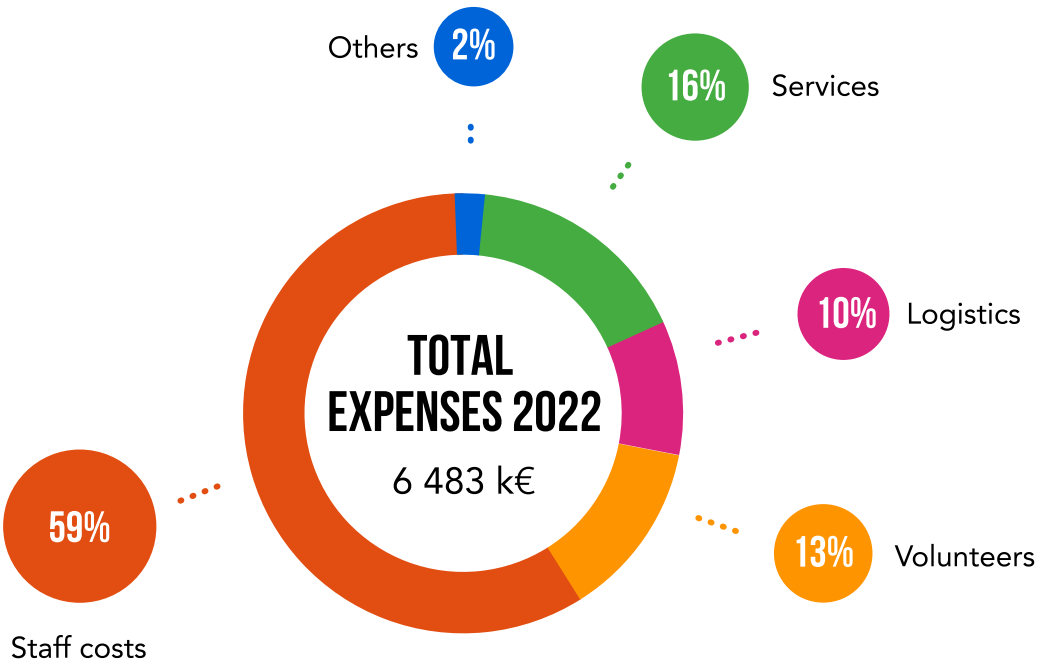
Renewed

Specific Grant Agreement (Operating Grant) for year 2022 (SGA 2022), single beneficiary, DG SANTE, 12 months

PROJECT REFERENCE NO AND TITLE, FUNDING PROGRAMME	PERIOD (START – END DATE)	ROLE	AMOUNT (EUR)
European Joint Programme on Rare Diseases Grant Agreement N° 825575 – EJP RD Horizon 2020	01/01/2019 31/12/2023	Partner, beneficiary	EURORDIS: 930,665 € Project: 100,362,308.32 €
Solving the unsolved rare diseases – Solve RD Grant Agreement N° 779257 – Solve-RD Horizon 2020	01/01/2018 30/06/2023	Partner, beneficiary	EURORDIS: 375,000 € Project: 15,361,621 €
"Next Generation Health Technology Assessment to support patient-centred, societally oriented, real time decision making on access and reimbursement for health technologies throughout Europe" – "HTx" Grant Agreement N° 825162 – HTX Horizon 2020	01/01/2019 31/12/2023	Partner, beneficiary	EURORDIS: 625,097.50 € Project: 9,640,775 €
FrAmework for CIInicaL trIal participants' daTA reutilization for a fully Transparent and Ethical ecosystem: FACILITATE Grant Agreement N° 101034366 – FACILITATE IMI2	01/01/2022 31/12/2025	Partner, beneficiary	EURORDIS: 140,250 € Project: 3,260,000 €
"Shortening the path to rare disease diagnosis by using newborn genetic screening and digital technologies": SCREEN4CARE Grant Agreement N° 101034427 – SCREEN4CARE IMI2	01/10/2021 30/09/2026	Partner, beneficiary	EURORDIS: 808,000 € Project: 11,938,568.75 €
European Rare Disease research coordination and support action: ERICA Call reference – N° SC1-HCO-20-2020 – ERICA Horizon 2020	01/03/2021 28/02/2025	Partner, beneficiary	EURORDIS: 143,675.00 € Project: 2,313,808.75 €
Conect4Children: C4C Grant Agreement N° 777389 – C4C IMI2	01/05/2018 01/04/2024	Partner, beneficiary	EURORDIS: 578 750,00 € Project: 182,018,216.00 €
REMEDI4ALL Call reference – HORIZON-HLTH-DISEASE-2021-04-02 – REMEDI4ALL Horizon Europe	01/09/2022 31/08/2027	Partner, beneficiary	EURORDIS: 1,351,187.50 € Project: 25, 000,000.00 €

EURORDIS REVENUE AND EXPENSES 2022





"My Faithful Companion", Pulmonary Arterial Hypertension, Ukraine

BOARD OF DIRECTORS
MAY 2022 – MAY 2023

PRESIDENT

MR TERKEL ANDERSEN [1]
Danish Haemophilia Society
Denmark

MS AVRIL DALY [2]
Retina International
Ireland

DIRECTORS

MS ALBA ANCOCHEA [3]
Spanish Federation of Rare Diseases (FEDER)
Spain

MS ANNA ARELLANESOVA
Rare Diseases Czech Republic
Czechia

MS SIMONA BELLAGAMBI
UNIAMO – Rare Diseases Italy
Italy

MR ALAIN CORNET
Lupus Belgium
Belgium

MR ALEXANDRE MEJAT
AFM - Téléthon
France

MS BIRTHE BYSKOV HOLM
Rare Diseases Denmark
Denmark

MS DORICA DAN
Romanian Prader Willi Association
Romania

MS MARIA MONTEFUSCO
Rare Diseases Sweden
Sweden

MS ELIZABETH VROOM
World Duchenne Organisation
Netherlands

MS GESKE WEHR
European Network for Ichthyosis e.V
Germany

MS KIRSTEN JOHNSON
The Fragile X Society
UK

MS REBECCA TVEDT SKARBERG
Osteogenesis Imperfecta Federation Europe (OIFE)
Norway

[1] Resigned as President in November 2022 for health reasons and remains in the Board of Directors
[2] Acting President between June and November 2022, elected President as of November 2022
[3] Resigned in June 2022

BOARD OF OFFICERS
MAY 2022 – NOVEMBER 2022

MR TERKEL ANDERSEN
PRESIDENT
Denmark

MS AVRIL DALY
VICE-PRESIDENT
Ireland

MS GESKE WEHR
GENERAL SECRETARY
Germany

MR ALAIN CORNET
TREASURER
Belgium

MS DORICA DAN
OFFICER
Romania

MS MARIA MONTEFUSCO
OFFICER
Sweden

BOARD OF OFFICERS
NOVEMBER 2022 – MAY 2023

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Romania

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VICE-PRESIDENT
Sweden

MS GESKE WEHR
GENERAL SECRETARY
Germany

MS SIMONA BELLAGAMBI
DEPUTY GENERAL SECRETARY
Italy

MR ALAIN CORNET
TREASURER
Belgium

EURORDIS MEMBERS

DECEMBER 2022

"Brave"
Cockayne Syndrome
Taken by Sarah
Matthews, Australia

ALBANIA		
Shoqata e Semundjeve te Rralla / Rare Disease Association Albania	http://www.rda-al.com	Associate Member
ALGERIA		
Association Elamani pour venir en aide aux Malades souffrant de l'Anémie Héréditaire		Associate Member
ANDORRA		
Associació de Malalties Minoritàries d'Andorra	https://amma.ad/	Full Member
ARGENTINA		
Alianza Argentina de Pacientes	http://alianzapacientes.org/	Associate Member
Federación Argentina de Enfermedades Poco Frecuentes	http://fadepof.org.ar/	Associate Member
ARMENIA		
Doctors And Children Health Care	http://www.rambler.ru	Associate Member
Neurohereditary Diseases Charity Association	http://www.arminco.com	Associate Member
AUSTRALIA		
Cystic Fibrosis Australia	http://www.cysticfibrosis.org.au	Associate Member
Genetic Alliance Australia	http://www.geneticalliance.org.au	Associate Member
Genetic Support Network of Victoria	https://www.gsnv.org.au/	Associate Member
Muscular Dystrophy WA	https://www.mdwa.org.au/	Associate Member
Rare Voices Australia	http://www.rarevoices.org.au	Associate Member
Save our Sons	https://www.saveoursons.org.au/	Associate Member
AUSTRIA		
Angelman Verein Österreich	http://www.angelman.at	Full Member
Childhood Cancer International-Europe	https://ccieurope.eu/	Full Member
Debra International	http://www.debra-international.org	Full Member
Hand in Hand Für Tay-Sachs & Palliativkinder	http://www.tay-sachs.net	Associate Member
ICA-Österreich	http://www.ica-austria.at	Full Member
NF Kinder – Verein zur Förderung der Neurofibromatoseforschung Österreich	http://www.nfkinder.at	Full Member
NF Patients United	http://www.nf-patients.eu	Associate Member
PH Austria - Initiative Lungenhochdruck	http://www.phaustria.org	Full Member
PHA Europe	http://www.phaeurope.org	Associate Member
Pro Rare Austria, Allianz für seltenen Erkrankungen	http://www.prorare-austria.org	Full Member

Smith-Magenis-Syndrom Österreich	https://www.smith-magenis.at/	Full Member
Usher Deafblind Forum Austria	http://www.usher-taubblind.at	Associate Member

BELARUS

Belarussian Organization of Patients with MPS and other Rare Genetic Disorders	http://www.mpssociety.by	Full Member

BELGIUM

22Q11 Europe	https://22q11europe.org/	Associate Member
Alpha-1 Plus Asbl	http://www.alpha1plus.be/	Associate Member
ALS Liga België	http://www.alsliga.be	Full Member
Association Belge du Syndrome de Marfan Asbl	https://www.marfan.be/	Full Member
Association des Patients Sclérodermiques de Belgique	https://www.sclerodermie.be/	Full Member
Association Lupus Erythematosus	http://www.lupus-belgium.org	Full Member
Association pour l'Information et la Recherche sur les maladies rénales génétiques	http://www.airg-belgique.org	Associate Member
Belgische Organisatie Voor Kinderen En Volwassenen Met Een Stofwisselingsziekte	http://www.boks.be	Full Member
Belgische Vereniging voor Longfibrose VZW	http://www.longfibrose.org	Associate Member
BE-TSC VZW	http://www.betsc.be	Full Member
CF Europe	http://www.cf-europe.eu	Full Member
Children's Tumor Foundation Europe	https://www.ctfeurope.org/	Associate Member
Contactgroep Myeloom En Waldenström Patiënten	http://www.cmp-vlaanderen.be	Full Member
Debra Belgium	http://www.debra-belgium.org	Full Member
Euro Ataxia - European Federation of hereditary Ataxias	http://www.euroataxia.org	Full Member
European Chromosome 11 Network	http://www.chromosome11.eu	Full Member
European CMT Federation	http://www.ecmtf.org	Associate Member
European Federation of Williams Syndrome	http://www.eurowilliams.org	Full Member
European Haemophilia Consortium	http://www.ehc.eu	Full Member
European Idiopathic Pulmonary Fibrosis & Related Disorders Federation	http://www.eu-ipff.org	Associate Member
European Myasthenia Gravis Association	http://www.eumga.eu	Associate Member
European Network For Research On Alternating Hemiplegia	http://www.enrah.net	Full Member
European Polio Union	http://www.europeanpolio.eu	Full Member
Eye Hope Foundation	http://www.eyehopefoundation.org	Associate Member
Familial Adenomatous Polyposis Association	http://www.belgianfapa.be/fr	Associate Member
Federation of European Scleroderma Associations	http://www.fesca-scleroderma.eu	Associate Member
FEDERG - Federation of European Associations of Patients affected by Renal Diseases	http://federg.org	Full Member
HTAP Belgique asbl	http://test.ph-vzw.be/fr_FR/	Full Member
Ichthyose Belgique - Ichthyosis België	http://www.ichthyosis.be	Full Member
International Federation for Spina Bifida and Hydrocephalus	http://www.ifglobal.org	Full Member
Liver Patients International	https://liverpatientsinternational.org/	Associate Member
Lupus Europe	http://www.lupus-europe.org	Associate Member
Myeloma Patients Europe	http://www.mpeurope.org	Associate Member
OIFE - Osteogenesis Imperfecta Federation Europe	http://www.oife.org	Full Member
Patiëntenvereniging Zeldzame Leverziekten	https://www.mijnlever.be/	Full Member
RaDiOrg - Rare Diseases Belgium asbl/vzw	http://www.radiorg.be	Full Member
Rare Disorders Belgium	http://www.rd-b.be	Associate Member
Relais 22 Asbl	http://www.relais22.be	Full Member
Siop Europe - European Society For Paediatric Oncology	http://www.siope.eu	Associate Member
Spierziekten Vlaanderen VZW	https://spierziektenvlaanderen.be	Full Member
Steunpunt Kinderepilepsie vzw	https://kinderepilepsie.be/	Full Member
Vascular Anomaly Patient Association	http://www.vascapa.org	Full Member
Vlaams Patiëntenplatform vzw	http://www.vlaamspatientenplatform.be	Associate Member
Vlaamse Vereniging voor erfelijke Bindweefselaandoeningen	http://bindweefsel.be	Full Member
vzw GEN	https://www.vzw-gen.be/	Associate Member
Zebrapad VZW	https://www.zebrapadvzw.be	Full Member

BENIN

Albinos Sans Frontières		Associate Member

BOSNIA AND HERZEGOVINA

Alliance for rare diseases of Republic of Srpska, Bosnia and Herzegovina	https://savezzarijetke.org/	Full Member

BRAZIL

Associacao Brasileira de Enfermedades Raras	http://feberraras.wixsite.com/feber-raras	Associate Member
Associação Brasileira de Paramiloidose	http://www.abpar.org.br/	Associate Member
Instituto Vidas Raras	http://www.vidasraras.org.br	Associate Member

BULGARIA

Assocation Of People Suffering By Acromegaly In Bulgaria		Associate Member
Association of Tarlov Cyst patients in Bulgaria	http://tarlov-bg.eu/	Full Member
Bulgaria society of patients with pulmonary hypertension	http://www.bspph.net	Full Member
Bulgarian Association Wilson Disease	https://wilsonbg.org	Full Member
Bulgarian Cystic Fibrosis Association	http://lifewithcf.org/	Full Member
Bulgarian Huntington Association	http://huntington.bg	Associate Member
Bulgarian National Alliance of People with Rare Diseases	http://rare-bg.com/	Associate Member
Nas - National Association Sarcoidosis Bulgaria	http://sarcoidosisbulgaria.alle.bg/	Full Member
National Association For Child Support Congenital Hypothyroidism	http://www.abv.bg	Associate Member
National Association of Patients With Growth Hormone Deficiency	https://www.facebook.com/profile.php?id=100069309093196	Full Member
National Association of Patients with Mitochondrial Diseases in Bulgaria	http://www.mitobg.com/	Full Member
National Association of Syringomyelia		Full Member
National Gaucher Organization	http://gaucher-bg.org/	Associate Member
National Patients' Organisation	https://npo.bg/	Associate Member
PHA Bulgaria	http://www.phabulgaria.eu	Full Member
Retina Bulgaria	http://www.retinabulgaria.bg	Associate Member
Together Fighting Sarcoma	https://sarcoma.bg/	Associate Member

BURKINA FASO

Fondation Internationale Tierno et Mariam	http://www.fitima.org	Associate Member

CANADA

Canadian Organization For Rare Disorders	http://www.raredisorders.ca	Associate Member
Lymphoma Coalition	http://www.lymphomacoalition.org/	Associate Member

PVNH Support & Awareness	http://www.pvnhsupport.com	Associate Member
CHINA		
Chinese Organization for Rare Disorders	http://www.hanjianbing.org	Associate Member
Illness Challenge Foundation	http://www.chinaicf.org/	Associate Member
COLOMBIA		
Asociación Colombiana De Pacientes Con Enfermedades De Depósito Lisosomal	http://www.acopel.org	Associate Member
Foundation Diana Garcia de Olarte for PID	http://www.fundacion-fip.org	Associate Member
CROATIA		
Debra Croatia	http://www.debra-croatia.com	Full Member
Dravet sindrome Croatia	http://dravet-sindrom-hrvatska.hr	Associate Member
Rare Diseases Croatia	https://rijetke-bolesti.com/	Full Member
CYPRUS		
Cyprus Alliance For Rare Disorders	http://raredisorderscyprus.com/	Full Member
Cyprus Association of Inherited Metabolic Diseases 'Aspida Zois'	http://www.aspidazois.com	Full Member
Pancyprian Association For Rare Genetic Diseases "Unique Smiles"	http://www.monadikaxamogela.com	Full Member
Thalassaemia International Federation	http://www.thalassaemia.org.cy	Full Member
The Association of People with Primary Immunodeficiency and Friends		Full Member
CZECH REPUBLIC		
Association of Atypical Parkinsonian Syndromes		Associate Member
Czech Huntington Association	http://www.huntington.cz	Associate Member
HAE Junior	https://haejunior.cz/	Associate Member
Klub Nemocnych Cystickou Fibrozou	http://www.cfklub.cz	Full Member
Meta, Association of Patients with Lysosomal Storage Diseases	http://www.sdruzenimeta.cz	Full Member
Narodni Sdruzeni Pku A Jinych Dmp (Czech Pku Association)	http://www.nspku.cz	Full Member
Rare Diseases Czech Republic (Ceska Ascociace Pro Vzacna Onemocneni)	http://www.vzacna-onemocneni.cz	Full Member
DENMARK		
22Q11 Danmark	http://www.22q11.dk	Full Member
Addison Foreningen i Danmark	http://www.addison.dk	Full Member
Blæreekstrofiforeningens	http://www.lfmb.dk	Full Member
CCHS Danmark	http://cchsdanmark.wordpress.com/	Associate Member
Danish Apert Syndrome Association /Danmarks Apertforening	http://www.apertforening.dk	Full Member
Danmarks Bloderforening / Danish Haemophilia Society	http://www.bloderforeningen.dk	Full Member
Ehlers-Danlos Foreningen I Danmark	http://www.ehlers-danlos.dk	Full Member
Foreningen for Ataksi / HSP	http://www.sca-hsp.dk	Full Member
HAE Scandinavia	https://haescan.org/	Full Member
Ichtyosis Association in Denmark	http://www.iktyosis.dk	Full Member
MCADD-Foreningen	http://www.mcadd.dk	Full Member
Mitokondrie-Foreningen I Danmark	http://www.mitokondrie.dk/index.php	Full Member
Möbius Syndrom Foreningen / Moebius Syndrome Association in Denmark	http://www.moebiussyndrom.dk	Full Member
Porfyriforeningen Danmark - Porphyria Association Denmark	http://www.porfyriforeningen.dk	Full Member
Rare Diseases Denmark (Sjaeldne Diagnoser)	http://www.sjaeldnediagnoser.dk/	Full Member
The Danish Osteogenesis Imperfecta Society	http://www.dfoi.dk	Full Member
Wilson Patientforeningen	http://www.wilsons.dk	Full Member
XLH, arvelig rakitis (Hereditary Rickets Patient Association)	https://xlh-patientforeningen.dk/	Associate Member
ESTONIA		
Estonian Prader Willi Syndrome Association	http://www.pws.ee	Full Member
FINLAND		
Association of Cancer Patients in Finland (Suomen Syöpäpotilaat Ry,)	http://www.syopapotilaat.fi	Full Member
Finnish Acoustic Neuroma Association	https://www.akustikusneurinoomayhdistys.com/	Full Member
Finnish Allergy, Skin And Asthma Federation	http://www.allergia.fi	Full Member
Finnish Association for Ultra-Rare Diseases	http://www.ultraharvinaiset.fi	Full Member
Finnish Association of People with Physical Disabilities	http://www.invalidiliitto.fi	Full Member
Finnish Neuro Society	http://www.ms-liitto.fi	Full Member
Harso-Rare Disease Alliance Finland	https://www.harso.fi/en/home/	Full Member
Harvinaiset-Verkosto - Finnish Network For Rare Diseases	http://www.harvinaiset.fi	Associate Member
Inclusion Finland Kvtl	http://www.kvtl.fi	Full Member
Rinnekoti Foundation, Norio Centre	http://www.rinnekoti.fi	Associate Member
SMA Finland ry	http://www.smafinland.fi/	Full Member
Suomen Vaskuliittiyhdistys Ry / Finnish Vasculitis Association	https://www.vaskuliittiyhdistys.fi/	Full Member
FRANCE		
ACO2 Gene Association	https://aco2gene.wixsite.com/asso	Associate Member
afa Crohn RCH France	http://www.afa.asso.fr	Associate Member
AFMKT-France	http://www.associationkystedetarlov.com	Full Member
AFM-Téléthon	http://www.afm-telethon.com/	Full Member
Agir pour la Malformation Lymphatique en Alliance	http://www.asso-amla.org	Full Member
Alliance Maladies Rares	http://www.alliance-maladies-rares.org	Full Member
Alliance Syndrome de Dravet	http://www.dravet.fr	Full Member
Amis de ADNP France	https://www.alliance-maladies-rares.org/association/amis-de-adnp-france/	Associate Member
Amyloidosis Alliance the Voice of Patients	https://www.amyloidosisalliance.org/	Associate Member
ASL HSP-France	http://www.asl-hsp-france.org	Full Member
Ass. Internationale De Dystrophie Neuro Axonale Infantile	http://www.nbiaalliance.org	Full Member
Assedea	http://www.assedea.fr	Full Member
Association ISIS	http://www.isis-asso.com	Full Member
Association "Turner et Vous"	http://turneretvous.org	Full Member
Association AMS-ARAMISE	http://www.ams-aramise.fr	Full Member
Association Anémies Dyserythropoïétiques Congénitales	https://www.petitcoeurdebeurre.fr/	Full Member
Association Bernard Pépin pour la Maladie de Wilson	http://www.abpmaladiewilson.fr	Full Member
Association BPAN France	https://bpanfrance.fr/	Full Member
Association Contre Les Maladies Mitochondriales	http://www.association-ammi.org	Full Member
Association Craniopharyngiome Solidarité	http://www.cranio.fr	Full Member
Association des Groupes Amitié Turner	http://www.agat-turner.org	Full Member
Association des Malades du Syndrome de Mccune-Albright	http://www.assymcal.org	Full Member

Association des Malades Souffrant d'Angio Oedème par déficit en C1 Inhibiteur	http://www.amsao.fr	Full Member
Association des Pancréatites Chroniques Héréditaires	http://www.association-apch.org	Full Member
Association des Patients de la maladie de Fabry	http://www.apmf-fabry.org	Associate Member
Association des personnes concernées par le Tremblement Essentiel	http://www.aptes.org	Associate Member
Association du Naevus Géant Congénital	http://naevus.fr	Full Member
Association Française contre l'Amylose	http://www.amylose.asso.fr	Full Member
Association Française de Gilles de la Tourette	http://www.france-tourette.org	Full Member
Association Française de La Maladie de Fanconi	http://www.fanconi.com	Full Member
Association Française de l'Ataxie de Friedreich	http://www.afaf.asso.fr/	Full Member
Association Française de l'Osteodystrophie Hereditaire d'Albright	http://www.afoha.org	Full Member
Association Française de l'Ostéogenèse Imparfaite	http://www.aoi.asso.fr	Full Member
Association Française de Narcolepsie-Cataplexie et Hypersomnie	http://www.anc-narcolepsie.com	Full Member
Association Française des Dysplasies Ectodermiques	http://www.afde.net	Full Member
Association Française des Hémophiles	http://www.afh.asso.fr	Full Member
Association Française des Malades atteints de Porphyries	http://www.porphyries-patients.org	Associate Member
Association Française des Maladies Héréditaires du Rythme Cardiaque	http://www.afmhrc.org	Full Member
Association Française des Syndromes d'Ehlers-Danlos	http://www.afsed.com	Full Member
Association Française du Gougerot-Sjögren	http://www.afgs-syndromes-secs.org	Full Member
Association Française du Lupus et autres Maladies Auto-Immunes	http://www.lupusplus.com	Full Member
Association Française du Syndrome de Cornelia de Lange	http://afscdl.fr	Full Member
Association Française du Syndrome de Klippel-Feil	https://afskf.fr/	Full Member
Association Française du Syndrome de Lowe	http://www.syndrome-lowe.org	Full Member
Association Française du Syndrome de Rett	https://afsr.fr/	Full Member
Association Française du Syndrome d'Ondine	https://afsondine.org/	Full Member
Association Française du Syndrome Phelan-Mcdermid	http://22q13.fr	Full Member
Association Française Lesch-Nyhan Action	http://www.lesch-nyhan-action.org	Full Member
Association Française Niemann Pick	https://www.niemannpick-france.org/	Associate Member
Association Francaise Sturge Weber "Vanille-Fraise"	http://www.vanille-fraise.org	Associate Member
Association France Vascularites	http://www.association-vascularites.org	Full Member
Association Francophone contre la Polychondrite Chronique Atrophiante	http://www.afpca.fr	Associate Member
Association Francophone de la Maladie de Blackfan Diamond	http://www.afmbd.org	Full Member
Association Francophone des Glycogénoses	http://www.glycogenoses.org	Full Member
Association Francophone du Syndrome d'Angelman	http://www.angelman-afsa.org	Full Member
Association Gitelbart	https://gitelbart.fr	Full Member
Association Grandir	http://www.grandir.asso.fr	Full Member
Association Hemochromatose France	http://www.hemochromatose.fr	Associate Member
Association Histiocytose France	http://www.histiocytose.org	Full Member
Association Huntington France	https://huntington.fr/	Full Member
Association Hypoparathyroidisme France	http://www.hypopara.fr	Full Member
Association Ichtyose France	http://www.ichtyose.fr	Full Member
Association Internationale Maladies Kystes Tarlov	http://www.aimktarlov.org	Associate Member
Association Kourir	http://www.kourir.org	Full Member
Association Maladies Foie Enfants	http://www.amfe.fr/	Full Member
Association Marfans	http://www.assomarfans.fr	Full Member
Association Microphtalmie France	http://asso-microphtalmie.org	Full Member
Association Naevus 2000	https://www.naevus2000.com/	Full Member
Association Nationale des Cardiaques Congénitaux	http://www.ancc.asso.fr	Full Member
Association Neurofibromatoses & Recklinghausen	http://www.anrfrance.fr	Full Member
Association Noonan	http://www.assonoonan.fr/	Full Member
Association Ollier Maffucci Europe	http://www.olliermaffucci-asso.fr	Full Member
Association Pemphigus – Pemphigoïdes France	http://www.pemphigus.asso.fr	Full Member
Association pour aider et informer les Syringomyéliques Européens Réunis	https://www.apaiser.org/	Full Member
Association pour la lutte contre l'Alcaptonurie	http://www.alcap.fr	Full Member
Association pour la Lutte contre les maladies Inflammatoires du Foie et des voies biliaires	http://www.albi-france.org	Full Member
Association pour la recherche sur la Sclérose Latérale Amyotrophique	https://www.arsla.org/	Associate Member
Association pour la Sensibilisation aux Maladies Rares, Orphelines et Auto-immunes à Saint-Pierre-et-Miquelon	https://maladiesrarespm.simdif.com/index.html	Full Member
Association pour l'aide aux personnes concernées par les Maladies Rares Muckle Wells Syndrome et CINCA	http://www.amws-caps.org/	Full Member
Association pour l'information et la prévention de la Drépanocytose	http://www.apipd.fr	Full Member
Association pour l'information et la recherche sur les Maladies Rénales Génétiques	http://www.airg-france.fr	Full Member
Association Sans Diagnostic et Unique		Associate Member
Association Sclérose Tubéreuse de Bourneville	http://www.astb.asso.fr	Full Member
Association SED1+	http://www.assosed1plus.com/	Full Member
Association SOS Desmoïde	http://www.sos-desmoide.asso.fr	Full Member
Association Spina Bifida et handicaps associés	http://www.spina-bifida.org	Associate Member
Association Surrénales	http://www.surrenales.com	Full Member
Association syndrome de Kleine-Levin	http://kls-france.org	Full Member
Association Syndrome de Moebius France	http://www.moebius-france.org	Full Member
Association Syndrome PACS1 - Schuurs-Hoeijmakers	http://www.pacs1.org/	Associate Member
Association Tanguy Moya Moya	http://www.tanguy-moya-moya.org	Full Member
AVML Vivre Mieux le Lymphœdème	http://www.avml.fr/	Full Member
Charcot-Marie-Tooth France	http://www.cmt-france.org	Full Member
Cutis Laxa Internationale	http://www.cutislaxa.org	Full Member
Debra France	http://www.debra.fr	Full Member
Diabete Insipide France		Associate Member
Dravet Syndrome European Federation	http://www.dravet.eu	Full Member
Dup15q France	https://www.dup15qfrance.fr	Full Member
Enfants de la Lune Association pour le Xeroderma Pigmentosum	http://www.enfantsdelalune.org	Full Member
Ensemble Leucemie Lymphomes Espoir	https://www.francelymphomeespoir.fr	Full Member
Euro-Dyma	https://euro-dyma.eu/	Associate Member
European Federation for Hereditary Spastic Paraplegia	http://eurohsp.eu/	Full Member
European Federation Lesch-Nyhan Disease	http://www.LNDE.org	Associate Member
European Federation of Associations of Patients with Haemochromatosis	http://efaph.eu/	Full Member
European Patient Organisation for Dysimmune and Inflammatory Neuropathies	https://www.epodin.org/	Associate Member
EwenLife Rare Diseases	https://www.ewenlife.org/	Associate Member
Fédération SOS Globi	https://sosglobi.fr/	Full Member
Fédération Williams France	http://www.williams-france.org	Full Member
Fitima Europe - Fondation International Tierno et Mariam	http://www.fitima.org	Full Member
FOP France	http://www.fopfrance.fr	Full Member

Fragile X France	http://www.xfra.org	Full Member
France Lymphangioleiomyomatose	http://www.francelam.org	Full Member
Fructos'Amis pour la Vie	http://www.fructosamispourlavie.org/	Full Member
Génération 22	http://www.generation22.fr	Full Member
Genespoir: Association Française des Albinismes	http://www.genespoir.org	Full Member
Geniris	http://associationgeniris.free.fr/	Full Member
HTaPFrance	http://www.htapfrance.com	Full Member
Hypophosphatasie Europe	http://www.hypophosphatasie.com	Full Member
Incontinentia Pigmenti France	http://incontinentia-pigmenti.fr/	Full Member
Inflam'Œil	http://www.inflamoeil.org	Full Member
La Maison 8p	https://www.lamaison8p.fr	Associate Member
LAMA2 France, Contre les dystrophies musculaires par déficit en mérosine	https://www.lama2.fr/	Full Member
Les Amis de Remi	http://www.lesamideremi.fr	Full Member
Les Petits Bourdons	http://www.lespetitsbourdons.org	Associate Member
Ligue contre la Cardiomyopathie	http://www.ligue-cardiomyopathie.com/	Associate Member
Ligue Nationale Contre Le Cancer	http://www.ligue-cancer.net	Full Member
Lupus France	http://www.lupusfrance.com/	Full Member
MED13L Syndrome association	http://www.med13lsyndrome.eu/	Associate Member
Mosaïques - Association Des "X Fragile"	http://www.xfragile.org	Full Member
Ouvrir Les Yeux	http://www.ouvrirlesyeux.org	Full Member
Petit Coeur de Beurre	https://www.petitcoeurdebeurre.fr/page/43746-l-association	Full Member
Polyposes Familiales - APTEPF	http://www.polyposes-familiales.fr	Full Member
Prader Willi France	http://www.prader-willi.fr	Full Member
Retina France	http://www.retina.fr	Full Member
Sanfilippo Sud	https://www.facebook.com/sanfilipposud/	Full Member
Solidarité Handicap autour des Maladies Rares	http://www.solhand-maladiesrares.org	Full Member
Tintamarre	https://www.asso-tintamarre.org/	Full Member
Union Huntington Espoir	http://www.huntington.asso.fr	Full Member
Union Nationale des Associations Parents et Amis Personnes Handicapées Mentales	http://www.unapei.org	Associate Member
Union Nationale des Syndromes d'Ehlers-Danlos	http://www.unsed.org	Full Member
Vaincre Dunnigan	http://vaincre-dunnigan.org/	Associate Member
Vaincre la Mucoviscidose	http://www.vaincrelamuco.org	Full Member
Vaincre La Papillomatose Respiratoire Récurrente	http://www.vaincreprr.fr	Full Member
Vaincre Les Maladies Lysosomales	http://www.vml-asso.org	Full Member
Valentin APAC - Association de Porteurs d'Anomalies Chromosomiques	http://www.valentin-apac.org	Full Member
VHL France	http://www.vhlfrance.org	Full Member
Vivre sans Thyroide	https://www.forum-thyroide.net/	Full Member
White Sutton France	https://whitesuttonfrance.wixsite.com/	Associate Member
Xtraordinaire	http://www.xtraordinaire.org	Full Member

GEORGIA

Georgian Foundation for Genetic and Rare Diseases		Full Member
SCN2A Georgia		Associate Member

GERMANY

AHC-Deutschland e.V.	http://www.ahckids.de	Full Member
Allianz Chronischer Seltener Erkrankungen e.V.	http://www.achse-online.de	Full Member
Angelman e.V.	http://www.angelman.de	Full Member
ARVC-Selbsthilfe e.V.	http://www.arvc-selbsthilfe.org	Full Member
BSHV für Kinder, Jugendliche und Erwachsene mit seltenen,chronischen Skeletterkrankungen e.V.	http://bshv-seltene-skelett-erkrankungen.com	Full Member
Bundesverband Angeborene Gefäßfehlbildungen e.V.	http://www.angiodysplasie.de	Full Member
Bundesverband der Clusterkopfschmerzen-Selbsthilfe-Gruppen	http://www.clusterkopf.de	Full Member
Bundesverband für PFFD, FFU, Fibula- und Tibiadefekte e.V.	https://bupft.de/	Associate Member
Bundesverband Kleinwüchsige Menschen und ihre Familien e.V.	http://www.bkmf.de	Full Member
Bundesverband Niere e.V	https://www.bundesverband-niere.de/	Associate Member
Bundesverband Poliomyelitis e.V.	http://polio-selbsthilfe.de/willkommen	Full Member
Bundesverband Schilddrüsenkrebs - Ohne Schilddrüse Leben e.V.	https://www.sd-krebs.de/	Full Member
Charge Syndrom e.V.	http://www.charge-syndrom.de	Full Member
Cholesterin & Co e.V.	http://cholco.org	Associate Member
CRPS Bundesverband Deutschland e.V.	https://crps-netzwerk.org/cms/	Full Member
Cystinose Selbsthilfe e.V.	http://www.leben-eben.com	Full Member
Deutsche Duchenne Stiftung	https://www.duchenne-deutschland.de/	Full Member
Deutsche Ehlers-Danlos Initiative e.V.	http://www.ehlers-danlos-initiative.de	Full Member
Deutsche GBS CIDP Selbsthilfe e.V.	http://gbs-selbsthilfe.org	Full Member
Deutsche Interessengemeinschaft PKU	http://www.dig-pku.de	Full Member
Deutsche Klinefelter-Syndrom Vereinigung e.V.	http://www.klinefelter.de	Full Member
Deutsche Sarkoidose Vereinigung e.V.	http://www.Sarkoidose.de	Full Member
Deutsche Syringomyelie und Chiari Malformation DSCM e.V.	http://www.deutsche-syringomyelie.de	Full Member
Deutsche Uveitis-Arbeitsgemeinschaft e.V.	http://www.duag.org	Full Member
Deutschsprachige Selbsthilfegruppe für Alkaptonurie e.V.	http://www.dsaku.de	Full Member
dsai e.V. - Patientenorganisation für angeborene Immundefekte	http://www.dsai.de	Full Member
Dup15q e.V.	https://dup15q.de/	Full Member
EAT – Esophageal Atresia Global support groups	http://www.we-are-eat.org	Full Member
European Association of Patient Organisations of Sarcoidosis	http://www.sarcoidosis.biz	Full Member
European Congenital Heart Disease Organisation	http://echdo.eu	Associate Member
European MEN Alliance	http://www.Emena.eu	Associate Member
European Network for Ichthyosis	https://ichthyosis.info/	Full Member
Faun Stiftung	http://faun-stiftung.de	Associate Member
FOP Germany (Förderverein für an Fibrodysplasia Ossificans Progressiva Erkrankte)	http://www.fop-ev.de	Full Member
Gaucher Gesellschaft Deutschland e.V.	http://www.ggd-ev.de	Full Member
Gesellschaft für Mukopolysaccharidosen e.V.	http://www.mps-ev.de	Full Member
HAE Vereinigung e.V. (Hereditary Angioedema)	http://www.angiooedem.de	Full Member
Hand in Hand gegen Tay-Sachs und Sandhoff e.V.	http://tay-sachs-sandhoff.de/	Full Member
Hoffnungsbaum e.V. - Verein zur Förderung der Erforschung und Behandlung von NBIA-Erkrankungen	http://www.hoffnungsbaum.de	Full Member
HSP-Selbsthilfegruppe Deutschland e.V.	http://www.hsp-selbsthilfegruppe.de	Full Member
IEB e.V. DEBRA Deutschland	http://www.ieb-debra.de	Full Member
IMBS Alliance	https://www.imbs-alliance.org/	Associate Member

Interessengemeinschaft Fragiles-X e.V.	http://www.frax.de	Full Member
Kinder-Augen-Krebs-Stiftung	http://www.kinderaugenkrebsstiftung.de	Associate Member
Kindernetzwerk e.V.	http://www.kindernetzwerk.de	Associate Member
Kindness for Kids Foundation	http://www.kindness-for-kids.de	Associate Member
Leben mit Behcet in Deutschland	http://www.behcet-selbsthilfe.de	Associate Member
Leona e.V.	http://www.leona-ev.de	Full Member
Marfan Europe Network	http://www.marfan.eu	Full Member
Marfan Hilfe Deutschland e.V.	http://www.marfan.de	Associate Member
Morbus-Osler Selbsthilfe e.V	http://www.morbus-osler.de	Full Member
MPN-Netzwerk E.V.	http://www.Mpn-netzwerk.de	Full Member
MPS Europe	http://mps-europe.org	Full Member
Myelitis e.V.	http://www.myelitis.de	Full Member
NCL-Gruppe Deutschland e.V.	http://www.ncl-deutschland.de	Full Member
Nephie e.V. - Selbsthilfe nephrotisches Syndrom	http://www.nephie.de	Full Member
Netzwerk Hypophysen- und Nebennierenerkrankungen e.V. - Network pituitary and adrenal disorders	http://www.glandula-online.de	Full Member
Patienten- und Selbsthilfeorganisation für Kinder und Erwachsene mit Kranker Speiseröhree Mit Kranker Speiseröhre	https://keks.org/	Full Member
Patientenverband Familiäre Amyloid Polyneuropathie	http://patientenverband-fap.de	Associate Member
PCH-Familie e.V.	https://pch-familie.de/	Full Member
Peutz-Jeghers-Germany e.V.	http://peutz-jeghers.eu	Full Member
PKD Familiäre Zystenniere e. V.	http://www.pkdcure.de	Full Member
Pro Retina Deutschland e.V.	http://www.pro-retina.de	Associate Member
Pulmonale Hypertonie e.V.	http://www.phev.de	Full Member
Sanfilippo Initiative e.V.	http://www.sanfilippoinitiative.org	Associate Member
Selbsthilfe EPP e.V.	http://www.epp-deutschland.de	Full Member
Selbsthilfe Ichthyose e.V.	http://www.ichthyose.de	Full Member
Selbsthilfegruppe Ektodermale Dysplasie e.V.	http://www.ektodermale-dysplasie.de	Full Member
Selbsthilfegruppe Für PXE-Erkrankte Deutschlands e.V.	http://www.pxe-shg.de	Full Member
Selbsthilfegruppe Glykogenose Deutschland e.V.	http://www.glykogenose.de	Full Member
Selbsthilfeorganisation für Menschen mit Anorektalfehlbildungen	http://www.soma-ev.de	Full Member
Sklerodermie Selbsthilfe e.V.	https://www.sklerodermie-sh.de/	Full Member
SMA Europe	http://www.sma-europe.eu	Full Member
SSADH-Defizit e.V.	http://ssadh.wordpress.com	Associate Member
Tom Wahlig Stiftung	http://www.hsp-info.de	Full Member
Verein Ahc18+	https://www.facebook.com/profile.php?id=100070945728878	Associate Member
Verein VHL (Von Hippel - Lindau) betroffener Familien e.V.	http://www.hippel-lindau.de	Full Member
Wir sind 22Q e.V.	http://www.wirsind22q.de/	Full Member
XP – Freu(n)de Mondscheinkinder	http://www.xerodermapigmentosum.de	Full Member
ZNM - Zusammen Stark! e. V.	http://www.znm-zusammenstark.org	Associate Member

GREECE

"95" - Rare Alliance Greece	http://www.rarealliance.gr	Full Member
AGORA – federation of associations for patients with rheumatic and musculoskeletal diseases of Southern Europe	https://www.agora-platform.eu/	Associate Member
Angelman syndrome Greece	https://angelman.gr/	Full Member
Association of Greek Friends for Paediatric Immunology Primary Immunodeficiencies "Harmony"	http://www.paed-anosia.gr	Associate Member
Child's Heart	http://www.kardiapaidiou.gr	Full Member
Hellenic Cystic Fibrosis Association	http://www.cysticfibrosis.gr	Full Member
Hellenic Friedreich's Ataxia Association	https://www.hefaa.org	Full Member
Hellenic League against Rheumatism	http://www.arthritis.org.gr	Associate Member
Hellenic Myasthenia Gravis Association	http://www.myasthenia.gr	Full Member
Immune Deficiency Association GALINOS	https://galinossilogos.wixsite.com/galinos	Associate Member
Karkinaki Awareness for childhood and adolescent Cancer	http://www.karkinaki.gr	Associate Member
KRIKOS ZOIS Society for Patients and Friends of Patients with Inherited Metabolic Disease	http://www.krikoszois.gr	Full Member
Muscular Dystrophy Association Hellas	http://mdahellas.gr	Full Member
Panhellenic Association of Patients & Friends with Neurofibromatosis "Life With NF"		Associate Member
Panhellenic Association of Patients with Lysosomal Disorders	http://greeklysosomal.gr	Full Member
Parents and Friends of People with Rett Syndrome Association	http://www.rettgreece.gr	Associate Member
Pespa (Greek Alliance for Rare Diseases)	http://www.pespa.gr	Associate Member
Prader Willi Syndrome Association Hellas		Full Member
Pulmonary Hypertension of Greece – Hellenic Pulmonary Hypertension	http://www.hellenicpulmonaryhypertension.gr/	Associate Member
Rare Diseases Greece		Associate Member
To Mellon- Association of People with Genetic Disorders	http://www.tomellon.com.gr	Full Member
Together for Life		Full Member
VHLFA Alliance in Greece	https://www.vhlgr.org/	Associate Member

GUATEMALA

Asociación Nacional Guatemalteca para las Enfermedades de depósito Lisosomal	http://angel-lisosomal.org/	Associate Member
Procrece	https://www.facebook.com/asociacionprocreceguatemala/	Associate Member

HONG KONG, SAR OF CHINA

Joshua Hellmann Foundation For Orphan Disease	http://www.jhforphandisease.org/	Associate Member

HUNGARY

Hungarian Haemophilia Society - Magyar Hemofília Egyesület	http://www.mhe.hu	Full Member
Kéregtest rendellenesség AgCC Magyarországi Alapítványa / Disorder of the Corpus Callosum Hungary Fundation	https://www.agykeregtest.hu/	Associate Member
Magyaórszági Mitochondriális Betegek Alapítványa	http://www.nbia.hu	Full Member
Primer Immunhianyos Betegek Egyesülete	http://www.pibe.hu	Full Member
Rare Diseases Hungary - HUFERDIS	http://www.rirosz.hu	Full Member

ICELAND

AHC Federation of Europe	http://www.ahcfe.eu	Associate Member
Alternating Hemiplegia of Childhood Association of Iceland	http://www.ahc.is	Full Member
Einstök Börn - Support Group for Children with Rare Disorders	http://www.einstokborn.is	Full Member

Gudrun's Rett Syndrome Research Trust	http://rettenglar.yolasite.com	Associate Member
INDIA		
Indian Organization for Rare Diseases	https://www.rarediseases.in/	Associate Member
Organization For Rare Diseases India	https://ordindia.in/	Associate Member
IRAN (ISLAMIC REPUBLIC OF)		
Rare Disease Foundation of Iran	http://radoir.org/fa/	Associate Member
IRELAND		
22Q11 Ireland Support Group	http://www.22q11ireland.org	Full Member
Alpha One Foundation	http://www.alpha1.ie	Full Member
Barretstown Serious Fun	http://www.barretstown.org	Associate Member
Cystic Fibrosis Ireland	http://www.cfireland.ie	Associate Member
Cystinosis Ireland	http://cystinosis.ie	Full Member
Debra Ireland	http://www.debraireland.org	Full Member
European Sickle Cell Federation	https://escfederation.eu/	Associate Member
Fighting Blindness	http://www.fightingblindness.ie	Full Member
Friedreich's Ataxia Research Alliance	http://www.faraireland.ie	Full Member
Irish Cancer Society	http://www.cancer.ie	Associate Member
Irish MPS Society	http://www.mpssociety.ie/wordpress/	Full Member
Neurofibromatosis Association of Ireland	http://www.nfaireland.ie	Full Member
Popsycle Foundation	https://www.facebook.com/Popsycle01/	Associate Member
Rare Diseases Ireland	http://rdi.ie/	Full Member
Rare Ireland	https://www.rareireland.ie/	Associate Member
Retina International	http://www.retina-international.org	Full Member
Sickle Cell and Thalassaemie Ireland	http://www.sicklecellireland.ie	Full Member
The Cavan Tommy Hoey Trust	https://www.facebook.com/the.cavan.tommy.hoey.trust/	Associate Member
The Irish Fragile X Society	http://fragilexireland.org	Full Member
Usher syndrome Ireland	https://usherireland.org/	Associate Member
Vasculitis Ireland Awareness	http://www.vasculitis-ia.org	Full Member
ISRAEL		
CdLS Israeli Foundation	https://www.cdlsisrael.org.il/	Associate Member
Coalition of Rare Diseases in Israel		Associate Member
GRIN Disorders Research Foundation	http://www.gringn.com/	Associate Member
Israel Adult Polyglucosan Body Disease		Associate Member
Little Steps Association	https://www.littlesteps.org.il/	Associate Member
ITALY		
ABC Associazione Bambini Cri Du Chat	http://www.criduchat.it	Full Member
ACMT- Rete per la malattia di Charcot-Marie-Tooth OdV	http://www.acmt-rete.it	Full Member
Acondroplasia - Insieme Per Crescere - Onlus	http://www.acondroplasiaonlus.com	Full Member
AIBWS ODV	http://www.aibws.org	Full Member
Amici Della Porfiria - San Pio Da Pietrelcina Onlus	http://www.porfiria.it	Associate Member
Angeli Noonan	http://www.angelinoonan.it	Full Member
Aniridia Italiana APS	http://www.aniridia.it/	Full Member
Assi Gulliver - Associazione Sindrome di Sotos Italia	https://www.assigulliver.it/	Full Member
Associaizone Nazionale Pemfigo/Pemfigoide	https://www.pemfigo.org/	Full Member
Associazione conto alla Rovescia, the Patient Association for Multiple Osteochondromas, Ollier disease and Maffucci syndrome.	http://www.acar2006.org	Full Member
Associazione Famiglie COL4A1-A2 APS	http://www.col4a1.it/	Associate Member
Associazione Famiglie di Soggetti con Deficit dell'Ormone della Crescita ed altre Patologie	http://www.afadoc.it	Full Member
Associazione HHT Onilde Carini APS	http://www.hht.it/	Full Member
Associazione Italiana Adrenoleucodistrofia Onlus	https://www.adrenoleucodistrofia.it/	Associate Member
Associazione Italiana Cistite Interstiziale	http://www.aici-onlus.it	Full Member
Associazione Italiana Dei Cardiopatici Congeniti Adulti - Italian Guch Association	http://www.aicca.eu	Associate Member
Associazione Italiana Estrofia Vescicale-Epispadia ODV	http://www.estrofiavescicale.it	Full Member
Associazione Italiana Gaucher Onlus	http://www.gaucheritalia.org	Full Member
Associazione Italiana Glicogenosi	http://www.aig-aig.it	Full Member
Associazione Italiana Laryngectomizzati	http://www.ailar.it	Associate Member
Associazione Italiana Leucodistrofie Unite	http://digilander.libero.it/ailu/	Full Member
Associazione Italiana Linfoistiocitosi Emofagocitica	https://www.aileonlus.org/	Associate Member
Associazione Italiana Malati di Alcaptonuria	http://www.aimaku.it	Full Member
Associazione Italiana Miastenia e Malattie Immunodegenerative - Amici del Besta Onlus	http://www.miastenia.it	Full Member
Associazione Italiana Miastenia Onlus	http://www.viverelamiastenia.it	Full Member
Associazione Italiana Morbo di Hirschsprung	http://amorhi.org	Associate Member
Associazione Italiana Mucopolisaccaridosi e Malattie Affini	http://www.aimps.it	Full Member
Associazione Italiana Niemann Pick Onlus	http://www.niemannpick.org	Full Member
Associazione Italiana per la lotta alle PHTS	http://www.ptenitalia.org	Full Member
Associazione Italiana per le Malformazioni Anorettali	http://www.aimar.eu	Full Member
Associazione Italiana Rene Policistico Onlus	https://www.renepolicistico.it/	Full Member
Associazione Italiana Sindrome di Ehlers-Danlos	https://www.aised.it/	Full Member
Associazione Italiana Sindrome di Poland ODV	http://www.sindromedipoland.org	Full Member
Associazione Italiana Sindrome e malattia di Behçet	http://www.behcet.it	Full Member
Associazione Italiana Sindrome X Fragile	http://www.xfragile.net	Full Member
Associazione Italiana Siringomielia e Arnold Chiari	http://www.aismac.org	Full Member
Associazione Italiana Sostegno Malattie Metaboliche Ereditarie Onlus	http://www.aismme.org	Full Member
Associazione LAM Italia Onlus	http://www.lam-italia.org	Full Member
Associazione Ligure Thalassemici Onlus	http://www.althonlus.org	Full Member
Associazione Malattie Rare Dell'Alta Murgia Onlus	http://www.amaram.it	Associate Member
Associazione Nazionale Angioma Cavernoso Cerebrale	https://www.anaccaps.org/	Full Member
Associazione Nazionale Malattie Rare Dermatologiche Vascolari ODV	https://mrdv.org/	Associate Member
Associazione Nazionale Persone con Malattie Reumatologiche	http://www.apmar.it	Full Member
Associazione p63 Sindrome E.E.C. International APS	http://www.sindrome-eec.it	Associate Member
Associazione per le Immunodeficienze Primitive ODV	http://www.aip-it.org	Full Member
Associazione Per L'Informazione e lo Studio della Acondroplasia	http://www.aisac.it	Full Member
Associazione Persone Williams Italia Onlus	http://www.apwitalia.org	Full Member
Associazione Poic e dintorni Onlus	https://www.poic-e-dintorni.org/	Full Member

Associazione S.P.R.IN.T.	https://www.associazionesprint.org/	Associate Member
Associazione Sclerosi Tuberosa	http://www.sclerosituberosa.org	Full Member
Associazione Sindrome di Alström / Italian Association for Alstrom Syndrome	https://www.alstrom.it/	Full Member
Associazione Sindrome Nefrosica Italia	http://www.asnit.org	Full Member
Associazione Spina Bifida Italia	https://www.spinabifidaitalia.it/	Full Member
Associazione Studio Malattie Metaboliche Ereditarie Onlus	http://www.cometaasmme.org	Full Member
Associazione Veneta per la Lotta alla Talassemia	http://www.avlt.it	Full Member
CIDP Italia Onlus	http://www.cidp.it	Full Member
Collagene Vi Italia Onlus - Col6	http://www.col6.it	Full Member
Costello.Cfc - Associazione Italiana Sindrome Di Costello - Cardiofaciocutanea - Rasopatie - Onlus	http://www.sindromedicostello.it	Full Member
Debra Italia Onlus	http://www.debraitalia.com/	Full Member
Dravet Italia Onlus	http://www.dravet.it	Full Member
Federazione Sindrome di Prader Willi Italia	http://www.praderwilli.it	Full Member
Fondazione Alessandra Bisceglia ViVa Ale Onlus	http://www.fondazionevivaale.org	Associate Member
Fondazione Lega Italiana Ricerca Huntington	https://lirh.it/en	Full Member
FOP Italia Onlus	http://www.fopitalia.it	Full Member
Gli Amici di Daniela	http://www.amicididaniela.it	Associate Member
Gli Equilibristi HIBM	http://www.gliequilibristi-hibm.org/	Full Member
Gruppo "Italia - Glioblastoma Multiforme - Cancro al Cervello"	https://www.facebook.com/groups/italia.glioblastoma.multiforme/	Associate Member
Gruppo Famiglie Dravet Associazione Onlus	http://www.sindromedidravet.org/	Full Member
Gruppo Familiari Beta-Sarcoglicanopatie	http://www.beta-sarcoglicanopathy.org/	Full Member
Gruppo Italiano per la lotta alla Sclerodermia Onlus	http://www.sclerodermia.net	Full Member
HHT Europe	http://www.hhteurope.org	Full Member
HHT Onlus	http://www.hhtonlus.com	Full Member
Incontinentia Pigmenti Associazione Italiana Onlus	http://www.incontinentiapigmenti.it	Associate Member
La Strada per l'Arcobaleno	http://www.stradaperlarcobaleno.com	Full Member
Linfa Odv	https://www.linfaneurofibromatosi.com/	Full Member
LND Famiglie Italiane odv	http://www.lesch-nyhan.eu	Full Member
Mitocon- Insieme per lo Studio e la Cura delle Malattie Mitocondriali - Onlus	http://www.mitocon.it	Associate Member
Nana Onlus	https://www.nanaonlus.org/	Associate Member
PANDAS Italia	http://www.pandasitalia.com	Full Member
Parent Project aps	http://www.parentproject.it	Full Member
PKS Kids Italia Onlus	http://www.pksitalia.org	Full Member
Retina Italia Onlus	http://www.retinaitalia.org/	Full Member
Ring 14 International	http://www.ring14.org	Full Member
SCN2A ITALIA Famiglie in Rete APS	https://scn2a-italia.it/	Full Member
SCN8A Italia	https://www.scn8a.it/#	Full Member
SOD Italia - Associazione Septo Optic Dysplasia and Other Neuroendocrine Disorders	http://www.soditalia.it	Full Member
Un Filo per la Vita ANAD IICB	http://www.unfiloperlavita.it	Full Member
UNIAMO- Rare Diseases Italy	http://www.uniamo.org	Full Member
Unione Italiana Ittiosi	http://www.ittiosi.it	Full Member
Unione Italiana Lotta Alla Distrofia Muscolare	http://www.uildm.org/	Full Member
Unione Trapiantati Polmone - Padova ODV	https://oltrelamalattia.it/	Full Member
United Onlus	http://www.unitedonlus.org	Full Member
Uniti per la P.I.P.O.	https://www.unitiperlapipo.it/	Associate Member
World Federation of Incontinence Patients	http://www.wfip.org	Associate Member
XLPDR International Association	http://www.xlpdr.com	Associate Member
JAPAN		
Japan Patient Association	http://www.nanbyo.jp	Associate Member
KAZAKHSTAN		
Association of Support to Patients with Orphan Diseases in the Republic of Kazakhstan		Associate Member
Patients with Cancer and Rare Diseases Support Association		Associate Member
KOSOVO		
Rare Diseases Kosovo (Shoqata e Semundje te Rralla Kosove)	https://www.facebook.com/Shoqata-e-S%C3%ABmundjeve-t%C3%AB-Rralla-Kosov%C3%AB-2034022230042375/	Associate Member
LATVIA		
Association of People with Special Needs "Motus Vita"	http://www.motusvita.lv/index.php?lang=en	Associate Member
Latvian Alliance for Rare Diseases	http://www.retasslimibas.lv	Full Member
Rare Disease Association "Caladrius"	http://www.inbox.lv	Associate Member
LEBANON		
Lebanese Association for Neuromuscular Diseases	http://landforhope.org	Associate Member
LITHUANIA		
Community of Oncohematological Patient Organisations Kraujas	http://www.kraujas.lt	Full Member
Rare Diseases Lithuania (Vaikų retų ligų asociacija)	http://retosligos.lt/	Associate Member
LUXEMBOURG		
Alan - Maladies Rares Luxembourg	http://www.alan.lu	Full Member
EDS Lëtzebuerg a.s.b.l.	https://edsletzebuerg.lu/2/	Associate Member
Een Häerz Fir Kriibskrank Kanner Asbl	http://www.kriibskrankkanner.lu	Full Member
Fondatioun Kriibskrank Kanner	http://www.fondatioun.lu	Associate Member
Rett Syndrome Europe	http://www.rettsyndrome.eu	Full Member
Syndrome de Matfan den-i.lu asbl	http://www.den-i.lu/	Associate Member
MALAYSIA		
Malaysia Metabolic Society	http://www.mms.my	Associate Member
Malaysian Rare Disorders Society	http://www.mrds.org.my	Associate Member
MALTA		
National Alliance For Rare Diseases Support - Malta	http://www.rarediseasesmalta.com	Full Member
MEXICO		
Asociacion de Gaucher de Mexico		Associate Member
Proyecto Pide Un Deseo Mexico Iap	http://www.pideundeseo.org	Associate Member

Red Sanfilippo	http://www.redsanfilippo.org	Associate Member
MONTENEGRO		
National Organisation for Rare Diseases	http://www.rijetkebolesti.com	Associate Member
MOROCCO		
Association Marocaine de la Fièvre Méditerranéenne Familiale et des autres Fièvres Récurrentes		Associate Member
Sun'Hop		Associate Member
NEPAL		
GBS/CIDP Foundation Nepal		Associate Member
Muscular Dystrophy Organization Nepal	https://www.facebook.com/MuscularDystrophyOrganizationNepalPanautKavre/	Associate Member
NETHERLANDS		
Acanthamoeba Keratitis Eye Foundation	https://akeyefoundation.com/	Associate Member
ALS Patients Connected	http://www.alspatientsconnected.com	Full Member
Amyloidose Nederland	http://amyloidose.nl	Full Member
Ataxie Vereniging Nederland	http://www.ataxie.nl	Full Member
Bijniervereniging (Dutch Adrenal Patient Society)	http://www.nvacp.nl	Full Member
CMTC-OVM	http://www.cmtc.nl	Full Member
Cornelia de Lange Syndrome World Federation	http://www.cdlsworld.org	Full Member
European Cleft Organisation	http://europeancleft.org/	Associate Member
European Sarcoidosis Foundation	https://sarcoidosis.eu/	Associate Member
European Society for Phenylketonuria	http://www.espku.org	Associate Member
European VHL (Von Hippel-Lindau) Federation	http://www.vhl-europa.org	Associate Member
Fabry Support & Informatie Groep Nederland	http://www.fabry.nl	Full Member
Fibrodysplasia Ossificans Progressiva Stichting Nederland	http://www.fopstichting.nl	Full Member
FSHD Europe	http://fshd-europe.info	Full Member
International Mito Patients	http://www.mitopatients.org	Full Member
International Painful Bladder Foundation	http://www.painful-bladder.org	Associate Member
International Porphyria Patient Network	https://porphyria.network/IPPN/	Associate Member
Interstitiële Cystitis Patientenvereniging	http://www.icpatienten.nl	Full Member
ITP Patientenvereniging	http://www.itp-pv.nl	Full Member
KAISZ - Children with a Autoinmuum or Autoinflammatory Disease	http://www.kaisz.nl	Full Member
LMNA Cardiac Foundation	https://www.lmnacardiac.org/	Associate Member
MSS (Marshall-Smith Syndrome) Research Foundation	http://www.marshallsmith.org	Full Member
Naevus Global	http://www.naevusglobal.org	Associate Member
National Association ReumaZorg Nederland	https://reumazorgnederland.nl/	Associate Member
Nationale Vereniging L.E. patiënten	http://www.nvle.org/	Full Member
Nederlands Netwerk voor Lymfoedeem en Lipoedeem	http://www.lymfoedeem.nl	Associate Member
Nederlandse Hypofyse Stichting (Dutch Pituitary Foundation)	http://www.hypofyse.nl	Full Member
Nederlandse Leverpatienten Vereniging	http://www.leverpatientenvereniging.nl	Full Member
Nederlandse Phenylketonurie Vereniging / Dutch Pku Association	http://Www.pkuvereniging.nl	Full Member
Nederlandse Vereniging van Hemofilie-Patiënten/Netherlands Haemophilia Society	http://www.nvhp.nl	Full Member
Nephceurope	http://nephceurope.eu/nc/	Associate Member
Neurofibromatose Vereniging Nederland	http://www.neurofibromatose.nl	Full Member
Nevus Netwerk Nederland	https://nevusnetwerk.nl/	Full Member
Oscar Nederland	http://www.oscarnederland.nl	Full Member
Patientenorganisatie Fibreuze Dysplasie	http://www.fibreuzedysplasie.eu	Full Member
Patiëntenvereniging voor Blaasextrophie Nederland	https://www.blaasextrophy.nl	Full Member
Sarcoidose.NI	http://www.sarcoidose.nl	Full Member
Spierziekten Nederland - Dutch Association for Neuromuscular Disease	http://www.vsn.nl	Full Member
Stichting AA & PNH Contactgroep	http://www.aaenpnh.nl	Full Member
Stichting Christianson Syndrome Europe	https://cs-europe.org/	Associate Member
Stichting Hart4Onderzoek / Heart4Research	https://hart4onderzoek.nl/	Full Member
Stichting Histiocytose Nederland	http://www.histio.nl	Full Member
Stichting Ijzersterk	http://www.stichtingijzersterk.nl	Associate Member
Stichting Kans voor PKAN Kinderen	https://kansvoorpkankinderen.nl	Full Member
Stichting Net-Groep	http://www.net-kanker.nl	Full Member
Stichting Overdruksyndroom NL	https://sosnl.nl	Full Member
Stichting Pierre Robin Europe - Pierre Robin Europe Foundation	http://pierrerobineurope.com	Full Member
Stichting RPF Nederland / Dutch RPF Foundation	https://rpf.nl/	Full Member
Stichting Shwachman Diamond Syndrome Support Holland	http://www.shwachman.nl	Full Member
Stichting Spierkracht	http://www.stichtingspierkracht.com/	Associate Member
Stichting TAPS Support/ TAPS Support Foundation	https://www.stichtingtapssupport.com/	Associate Member
Stichting Terre - Rett Syndrome Foundation	http://www.stichtingterre.nl	Full Member
Stichting voor Afweerstoornissen	http://www.stichtingvoorafweerstoornissen.nl	Full Member
Stichting Zeldzame Bloedziekten	http://www.bloedziekten.nl	Associate Member
Thyroid Cancer Alliance	http://www.thyroidcanceralliance.org	Full Member
Vasculitis Stichting	http://www.vasculitis.nl	Full Member
Vereniging van Ehlers Danlos Patienten	http://www.ehlers-danlos.nl/	Full Member
Vereniging voor Ichthyosis Netwerken	https://ichthyosisnetwerken.nl/	Full Member
Volwassenen, Kinderen En Stofwisselingsziekten	http://www.stofwisselingsziekten.nl	Full Member
VSOP - Vereniging Samenwerkende Ouder En Patiëntenorganisaties	http://www.vsop.nl	Full Member
World Alliance of Pituitary Organizations	http://www.wapo.org	Associate Member
World Duchenne Organization	http://worldduchenne.org	Full Member
NEW ZEALAND		
Rare Disorders NZ	http://www.nzord.org.nz	Associate Member
NORTH MACEDONIA		
Association for help and support of patients and their caregivers with Haematological Diseases	http://www.hema.org.mk	Associate Member
Association for persons with Spinal Muscular Atrophy- Stop SMA	https://www.facebook.com/STOP-SMA-Macedonia-%D0%A1%D0%A2%D0%9E%D0%9F-%D0%A1%D0%9C%D0%90-1916753535041997/	Associate Member
Life With Challenges	http://challenges.mk/?p=2288&lang=en	Full Member
National Alliance For Rare Diseases of North Macedonia		Full Member

Save Liver Association of Patients with Liver Diseases	https://www.facebook.com/udrugaslap/	Full Member
NORWAY		
Aniridia Europe	http://www.aniridia.eu	Full Member
European Huntington Association	http://eurohuntington.org/	Full Member
Frambu - Resource Centre For Rare Disorders	http://www.frambu.no	Full Member
Hjernesvulstforeningen (Norwegian Brain Tumour Association)	https://hjernesvulst.no/	Full Member
Hypopara Norge	http://www.hypopara.no	Full Member
International Huntington Association	https://huntington-disease.org/	Associate Member
Morbus Addison Association Norway	http://www.addison.no	Full Member
MPS-Foreningen I Norge	http://mpsforeningen.no	Associate Member
Norsk Forening For Arvelig Spastisk Paraparese / Ataksi	https://www.naspa.no/	Full Member
Norsk Forening For Ehlers-Danlos Syndrom	http://www.eds-foreningen.no	Full Member
Norsk Forening for Osteogenesis Imperfecta/The Norwegian Osteogenesis Imperfecta Association	http://www.nfoi.no	Full Member
Norsk Forening For Tuberos Sklerose	http://www.nfts.no	Full Member
Norwegian Federation of Organsiations of Disabled People (Funksjonshemmedes Fellesorganisasjon)	https://www.ffo.no/	Full Member
Norwegian organisation for Prader Willis syndrome	https://www.prader-willis.no/	Full Member
Oslerforeningen Norge	http://www.osler.no	Full Member
POLAND		
Debra Polska	http://www.debra-kd.pl	Full Member
Foundation of Borys the Hero /Fundacja Bohatera Borysa	http://bohaterborys.pl/	Associate Member
Fundacja Sanfilippo	http://sanfilippo.org.pl/en/home/	Associate Member
Fundacja SMA (SMA Foundation Poland)	http://www.fsma.pl	Full Member
Fundacja Umiec Pomagac (Foundation For RD MPS)	http://www.umiecpomagac.org	Full Member
Matio Fundacji Pomocy Rodzinom I Chorym Na Mukowiscydozę/MATIO Polish CF Foundation	http://www.mukowiscydoza.pl	Full Member
Polish National Forum on the treatment of Orphan Diseases - Orphan	http://www.rzadkiechoroby.pl	Full Member
Polish PKU and RD Association "Ars Vivendi"	http://www.fenyloketonuria.org	Full Member
Polish Society of MPS and Related Diseases	http://www.mps-society.pl	Full Member
Polskie Stowarzyszenie Na Rzecz Osób Z AHC	http://www.stow.ahc-pl.org/	Associate Member
Rett Syndrome Poland - Ogólnopolskie Stowarzyszenie Pomocy Osobom Z Zespołem Retta	http://www.rettsyndrome.pl	Full Member
The Dina Radziwillowa Child's Heart Foundation	http://www.sercedziecka.org.pl	Full Member
PORTUGAL		
andLINFA|Associação Nacional de Doentes Linfáticos / National Association of Sufferers of Lymphatic Disorders	https://andlinfa.pt/	Full Member
Associação Nacional De Displasias Ósseas	http://www.andoportugal.org	Associate Member
Associação Nacional para Divulgar e Orientar para Combater e Enfrentar a Tay Sachs e outras Gangliosidoses	http://doce.pt/wp/	Associate Member
Associacao Portuguesa CDG e Outras Doencas Metabolicas	http://www.apcdg.com	Associate Member
Associação Portuguesa De Charcot-Marie-Tooth	https://www.facebook.com/associacao.portuguesa.charcot.marie.tooth/	Full Member
Associação Portuguesa De Insuficientes Renais	http://www.apir.org.pt	Full Member
Associaçao Portuguesa de Leucemias e Linfomas	http://www.apll.org	Full Member
Associaçao Portuguesa De Neuromusculares	http://www.apn.pt	Full Member
Associação Portuguesa de Osteogénese Imperfeita	http://www.apoi.pt	Full Member
Associação Sanfilippo Portugal	http://www.sanfilippoportugal.com	Full Member
EVITA	http://www.evitacancro.org/	Associate Member
Fedra - Federaçao Portuguesa De Doenças Raras	http://www.fedra.pt	Associate Member
Liga Portuguesa contra as Doenças Reumáticas	http://www.lpcdr.org.pt	Full Member
Rarissimas - Associaçao Nacional De Deficiencias Mentais E Raras	https://rarissimas.pt/	Full Member
RD-Portugal - União de Associações das Doenças Raras de Portugal	https://raras.pt/	Associate Member
REPUBLIC OF MOLDOVA		
Copiii Ploii	https://www.facebook.com/AO-Copiii-Ploii-686485228086936/	Full Member
ROMANIA		
Asociatia Copiilor Cu Boli Mitocondriale/Association of Children with Mitochondrial Diseases	https://cure-echs1.com/	Associate Member
Asociatia Copilul Meu-Inima Mea	http://www.acmim.ro	Full Member
Asociatia Nationala Miastenia Gravis Romania /Romanian Myasthenia Gravis Association	http://www.miastenie.ro	Full Member
Asociatia Persoanelor Cu Glicogenoza (APG Romania)	https://glicogenoza.ro/	Associate Member
Asociatia Persoanelor Cu Talasemie Majora	http://www.talasemia.ro	Full Member
Asociația Română De Cancere Rar	http://www.arcrareromania.ro	Associate Member
Asociatia Romana pentru Boli Neurologice Periferice/Romanian Association of Perypheral Neurological Diseases	https://arbnp-cidp.ro/	Full Member
Asociatia Romana Spina Bifida si Hidrocefalie	https://www.facebook.com/ARSBH/	Associate Member
Asociatia Sindromul Coffin-Lowry	http://www.coffin-lowry.ro/	Associate Member
Asociatia Smacare	http://www.amiotrofie-spinala.ro	Full Member
Asociatia Werdnig Hoffman Awh	https://www.facebook.com/werdnighoffman/	Full Member
Autoimmune Diseases Patients Association	http://www.apaa.ro	Associate Member
Charcot Marie Tooth Romania Association	http://www.asociatiacmt.ro	Associate Member
DMD Care	https://dmdcare.org	Full Member
Mastocytosis Support Association Romania	http://www.mastocitoza.ro	Associate Member
Neuro Move CMT Association	https://neuromovecmt.ro/	Associate Member
Romanian National Alliance For Rare Diseases	http://bolirareromania.ro	Full Member
Romanian Prader Willi Association	http://www.apwromania.ro	Full Member
RUSSIAN FEDERATION		
Help to Cystic Fibrosis Patients		Full Member
Interregional Public Organisation For Gaucher Disease	http://www.gaucher.ru	Full Member
Interregional Public Organisation Of Patients With Fabry Disease "The Road To Life"	http://www.fabryufa.ru	Full Member
Inter-Regional Support Centre dor Patients with Aniridia " Iris"	http://www.aniridia.ru	Associate Member
Look to see	http://looktosee.ru	Full Member
MPS Russia	http://www.mps-russia.org	Full Member
National Association of Organization of Patient with Rare Diseases "Genetics"	http://www.nacgenetic.ru/	Associate Member
NGO "Fragile Children"	http://www.osteogenez.ru	Full Member

Russian Association of Rare Diseases	http://www.rare-diseases.ru	Full Member
Russian Patient Association	http://www.patients.ru/en	Associate Member
Russian Rett Syndrome Asociation	http://www.rettsyndrome.ru	Associate Member
SPIPORZ Union of patients with rare diseases and rare disease patients organsiations	http://www.spiporz.ru	Associate Member
The Association of Primary Immunodeficiency Patients	http://www.oppid.ru	Full Member

SERBIA

Child Rare Disease Support and Research Association Life	http://www.zivotorg.org	Associate Member
Citizens Association "Bromologos"	https://fenilketonurija.com/	Associate Member
DMD Serbia	http://dmdsrbija.rs/	Full Member
Lymphoma Patients' Association	http://www.lipa.org.rs	Associate Member
National Organization For Rare Diseases of Serbia	http://www.norbs.rs	Full Member

SINGAPORE

Rare Disorders Society (Singapore)	http://www.rdss.org.sg	Associate Member

SLOVAKIA

Debra SR	http://www.debra-slovakia.org	Full Member
Organisation of Muscular Dystrophy in the Slovak Republic/Organizácia muskulárnych dystrofikov v SR	http://www.omdvsr.sk	Full Member
Slovak Alliance of Rare Diseases	http://www.sazch.sk	Full Member
Slovak Cystic Fibrosis Association	http://www.cfasociacia.sk	Full Member
Zdruzenie Ojedlinelych Genetickych Ochorenie	http://www.zogo.sk/	Associate Member

SLOVENIA

Association of Patients with Blood Diseases - Drustvo Bolnikv S Krvnimi Boleznimi	http://www.drustvo-bkb.si	Full Member
Debra Slovenia - Drustvo Debra Slovenija	http://www.debra-slovenia.si	Full Member
EAMDA - European Alliance of Neuromuscular Disorders Associations	http://www.eamda.eu	Full Member
European Foundation for SATB2-Associated Syndrome (SATB2 Europe)	https://www.satb2europe.org/	Associate Member
Fabry Patients Association Slovenia / Društvo Bolnikov S Fabryjevo Boleznijo Slovenije	http://www.sb-sg.si	Full Member
Foundation of Child Neurology	http://pednevro.pedkl.si/english/foundation/	Associate Member
IDefine Europe - Foundation for the Advanced Treatment of Rare Genetic Diseases	https://redkegenetskebolezni.ijs.si/	Associate Member
Viljem Julijan Association for Children with Rare Diseases	http://viljem-julijan.si/	Full Member
Zavod Bernardi Ventrella	https://www.zavodbv.org/	Associate Member

SOUTH AFRICA

Primary Immunodeficiency Network of South Africa	http://www.pinsa.org.za	Associate Member
Rare Diseases South Africa NPC	http://www.rarediseases.co.za	Associate Member

SPAIN

Acción y Cura Para Tay-Sachs	http://www.actays.org	Full Member
AHUCE - Asociación Nacional Huesos de Cristal	http://www.ahuce.org	Full Member
Alianza Española de Familias de von Hippel Lindau	http://www.alianzavhl.org	Full Member
Asociació Catalana de las Neurofibromatosis	http://www.acnefi.com/castella/gamhome.htm	Full Member
Asociacion Afectados Cdkl5	https://aacdkl5.org	Full Member
Asociacion Albi España	http://www.asociacionalbi.com/	Full Member
Asociacion Andaluza de Fibrosis Quística	http://fqandalucia.org/	Full Member
Asociación Andaluza de pacientes con Síndrome de Tourette y Trastornos Asociados	http://www.tourette.es	Full Member
Asociación Artrogriposis Múltiple Congénita	https://artrogriposis.org/	Full Member
Asociación Chiari y Siringomielia del Principado de Asturias	http://www.chyspa.org	Full Member
Asociación Ciudadana de Afectados de Cistitis Intersticial	http://www.acaci.es/	Full Member
Asociación de Afectados de Neurofibromatosis	http://www.neurofibromatosis.es	Full Member
Asociación de Afectados por Displasia Ectodérmica	http://www.displasiaectodermica.org	Full Member
Asociación de Afectados por Hiperinsulinismo Congénito	http://hiperinsulinismocongenito.org/	Full Member
Asociación de Atrofia de Nervio Optico de Leber	http://www.asanol.com	Full Member
Asociacion De Enfermedades Raras D'genes	http://www.dgenes.es/	Full Member
Asociación de Epidermolisis Bullosa de España (Debra Spain)	http://www.pieldemariposa.es	Full Member
Asociación de Esclerodermia Castellon	https://www.esclerodermia.es/	Full Member
Asociación de Familiares y Afectados por Lipodistrofias	http://www.aelip.org	Full Member
Asociación De Hemoglobinuria Paroxística Nocturna	http://www.hpne.org	Associate Member
Asociación De Nevus Gigante Congénito	http://asonevus.org/	Full Member
Asociación de pacientes ASMD España	https://www.asmd.es/	Full Member
Asociación de Pacientes de Uveítis	https://www.asociacionauvea.es/	Full Member
Asociación Enfermedad de Kawasaki	https://asenkawa.org/	Full Member
Asociación Española Aniridia	http://www.aniridia.es	Full Member
Asociación Española de Afectados por Sarcoma	http://www.aeasarcomas.org	Full Member
Asociación Española de Amiloidosis	http://www.amilo.es/	Associate Member
Asociación Española de Angioedema Familiar	http://www.angioedema-aedaf.org	Full Member
Asociación Española de Enfermos de Glucogenosis	http://www.glucogenosis.org	Full Member
Asociación Española de Enfermos de Pompe	http://www.asociaciondepompe.org	Associate Member
Asociación Española de Enfermos y Familiares de la Enfermedad De Gaucher España	http://www.aeefegaucher.es	Full Member
Asociación Española de Esclerodermia	https://esclerodermia.com	Full Member
Asociación Española de Familiares y Enfermos de Wilson	http://www.enfermedaddewilson.org	Full Member
Asociación Española de Fibrodisplasia Osificante Progresiva	http://www.aefop-es.org	Full Member
Asociación Española De Fiebre Mediterranea Familiar	http://fmf.org.es	Full Member
Asociación Española de Ictiosis	http://www.ictiosis.org	Full Member
Asociación Española de Mastocitosis y Enfermedades Relacionadas	http://www.mastocitosis.com	Full Member
Asociación Española de Pacs1	http://pacs1.es/	Associate Member
Asociación Española de paraparesia espástica familiar Strümpell-Lorrain	http://www.aepef.org	Full Member
Asociación Española de Porfiria	http://www.porfiria.org	Full Member
Asociación Española de Raquitismos y Osteomalacia Heredados	https://aeryoh.org/	Associate Member
Asociación Española de Síndrome De Poland	http://www.aesip.es	Full Member
Asociación Espanola deficit de Lipasa Acida Lisosomal	http://www.aelald.org	Full Member
Asociación Española del sindrome CDG	http://aescdg.com/	Associate Member
Asociación Española del Síndrome de Schaaf-Yang	https://www.aesys.org/	Full Member
Asociación Española Sindrome de Sjögren	http://www.aesjogren.org	Full Member
Asociación HHT España	http://www.asociacionhht.org	Full Member
Asociación KIF1A España	https://kif1a.es/	Full Member
Asociación Madrileña de Pacientes con Sindrome de Gille de la Tourette y Trastornos Asociados	https://www.ampastta.com/	Full Member
Asociación Nacional Amigos De Arnold Chiari	http://www.arnoldchiari.es/	Full Member

Asociacion Nacional de Dermatomiositis Juvenil	https://anadeju.org/	Full Member
Asociación Nacional de Hipertensión Pulmonar	http://www.hipertensionpulmonar.es	Full Member
Asociación Nacional Síndrome de Apert y otras Craneosinostosis Sindrómicas	http://www.ansapert.org	Full Member
Asociación Retina Murcia	https://www.retimur.org/	Full Member
Asociación Síndrome de Angelman	http://www.angelman-asa.org	Full Member
Asociación Síndrome de Lowe de España	http://www.sindromelowe.es	Full Member
Asociación Síndrome Lesch Nyhan España	https://www.facebook.com/asociacion.sindromeleschnyhanespana	Full Member
Asociación Stop Sanfilippo	http://www.stopsanfilippo.org	Associate Member
Asociación Xeroderma Pigmentosum	https://xerodermapigmentosum.es/	Full Member
Associacio Catalana de la Deleccio 22Q	http://www.22q.cat	Full Member
Associació d'Apràxia Ocular i Malalties Associades	http://apraxiaocular.blogspot.com	Associate Member
Asociación Catalana De Enfermedades Neuromusculares	http://www.asemcatalunya.com	Full Member
De Neu - Asociación De Enfermedades De Los Neurotransmisores	http://www.deneu.org	Full Member
Duchenne Parent Project España	http://www.duchenne-spain.org	Full Member
European Network For Rare And Congenital Anaemias	http://www.enerca.org	Associate Member
FEDER - Federación Española De Enfermedades Raras	http://www.enfermedades-raras.org	Full Member
Federació Catalana de Malalties Minoritàries	https://www.fecamm.org/portal1/m_index.asp?idioma=1	Full Member
Federación de Asociaciones de Retinosis Pigmentaria de España	http://www.retinosisfarpe.org	Full Member
Federación de Ataxias de España	http://www.fedaes.org	Full Member
Federación Española de Enfermedades Neuromusculares	http://www.asem-esp.org	Full Member
Federación Española De Fibrosis Quistíca	http://www.fibrosis.org/	Full Member
Federación Española De Hemofilia	http://www.hemofilia.com	Full Member
Federacion Espanola de Padres con Ninos con Cancer	http://www.cancerinfantil.org	Full Member
Federación Española del Síndrome X Frágil	http://www.xfragil.org	Full Member
Fundació Catalana d'ELA Miquel Valls (Miquel Valls Foundation)	http://www.elacat.org	Associate Member
Fundación ALPE Acondroplasia	https://www.fundacionalpe.org	Full Member
Fundación Andrés Marcio, Niños Contra La Laminopatía	http://www.fundacionandresmarcio.org	Full Member
Fundación Contra La Hipertensión Pulmonar	https://www.fchp.es/	Associate Member
Fundacion Libellas	https://fundacionlibellas.org	Associate Member
Fundación Mari Paz Jiménez Casado	https://www.fundacionmaripazjimenez.org	Full Member
Fundación Niemann-Pick de España	http://www.fnp.es	Full Member
Fundacion Noelia, Collagen VI Deficiency Muscular Dystrophy	https://fundacionnoelia.org/	Full Member
Fundación Síndrome 5P Menos	http://www.fundacionsindrome5p.org	Full Member
Fundación Síndrome de Dravet - Dravet Syndrome Foundation	http://www.dravetfoundation.eu	Full Member
Fundacion Síndrome De West	http://www.sindromedewest.org	Full Member
Fundación Síndrome Wolf Hirschhorn 4P-	http://www.4pmenos.org	Full Member
Hipertension Pulmonar España Organizacion De Pacientes	http://www.hipertension-pulmonar.com	Associate Member
Instituto de Investigación y Desarrollo Social de Enfermedades Poco Frecuentes	http://www.pocofrecuentes.org	Associate Member
Menkes International Association	https://menkesinternational.com/	Associate Member
MPS Lisosomales	http://www.mpsesp.org/	Full Member
Pequeños Superheroes	https://pequenossuperheroes.org/	Associate Member
SAF España	https://www.antifosfolipido.es/	Full Member
SAMS - Asociacion para la lucha contra los Sindromes Arrítmicos Relacionados con la Muerte Súbita	http://samsasociacion.com/	Full Member
Sense Barreres de Petrer	https://sensebarreres.es/	Associate Member
SIMA Asociación de afectados Síndrome de Marfan	http://www.marfan.es/	Full Member

SWEDEN

Acción y Cura Para Tay-Sachs	http://www.actays.org	Full Member
Agrenska	http://www.agrenska.se	Full Member
Aorta Dissektion Föreningen Skandinavien	http://www.aortadissektion.com	Full Member
DysNet	https://www.dysnet.org/	Full Member
European Cavernoma Alliance	https://sites.google.com/cavernostangiomsverige.org/eca/home	Associate Member
Neuroförbundet / Swedish Association Of Persons With Neurological Disabilities	http://neuroforbundet.se	Associate Member
Prader Willi Syndrome Association In Sweden	http://www.prader-willi.se	Full Member
Primär Immunbrist Organisationen	http://www.pio.nu	Full Member
Rare Diseases Sweden (Riksförbundet Sällsynta Diagnoser)	http://www.sallsyntadiagnoser.se	Full Member
Svensk förening för Hypoparatyreoidism	https://www.hypopara.se/	Associate Member
Svenska Marfanföreningen - Swedish Marfan Assciation	http://www.marfanforeningen.se/	Full Member
Svenska Ödemförbundet (Swedish Assocation of Chronic Oedema)	https://www.svenskaodemforbundet.se/	Associate Member
Swedish Cystic Fibrosis Association	http://www.rfcf.se	Full Member
Swedish EDS Association (EDS Riksförbund)	http://www.ehlers-danlos.se	Full Member
Swedish MPS Society	http://www.mpsforeningen.se	Full Member
Thyroid Federation International	http://www.thyroid-fed.org	Associate Member
Wilhelm Foundation	http://www.wilhelmfoundation.org	Full Member

SWITZERLAND

AGO2 Association	https://ago2.org/en	Associate Member
Association Enfance et Maladies Orphelines	http://www.aemo.ch	Associate Member
Association Suisse Romande Intervenant contre les maladies Neuro-Musculaires	http://www.asrimm.ch	Full Member
Blackswan Foundation	http://www.blackswanfoundation.ch	Associate Member
CML Advocates Network	http://www.cmladvocates.net	Associate Member
Esperare	http://www.esperare.org	Associate Member
FMF & AID Global Association	http://www.fmfandaid.org	Associate Member
Fondation Sanfilippo Suisse	https://www.fondation-sanfilippo.ch/	Associate Member
Foundation Orphanhealthcare	http://www.orphanhealthcare.org/	Associate Member
Fraxas - Association X Fragile Suisse	http://www.fraxas.ch	Full Member
HAEi - Hereditary Angioedema International Association	http://www.haei.org	Associate Member
HHT-Swiss	http://www.hhtswiss.org	Associate Member
MaRaVal – maladies rares valais – seltene krankheiten wallis.	http://www.maraval.ch/	Full Member
Marfan Foundation Switzerland (Marfan Stiftung Schweiz)	http://www.marfan.ch	Full Member
Prader Willi Syndrom Vereinigung Schweiz	http://www.prader-willi.ch	Full Member
Proraris	http://www.proraris.ch	Full Member
Save sight now Europe	https://www.savesightnoweurope.org/	Associate Member
Schweizerische Gesellschaft Für Porphyrie	http://www.porphyria.ch	Full Member
Sjögren Europe	http://sjogreneurope.org/	Associate Member
SMA Schweiz	http://www.sma-schweiz.ch	Associate Member

Swiss FH/Schweizerische Gesellschaft für familiäre Formen der Hypercholesterinämie	http://www.sgfh.ch	Associate Member
TAIWAN, PROVINCE OF CHINA		
Taiwan Foundation For Rare Disorders	http://www.tfrd.org.tw	Associate Member
TURKEY		
DMD Aileleri Dernegi	https://www.dmdaileleri.org/	Full Member
Duchenne Kas Hastalığı Ile Mucadele Dernegi	http://dmdturkiye.org/	Full Member
Kifder	http://www.kifder.org.tr	Associate Member
Mukopolisakkaridoz ve Benzeri Lizozomal Depo Hastaliklari Dernergi / MPS Turkey	http://www.mpsturk.org	Associate Member
Pulmoner Hipertansiyon ve Skleroderma Hasta Derneği / PHA Turkey	http://www.pahssc.org.tr	Full Member
Sistinozis Hastaları Dernegi / Cıstınosıs Patıents Assocıatıon	https://www.sistinder.org/	Associate Member
SMA Hastalıgı Ile Mücadele Dernegi	http://www.sma.org.tr	Full Member
Yuzumle Mutluyum Dernegi/Happy Faces	http://www.yuzumlemutluyum.org.tr	Associate Member
UKRAINE		
Association of Patients with Pulmonary Hypertension	http://www.pha.org.ua	Full Member
Children with Spinal Muscular Atrophy, Charitable Foundation	http://www.csma.org.ua	Full Member
LCCF "Sister Dalila" Pulmonary Hypertension Ukrainian Rare Disease Association	http://poryatunok.info/uk/	Full Member
NGO "Rare Diseases of Ukraine"	https://www.facebook.com/orphandisua/	Associate Member
Ukrainian Association Crystal People	http://ostimperfecta.wix.com/oiua	Associate Member
Ukrainian Association of Help for Patients with CF	http://facebook.com/cysticfibrosisua	Full Member
Ukrainian National Charitable Fund Zaporuka		Associate Member
Ukrainian Union Of Patients' Organisations	https://patients.org.ua/en/	Associate Member
Ukranian Parent Project "Mio-Life"	http://miolife.org	Associate Member
UNITED KINGDOM OF GREAT BRITAIN AND NORTHERN IRELAND		
Acrodysostosis Support and Research	http://www.acrodysostosis.com	Associate Member
Action Duchenne	http://www.actionduchenne.org	Full Member
Advocacy For Neuroacanthocytosis Patients	http://www.naadvocacy.org	Associate Member
AKU Society	http://www.akusociety.org	Full Member
Alex The Leukodystrophy Charity	https://www.alextlc.org/	Associate Member
Alstrom Syndrome Europe	https://www.alstrom.org.uk/as-europe/	Associate Member
Alstrom Syndrome UK	http://www.alstrom.org.uk	Full Member
Ann Edgar Charitable Trust - Neuroendocrine Tumour Support	http://www.taect.scot	Full Member
Annabelle's Challenge	http://www.annabelleschallenge.org	Full Member
Association for Glycogen Storage Disease	http://www.agsd.org.uk	Full Member
Association for Multiple Endocrine Neoplasia Disorders	http://www.amend.org.uk	Full Member
Ataxia UK	http://www.ataxia.org.uk	Full Member
Batten Disease Family Association	http://www.bdfa-uk.org.uk/about-batten-disease	Full Member
Beacon!	https://www.rarebeacon.org/	Associate Member
Behcet's UK	http://www.behcets.org.uk	Full Member
Brittle Bone Society	http://www.brittlebone.org	Associate Member
Cambridge Rare Disease Network	http://www.camraredisease.com	Associate Member
Cancer 52	http://www.cancer52.org.uk	Associate Member
Cavernoma Alliance UK	http://www.cavernoma.org.uk	Full Member
CDH International	https://cdhi.org/	Associate Member
CDH Uk	http://www.cdhuk.org.uk/	Full Member
Child Growth Foundation	http://www.childgrowthfoundation.org	Full Member
Child Lung Foundation	http://www.childlungfoundation.org	Associate Member
Childhood Tumor Trust	https://www.childhoodtumourtrust.org.uk/	Full Member
Children's Liver Disease Foundation	http://www.childliverdisease.org	Associate Member
Ciliopathy Alliance	http://www.ciliopathyalliance.org	Full Member
Contact A Family	http://www.cafamily.org.uk	Full Member
Cure CLCN4	https://cureclcn4.org/	Associate Member
Cystinosis Foundation of the UK	http://www.cystinosis.org.uk	Full Member
Dancing Eye Syndrome Support Trust	http://www.dancingeyes.org.uk	Full Member
Ectodermal Dysplasia Society	https://edsociety.co.uk/	Full Member
European Lung Foundation	http://www.european-lung-foundation.org/	Associate Member
European Tuberous Sclerosis Complex Association	http://www.e-tsc.eu	Full Member
Eyes on the Future	https://eyesonthefuture.org.uk/	Associate Member
Fabry International Network	http://www.fabrynetwork.org/	Associate Member
Fetal Anti Convulsant Syndrome Assocation		Associate Member
FH Europe	https://www.fheurope.org/	Associate Member
FOP Friends	http://www.fopfriends.com	Full Member
Gauchers Association UK	http://www.gaucher.org.uk	Full Member
Genetic Alliance UK	http://www.geneticalliance.org.uk	Full Member
Glut1 Deficieny UK	http://www.glut1deficiency.org.uk	Associate Member
HBA Support	https://www.hbasupport.org/	Associate Member
Hope for Hasti	https://www.hopeforhasti.org/	Associate Member
Hope for Hypothalamic Hamartomas UK	http://www.hopeforhh.org	Full Member
Huntington's Disease Youth Organisation	https://en.hdyo.org/	Full Member
International Brain Tumour Alliance	http://www.theibta.org	Associate Member
International Gaucher Alliance	https://gaucheralliance.org/	Full Member
International Niemann-Pick Disease Alliance	http://www.inpda.org	Full Member
International Patient Organization for Primary Immunodeficiencies	https://ipopi.org/	Full Member
International Prader-Willi Syndrome Organisation	http://www.ipwso.org	Full Member
Joining Jack	http://www.joiningjack.org	Full Member
Krabbe UK	https://www.krabbeuk.org/	Full Member
Leber's Hereditary Optic Neuropathy Society	http://www.lhonsociety.org	Full Member
LGD Alliance Europe	http://www.lgda.eu	Full Member
Max Appeal	http://www.maxappeal.org.uk	Full Member
Mebo Research	http://www.meboresearch.org	Associate Member
Metabolic Support UK	http://www.metabolicsupportuk.org	Full Member
MPS Society	https://www.mpssociety.org.uk/	Full Member
Myotubular Trust	http://www.myotubulartrust.org	Full Member
Niemann-Pick UK	http://www.npuk.org	Full Member
Northern Ireland Rare Disease Partnership	http://www.nirdp.org.uk	Full Member
Organisation For Anti-Convulsant Syndrome	http://www.oacscharity.org	Full Member
PIP-UK Poland Syndrome Support	https://pip-uk.org/	Full Member

Pitt Hopkins UK	http://pitthopkins.org.uk/	Full Member
Polycystic Kidney Disease Charity	https://pkdcharity.org.uk	Full Member
Pompe Support Network	https://pompe.uk/	Full Member
PSC Support	http://www.pscsupport.org.uk	Full Member
Pseudomyxoma Survivor	http://www.pseudomyxomasurvivor.co.uk	Associate Member
Pten Research Foundation	http://www.ptenresearch.org	Associate Member
Rare Autoinflammatory Conditions Community- UK	http://www.raccuk.com	Associate Member
Rare Disease UK	http://www.raredisease.org.uk	Associate Member
Reverse Rett UK	http://www.reverserett.org.uk/	Full Member
Ring 20 Research and Support UK CIO	http://ring20researchsupport.co.uk/	Associate Member
Salivary Gland Cancer UK	https://www.salivaryglandcancer.uk/	Associate Member
Schinzel-Giedion Syndrome Foundation	https://sgsfoundation.org/	Associate Member
Smile with Shiv	http://smilewithshiv.org/	Associate Member
Stiff Person Support Group	http://www.smssupportgroup.co.uk	Full Member
Sturge-Weber UK	http://www.sturgeweber.org.uk	Full Member
The AADC Research Trust Children's Charity	http://www.aadcresearch.org	Full Member
The Aarskog Foundation	http://www.aarskogsyndromefoundation.co.uk	Associate Member
The Chromosome 18 Registry And Research Society (Europe)	http://www.chromosome18eur.org	Associate Member
The Cure & Action For Tay-Sachs (Cats) Foundation	http://www.cats-foundation.org	Full Member
The EHE Rare Cancer Charity	https://www.ehercc.org.uk/	Full Member
The Ehlers-Danlos Society	http://ehlers-danlos.com/	Full Member
The Fragile X Society	http://www.fragilex.org.uk/	Full Member
The Maddi Foundation	http://themaddifoundation.com/	Associate Member
The PBC Foundation (UK) Ltd	http://www.pbcfoundation.org.uk	Associate Member
The Ultra Rare Diseases, Disorders & Disabilities Foundation	http://www.ultrarare-disease.uk	Associate Member
Timothy Syndrome Alliance	https://timothysyndrome.org.uk/	Full Member
Tuberous Sclerosis Association	http://www.tuberous-sclerosis.org	Full Member
UK ATTR Amyloidosis Patients' Association	https://ttramyloidosis.uk/	Associate Member
UK Mastocytosis Support Group	http://www.ukmasto.org	Full Member
Unique - Rare Chromosome Disorder Support Group	http://www.rarechromo.org	Full Member
United Kingdom Thalassaemia Society	http://www.ukts.org	Full Member
Vasculitis UK (The Vasculitis Trust)	http://www.vasculitis.org.uk	Full Member
VHL UK/Ireland	https://vhl-uk-ireland.org	Full Member

UNITED STATES OF AMERICA

Alagille Syndrome Alliance	http://www.alagille.org	Associate Member
Alstrom Syndrome International	http://www.alstrom.org	Associate Member
APS Foundation of America, Inc	http://www.apsfa.org	Associate Member
Association for Creatine Deficiencies	https://creatineinfo.org/	Associate Member
BCM Families Foundation	http://www.blueconemonochromacy.org/fr/	Associate Member
CACNA1A Foundation	https://www.cacna1a.org/	Associate Member
Chordoma Foundation	http://www.chordoma.org	Associate Member
Cure Mucolipidosis	https://www.curemucolipidosis.org/	Associate Member
Cure PSP	http://www.curepsp.org	Associate Member
Defeat MSA Alliance	https://defeatmsa.org	Associate Member
FMD Chat	http://fmdchat.blogspot.fr/	Associate Member
GBS/CIDP Foundation International	https://www.gbs-cidp.org/	Associate Member
Gould Syndrome Foundation	http://www.gouldsyndromefoundation.org/	Associate Member
International Foxg1 Foundation	http://foxg1.org	Associate Member
International Pemphigus & Pemphigoid Foundation	http://www.pemphigus.org	Associate Member
International Sacral Agenesis/Caudal Regression Association	http://isacra.org/	Associate Member
International WAGR Syndrome Association	http://wagr.org/	Associate Member
International Waldenstrom's Macroglobulinemia Foundation	http://www.iwmf.com	Associate Member
Malan Syndrome Foundation	https://www.malansyndrome.org/	Associate Member
MCT8-AHDS Foundation Inc.	http://www.mct8.info	Associate Member
Myhre Syndrome Foundation	https://www.myhresyndrome.org/	Associate Member
Neuromuscular Disease Foundation	https://curegnem.org/	Associate Member
Nord National Organization for Rare Disorders	http://www.rarediseases.org	Associate Member
NTM Info & Research	http://www.ntminfo.org	Associate Member
Project 8p Foundation	https://project8p.org/	Associate Member
PTEN Hamartoma Tumor Syndrome Foundation	http://www.ptenfoundation.org	Associate Member
Pura Syndrome Foundation	http://www.purasyndrome.org	Associate Member
Remember The Girls	https://rememberthegirls.org/	Associate Member
SATB2 Gene Foundation	https://satb2gene.org/	Associate Member
Sisters' Hope Foundation	https://sistershopefoundation.com/	Associate Member
STXBP1 Foundation	https://www.stxbp1disorders.org/	Associate Member
Superficial Siderosis Research Alliance	https://ssra.livingwithss.com/	Associate Member
SYNGAP1 Foundation	http://www.bridgesyngap.org	Associate Member
The Cushing Support & Research Foundation	http://www.csrf.net	Associate Member
The Cute Syndrome Foundation	https://www.thecutesyndrome.com/	Associate Member
The Oxalosis & Hyperoxaluria Foundation	http://www.ohf.org	Associate Member
The Snyder-Robinson Foundation	https://snyder-robinson.org/	Associate Member
Usher Syndrome Coalition	http://www.usher-syndrome.org	Associate Member

URUGUAY

Asociacion Acondroplasia Uruguay	https://www.facebook.com/acondroplasia.uruguay/	Associate Member
Fundacion Uruguaya para la Investigacion de las Enfermedades Raras	http://www.fupier.org/	Associate Member

VENEZUELA, BOLIVARIAN REPUBLIC OF

Fundación Fura	http://fundacion-fura.mozello.com	Associate Member

ZIMBABWE

Child & Youth Care, Zimbabwe	http://www.cyc.org.zw/	Associate Member

PARTICIPATION OF EURORDIS' REPRESENTATIVES IN PUBLIC EUROPEAN / INTERNATIONAL

CONFERENCES & WORKSHOPS

2022

ROUND TABLE "TOWARDS A EUROPEAN HEALTH SYSTEM", 12 JANUARY

Francois Houÿez represented EURORDIS

10TH INTERNATIONAL CONFERENCE ON RARE AND UNDIAGNOSED DISEASES (UDNI), 31 JANUARY

Gulcin Gumus: Patient Engagement Plus

OD EXPERT GROUP WORKSHOP - A PROCESS FOR UNMET NEEDS, 1 FEBRUARY

Maria Cavaller represented EURORDIS

VIRTUAL HIGH-LEVEL EUROPEAN CONFERENCE: CITIZENSHIP, ETHICS AND HEALTH DATA, 2 FEBRUARY

Yann Le Cam contributed to the discussion

BEACON (FINDACURE) INTERNATIONAL RARE DISEASE SHOWCASE, 2 FEBRUARY

Edith Gross: Harnessing the power of technology to drive diagnosis and rare disease understanding

ASSISES DE GÉNÉTIQUE HUMAINE ET MÉDICALE, 2 FEBRUARY

Virginie Bros-Facer: Patients, Famille, Chercheurs, Médecins: Travailler ensemble grâce aux réseaux & projets Européens

A CRUCIAL TIME FOR THE EUROPEAN RARE CANCER POLICY LANDSCAPE: EUROPE'S BEATING CANCER PLAN AND CANCER MISSION, 8 FEBRUARY

Ariane Weinman: Europe's Beating Cancer Plan and its implementation roadmap: what will this change for rare cancers?

ONE SUSTAINABLE HEALTH FORUM, BUILDING EUROPEAN HEALTH UNION, 8 FEBRUARY

Yann Le Cam contributed to the discussion

INNOVATION IN RARE DISEASE: HOW CAN EUROPE BE A GLOBAL LEADER? 15 FEBRUARY

Yann Le Cam contributed to the discussion

CARE 2022, CENTRES DE RÉFÉRENCE ET DE COMPÉTENCE MALADIES RARES : UNE EXCELLENCE FRANÇAISE À DÉFENDRE, 15 FEBRUARY

Yann Le Cam contributed to the discussion

SCIENTIFIC SYMPOSIUM ON RARE DISEASES, 28 FEBRUARY

Yann Le Cam contributed to the discussion

HIGH-LEVEL CONFERENCE: CARE AND INNOVATION PATHWAYS FOR A EUROPEAN RARE DISEASES POLICY, 28 FEBRUARY

Avril Daly and Yann Le Cam contributed to the discussion

C4C (VIRTUAL) MULTI-STAKEHOLDER MEETING ON ATOPIC DERMATITIS IN CHILDREN AND ADOLESCENTS, 1 MARCH

Maria Cavaller contributed to the discussion

2ND INTERNATIONAL RARE DISEASES CONFERENCE 2022, 1 MARCH

Yann Le Cam, Anna Kole, Simona Bellagambi, and Elizabeth Vroom contributed to the discussion

COOPERATION OF HTA AGENCIES IN EUROPE: FROM SHARING EXPERIENCE TO CARRYING OUT JOINT ASSESSMENTS, 7 MARCH

Francois Houÿez contributed to the discussion

CENTRE FOR INNOVATION IN REGULATORY SCIENCE VIRTUAL WORKSHOP (CIRS), 10 MARCH

Francois Houÿez: What is the value of RWE from the patient perspective and how should this be factored into decision making?

EXPLORING POTENTIAL FOR COLLABORATION BETWEEN EUROPEAN REFERENCE NETWORKS AND THE PHARMACEUTICAL INDUSTRY, 15 MARCH

Yann Le Cam contributed to the discussion

POLITICO LIVE "THE SHAKE-UP OF ORPHAN AND PEDIATRIC RULES", 17 MARCH

Virginie Hivert contributed to the discussion

DIA EUROPE 2022, 29-31 MARCH

Yann Le Cam: How to Ensure that the Patient Voice is Systematically Incorporated Throughout Drug Development, Associated Evidence Generation and Decision-Making?

François Houÿez: Addressing the Medicine Shortages: A Multistakeholder Overview

WEBINARS OF THE EUROPEAN COMMISSION'S HPP NETWORK "SUPPORTING UKRAINE, NEIGHBOURING EU MEMBER STATES AND MOLDOVA", 3 APRIL

Anastasiia Saliuk: co-chair of the Network; a series of presentations on the needs of Ukrainians living with a rare disease

RARE CONVERSATION, 6 APRIL

François Houÿez: Game-changing opportunities for the R&D community to address existing challenges in rare diseases

CHALLENGES OF HEALTH DATA IN EUROPE – ARE WE PREPARING? BUILDING TRUST - ENABLING SCIENCE, 6 APRIL

Edith Gross represented EURORDIS

C4C WEBINAR FOR PATIENTS, 28 APRIL

Maria Cavaller contributed to the discussion

UNDIAGNOSED DAY, 29 AVRIL

Gulcin Gumus: EURORDIS' involvement in Newborn Screening

ISPOR CONFERENCE, 18 MAY

François Houÿez: HTA in a Public Crisis: Communication Challenges

EAPM EXPERT PANEL SECURING EUROPEAN CITIZENS´ TRUST IN THE EUROPEAN HEALTH DATA SPACE AS AN AID TO HEALTHCARE, 7 JUNE

Edith Gross: Patient Engagement and Data Sharing

3RD ANNUAL CONGRESS OF THE FRENCH ALLIANCE OF RARE DISEASES, 10 JUNE

François Houÿez: Gene therapy: what opportunities for patients and their organisations?

EUROPEAN SOCIETY FOR HUMAN GENETICS, 10-12 JUNE

Edith Gross and Gulcin Gumus represented EURORDIS

DIGITALEUROPE SUMMER SUMMIT, 20 JUNE

Yann Le Cam represented EURORDIS

EUROPEAN CONFERENCE ON HTA, EUROPEAN COMMISSION, 22 JUNE

François Houÿez: Patients and healthcare professionals as external experts in HTA

2022 HTAI ANNUAL MEETING, 27 JUNE

François Houÿez contributed to the discussion

ESTRATEGIA FARMACÉUTICA EUROPEA: NUEVOS HORIZONTES PARA LOS MEDICAMENTOS HUÉRFANOS, 28 JUNE

Maria Cavaller contributed to the discussion

11TH EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS (ECRD), 29 JUNE

Anastasiia Saliuk, Michael Wilbur, Sandra Courbier, Matt Bolz-Johnson, Edith Gross, Jelena Malinina, François Houÿez, Yann Le Cam contributed to the discussion

EURORDIS Board Members **Elizabeth Vroom, Maria Montefusco, Dorica Dan** also actively participated in the debates

11TH EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS (ECRD), 30 JUNE

Jelena Malinina: Building a seamless health data ecosystem: patient perspective

WORLD ORPHAN DRUG CONGRESS, BOSTON, 11-14 JULY

Edith Gross: Public-Private-Patient Partnerships, Virginie Hivert represented EURORDIS

THE TECHNICAL MEETING "EARLY DIAGNOSIS OF PATIENTS WITH RARE DISORDERS IN THE EU: CRUCIAL ROLE OF THE NEWBORN SCREENING", 23 JULY

Gulcin Gumus: Newborn Screening (NBS): a Gateway to Early Diagnosis

HARMONISING NBS APPROACHES IN THE EU, 4 AUGUST

Gulcin Gumus contributed to the discussion

SSIEM ANNUAL SYMPOSIUM 2022, 31 AUGUST

Gulcin Gumus: A Snapshot of RD Patient Organizations' Involvement in Research & Rare Disease Policies

Edith Gross: Plenary panel on ethical and social issues in newborn screening

Edith Gross: (talk) Patient Engagement and Partnership in IMI project Screen4Care

DIA DIRECT WEBINAR: PROTOCOLS, RESEARCH PARTICIPANTS, AND SAFETY IN THE CONTEXT OF WAR, 31 AUGUST

Anastasiia Saliuk: Working together to support Ukrainians living with a rare disease`

14TH CROATIAN COCHRANE SYMPOSIUM, 1 SEPTEMBER

François Houÿez: Appropriate patient involvement in HTA

EJP RD - EUROPEAN JOINT PROGRAMME ON RARE DISEASES GENERAL ASSEMBLY AND CONSORTIUM MEETING, 13-15 SEPTEMBER

Roseline Favresse contributed to the discussion

STUDY VISITS: LIGHT IN THE DARK - CIVIL SOCIETY STANDS WITH UKRAINE (ORGANIZED BY SOCIAL PLATFORM), 22 SEPTEMBER

Anastasiia Saliuk: Presentation on the EURORDIS' project in Poland "Razem z Ukrainą"

BIOTECH ATELIER 2022, "ALL FOR ONE, ONE FOR ALL: PATIENT ADVOCACY IN DIFFERENT DISEASE AREAS", 29 SEPTEMBER

Edith Gross: Patient Engagement in Research Partnerships

DUKE-NUS CENTRE OF REGULATORY EXCELLENCE (CORE) SCIENTIFIC CONFERENCE ON PATIENT ENGAGEMENT, 5 OCTOBER

François Houÿez: Patient involvement in the R&D, regulation and safe use of medicines

DIA PHARMACOVIGILANCE STRATEGIES WORKSHOP 12-13 OCTOBER

Virginie Hivert: The role of a patient representative in PRAC

EUROPE BIOBANK WEEK 2022, 13-14 OCTOBER

Roseline Favresse: Paediatric Biobanking and Minor Engagement

2022 WORLD HEALTH SUMMIT, 17 OCTOBER

Yann Le Cam represented EURORDIS

OPERATIONALISE EARLY ACCESS PROGRAMMES EUROPE SUMMIT, 18-19 OCTOBER

François Houÿez: Planning a Compassionate use programme and/or an early Access programme ahead

DIA CLINICAL TRIALS AND DATA SCIENCE CONFERENCE, 20 OCTOBER

François Houÿez contributed to the discussion

WHAT SHOULD THE NEW REGULATION ON ORPHAN AND PAEDIATRIC MEDICINES LOOK LIKE, 21 OCTOBER

Yann Le Cam represented EURORDIS

REGIONAL CONFERENCE ON RARE DISEASES, SERBIA, 25 OCTOBER

François Houÿez: Clinical trials for rare diseases, from the perspective of EURORDIS

CZECH PRESIDENCY EXPERT CONFERENCE ON RARE DISEASES "TOWARDS A NEW EUROPEAN POLICY FRAMEWORK: BUILDING THE FUTURE TOGETHER FOR RARE DISEASES", 25-26 OCTOBER

Virginie Hivert and Yann Le Cam contributed to the discussion

POLITICO'S HEALTHCARE SUMMIT, 27-28 OCTOBER

Jelena Malinina: Digital Health - examining Europe's next steps in unleashing a health data economy

Yann Le Cam contributed to the discussion

TOGETHER FOR RARE DISEASES (TOGETHER4RD) POLICY ASKS LAUNCH, 10 NOVEMBER

Yann Le Cam represented EURORDIS

WORLD ORPHAN DRUG CONGRESS EUROPE, 14-17 NOVEMBER

Gulcin Gumus, Yann Le Cam, Maria Cavaller represented EURORDIS

COLLOQUE SCIENTIFIQUE : 10 ANS DE CONTRIBUTION DE L'ANR AU DOMAINE DES MALADIES RARES, 24 NOVEMBER

Roseline Favresse contributed to the discussion

EUROPEAN HEALTH POLICY SUMMIT, REIMAGINING HEALTH SYSTEMS: GREEN, AGILE, AND CITIZEN-CENTRED, 29 NOVEMBER

Yann Le Cam contributed to the discussion

DIA/EUCOPE INNOVATIVE THERAPIES IN EUROPE WORKSHOP, 30 NOVEMBER

François Houÿez: More than 10 Years of ATMPs in Europe

CLINICAL RESEARCH NETWORKS FOR RARE DISEASES, 1-2 DECEMBER

Virginie Hivert contributed to the discussion

INTERNET GOVERNANCE FORUM 2022, 28 NOVEMBER-2 DECEMBER

Jelena Malinina: Reducing Disparate Outcomes with Digital Health Tools

9TH EUROPEAN TAY-SACHS AND SANDHOFF FAMILY CONFERENCE, 1 DECEMBER

Maria Cavaller: Patient Engagement in regulatory activities; Maria also contributed to the panel discussion

CENTRES OF EXCELLENCE SYMPOSIUM "IMPACT OF RARE DISEASE PATIENT ORGANISATIONS IN DRIVING RESEARCH", 22 DECEMBER

Gulcin Gumus contributed to the discussion

JOURNAL PUBLICATIONS
2022

We count the years in children recovered, Leukemia, Italy

REPURPOSING OF MEDICINES IN THE EU: LAUNCH OF A PILOT FRAMEWORK, JANUARY 2022

Contributing author: **François Houÿez**

THE ADDED VALUE OF PATIENT ENGAGEMENT IN EARLY DIALOGUE AT EMA: SCIENTIFIC ADVICE AS A CASE STUDY, JANUARY 2022

Contributing author: **Maria Mavris**

MONITORING AND EVALUATION OF PATIENT ENGAGEMENT IN HEALTH PRODUCT RESEARCH AND DEVELOPMENT: CO-CREATING A FRAMEWORK FOR COMMUNITY ADVISORY BOARDS, JANUARY 2022

Contributing authors: **Rob Camp, François Houÿez**

THE VASCERN EUROPEAN REFERENCE NETWORK: AN OVERVIEW, JANUARY 2022

Contributing author: **Marine Hurard** (in her capacity as part of the AP-HP / VASCERN team)

ENHANCING THE VALUE OF CLINICAL NETWORKS FOR RARE DISEASES, APRIL 2022

Contributing author: **Matthew Bolz-Johnson**

DESIGNING RARE DISEASE CARE PATHWAYS IN THE REPUBLIC OF IRELAND: A CO-OPERATIVE MODEL, APRIL 2022

Contributing author: **Matthew Bolz-Johnson**

THE IMPORTANCE OF PSYCHOLOGICAL SUPPORT FOR PARENTS AND CAREGIVERS OF CHILDREN WITH A RARE DISEASE AT DIAGNOSIS

Contributing author: **Matthew Bolz-Johnson**

VALUING THE "BURDEN" AND IMPACT OF RARE DISEASES: A SCOPING REVIEW, JUNE 2022

Contributing authors: **Julien Delaye, Anna Kole**

TOWARDS ACHIEVING EQUITY AND INNOVATION IN NEWBORN SCREENING ACROSS EUROPE, INTERNATIONAL JOURNAL OF NEONATAL SCREENING, MAY 2022

Contributing authors: **Yann Le Cam, Gulcin Gumus, Valentina Bottarelli**

REAL-WORLD EVIDENCE (RWE): A CHALLENGE FOR REGULATORY AGENCIES DISCUSSION OF THE RWE CONFERENCE WITH THE NETWORK OF THE EUROPEAN MEDICINE AGENCIES, PATIENTS, AND EXPERTS, JULY 2022

Contributing author: **Virginie Hivert**

CONTRIBUTION OF PATIENT REGISTRIES TO REGULATORY DECISION MAKING ON RARE DISEASES MEDICINAL PRODUCTS IN EUROPE, AUGUST 2022

Editor: **Virginie Hivert**

PAEDIATRIC INFLAMMATORY BOWEL DISEASE: A MULTI-STAKEHOLDER PERSPECTIVE TO IMPROVE DEVELOPMENT OF DRUGS FOR CHILDREN AND ADOLESCENTS, JOURNAL OF CROHN'S AND COLITIS, SEPTEMBER 2022

Contributing author: **Maria Cavaller-Bellaubi**

IDENTIFYING OBSTACLES HINDERING THE CONDUCT OF ACADEMIC-SPONSORED TRIALS FOR DRUG REPURPOSING ON RARE-DISEASES: AN ANALYSIS OF SIX USE CASES, SEPTEMBER 2022

Contributing author: **Virginie Hivert**

EDITORIAL: THE USE OF REAL WORLD DATA FOR REGULATORY PURPOSES IN THE RARE DISEASES SETTING, NOVEMBER 2022

Contributing author: **Virginie Hivert**

PATIENT JOURNEY EXPERIENCES MAY CONTRIBUTE TO IMPROVE HEALTHCARE FOR PATIENTS WITH RARE ENDOCRINE DISEASES, DECEMBER 2022

Contributing authors: **Matthew Bolz-Johnson, Lenja Katharina Wiehe**

RARE DISEASE EDUCATION IN EUROPE AND BEYOND: TIME TO ACT, DECEMBER 2022

Contributing author: **Sharon Ashton**

ACKNOWLEDGEMENTS

"GRIN2B under the Sea"
GRIN2B
Taken by Sandra Silva, Spain

EURORDIS WOULD LIKE TO THANK THE FOLLOWING ORGANISATIONS AND COMPANIES FOR THEIR FINANCIAL SUPPORT IN 2022:

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.	AFMTÉLÉTHON CURE THROUGH INNOVATION
EUROPEAN COMMISSION DG Health and Food Safety	• The Operating Grant for year 2022	Co-funded by the Health Programme of the European Union

EUROPEAN COMMISSION
DG Research and Innovation
	• The European Joint Programme on Rare Diseases (EJP RD)
	• ERICA: European Rare Disease Research Coordination and Support Action consortium
	• The Next Generation Health Technology Assessment (HTx)
	• Solve-RD – Solving the Unsolved Rare Diseases
	• More-EUROPA – More Effectively Using Registries to suppOrt PAtient-centered Regulatory and HTA decision-making
	• Remedi4All
	• The Innovative Medicines Initiative Joint Undertaking (IMI JU) projects:
	  • conect4children (c4c)
	  • Screen4Care
	  • FACILITATE – FrAmework for CIInicaL trIal participants' daTA reutilization for a fully Transparent and Ethical ecosystem
		Co-funded by the Horizon 2020 Framework Programme of the European Union
		imi innovative medicines initiative

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 corporate donors in 2022. These companies have supported EURORDIS through the EURORDIS Round Table of Companies, the European Conference on Rare Diseases and Orphan Products, 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, the EURORDIS Ukraine Response programme and unsolicited donations. The breakdown of each company's contributions by project is detailed on the EURORDIS website on the "Corporate revenue" tab of the "Our Funding" section.

TOP FIVE DONORS

1	TAKEDA	Takeda
2	PFIZER	Pfizer
3	ROCHE	Roche
4	ILLUMINA	illumina
5	HORIZON THERAPEUTICS and NOVARTIS	HORIZON NOVARTIS

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

FOUNDATIONS AND NON-FOR-PROFIT CONTRIBUTORS

Agrenska	Bertarelli Fondation
Be your Possible	Boston Scientific Foundation Europe
Chan Zuckerberg Initiative	Everylife Fondation
Fondation Ipsen, under the aegis of Fondation de France	Global Genes
International Federation of Pharmaceutical Manufacturers & Associations	The Marigold Fondation
Singapore Red Cross	Special Mention

We also would like to sincerely thank all the generous individuals, organisations and companies from across many countries who made a gift to EURORDIS in 2022.

ACRONYMS AND DEFINITION

EURORDIS INTERNAL AND TASKFORCES

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

EURORDIS PROJECTS/INITIATIVES OR IN WHICH EURORDIS IS INVOLVED

ERTC	EURORDIS Round Table of Companies (with pharma & biotech developing Orphan Medicinal Products)
OA	EURORDIS Open Academy
ePAG	European Patient Advocacy Group
ECRD	European Conference on Rare Diseases and Orphan Products
NGO Committee for Rare Diseases	The NGO Committee for Rare Diseases engages at the UN level to elevate rare diseases to a priority within global public health.
RDD	Rare Disease Day
RDI	Rare Diseases International
EMM	EURORDIS Membership Meeting
AGA	Annual General Assembly
BPA	Black Pearl Awards
RDW	Rare Disease Week
RareConnect	A safe, easy to use platform where rare disease patients, families and patient organizations can develop online communities and conversations across continents and languages.
Rare Barometer	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.
Rare 2030	Rare 2030 was a foresight study that gathered the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations that would lead us to improved policy and a better future for people living with a rare disease in Europe.
BBMRI Stakeholders' Forum	Biobanking and Biomolecular Resources Research Infrastructure
ECRIN	European Clinical Research Infrastructures Network
E-Rare	Network of ten partners – public bodies, ministries and research management organisations – from eight countries, responsible for the development and management of national/ regional research programs on rare diseases

EUnetHTA Forum	Support effective HTA collaboration in Europe that brings added value at the European, national and regional levels
EuroBioBank	European Network of DNA, cell and tissue banks for rare diseases – EU project 2003 – 2006 for which EURORDIS was administrative coordinator. It is now the biobank network of RD-Connect
EUROPLAN	Fostering National Plans in Europe (project ended in 2018)
EUPATI	Innovative Medicines Initiatives Joint Undertaking "Fostering Patient Awareness on Pharmaceutical Innovation"
EJA	"Joint Action on Rare Diseases of the EU Committee of Experts on Rare Diseases: Funded by EC and by Member States, divided in work packages corresponding to specific activities, e.g. continuity of Europlan (Work Package 4); developing guidelines for social services dedicated to RDs (Work Package 6)"
FACILITATE	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.
Global Commission	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.
GCOF	Genetic Clinics of the Future: To map the opportunities and challenges that surround the clinical implementation of next generation sequencing technologies, Horizon 2020, 2015-2017
HTx Project	HTx is a project funded by the European Union which aims to take this to the next level.
InnovCare	Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions, DG Employment and Social Innovation (EaSI), 2015-2018
IMI	Innovative Medicines Initiative
IRDiRC	International Rare Disease Research Consortium
PARADIGM	PARADIGM's mission is to provide a unique framework that enables structured, effective, meaningful, ethical, innovative, and sustainable patient engagement (PE).
SCOPE	The Strengthening Collaboration for Operating Pharmacovigilance in Europe (SCOPE) Joint Action
SCREEN4CARE	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.
SOLVE-RD	"Solve-RD – solving the unsolved rare diseases" is a research project funded by the European Commission for five years (2018-2022)
TREAT-NMD	Translational Research in Europe – Assessment and Treatment of Neuromuscular diseases
Web-RADR	Development of tools for patients and healthcare professionals to report suspected adverse drug reactions to national EU regulators, Innovative Medicines Initiative (IMI), 2014-2017
RD-Action	Joint Action to expand and consolidate the achievements of the former EUCERD JA, DG Sanco, 2015-2018
EJP RD	European Joint Programme for Rare Diseases

EURORDIS & EUROPEAN REGULATORY NETWORK

CAT	Committee for Advanced Therapies
CHMP	Committee for Human Medicinal Products
SAG	Scientific Advisory Group at the Committee for Human Medicinal Products

COMP	Committee of Orphan Medicinal Products
EMA	European Medicines Agency
HMA	Heads of Medicines Agencies
PCWP	Patients and Consumers Working Party
PDCO	Paediatric Drugs Committee
PRAC	Pharmacovigilance and Risk Assessment Committee
SAWP	Scientific Advice Working Party
EPAR	European Public Assessment Report
SPC / SmPC	Summary of Product Characteristics
MA	Marketing authorisation

EUROPEAN COMMISSION

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

NON GOVERNMENTAL PARTNERS

AFM-Téléthon	French Muscular Dystrophy Association
DIA	Drug Information Association
CORD	Canadian Organization for Rare Disorders / Chinese Organization for Rare Disorders
EFPIA	European Federation of Pharmaceutical Industries and Associations
EPF	European Patients' Forum
EPPOSI	European Platform for Patients' Organisations, Science and Industry
EuropaBio	The European Association for Bioindustries
EUCOPE	European Confederation of Pharmaceutical Entrepreneurs
ESHG	European Society of Human Genetics

IAPO	International Alliance of Patients' Organizations
IFSW-Europe	International Federation of Social Workers
Inserm	French National Institute for Health and Medical Research
ISPOR	International Society for Pharmacoeconomics and Outcomes Research
MRIS	Maladies Rares Info Services (French helpline for rare diseases)
NORD	National Organization for Rare Disorders (USA) – EURORDIS' counterpart in the US
RDI	Rare Diseases International
Orphanet	The online portal for rare diseases and orphan drugs: orpha.net
NORBS	The National Organisation for Rare Diseases Of Serbia
HUFERDIS	Hungarian Federation of People with Rare and Congenital Diseases
RADOIR	Rare Disease Foundation of Iran
San Pau	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

PLWRD	Persons living with a rare disease
CoE / CE	Centre of Expertise / Excellence
ERN	European Reference Network
EU MS	Member State (of the European Union)
EUNRDHL	EU Network for Rare Diseases Helplines
NP (RD)	National Plan / Programme (for Rare Diseases)
HTA	Health Technology Assessment
MAPPS	Medicine Adaptive Pathways to Patients
MEP	Member of the European Parliament
MoCA	Mechanism of Coordinated Access to orphan medicinal products
PACE-ERN	Partnership for Assessment of Clinical Excellence in European Reference Network (PACE-ERN) Consortium
PE	Patient Engagement
PLWRD	Persons Living with a Rare Disease
TRP	Therapeutic Recreation Programme
CAVOD	Clinical Added Value of Orphan Drugs
OMP	Orphan Medical Product
Orphan drug	"Orphan drugs" are medicinal products intended for diagnosis, prevention or treatment of life-threatening or very serious diseases or disorders that are rare.
MA	Marketing Authorisation (for a medical product)
PV	Pharmacovigilance
EudraVigilance	EudraVigilance is a system designed for collecting reports of suspected side effects
ADR	Adverse Drug Reaction

CUP	Compassionate Use Programme
ATMP	Advanced Therapy Medicinal Product
NBS	Newborn Screening
NGS	Next-Generation Sequencing
UN	United Nations
CoNGO	Conference of Non-Governmental Organizations in Consultative Relationship with the United Nations
#Resolution4Rare	#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.

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