EURORDIS
RARE DISEASES EUROPE

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ACTIVITY REPORT 2023

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COVER PHOTO: "I AM SHORT OF OXYGEN BUT OVERWHELMED WITH HOPE!"
PULMONAR HYPERTENSION, UKRAINE

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FOREWORD BY PRESIDENT AND CEO

AVRIL DALY
President of the Board of Officers

VIRGINIE BROS-FACER
Chief Executive Officer

We can be proud of how far we've all come over the past year. Our community, which reached the important milestone of 1,000 members in 2022, continued to grow and this year could gather again face-to-face to exchange knowledge and share experiences after several years of online meetings. The EURORDIS Membership Meeting, focusing this year on a holistic approach to care, took place in Stockholm with over 200 participants from our community representing 38 countries.

On this occasion, the call to join the Mental Health & Wellbeing Partnership Network, a new EURORDIS initiative addressing the mental wellbeing of people with rare diseases, was launched. This call resulted in the registration of over 100 members from 25 countries. The Partnership Network held its inaugural meeting on World Mental Health Day, 10 October 2023, to celebrate and stand in solidarity with all those affected by mental health issues.

We celebrated Rare Disease Day in over 100 countries, broadly disseminated the results of the largest-ever Rare Barometer survey on diagnosis and launched a new survey on newborn screening. We also organised the Brussels Rare Disease Week in February, which comprised an online training programme and a series of in-person advocacy events in Brussels. 38 participants from 21 countries benefited from this programme aiming at creating a direct connection between rare diseases patient advocates and their MEPs and other EU policymakers, allowing for solid relationship-building.

EURORDIS continued to strive to empower its members and community through the Open Academy Schools, offering over 70 hours of eLearning, as well as new alumni programmes, including Meetups and Masterclasses. Over 2,600 registered users from approximately 160 countries participated in our eLearning courses.

2023 was also marked by an important evolution in EURORDIS leadership, with Yann Le Cam, who had served as the EURORDIS Chief Executive Officer for the past 25 years, announcing his intention to step down as CEO in March 2024. Virginie Bros-Facer was appointed as the new EURORDIS CEO in November 2023 and took office in March 2024.

In 2023, the Rare Cancer Advocate Network was launched. It is the only network that brings together rare cancer patient advocates and ePAGs across all types of rare cancers and all ages. This network includes members from national, European, and international federations of patients and is connected with EURORDIS and the ERNs.

As 2024 unfolds, EURORDIS stands at a critical juncture of transformative change. This year, against the backdrop of the EU elections, is pivotal in steering our path towards the ambitious objectives set for 2030. Our commitment deepens as we aim to thoroughly integrate the narrative of rare diseases into the broader context of healthcare, economic, and social considerations at national, European, and international levels.

Strategic Focus in Europe:
The year 2024 in Europe is earmarked for intensified policy advocacy, addressing the unique challenges and opportunities presented by rare diseases. With the EU elections in sight, our focus is on shaping an all-encompassing European action plan for rare diseases, with our strengthened Brussels public affairs team leading the charge.

Calling on the New EU Parliament and Commission:
At the heart of our agenda is advocating for a European action plan, integrating rare diseases into the broader healthcare, social, research and economic fabrics. Our dual focus includes shaping new comprehensive policies, such as the European Health Data Space, and actively engaging in the review of existing legislation, such as the Orphan Medicinal Products Regulation.

The 'Championing the Rare' Campaign (#ActRare2024):
This campaign, commencing on Rare Disease Day at the EU Parliament and continuing through to the EU elections, is designed to rally EU candidates around our cause, amplifying the voice of the rare disease community in pivotal political spheres.

Strengthening Networks for Greater Impact:
2024 is a year when we reinforce our support for member organisations, particularly focusing on regions requiring our utmost attention, including South-East and Eastern Europe. Additionally, we will concentrate on countries such as Spain, Italy, Greece, Luxembourg, Ireland, Sweden, and Finland, where emerging national plans for rare diseases necessitate a harmonised approach at both national and European levels.

Advancing Healthcare and Research:
A foundational element of our strategy is to minimise diagnostic delays and improve access to specialised care, especially for ultra-rare conditions. Pioneering initiatives like the European Rare Disease Research Alliance (ERDERA) and the Rare Disease Moonshot are pivotal in propelling our understanding of rare diseases through cutting-edge research and clinical networks.

Global Reach and Collaboration:
In 2024, EURORDIS will strengthen its action to elevate rare diseases as a global health priority, engaging with international stakeholders, first of which Rare Diseases International.

Advocacy at the United Nations:
Leveraging the United Nations system, we aim to build upon our past achievements, including the Political Declaration on UHC in 2019 and 2023, and the UN Resolution on Persons Living with a Rare Disease upgraded in 2023. Our priority 2024-27 is on advocating for the inclusion of rare diseases in a WHO resolution on RDs and Universal Health Coverage and a WHO Global Action Plan on RDs.

Fostering International Alliances and Partnerships:
Central to our global strategy is building and nurturing international networks and collaborations on research initiatives. By sharing experiences and learning from global partners, we aim to address the challenges faced by the rare disease community on an international scale.

Global Healthcare Pathways:
In advocating for comprehensive healthcare frameworks globally, our aim is to be at the forefront of developing tools for early diagnosis, and access to expertise, preparing the global healthcare system for the upcoming wave of diagnostic and therapeutic innovations. 2024 is a year of unprecedented opportunity to forge a future where rare diseases are not just recognised but integrated into the national, European and global agenda. Our mission is clear: to ensure that no one with a rare disease is left behind in our collective pursuit of a healthier, more inclusive world.

EURORDIS IN BRIEF

AS OF DECEMBER 2023

EURORDIS - Rare Disease 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.

1997
Year of Establishment

1051
Member Patient Organisations

74
Countries

27
EU Countries

55
National Alliances of RD Patients Organisations

80
European Federations of Specific Rare Diseases

55+
Staff, Offices in Paris, Brussels, Barcelona

7 M€
Budget

OVER 60
Volunteers

OVER 2500
Patient Groups Outreach

“WITH A BIG HEART”
BARTH SYNDROME / RUSSIA

"AMELIA WITH BEAUTIFUL, BLUE EYES"
HYPOPLASTIC LEFT HEART SYNDROME / LATVIA

OUR STRATEGY & IMPACT 2021-2030

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 their strategic review throughout 2022 and adopted the revised strategy in November of that year.

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 01

Diagnosed timely and accurately 02

Supported with state-of -the-art medical and social care, or cured 03

Included in society in all aspects of life and enabled to live independently 04

MISSION

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

OUR STRATEGY 2021-2030

RESEARCH POLICY & ACTIVITIES

DEVELOPMENT & ACCESS TO DIAGNOSTIC & THERAPIES

DATA & DIGITAL HEALTH

HEALTHCARE POLICY & SERVICES

SOCIAL POLICY & SERVICES

PEOPLE FIRST

Based on evidence and experience of unmet needs and preferences

PARTNER

To establish and facilitate networks with strategic partners and key stakeholders.

EMPOWER

To empower organisations and advocates representing people living with rare diseases across all rare diseases and all European countries.

ADVOCATE

To advocate for policies and innovative solutions driven by the needs of people living with rare diseases.

STRATEGIC OBJECTIVES

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 Foresight Study "Rare 2030", guaranteeing that rare diseases are a **sustainable public health priority** and integrating **European policies and national strategies** across all countries in Europe, to a**chieve measurable goals improving the health & well-being** of people living with rare diseases and their i**nclusion in society, as well as reducing inequalities across Europe.**

"HE IS OUR ANGEL"
HYDROCEPHALUS / INDIA

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 a rare disease and a collective responsibility to prioritise solidarity and equity.

DELIVERING ON 6 PRIORITY AREAS

By 2030, EURORDIS will have made contributions to the goals of (Based on the Foresight Study Rare 2030):

- Earlier, faster and more accurate diagnosis – the goal of diagnosis within 6 months;
- High-quality national and European healthcare pathways, including cross-border healthcare – a goal of improving survival by 3 years on average over 10 years and reducing the mortality of children under 5 years of age by one third;
- Integrated medical and social care with a holistic life-long approach and inclusion in society – a goal of reducing the social, psychological and economic burden by one third;

- Research and knowledge development that is 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;
- Development and availability, accessibility, and affordability of treatments, particularly transformative or curative therapies – a goal of 1000 new therapies within 10 years.

INCLUSIVE OF ALL RARE DISEASES, ALL REGIONS, “LEAVING NO ONE BEHIND”

By 2030, EURORDIS has consolidated its scope to “leave no one behind” in the rare disease community by covering:

- All therapeutic areas, including genetic or non-genetic rare diseases, and rare cancers, with progress to be made with regard to rare infections and rare health hazards;
- All countries in geographical Europe prioritising Eastern and Southern Members of the EU, European Economic Area & EU Accessing Countries;
- All rare disease prevalence and incidence levels, particularly the ones affecting fewer than 1 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.

“FRIENDS”
SPINA BIFIDA, VESICOURETERAL REFLUX / NEPAL


"WATERMELON MOOD"
MUCOPOLYSACCHARIDOSIS TYPE 2 (HUNTER DISEASE) / RUSSIA

HIGHLIGHTS

The EURORDIS membership grows steadily every year, a trend that continued in 2023 as EURORDIS developed and engaged its network of members. **Fifty-seven new members (patient organisations) joined in 2023.**

2023 was also marked by a significant evolution in EURORDIS leadership, with Yann Le Cam, who had served as EURORDIS CEO for the past 25 years, announcing he would step down in March 2024. Virginie Bros-Facer was appointed as the new EURORDIS CEO in November 2023 and assumed the role in March 2024.

In 2023, more than **60 EURORDIS volunteer patient advocates,** belonging to various groups or task forces managed by EURORDIS Patient Engagement Managers, contributed their efforts. These included the Therapeutic Action Group, the Drug Information, Transparency and Access Task Force, the Health Technology Assessment Task Force, the Digital and Data Advisory Group, and the Social Policy Action Group, which was relaunched in 2023.

In 2023, EURORDIS continued to collaborate with experts, including clinicians and researchers, along with **over 300 partners** and stakeholders – ranging from organisations and institutes to private sector bodies and governments. This collaboration occurred within co-funded European projects where we are either a partner or leader[1], as well as through our memberships in **70 European and international networks**[2], and within the EURORDIS Round Table of Companies, which continues to grow. EURORDIS acknowledges the support and collaboration from all our partners as we work towards our mutual goals for 2030.

For its 16th edition, Rare Disease Day 2023 events took place in **over 110 countries** and regions across every corner of the globe, and we welcomed **three new countries** to the campaign. In addition to holding events, individuals and organisations worldwide downloaded and shared our branded materials. We also built momentum throughout February by launching the #RareDiseaseDay challenge. Despite energy constraints, **buildings, monuments, homes,**

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[1] **Visit our Projects & Partnerships webpage here:**
https://www.eurordis.org/projects-and-partnerships/

[2] **External representation on the website:**
https://www.eurordis.org/external-representation/

and offices worldwide were illuminated in the colours of Rare Disease Day in the Global Chain of Lights, demonstrating solidarity for people living with rare diseases.

The Rare Barometer programme launched a survey in March 2022 to explore the diagnostic **journey of rare disease patients,** with results analyzed in 2023. EURORDIS members received **24 dashboards in 27 languages** and a key results factsheet. The comprehensive European results were submitted to the European Journal of Human Genetics, featuring several EURORDIS members as co-authors. Additionally, **a new survey on newborn screening** was launched in May 2023.

The EURORDIS Membership Meeting (EMM) 2023 in Stockholm took place in May, with **216 patient representatives from 38 countries.** It focused on a **'lifelong holistic approach to people's needs and their full inclusion in society,'** aligning with our strategic objective of social inclusion. EURORDIS members shared experiences, identified gaps, and formulated recommendations to help ensure that people with rare diseases achieve the highest standard of health and well-being, and access equal opportunities and rights throughout their lives.

Rare Disease Week (RDW) 2023 was held from 6 to 9 February in Brussels, with **38 participants from 21 countries.** Throughout the week, 29 individual meetings with MEPs and **two additional meetings with other EU policymakers** (Commission and Council) were organised. A high-level conference titled **'For a patient-driven evolution of the Orphan Medicinal Product Regulation: Addressing unmet needs'** took place at the European Parliament, engaging RDW participants and a wider audience. Preparations for **RDW 2024** are underway, with the event scheduled for October, following the EU elections.

EURORDIS actively contributed to the EU debate on **revising the General Pharmaceutical Legislation,** including regulations for special populations (rare diseases and paediatric). Key activities included analysing proposals from the European Commission and amendments by the European Parliament's committees (ENVI and ITRE); **regularly updating and consulting with EURORDIS members** to gather their views on the upcoming EU legislation; conducting over **50 meetings with key figures** from the European Commission, European Parliament, and Council; and **organising trainings and ad hoc meetings**, particularly for National Alliances, on critical aspects of the proposals and debates. With the support of its National Alliances, EURORDIS also monitored national debates in EU Member States.

By the end of 2023, the **EURORDIS Open Academy's eLearning platform had over 3,138 registered users from more than 119 countries.** The year's trainings included the School on Medicines Research & Development and the School on Scientific Innovation and Translational Research, featuring e-learning modules, pre-training webinars, and intensive in-person training days, all delivered in English. **Additionally, six Meetups and two Masterclasses were organised for the Open Academy Alumni.** These sessions, led by experts from industry, academia, and the patient community, covered topics like European Pharmaceutical Legislation, research initiation, digital rights, establishing a scientific committee, survey design, introduction to N-of-1 trials, and quality of life and patient reported outcomes.

"JOY AND STRUGGLE OF MOON CHILDREN"
OCULOCUTANEOUS ALBINISM / BANGLADESH

During 2023, **EURORDIS continued its response programme in Ukraine.** EURORDIS was actively engaged in the coordination of patient organisations in Ukraine and supported the National Alliance and other organisations with direct grants to support Ukrainians living with rare diseases. In May 2023, EURORDIS completed the 1-year "Razem z Ukraina" project in partnership with 14 patient-oriented non-profit organisations which was able to help 1188 Ukrainians living with rare diseases and their families. 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, EURORDIS supported the work of the Rare Disease Hub, an organisation whose mission is to ensure a very close partnership with the ERNs to provide medical support for families coming from Ukraine.

The Rare Cancer Advocates Network (RCAN) was officially launched in January 2023. This network is made up of **rare cancer ePAGs** across the four ERNs relevant to rare cancers, as well as ePAGs in the sub-network "endocrine tumours" of ENDO-ERN, and Dorica Dan as EURORDIS Board member and Chair of the Romanian association for rare cancers and the Romanian national alliance for rare diseases. **RCAN is the only network that brings together rare cancer patient advocates/ ePAGs across all types of rare cancers and all ages, from national, European, international federations of patients, connected with EURORDIS and the ERNs.** RCAN members assessed and ranked the most pressing priorities to work on which are raising the profile of rare cancer patients; fostering drug development and access; integrating rare adult cancers and paediatric cancers (distinct sections) in National Cancer Plans.

Following a call from the rare disease community to address the psychological impacts of rare diseases, EURORDIS coordinated the creation of the **Mental Health & Wellbeing Partnership Network**. Launched during the EMM 2023 Satellite Workshop - Enhancing Mental Wellbeing in the Rare Disease Community, it attracted over 100 members from **25 countries. The network connects with more than 40 medical experts, psychologists, and researchers across 12 European Reference Networks and other international centres. Its inaugural meeting took place on World Mental Health Day, 10 October 2023,** to support and show solidarity with those affected by mental health issues. EURORDIS has also developed an action plan to guide the Network's activities from 2024 to 2026.

In 2023, EURORDIS appointed a new Human Resources Director to undertake a comprehensive HR diagnosis. This included 22 qualitative staff interviews, a 360° assessment of the Core Leadership Team, and the initiation of an annual Social Barometer in collaboration with 'Great Place to Work.' This three-year initiative aims to assess employee mental wellbeing and align all staff with the goal of achieving 'Great Place to Work' certification through a People-Centric Approach. A people management system will be developed over 2024-2025 to enhance the people-centric approach, core organisational culture, work processes, and performance measurement, while aligning HR processes and tools with strategic objectives.

“AN INJECTION A DAY, KEEPS MY FLARE AWAY”
RELA HAPLOINSUFFICIENCY / NETHERLANDS

STRATEGIC OBJECTIVE 01:

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

Throughout 2023, EURORDIS has supported this strategic objective through advocacy, empowerment and partnership activities and continued to promote rare diseases as a policy priority at national, European, and international levels.

ADVOCATE

EMPOWER

PARTNER

“WONDER AND LIGHT”
ORNITHINE TRANSCARBAMYLASE DEFICIENCY, A UREA CYCLE DISORDER / USA

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

In 2023, efforts to support the development of a European Action Plan for Rare Diseases gained momentum following the primary recommendation of the Rare 2030 foresight study, co-funded by the European Commission. Key activities included:

- **Facilitating the support of 48 Members of the European Parliament (MEPs) who, on Rare Disease Day (28 February 2023), sent a letter to Ms Ursula von der Leyen, President of the European Commission; Ms Stella Kyriakides, Commissioner for Health and Food Safety; and Mr Nicolas Schmit, Commissioner for Jobs and Social Rights. The letter advocated for the adoption of a European strategy on rare diseases.**
    - The MEPs, many from the Network of Parliamentary Advocates for Rare Diseases affiliated with EURORDIS, emphasised the urgent need to tackle challenges such as delayed diagnosis, limited access to transformative treatments, and inconsistent availability of rare disease medicines across EU member states.
    - The letter proposed that the European Commission establish a comprehensive European strategy on rare diseases. This strategy would integrate national and European legislation, policies, and programmes, encompassing the Pharmaceutical Package, the European Health Data Space, European Reference Networks, Europe's Beating Cancer Plan, EU4Health, Horizon Europe, the Long-Term Care Strategy, the European Disability Strategy, and more.
- **Contributing to, and attending as speakers, the Conference on Rare Diseases and the European Reference Networks in Bilbao, Spain (10–11 October 2023). Organised by the European Economic and Social Committee (EESC) under the Spanish EU Council Presidency, the conference titled 'How to guarantee European solidarity for patients?' highlighted the critical need for a European Action Plan for Rare Diseases.**

  - Yann Le Cam, CEO of EURORDIS – Rare Diseases Europe, represented the European rare disease patient community on a multi-stakeholder panel. The discussion focused on the necessity of listening to Member States and citizens and urgently adopting a European Action Plan for Rare Diseases.

  - The conference emphasised the collective commitment to this action plan, advocating for harmonised policies, increased resources, and collaborative efforts to support the 30 million Europeans living with a rare disease.

- **Contributing to the organisation of the "Hacia un Plan Europeo Integral: la implicación de España en Enfermedades Raras" conference led by FEDER (the Spanish Alliance for Rare Diseases) during the Spanish Presidency of the EU Council. Held on 20 November 2023 at the Congreso de los Diputados in Madrid, Spain, this event demonstrated strong support from Spanish institutional players for a European Action Plan for Rare Diseases.**

  - FEDER organised the event, with EURORDIS enhancing the agenda and ensuring the involvement of prominent European speakers. The conference highlighted the support of Spanish institutions for both advancing Spain's national policy on rare diseases and a reinforced EU policy on rare diseases underpinned by a comprehensive action plan that spans multiple policy areas.

- **Drafting EURORDIS's Manifesto, "Championing the Rare," ahead of the 2024 EU elections, aimed at making rare diseases a priority for European Parliament candidates, the future European Commission, and other political leaders. This effort formed part of the broader #ActRare2024 campaign, which seeks to influence the European debate in a transformative year (Q2-Q4 2023).**

  - The manifesto outlines recommendations across eight key policy areas, advocating for an inclusive European Health Union that leaves no one behind. The primary recommendation is for a comprehensive and goal-oriented European Action Plan for Rare Diseases to meet the substantial unmet needs of those living with a rare disease and to integrate various legislative aspects concerning data, research, treatment, healthcare, social protection, and wellbeing, alongside both national and EU initiatives.

  - EURORDIS also disseminated the manifesto to national organisations and provided a template email for engaging with national parties. Additionally, it was sent to EU political groups to inform the drafting of their own manifestos.

Securing proposals impacting rare diseases within the EU Multi-annual Financial Framework 2021–2027

In 2023, EURORDIS supported several initiatives under the EU4Health Work Programme 2024, including support for national plans and strategies for rare diseases, EU-level collaboration on newborn screening, enhanced EU cooperation for the delivery of highly specialised healthcare services and innovative therapies, and mental health action for individuals in vulnerable situations. It also proposed a Joint Action for European procurement of therapies for rare diseases and pilots to support WHO-led Global Networks for Rare Diseases, aiming to enhance coordination and access to treatments across Europe.

Involving the European Network of National Alliances in Advocacy Activities

EURORDIS engaged the European Network of National Alliances in advocacy through the CNA, including through in-person meetings and monthly "hot topics" calls. Throughout the year, EURORDIS provided continuous support to its National Alliances by sharing medium-term advocacy plans and supplying tools and information to enhance Member State engagement with relevant EU initiatives and legislation. These activities, aligned with the EURORDIS-CNA Common Goals and Mutual Commitments, included enhancing the access of National Alliances to the Rare Barometer surveys to facilitate more coordinated, evidence-based advocacy.

Convening the European Network of Parliamentary Advocates for Rare Diseases

In 2023, EURORDIS expanded the European Network of Parliamentary Advocates for Rare Diseases by organising 30 meetings with MEPs and patient advocates during Rare Disease Week (6–9 February 2023, Brussels, Belgium). These meetings bolstered the engagement of existing parliamentary allies and resulted in the recruitment of 10 new MEPs to the network.

EURORDIS also organised a high-level conference on 8 February 2023 at the European Parliament in Brussels, titled ‘For a patient-driven evolution of the Orphan Medicinal Product Regulation: Addressing unmet needs’. This conference focused on the revision of the OMP Regulation and was attended by over 100 key stakeholders. Throughout the year, EURORDIS continued to engage MEPs on various topics including mental wellbeing and the European Health Data Space (EHDS), effectively advocating for the rare disease community’s key requests during negotiations. Additionally, EURORDIS collaborated on the INI report on mental health, contributing to the inclusion of significant references in the final report.

PROMOTING RARE DISEASES AS AN INTERNATIONAL PUBLIC HEALTH PRIORITY

EURORDIS has been working on the promotion of rare diseases as an international public health priority for almost a decade. The aim has been to: raise awareness of rare diseases directly among relevant international institutions; equip patient groups with advocacy tools to engage their national authorities and drive local patient empowerment; and foster greater international cooperation in the field of rare diseases.

Rare Diseases International

During the annual meeting of Rare Diseases International (RDI) in May 2018 in Vienna, Austria, members voted to legally establish RDI as an independent entity. Subsequently, EURORDIS and RDI signed a five-year Memorandum of Understanding (MoU) for the period 2019-2023 to ensure continued collaboration.

EURORDIS has been an active participant in RDI:

- Yann Le Cam serves on RDI's Council, attending six meetings annually, and holds roles as an executive officer and treasurer, engaging in regular executive meetings.
- Yann Le Cam also chairs the RDI Policy Committee, which includes experienced patient advocates from RDI's membership worldwide, encompassing some national alliances and international federations based inEurope.
- EURORDIS has been involved in all RDI membership meetings and policy events.

Since its inception, EURORDIS has collaborated with RDI to promote rare diseases as an international public health initiative, contributing vision, strategy, and leadership. This includes engaging with the International Community through the UN System. All UN system policy actions are co-organised by the UN NGO Committee for Rare Diseases (EURORDIS is a founding member and Yann Le Cam its Vice President), RDI, and Agrenska.

Significant RDI policy achievements led by EURORDIS up to and during 2023 include:

- The explicit inclusion of people living with rare diseases in the UN Political Declaration on Universal Health Coverage (UHC), adopted in September 2019 and reaffirmed in September 2023.
- The unanimous adoption by the UN General Assembly of the resolution addressing the challenges of persons living with rare diseases and their families in December 2021, with an amendment reinforcing UHC diagnostics unanimously passed in November 2023.
- The continuation of the MoU with WHO for 2019-2022 and the submission of an application by RDI for Non-State Actor (NSA) status in 2023, which is set to be decided in 2024. This status would benefit EURORDIS in the WHO-Europe Region.

With the MoU between EURORDIS and RDI concluding at the end of 2023, and as RDI evolves into a natural platform for global collaborations on rare diseases, EURORDIS plans to reassess and update the MoU in 2024 to re-establish partnerships in an adjusted format.

World Health Organization (Geneva)

In 2023, EURORDIS-Rare Diseases Europe advanced its partnership with the World Health Organization (WHO) through an ongoing Memorandum of Understanding (MoU) and a new Non-State Actor (NSA) application. This collaboration led to several key achievements:

WHO Global Network for Rare Diseases (GNRD): The final phase of concept development was completed, including a final needs assessment, the establishment of a conceptual and operational framework, the adoption of Terms of Reference by WHO, and the implementation of a Hub & Spoke model across WHO regions. This initiative is pivotal in fostering global cooperation on rare diseases.

Operational Description of Rare Diseases: Following a consensus on a Core Definition of Rare Diseases and a Descriptive Framework in previous years, 2023 saw the completion of an impact assessment with very positive outcomes. This work helps standardise the understanding and reporting of rare diseases worldwide.

Inclusion in WHO Essential Lists: Significant progress was made towards including more rare disease therapies in the WHO Essential Medicines List and the Essential In Vitro Diagnostic List. Efforts included analysing current listings for rare indications, promoting application opportunities for rare diseases, and building the capacity of applicants. This phase was actively pursued in collaboration with the International Rare Diseases Research Consortium (IRDIRC) and supported by RDI, with significant contributions from EURORDIS.

Additionally, a Technical Officer dedicated to rare diseases was appointed at WHO's Geneva office within the UHC and Health Promotion Department. This appointment underscores the commitment to integrating rare diseases into global health agendas.

EURORDIS has played a crucial role in these developments, contributing directly through its leadership roles in RDI and the RDI Policy Committee. The organisation's credibility and strategic input have been instrumental in articulating interactions with WHO and driving forward the agenda for people living with rare diseases (PLWRDs), aligned with Universal Health Coverage (UHC) and Sustainable Development Goals (SDGs).

WHO Regional Committee for Europe

In 2023, as part of the ongoing collaboration between RDI and EURORDIS with WHO Geneva, EURORDIS initiated dialogue and engagement with WHO Europe. Building on the expert collaboration and outcomes from 2020 to 2022, WHO Europe launched the Novel Medicines Platform in September 2023. This Member State-led multi-stakeholder initiative aims to improve access to highly innovative, costly treatments for low prevalence diseases. EURORDIS was heavily involved in both the preparation and launch of this initiative.

Following an open call for candidates, EURORDIS secured appointments in two of the three working groups. Yann Le Cam, CEO, joined Working Group 2 (WG2) on Solidarity, and Simone Boselli, Director of European Public Affairs, joined Working Group 3 (WG3) on Sustainability. Work began in late 2023, with the goal of delivering initial proposals by June 2024. This initiative is planned to continue for three years, aiming to address and resolve challenges related to the availability and sustainability of novel medicines in Europe.

"CHOOSE LIFE"
SÍNDROME DE MARFAN / ESPAÑA

EMPOWER

BUILDING THE COMMUNITY, CAPACITY-BUILDING, INFORMING, SUPPORTING, AND EMPOWERING OUR NETWORKS

Membership

EURORDIS, a grassroots movement of patient organisations active in rare diseases, continued to see steady growth in its membership in 2023. **By December 2023, 57 new members had joined, bringing the total to 1,051 Members.** EURORDIS actively engages its membership in organisational activities and decision-making processes through various channels including the Member News (a bimonthly newsletter distributed in six languages), webinars, Rare Barometer surveys, direct mailings, and face-to-face meetings, notably the EURORDIS Membership Meeting (EMM) and the General Assembly.

EURORDIS also extends its outreach to over 2,900 European patient organisations, continually enhancing its contact database.

1,051 EURORDIS MEMBERS

EURORDIS Membership Meeting (EMM) 2023

Held biennially in different European cities, the EMM is a vital forum for patient representatives to connect and learn from each other. The 2023 meeting took place in Stockholm in May, drawing 216 patient representatives from 38 countries. The event issued 40 fellowships to support patient representatives from 17 countries. The 2023 EMM focused on advancing a holistic approach to meeting the needs of individuals living with a rare disease and ensuring their full participation in society.

The meeting included:

- Two pre-workshops focused on training in digital safety and literacy amidst ongoing healthcare digitalisation and enhancing mental wellbeing within the Rare Disease Community.
- A plenary session on the lifelong holistic approach to living with a rare disease, covering stages from parenting, growing up, adulthood, to ageing.
- Four thematic workshops addressing the challenges of living with a rare disease during childhood, adolescence, adulthood, and ageing.

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

The National Alliances consolidate numerous rare disease organisations within a specific country. The Council of National Rare Disease Alliances (CNA), established and coordinated by EURORDIS, facilitates collaboration among national representatives of rare disease patients on common European actions. Significant achievements of the CNA include:

- The creation and successful implementation of Rare Disease Day.
- National Alliances actively promoting and developing national plans for rare diseases.
- The adoption of the "Common Goals and Mutual Commitments between EURORDIS & National Alliances in Europe".
- Leading a European campaign for a new policy framework.
- Advocacy for integrating European Reference Networks (ERNs) at the national level.
- Development of EURORDIS' position on newborn screening.

Currently, the CNA includes members from 25 EU countries and 9 non-EU countries (Bosnia, Georgia, North Macedonia, Montenegro, Norway, Ukraine, Russia, Serbia, Switzerland). In 2023, the CNA conducted two workshops: the first in May, a one-day meeting preceding the EURORDIS Membership Meeting (EMM) 2023, and the second in December, spanning two days with the first day dedicated to the CNA and the second involving both CNA and CEF representatives. Additionally, CNA members convene monthly online for "CNA Hot Topics" discussions and receive the "CNA Update" monthly newsletter.

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

European Federations, coordinated by EURORDIS, unite national patient organisations specific to rare diseases at the European level. The CEF facilitates collaboration on shared European initiatives. In 2023, EURORDIS enhanced its coordination of the European Network of Disease-Specific European Federations and the CEF, focusing on key strategic issues. This included improving access to disease-specific data through the Rare Barometer surveys, aiding evidence-based advocacy across Europe. Furthermore, EURORDIS provided financial support for network meetings of 17 emerging or less established rare disease European Federations.

In December 2023, representatives from the European Rare Disease Federations met over two days. The agenda included a joint session with the Council of National Alliances (CNA) and a day dedicated to peer-to-peer discussions, enhancing collaboration and learning.

European Network of Rare Disease Helplines

Established in September 2006 and coordinated by EURORDIS, the ENRDHL aims to enhance service to callers by sharing resources, best practices, and knowledge among rare disease helplines. The network strives to raise awareness, increase efficiency, and improve practice standards across its membership.

In 2023, the ENRDHL held two online meetings and introduced a new initiative: the **Access Campaign**. This campaign, available in 19 languages, invites members to collect and analyse reports from individuals experiencing difficulties accessing care for rare diseases via an online form. Participating helplines, on a voluntary basis, may contact reporters to assist in finding solutions. Launched in September and running until November 2023, the campaign gathered 558 reports, with those consenting having their details forwarded to volunteer helplines in Ireland, the UK, Italy, Romania, Croatia, France, and Spain.

Another key activity was the **Caller Profile Analysis**, an annual review conducted each October, where enquiries from all helplines are consolidated to analyse 13 common items, enhancing understanding of callers' needs and trends.


"LOOK THE FUTURE IN THE EYE"
LEUKEMIA / ITALY

Rare Connect

RareConnect, an online platform initiated by EURORDIS and hosted by the Children's Hospital of Eastern Ontario (CHEO), connected individuals living with rare diseases. While the platform maintained vibrant communities, a 2022 user survey indicated that most users were no longer reliant solely on RareConnect, as digital support for rare diseases has expanded significantly on mainstream social media.

Following discussions by the RareConnect Steering Committee, which includes representatives from CHEO and EURORDIS, it was decided to retire the platform. Officially closed on 5 December 2023, RareConnect no longer allows new communities, posts, or private messages. However, registered users and visitors can still view existing posts in public communities, and translations of content posted before the closure remain accessible.

Brussels Rare Diseases Week 2023

Rare Disease Week (RDW), organised by EURORDIS-Rare Diseases Europe in Brussels, is a week-long event comprising preparatory webinars and various activities including training sessions, policy meetings, and networking events. The programme aims to connect rare disease patient advocates with their MEPs and other EU policymakers, fostering strong relationships, raising awareness about rare diseases, and delivering a unified message to policymakers. Additionally, it provides training in European policy, advocacy, and communication.

RDW 2023 included an online training programme and a series of in-person advocacy events held from 6 to 9 February 2023 in Brussels, with 38 participants from 21 countries. The week featured 29 individual meetings with MEPs, two meetings with other EU policymakers, and a high-level conference at the European Parliament titled "For a patient-driven evolution of the Orphan Medicinal Product Regulation: Addressing unmet needs." Participants also engaged in social activities, including a bus tour and a dinner in Brussels.

Post RDW 2023, EURORDIS began preparations for RDW 2024, scheduled for October after the EU elections. A campaigning toolkit was developed for participants, containing documents summarising election priorities ("Manifestos"), a pledge for candidates, a social media campaign, and ideas for national-level activities. This toolkit aims to educate RDW participants about the EU elections interactively and to promote EURORDIS' priorities before, during, and after the elections.

Black Pearl Awards (BPA) 2023

The EURORDIS Black Pearl Awards are presented annually in February to coincide with Rare Disease Day, recognising exceptional contributions to the rare disease community. The awards honour patient advocates, organisations, policymakers, scientists, companies, and media for their ground-breaking work and dedication. The 12th edition of the awards took place on 21 February 2023 at the DoubleTree by Hilton Brussels City, marking the first truly hybrid event with both online and onsite participation. It attracted over 385 attendees from 38 countries across all stakeholder groups.

2023 Award Winners:

Lifetime Achievement Award:
Terkel Andersen (Denmark) for his pioneering work across various organisations including the Danish Haemophilia Society and EURORDIS.

European Rare Disease Leadership Award:
Dr Holm Graessner (Germany) for his extensive contribution to rare disease projects at national and international levels.

Policy Maker Award:
Dr Stelios Kympouropoulos, MEP (Greece) for his legislative efforts benefiting people with rare diseases.

Scientific Award:
Dr Luisa-María Botella Cubells (Spain) for her twenty years of research and development of treatments for Hereditary Haemorrhagic Telangiectasia and Von Hippel-Lindau Syndrome.

EURORDIS Members Award:
Orphan Diseases of Ukraine for their significant support to the rare disease community, especially during the ongoing conflict.

Young Patient Advocate Award:
Adrian Goretzki (Poland) for his leadership and support to the primary immunodeficiency's community and the Ukrainian rare disease community.

EURORDIS Volunteer Award:
Michela Onali (Italy) for her contributions to patient-centred research and advocacy.

EURORDIS Media Award:
"Red Sandra" (Belgium), for its compassionate and moving depiction of a Belgian family's experience with their daughter's diagnosis with metachromatic leukodystrophy (MLD).

Company Awards:

Innovation:
PTC Therapeutics, for developing transformative treatments for muscular dystrophies and pioneering gene therapy for AADC deficiency.

Patient Engagement:
Amryt Pharma, recognised for its patient-centric trial design leading to the approval of a treatment for severe epidermolysis bullosa.

Health Technology:
Mendelian, for its digital tool that aids in the detection of rare diseases within primary care.

Each award reflects the highest standards of leadership and achievement in the rare disease field, aiming to inspire continued progress and advocacy.

“DAVIDE’S DREAM”
SANFILIPPO SYNDROME / ITALY

STRATEGIC OBJECTIVE 02:

DELIVERING ON PRIORITY AREAS

EURORDIS has undertaken a range of activities to achieve this Strategic Objective, in alignment with its overarching strategy to advocate for, empower, and partner with people living with rare diseases. These activities fall within key priority areas: Healthcare Policy and Services, Research Policy and Activities, Data and Digital Health, Development and Access to Diagnostics and Therapies, and Social Policy and Services.

- HEALTHCARE POLICY AND SERVICES
- RESEARCH POLICY AND ACTIVITIES
- DATA AND DIGITAL HEALTH
- DEVELOPMENT AND ACCESS TO DIAGNOSTICS AND THERAPIES
- SOCIAL POLICY AND SERVICES

"DANCE ON WATER"
ARTHROGRYPOSIS MULTIPLEX CONGENITA (AMC) / CROATIA

HEALTHCARE POLICY AND SERVICES

EARLIER, FASTER AND MORE ACCURATE DIAGNOSIS: THE GOAL OF DIAGNOSIS WITHIN SIX MONTHS

HIGH-QUALITY NATIONAL AND EUROPEAN HEALTHCARE PATHWAYS, INCLUDING CROSS-BORDER HEALTHCARE: THE GOAL OF IMPROVING SURVIVAL BY 3 YEARS ON AVERAGE OVER 10 YEARS AND REDUCING THE MORTALITY OF CHILDREN UNDER 5 YEARS OF AGE BY ONE THIRD.

ADVOCATE

ADVOCATING TO IMPROVE ACCESS TO QUALITY RARE DISEASE DIAGNOSIS

Newborn screening

The Newborn Screening Working Group (NBS WG), comprising representatives from patient organisations, international screening societies, and federations focused on newborn screening (NBS), reviews policies and practices in NBS to develop recommendations for harmonised programme uptake across countries. This aims to maximise benefits and improve outcomes for babies born with rare diseases.

In 2023, EURORDIS continued to advocate for a harmonised European approach to NBS, as outlined in its position paper on the 11 Key Principles for NBS. Efforts included proposing the creation of an EU-level expert working group to design and implement these strategies, enhancing collaboration among Member States.

EURORDIS led the EURORDIS Newborn Screening Working Group (NBS-WG), moderating activities on research and policy and promoting the 11 Key Principles internationally. The NBS WG contributed to the Targeted Stakeholder Consultation by the European Commission for the EU4Health Work Programme 2024, suggesting the co-funding of a Joint Action on NBS to coordinate EU-wide actions and optimise Member State efforts, respecting national competences. This proposal was included in EURORDIS's consultation submission.

Additionally, EURORDIS joined the IRDIRC Newborn Screening Task Force and co-authored the article "Patient Advocacy in Action for Newborn Screening: Role of Patient Organisations in the Timely Diagnosis of Rare Diseases and Quality of Life."

Collaboratively, EURORDIS and the SMA NBS Alliances (including SMA Europe, Screen4Rare, ERN-RITA, EURO-NMD, ISNS, Screen4Care) submitted a joint statement on EU-level NBS cooperation to Health Commissioner Stella Kyriakides, meeting with her and her team on 29 September 2023.

To raise awareness, EURORDIS featured NBS topics in a RareOnAir podcast and published an article in Rare Revolution Magazine: Spotlight Edition on NBS.

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

The Rare Barometer programme conducts surveys to gather both qualitative and quantitative data on the experiences, needs, and expectations of people living with a rare disease. Managed by EURORDIS, the programme has successfully enlisted a panel of 20,000 individuals impacted by rare diseases who regularly participate in these surveys. The data collected is crucial for shaping EURORDIS' advocacy positions.

All studies from the Rare Barometer are translated into 27 languages, ensuring accessibility for a broad audience, and are distributed to rare disease patient organisations to support their work and increase the impact of the findings on policy and practice.

Rare Barometer Survey: The journey of rare disease patients to diagnosis

In 2022, EURORDIS initiated an analysis of **survey results concerning the diagnostic journeys** of people with rare diseases, concluding this work in 2023. Members were engaged through a webinar focused on utilising these survey results for their organisations, attracting 29 participants. Additionally, members received detailed results specific to their communities, including 24 dashboards (450 pages each) available in 27 languages, and a concise factsheet highlighting key results (1–2 pages).

These findings were further discussed at the EURORDIS Membership Meeting in May 2023. To extend the reach and impact of this study, the full European results were submitted for publication to the European Journal of Human Genetics in December 2023, with several EURORDIS members contributing as co-authors.

Rare Barometer Survey: Newborn Screening

The Newborn Screening survey was initiated in May through a webinar that outlined the survey's content, its application, and dissemination strategies. The 20,034 participants in the Rare Barometer panel were invited via email in their respective languages to participate; those who did not respond received four reminders. Of these, 3,984 participated in the survey, with 3,293 completing at least 80% of the questions, achieving a 16% response rate comparable to other Rare Barometer surveys. Additionally, outreach through social media, Facebook ads, and patient organisations reached 4,698 respondents, with 2,872 completing 80% of the questionnaire. The respondents represented 80 different countries and over 1,300 rare diseases.

To facilitate participation, rare disease patient organisations were provided with a communication toolkit in 27 languages, including a template email, suggested social media posts, and images. EURORDIS members had access to an online dashboard to monitor the daily distribution of the questionnaire within their community and view preliminary results. National alliances and European federations automatically received these dashboards, while other members could request them. European Reference Networks (coordinators, project managers, and patient representatives) were also given the opportunity to create and share specific dashboards with preliminary results in their networks. All dashboards were available in 24 languages.

Disseminating Results of Past Surveys

The results of previous surveys, such as "Share and Protect Our Health Data," have been presented on multiple occasions and are continually used by staff to inform advocacy positions and contributions at conferences and events.

EMPOWER

SUPPORTING PATIENT PARTNERSHIP WITHIN THE EUROPEAN REFERENCE NETWORKS (ERNS)

Since 2016, EURORDIS has significantly contributed to patient involvement in the European Reference Networks (ERNs) through the establishment of 24 European Patient Advocacy Groups (ePAGs). Each ePAG corresponds to one of the 24 ERNs, fostering collaboration among patient organisations, clinicians, experts, and researchers focused on specific rare or complex diseases or highly specialised interventions. Comprising over **300 appointed patient representatives**, including some elected in 2016 and others co-opted later, ePAG advocates are pivotal in connecting the networks with the broader rare disease patient community and promoting diverse patient perspectives within each ERN.

In 2023, EURORDIS continued to **enhance patient partnership within the ERNs**, supporting their involvement in both operational and strategic decision-making. This commitment aligns with the implementation of the Cross-Border Healthcare Directive (Article 12) and contributes towards the EU4Health Programme's objective of strengthening health systems (Article 3, point (a)), as detailed in Regulation (EU) 2021/522 Article 4, point (g).

EURORDIS's efforts in 2023 included empowering ePAGs to ensure effective and meaningful patient participation in the ERNs, alongside ongoing enhancement of methods and tools for patient engagement, thus driving continuous learning and improvement within the networks.

Activities to conduct patient engagement in the ERNs.

EURORDIS has been instrumental in promoting effective patient engagement within the European Reference Networks (ERNs) through its support and training of ePAG (European Patient Advocacy Groups) representatives. In 2023, EURORDIS led and managed several transversal working groups (WGs) aimed at enhancing the skills, knowledge, and effectiveness of patient representatives in collaborating with clinicians and other stakeholders within the ERNs:

- **ePAG Steering Committee**: This WG focuses on strategic aspects of structuring and improving patient involvement in the ERNs, holding four calls in 2023.

- **Connecting Patients with ERNs Working Group**: Aims to facilitate collaboration between ePAG advocates and Rare Diseases National Alliances, developing communication resources to better disseminate information on ERNs at the national level. Activities included four calls and one webinar on National RD Networks and ERN Coordination Hubs in 2023.

- **ePAG Clinical Practice Guidelines Working Group**: Supports patient partnership in developing and implementing clinical practice guidelines (CPG) and other clinical decision support tools (CDST), with five calls in 2023.
- **ePAG Research and Registries Working Group**: Focuses on disseminating information about research activities and supporting patient involvement in clinical research, with three calls in 2023.
- **ePAG AMEQUIS Task Force**: This WG aids patient involvement in the ERNs' monitoring, evaluation, and quality improvement system. Activities included five calls and one webinar on good practice exchange and patient interviews in the ERN evaluation process in 2023.
- **Patient Partnership Working Group**: Enhances collaboration with ERN project managers and exchanges information on tools and processes for patient involvement, with three calls in 2023.

These activities are documented and summarised in a figure that outlines the main milestones of each working group.

Developing Tools and Resources for Patient Engagement

In 2023, EURORDIS developed the Patient Partnership Framework for the ERNs and curated a variety of resources to support patient engagement, including survey development. These resources will be accessible through an online repository, continuously updated, with a release planned for March 2024.

Additionally, resources supporting patient involvement in clinical practice guideline development were curated and reorganised. This included the creation of short videos explaining in simple terms how to facilitate patient involvement across various stages of guideline development, enhancing accessibility and understanding.

PARTNER

H-CARE

The H-CARE project, initiated in 2019 by ERNs ERKNet, eUROGEN, GENTURIS, and LUNG with support from the EURORDIS Rare Barometer Programme, focuses on developing a feedback mechanism through Patient Reported Experience Measures (PREMs) to assess the healthcare experiences of those living with rare diseases. A pilot survey conducted in 2020 highlighted the need to develop and validate PREMs that could effectively measure healthcare experiences across different diseases and countries.

Progress in 2023:

- A comprehensive literature review was undertaken in partnership with Genetic Alliance UK and Heidelberg University to collate existing data on healthcare experiences from empirical studies. This involved screening 4,766 studies, assessing 283 full-texts for eligibility, and including 61 papers in the final analysis. Data was extracted and harmonised by three reviewers.

- A preliminary manuscript was shared with partners in December 2023, with plans to submit the final version to the Orphanet Journal of Rare Diseases in early 2024.

Future Phases

The project is set to continue with several key phases over the coming years:

- 2024: Conducting focus groups with patients, family members, and experts to gather in-depth insights.

- 2024: Designing a comprehensive questionnaire based on the findings from the focus groups.

- 2025: Launching the questionnaire online in 25 languages and onsite in English-speaking ERN centres.

- 2025-26: Finalising the development of the PREM.

In support of these efforts, EURORDIS joined the Rare Disease Research Catalyst Consortium (RDCat), funded by the Irish Health Research Board, to enhance patient involvement in rare disease research and policy. Collaborating with University College Dublin and RD Ireland, EURORDIS will conduct face-to-face focus groups in Ireland and deploy the H-CARE questionnaire across five Irish ERN centres, ensuring robust patient engagement and comprehensive data collection.

Joint Action on the integration of European Reference Networks into national health systems (JARDIN, January 2024–2027, DG Sante, Subcontractor)

The Joint Action aims to develop structures enabling Member States to link their national health systems with ERNs, covering areas such as care pathways, data interoperability, national dissemination, quality assurance, centres of expertise, national networks, referral systems, and the revision of national rare disease plans.
The project proposal was developed and submitted to the European Commission for evaluation, with the project kick-off scheduled for March 2024. As a subcontractor, EURORDIS will contribute to three work packages, focusing on:

- Refining care pathway methodologies and developing models for 10 diseases.

- Developing guidelines for case managers.

- Addressing barriers to data sharing with ERN registries and enhancing data integration between national health systems and ERNs.

- Developing emergency response recommendations tailored to the needs of those with rare diseases.
- Designing models for patient involvement in clinical networks.
- Formulating strategies for national undiagnosed pathways and their integration with ERNs.
- Developing an ERN-overarching expert panel for undiagnosed cases requiring interdisciplinary input.
- Establishing patient organisations for undiagnosed patients in Member States.

National Alliances such as UNIAMO (Italy), RD Ireland, ACHSE (Germany), Greek NA, and Pro-Rare Austria are included in the proposal, receiving budgets for specific tasks. Patient representatives involved in care pathway development will also be compensated by the work package lead.

Collaborative project on diagnostic characterisation of rare diseases (Solve-RD, January 2018-March 2024, Horizon 2020, Partner)

Solve-RD is a research initiative funded by the European Commission, aimed at discovering the molecular causes of many rare diseases that remain undiagnosed. This project aligns with the ambitious goals of the International Rare Diseases Research Consortium (IRDiRC) to develop diagnostic tests for most rare diseases.

To date, Solve-RD has amassed 21,422 datasets, including phenotype and exome/genome sequencing data, for re-analysis. This extensive effort has involved systematic research-driven re-analysis and ad hoc expert reviews for about 10,000 undiagnosed individuals from over 6,000 families, achieving an overall diagnostic yield of 12.6%. Of these, systematic re-analysis has established genetic diagnoses in 8.5% of the cases.

EURORDIS serves on the Steering Committee for the work package focused on disseminating the results of Solve-RD and engaging stakeholders effectively. Additionally, EURORDIS leads the Community Engagement Task Force (CETF) within Solve-RD, which strives to foster a collaborative community among patients, scientists, genetic counsellors, and clinicians dedicated to improving diagnosis and care for ultra-rare diseases and supporting the undiagnosed community.

The EURORDIS-led CETF has developed an infographic detailing the patient journey to diagnosis, illustrating the diagnostic challenges faced daily and highlighting resources from CETF member organisations to aid patients. This infographic has been translated into 28 languages, broadening its accessibility.

In 2023, EURORDIS continued its active participation in the project by presenting at the Solve-RD Public Symposium: The Impact of Solve-RD on Research & Care of Rare Disease Patients. Moreover, EURORDIS contributed to the development of a policy paper by the Solve-RD consortium, aimed at enhancing rare disease diagnosis and fostering collaborative solutions across Europe.

SOLVE-RD FINAL MEETING

RESEARCH POLICY AND ACTIVITIES 02^{05}

> RESEARCH AND KNOWLEDGE DEVELOPMENT THAT ARE INNOVATIVE AND LED BY THE NEEDS OF PEOPLE LIVING WITH A RARE DISEASE. ”


“HELPING DADDY”
ORNITHINE TRANSCARBAMYLASE DEFICIENCY,
A UREA CYCLE DISORDER / USA

ADVOCATE

Rare Disease Moonshot

The Rare Disease Moonshot is a coalition involving seven organisations: Critical Path Institute (C-Path), European Infrastructure for Translational Medicine (EATRIS), European Clinical Research Infrastructure Network (ECRIN), European Federation of Pharmaceutical Industries and Associations (EFPIA), European Confederation of Pharmaceutical Entrepreneurs (EUCOPE), EuropaBio, and EURORDIS-Rare Diseases Europe. This initiative accelerates scientific discovery and drug development for rare and paediatric diseases without current treatments by pooling expertise, reducing research fragmentation, and fostering collaboration.

Launched in December 2022, the initiative focuses on enhancing public-private partnerships in research. In 2023, EURORDIS actively participated in shaping the initiative through bimonthly or monthly calls, identifying three key action areas:

- Optimising the translational research ecosystem to accelerate scientific translation.
- Modernising clinical trials to adapt to very small patient populations.
- Enhancing infrastructure to speed up diagnosis and treatment processes.

EURORDIS led the drafting of the Diagnostic Research Needs Recommendations Paper, coordinating two workshops with industry stakeholders and iterative review rounds involving over 100 participants. This paper was presented at the European Respiratory Therapy Congress (ERTC) in Barcelona in October and the World Orphan Drug Congress (WODC) Europe in November 2023. It outlines the value of public-private partnerships in optimising diagnostic research for rare diseases and provides guidance on establishing such collaborations.

Furthermore, EURORDIS contributed to recommendations on translational research and clinical trials. To disseminate the initiative’s work, several publications have been issued, including an upcoming article in the DIA Global Forum’s January 2024 edition. Ongoing consultations with other rare disease initiatives aim to prevent duplication and promote the necessity for more public-private partnerships in rare disease research.

EMPOWER

EURORDIS Open Academy

The EURORDIS Open Academy equips patient advocates with the skills and knowledge necessary for effective engagement in patient advocacy roles at both European and national levels for rare diseases.

The Open Academy's offerings include in-depth schools for EURORDIS members, Meetups and Masterclasses for alumni, and open access e-learning courses available to all on the Open Academy website. By the end of 2023, the platform had attracted over 3,138 registered users from 119 countries.

2023 Training Highlights:

School on Medicines Research & Development:

- Held from 19 to 23 June 2023 in Barcelona.
- Pre-training from January to June 2023 included e-learning modules and three webinars.
- 31 trainees and 7 researchers from 25 countries covered topics like market access, regulatory frameworks, ethics, and pharmacovigilance.

School on Scientific Innovation and Translational Research:

- Concurrently held from 19 to 23 June 2023 in Barcelona.
- Pre-training involved e-learning modules and webinars.
- 27 patient representatives from 14 countries explored genetics, genome editing, translational research, drug repurposing, and patient participation in research projects.

Both programmes aim to empower participants to effectively engage with researchers, policymakers, and industry leaders.

Additional training: International Course on Leadership and Communication Skills:

- Conducted online on 26–27 October 2023, hosted by GUMED in Poland.
- Focused on developing presentation, negotiation, and leadership skills through plenary presentations and role-playing.
- In 2023, the Open Academy also organised 6 Meetups and 2 Masterclasses for alumni, addressing

topics such as European Pharmaceutical Legislation, research initiation, digital rights, scientific committee formation, survey design, N-of-1 trials, and patient-reported outcomes.

PARTNER

TRANSLATIONAL RESEARCH

ERDERA (European Rare Diseases Research Alliance and Partnership, September 2024-August 2031, Horizon Europe, Partner)[1]

ERDERA, set to launch in September 2024 and run until August 2031 under Horizon Europe, represents a major collaboration involving EURORDIS and key European rare disease research stakeholders. EURORDIS played a pivotal role in co-designing ERDERA, contributing to the Strategic Research and Innovation Agenda (SRIA) and the project proposal submitted in September 2023.

The SRIA, drafted between December 2022 and January 2023 by a task force and a core group, included substantial input from EURORDIS, particularly in authoring the strategic objective focusing on empowering people living with rare diseases as equal partners. Following the drafting phase, the strategy document underwent a consultation phase in February 2023, promoted by EURORDIS through various channels and garnering over 100 pieces of feedback, many advocating for enhanced patient involvement.

Throughout the partnership's formation, EURORDIS actively participated in writing meetings with hundreds of potential partners and took a lead role in the Education & Training workstream, which includes tasks dedicated to patient training. In collaboration with patient groups like Genetic Alliance UK, World Duchenne Organisation, and AFM-Téléthon, EURORDIS ensured coordinated patient representation across all ERDERA workstreams.

The ERDERA proposal was submitted for Horizon Europe funding as a co-funded partnership between the European Commission, European Member States, and additional partners. The project involves 36 countries and 171 organisations as full partners, with an overall estimated budget of €385.5 million. The project, anticipated to last seven years, includes associated partners and a mix of in-kind and in-cash contributions, with a European Commission contribution request of €160 million across two instalments.

EURORDIS is involved in 10 of the 24 work packages and co-leads on Education and Training. It also leads the Patient and Public Involvement and Engagement

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[1] **Rare Disease Partnership,** a major research programme meant to start by late 2024.

(PPIE) Group and is involved in significant research activities, such as estimating the socioeconomic impact of rare diseases and identifying disease indications needing Advanced Therapy Medicinal Products (ATMPs). EURORDIS expects to dedicate three to four full-time equivalents to the project, collaborating with five patient organisations, including AFM-Téléthon, World Duchenne Organisation, Genetic Alliance UK, RDI, and the Thalassaemia International Foundation, among others.

European Joint Programme on Rare Diseases (EJP RD, January 2019 – August 2024, Horizon 2020, Partner)

The EJP RD unites over 130 institutions from 35 countries to foster a sustainable ecosystem that enhances the link between research, care, and medical innovation. The programme focuses on optimising existing funded tools and programmes by scaling them, linking them, and adapting them to end-user needs through real-setting implementation tests.

2023 highlights:

The **Medicines Research & Development (MRD) and Scientific Innovation and Translational Research (SITR)** schools, supported by EJP RD funding, saw participation from 38 individuals in the MRD and 27 in the SITR.

EURORDIS supported the organisation of the EJP RD paediatric training in Barcelona, Spain, in June 2023, held concurrently with the EURORDIS schools to facilitate faculty participation. EURORDIS co-organised the EJP RD Leadership School in Gdańsk, Poland, in October 2023, and participated as a trainer at the EJP RD International Summer School in September 2023.

MOOCs and online learning:

The MOOC "Diagnosing Rare Diseases: from the Clinic to Research and back" saw two facilitation windows in 2023, with over 5,800 learners enrolled from 140 countries since April 2021. EURORDIS plays a key role as an active mentor.

The "From Lab to Clinic: Translational Research for Rare Diseases" MOOC, launched in October 2022, has attracted 2,030 learners. It was co-developed by EURORDIS and partners including ERN EURO-NMD and Leiden University Medical Centre.

A new MOOC on “Rare Diseases Research Data: Ethical and Legal Considerations” is being finalised for launch in spring 2024, in collaboration with ERN EpiCARE, Fondazione Gianni Benzi, and the Foundation for Rare Diseases, with EURORDIS recruiting contributors and beta testers.

EURORDIS continues to offer dedicated support to patient organisations and representatives in applying for EJP RD calls, including joint transnational calls and networking support schemes.

Throughout the year, EURORDIS has maintained co-leadership of all EJP RD Pillar 3 training and capacity-building activities, delivering presentations at various high-level meetings, including those with the European Commission, General Assembly, and Executive Committee, as well as during the ERDERA development phase to highlight the impact of the EJP RD training programme

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

ERICA aims to enhance the individual European Reference Networks (ERNs) by creating an integrated platform for their collective research and innovation capacities. The consortium strives to establish new competitive networks within and between ERNs, implement effective data collection strategies, improve patient involvement, enhance the quality and impact of clinical trials, and raise awareness of the ERNs' innovation potential. ERICA's goal is to strengthen the research and innovation capacity of ERNs by integrating their activities and collaborating with European research infrastructures to maximise impact and innovation.

In 2023, EURORDIS initiated a working group comprising ERN clinicians and ePAG advocates with clinical trial experience to co-develop a Patient Partnership framework for clinical trials across the ERNs. This initiative kicked off in October 2023 with subsequent calls to pinpoint key elements for the framework and establish an outline. The drafting, final validation, and dissemination of this framework are scheduled for completion in 2024, with a delivery deadline of February 2025.

"RAY OF LIGHT"
ARTHROGRYPOSIS MULTIPLEX CONGENITA / CROATIA

Undiagnosed Diseases Network International

The Undiagnosed Diseases Network International (UDNI) unites clinicians, researchers, genetic counsellors, and medical professionals globally to tackle complex and unresolved cases. Central to UDNI's effectiveness is the participation of patients and patient representatives, who contribute insights on maintaining a patient-centric focus in all operations.

EURORDIS has been actively involved in UDNI, which was established in 2014 to meet the global needs of undiagnosed patients. Together with NORD and the Wilhelm Foundation, EURORDIS helped develop a patient engagement membership that was endorsed by UDNI's Board.

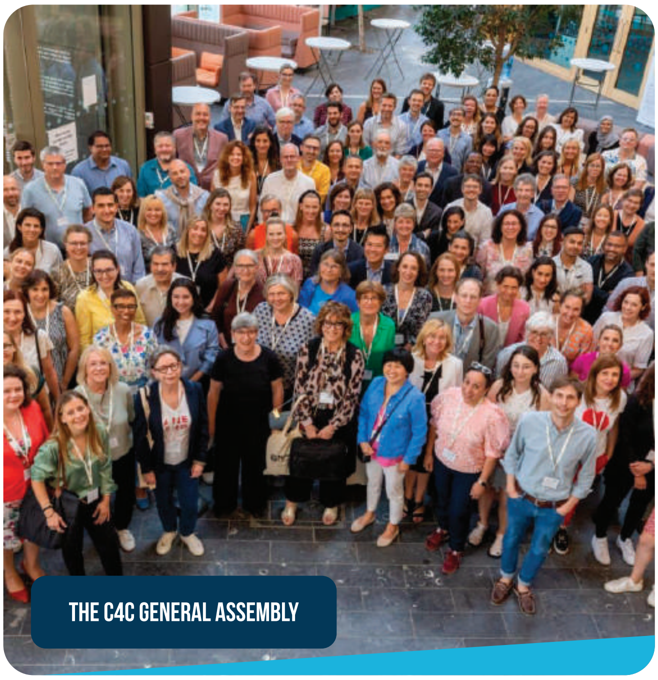
In 2023, EURORDIS, along with NORD and the Wilhelm Foundation, continued to lead the UDNI Patient Engagement Group, which included 22 member patient organisations from across all continents by year-end. EURORDIS played a key role in fostering international collaboration for Undiagnosed Day and delivered a presentation at the related webinar hosted by the Wilhelm Foundation.

Additionally, EURORDIS co-organised the inaugural pre-conference patient session held on October 21, the day before the UDNI Conference. This meeting focused on challenges related to undiagnosed diseases, covering topics such as diagnosis, newborn screening, diversity and inclusion, and the psychosocial impacts of having or lacking a diagnosis. Attended by over 50 participants from 23 patient organisations, the session saw contributions from EURORDIS, Wilhelm Foundation, National Organization for Rare Disorders, and the SCN2A Georgian Association on a range of pertinent issues.

PAEDIATRIC CLINICAL TRIALS

Conect4Children (C4C, May 2018 – April 2024, extended until April 2025, IMI 2, Partner)

Conect4Children (c4c) is a pan-European research network that includes 35 academic and 10 industry partners from 20 European countries, along with over 50 third parties and approximately 500 affiliated partners. The network brings together pharmaceutical companies, national paediatric networks, EU multinational subspecialty networks, major patient advocacy groups, children's hospitals, and other public research organisations. Its aim is to facilitate the development of new drugs and therapies for the paediatric population.


THE C4C GENERAL ASSEMBLY

Since 2019, EURORDIS has been an active member of the c4c Multi-stakeholder Programme Committee. EURORDIS has been deeply involved in organising c4c Multi-Stakeholder Meetings (MSM) on various paediatric conditions, including inflammatory bowel disease (2020), atopic dermatitis (2021), type 1 diabetes (2022), perinatal asphyxia (2023), and paediatric irritability (scheduled for March 2024). These meetings incorporate input from patient experts including young patients, parents/carers, and patient advocates, and focus on drafting peer-reviewed scientific publications on the MSM outcomes.

EURORDIS has also led workshops for patient organisations on paediatric drug development and follow-up webinars for patient experts on c4c activities, which are now available on the c4c website. Furthermore, EURORDIS is involved in drafting a white paper on paediatric patient engagement, supporting this effort with a workshop involving patient involvement experts planned for the end of 2024.

Additionally, EURORDIS contributes to c4c's work on data quality standards and is part of an education and training programme through its membership on the Education Board.

DATA AND DIGITAL HEALTH

03 05

> OPTIMISED DATA AND HEALTH DIGITAL TECHNOLOGIES FOR THE BENEFIT OF PEOPLE LIVING WITH A RARE DISEASE AND SOCIETY AT LARGE. ”

ADVOCATE

ADVOCATING FOR A EUROPEAN HEALTH DATA SPACE

EURORDIS actively advocates for the secure sharing and use of health data at the EU level, a vital initiative for the rare disease community, where data are often fragmented, and patient populations dispersed. EURORDIS plays a key role in highlighting the specific challenges and needs related to health data sharing for those with rare diseases.

Contributions to the European Health Data Space (EHDS):

- EURORDIS has injected the rare disease community's expectations into the EHDS proposal, based on a Position Paper adopted in 2022. Meetings with EU policymakers, especially MEPs involved in the ENVI and LIBE committees, have focused on the unique hurdles faced by individuals and families with rare diseases, underscoring the necessity for comprehensive health data and a strong commitment to protecting individual rights.

- Through participation in public forums, consultations, and discussions on EHDS, and as a member of the EC eHealth stakeholder group and the HealthData@EU Pilot project, EURORDIS offers valuable insights into the rare diseases' specific requirements.

EURORDIS's efforts aim to ensure inclusivity and equity in Europe's health data landscape, contributing to an enhanced health data ecosystem for the rare disease community.

EMPOWER

EURORDIS Digital Advisory Group (DAG)

The DAG played a crucial role in consultations for the FACILITATE project. To enhance the group's response to project needs, a call for volunteers in April 2023 led to three new members joining. The end of 2023 also marked the completion of the first mandate for the DAG's ten expert patient volunteers, who have diverse backgrounds and have significantly contributed to projects such as FACILITATE, Screen4Care, and CHEERS-AI. EURORDIS held nine consultations with the DAG, comprising four online and five written consultations. The DAG's input to CHEERS-AI was highly valued, and their contributions will be acknowledged in the project's published version.

PARTNER

FACILITATE (January 2022 – December 2025, IMI2, Project Partner)

FACILITATE is a four-year project under the Innovative Health Initiative (IHI) focused on returning clinical trial data to participants and developing a framework for the secondary use of such data. EURORDIS plays a pivotal role in ensuring the project addresses patient needs across ethical, legal, and technical dimensions. In 2023, the patient expert group DAG+ was established to enhance the patient centricity of the project, consisting of Digital Advisory Group members and ten other volunteers from non-rare disorder backgrounds. This group, coordinated by EURORDIS, contributed to discussions on ethical frameworks, data return and reuse, and patient consent processes, with their feedback influencing the FACILITATE semi-annual meeting discussions in June 2023.

Together4RD

Together4RD is a collaborative effort aimed at addressing the unmet medical needs of the 95% of rare diseases without dedicated treatments by fostering cooperation between European Reference Networks (ERNs) and the industry. In 2023, EURORDIS actively supported Together4RD by contributing to data collection and research efforts to advance new treatments for rare diseases. As a member of the Together4RD Steering Group, EURORDIS co-developed a position statement on ERN-industry collaboration, provided speakers for events and webinars, and produced communication videos to enhance awareness. Additionally, EURORDIS involved the ePAG Research & Registry Task Force in reviewing the existing ERN Board of Members Statement on ERN-Industry Collaboration, ensuring robust stakeholder feedback.

DEVELOPMENT AND ACCESS TO DIAGNOSTICS AND THERAPIES

04 05

> DEVELOPMENT AND AVAILABILITY, ACCESSIBILITY, AFFORDABILITY OF TREATMENTS, PARTICULARLY TRANSFORMATIVE OR CURATIVE THERAPIES – THE GOAL OF 1,000 NEW THERAPIES WITHIN 10 YEARS!

ADVOCATE

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 2023, EURORDIS played a crucial role in the EU debate on revising General Pharmaceutical Legislation, focusing on regulations for rare diseases and paediatrics. Key actions included:

- Analysing European Commission proposals from April 2023 and 3,000 amendments by the European Parliament's ENVI and ITRE committees.
- Engaging in consultations with EURORDIS staff and members to reflect their views on the proposed legislation, with findings shared online and via social media.
- Conducting over 50 meetings with European Commission, Parliament, and Council representatives to convey the rare disease community's expectations and concerns.
- Monitoring EU and national debates on pharmaceutical legislation, supported by National Alliances.
- Organising member training, especially for National Alliances, on the Commission's proposal and parliamentary amendments.

ACCELERATING CLINICAL TRIALS IN THE EU (ACT EU)

The ACT EU initiative, supported by the European Commission, HMA, and EMA, aims to bolster the EU as a leading hub for innovative clinical research. In 2023, EURORDIS joined the ACT EU Stakeholder Platform as an Advisor, influencing stakeholder engagement strategies and agenda setting. EURORDIS participated in workshops on the International Council on Harmonisation guidelines revision and methodologies to advance smarter clinical trials in the EU. Discussions covered complex trials, paediatric trials, pragmatic trials, digital endpoints, patient centricity, and decentralised trials, involving experts from patient organisations, regulatory bodies, HTA agencies, industry, and academia.

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ADVOCACY FOR EU COOPERATION ON HEALTH TECHNOLOGY ASSESSMENT (HTA)

EURORDIS continued advocating for the EU Cooperation on HTA through its HTA Task Force. The Task Force focused on treatment assessment, pricing, and reimbursement across Europe, advising on HTA methodologies and patient engagement. This effort included sharing insights on national HTA systems, expert opinions, and connecting with the scientific community, enhancing EURORDIS's contributions to EU-level HTA cooperation.

ADVOCATING FOR IMPROVED ACCESS TO TREATMENT FOR RARE DISEASES

EURORDIS tirelessly advocates for enhanced access to authorised therapies for rare diseases across the EU. Notably, the European Commission's EU4Health programme for 2024/2025 has introduced a call for Member States to support structured cooperation in pricing and reimbursement policies. In 2023, EURORDIS escalated its advocacy to the WHO Europe level, participating in the Novel Medicines Platform (WHO NMP) Working Group on solidarity and sustainability. Here, EURORDIS emphasises the need for a fund dedicated to evidence generation and a focus on the rarest diseases. Additionally, through advocacy related to the Pharmaceutical Package, EURORDIS proposed amendments supporting the centralised European procurement for centrally authorised Orphan Medicinal Products (OMPs) and underscored the benefits of early and structured dialogues via the Mechanism of Coordinated Action (MoCA). EURORDIS continues its involvement in established initiatives like the Expert Group on Orphan Drug Incentives and RWE4Decisions, and participation in European Commission-led and EU Council Presidency high-level conferences.

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

CUPs are critical for providing innovative medicines to patients, especially those in urgent need. Since adopting a position on compassionate use for medicines used in rare diseases in 2016, EURORDIS has actively participated in efforts to improve CUPs in Europe. In 2022, EURORDIS contributed to the European Commission's proposal for pharmaceutical legislation revision, advocating for an extension of Article 83 of Regulation 726/2004 to either adopt a European scheme or facilitate an Early Access programme akin to France's, which is among the most effective in the EU. Highlighting the urgency for compassionate use in emergency preparedness contexts, EURORDIS pointed out that over half of Member States lack a ready scheme for CUP implementation following EMA recommendations. While the European Commission proposes creating a European registry for all Member States' authorised CUPs, EURORDIS, at the World Orphan Drug Congress on 31 October, argued for the necessity of further initiatives to address these challenges comprehensively.

THE WORLD ORPHAN DRUG CONGRESS EUROPE 2023

SCIENTIFIC COMMITTEES AND WORKING PARTIES OF THE EUROPEAN MEDICINES AGENCY

EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

EURORDIS holds and has held representation in key European Medicines Agency (EMA) Committees and Working Parties, including the Committee for Orphan Medicinal Products (COMP), Paediatric Committee (PDCO), Committee for Advanced Therapies (CAT), Patients' and Consumers' Working Party (PCWP), Pharmacovigilance and Risk Assessment Committee (PRAC), Scientific Advice Working Party (SAWP), and the Committee on Herbal Medicinal Products (HMPC).

EURORDIS Therapeutic Action Group (TAG) Coordination:
The TAG, consisting of 13 members from diverse EMA committees, conducted nine monthly calls in 2023 to discuss the EU pharmaceutical legislation, patient data generation, and patient involvement at the EMA. TAG members were invited to attend the Black Pearl Awards and participated in various EURORDIS meetings and workshops.

EURORDIS plays a pivotal role in supporting rare disease patients' participation in European Medicines Agency (EMA) activities and scientific committees, ensuring the representation and involvement of patient perspectives in critical discussions and decisions.

Committee for Orphan Medicinal Products (COMP):
EURORDIS supports three rare disease patient representatives appointed to the COMP, with a EURORDIS expert attending monthly meetings and the EMA Strategic Learning Review Meetings (SLRM) in Uppsala (May 2023) and Madrid (October 2023). In Uppsala, EURORDIS facilitated a session on Major Contribution to Patient Care (MCPC) and in Madrid, collaborated with the Paediatric Committee (PDCO) on the agenda, providing speakers and presenting in the meeting. EURORDIS also backed three patient representatives for COMP appointments, including proposing a candidate for Civil Society representation.

Committee for Advanced Therapies (CAT):
Support for one rare disease patient representative (alternate) appointed in July 2022 to the CAT, participating in monthly meetings and attending the EMA SLRM.

EURORDIS supported two rare disease patient representatives (member and alternate) until July 2023, and from August 2023, supports four new patient representatives (two members, two alternates) for the Paediatric Committee (PDCO), facilitating their involvement in monthly meetings and the SLRM in Uppsala. Letters of support were issued for two patient representatives, with one endorsed as a EURORDIS candidate and appointed as an alternate.

Pharmacovigilance Risk Assessment Committee (PRAC):
EURORDIS supports two rare disease patient representatives (member and alternate) in PRAC, facilitating their participation in monthly meetings and the EMA Strategic Learning Review Meetings since June 2022.

Scientific Advice Working Party (SAWP):
EURORDIS played a key role in identifying, mentoring, and supporting patient experts for SAWP activities. In 2023, 122 dossiers were reviewed, with patient input required for 14, leading to the involvement of 12 patients (86%) in scientific advice and related activities.

Committee for Medicinal Products for Human Use (CHMP):
EURORDIS continued its involvement in CHMP's Early Contact with patient organisations in 2023, contributing to 8 product consultations and submitting 7 memos based on interviews with 23 individuals from 11 countries. EURORDIS also proposed more systematic patient involvement in CHMP's medicine benefit/risk evaluations.

Management Board (MB):
A EURORDIS staff member, representing patient organisations, participated in four plenary and two extraordinary MB meetings, contributing to consultations and the Annual Activity Report 2022 assessment. EURORDIS also played a leading role in the newly established Management Board Audits and Risks Group (MBARG), with an election as its Chair.

Patients' and Consumers' Working Party (PCWP):
As a PCWP member, EURORDIS ensures patient and consumer perspectives are considered in EMA discussions. François Houÿez and Russell Wheeler (Leber Hereditary Optic Neuropath UK Society) serve as alternate members, with EURORDIS actively participating in all 2023meetings. EURORDIS significantly contributed to the Patients' and Consumers Working Party (PCWP) at the EMA in 2023, with key activities including:

- ACT-EU Multi-stakeholder Platform: Achieved membership and an advisory role, enhancing stakeholder collaboration in clinical trials within the EU.

- DG SANTE/HaDEA Interview: Participated in discussions on hospital exemptions, influencing policy on access to treatments.

- CH E6 (R3) Guideline: Played a pivotal role in forming a common PCWP and Health Care Professionals Working Party (HCPWP) stance for the public consultation on the draft guideline on their opinion on the concept of High

High Unmet medical Needs and a method proposed by the NEEDS project.

Clinical Trials:
Contributed to the EMA consultation on the Reflection Paper on single-arm trials as pivotal evidence in marketing authorisation.

Followed up on Decentralised Clinical Trials (implementation) and participated in discussions on EMA Transparency rules for the Clinical Trial Information System (CTIS), emphasising the importance of maintaining clinical investigation site contact details public.

Shortages Workshop:
Participated in the HMA/EMA multi-stakeholder workshop on medicine shortages (1–2 March 2023), addressing supply chain issues.

Raw Data Network Advisory Group:
Contributed to the oversight and monitoring of pilot projects examining the use of raw data in EMA decisions. With up to 10 pilots considered and an interim report forthcoming, EURORDIS's involvement ensures the rare disease community's needs are represented in these discussions.

These efforts highlight EURORDIS's commitment to enhancing patient engagement and input in the regulatory processes, clinical trial design, and medicine accessibility, aiming to improve outcomes for the rare disease community.

ADDITIONAL ACTIVITIES

Review of EMA documents for public information

EURORDIS plays a crucial role in reviewing EMA-produced public information, including Medicine Overviews, Package Leaflets, and Q&A documents, especially for rare diseases not represented by other groups. This ensures information is clear and accessible to the intended audience.

Monthly Therapeutic Report

In 2023, 9 Therapeutic Reports were produced and distributed to around 500 stakeholders, including patients, regulators, and clinicians, with summaries also included in the EURORDIS Member news monthly.

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

The Task Force, comprising eight volunteers from member organisations across the EU, advises on assessment, pricing, and reimbursement of treatments. They share

“WALKING ON WATER”
NMO-SD / USA

They share expertise on national HTA systems, contribute to policy debates, and engage in public consultations. In 2023, discussions covered the use of real-world evidence, drug development attractiveness in the EU, fair pricing, and research and development costs for orphan drugs. A face-to-face meeting occurred in Paris in October.

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

EURORDIS supported two individuals with rare diseases in MoCA meetings in 2023, contributing to awareness and specific actions like updating the MoCA webpage and disseminating key papers on improving access to orphan medicinal products in Europe. MoCA was also presented at the WHO Novel Medicines Platform meeting in Prague.

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

The DITA TF, trained through EURORDIS Open Academy Schools, supports EURORDIS’s involvement in EMA committees by consulting on key papers and advising on therapeutic development and medicine evaluation. In 2023, the Task Force met ten times, addressing topics like the Pharma Package revision, single-arm trials, and patient engagement at the EMA, contributing to several critical consultations including the Pharma Package, concept of High Unmet Medical Needs, single-arm trials, and changes in clinical trials including decentralised trials.

The Task Force contributed to the following consultations:

- The Pharma Package Revision, both the Directive and the Regulation,

- Their opinion on the concept of High Unmet medical Needs and a method proposed by the NEEDS project,
- EMA consultation on Single-Arm trials,

- ACT-EU and changes patients would like to see happening in clinical trials,
- Decentralised Trials.

PARTNER

EUCAPA – HTA training (EU4Health, March 2023-March 2025, Project Lead)

EUCAPA aims to train patients and patient representatives in health technology assessment (HTA) ahead of the Regulation on HTA (EU) 2021/2282 (HTAR) taking effect in January 2025. This project will equip them with the knowledge and skills needed for active participation in HTA decision-making processes, advocating for equitable access to innovative treatments for rare diseases. It focuses on ensuring patients and patient organisations are well-versed in HTA processes to meaningfully engage in both scientific consultations and assessments as anticipated by the HTAR.

EURORDIS leads the EUCAPA project, coordinating WP1 for project management and WP2 for Training and Content Development. Responsibilities in WP1 extend to data governance, quality management, and ethics. WP2 involves developing curricula and content for introductory, fast-track, and extended training through co-development and co-creation among project partners.

The project commenced in March 2023 with a kick-off meeting in Hall in Tirol, Austria. Training programmes began in November 2023 with introductory sessions, with further Fast-Track Training scheduled for February 2024 and Extended Training for March 2024. Additional training programmes are planned throughout the project's duration.

More Effectively Using Registries to support Patient-centred Regulatory and HTA decision-making'(More-EUROPA, 2023–2028, Horizon Europe, Project Partner)

More-EUROPA aims to enhance the use of **Real-World Evidence (RWE)** by developing, implementing, and establishing standards and methods that meet the data and evidentiary requirements of regulatory authorities and HTA bodies in Europe. This initiative focuses on improving the development, registration, and assessment of medicinal products through efficient RWE utilisation.

In 2023, EURORDIS contributed to the development of a training programme led by the European Multiple Sclerosis Platform (EMSP). Additional project activities included identifying patient registries established by European Reference Networks, compiling a contact list of patient advocates involved in patient registry governance, and engaging in discussions with the French regulatory authority (ANSM) and the University of York (HTx project). These discussions centred on applying new analytical methods for data analysis from registries, particularly for medicine repurposing, in collaboration with France's "Filières Maladies Rares."

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

HTx aims to develop a framework for Next Generation Health Technology Assessment (HTA) to support patient-centred, societally oriented, and real-time decision-making on health technologies across Europe. This includes determining which patients could benefit most from high-cost new therapies, identifying the best treatment sequences or combinations, and determining optimal therapeutic strategies for specific populations or subgroups.

A key aspect of HTx is assessing the potential of real-world evidence in addressing these questions, with AI and machine learning being pivotal in cost-effectiveness predictions. The project's new methods are being tested across four disease areas: head and neck cancer, Diabetes 1 and 2, Multiple Sclerosis, and Myelodysplastic Syndrome.

EURORDIS, as a consortium member, is dedicated to keeping the patient community informed about the project's progress and potential, and training future patient advocates on HTx's core concepts and findings.

HTx
Next Generation Health Technology Assessment

In 2023, EURORDIS released videos within the HTx toolkit to educate patients on the project and related topics through recorded interviews, webinars, and presentations. These covered the development and use of predictive models, selection of patient-reported outcomes, challenges faced by HTA agencies in assessing COVID-19 technologies, and policies on real-world evidence data. Additionally, EURORDIS is leading a qualitative study on the acceptability and barriers to incorporating cost considerations into clinical shared decision-making. EURORDIS also contributed to a forthcoming publication on expanding the Consolidated Health Economic Evaluation Reporting Standards (CHEERS) to include AI evaluations (CHEERS-AI).

GetReal Institute

Building on the success of two Innovative Medicines Initiative (IMI) projects – GetReal and The GetReal Initiative – the GetReal Institute (GRI) aims to foster the sustainable development and adoption of tools, methods, and best practices for generating and using real-world evidence (RWE) to improve healthcare decision-making.

In 2022, EURORDIS joined GRI as a co-founder and Board member, focusing on reducing barriers to the use of secondary data, bridging the gap between Randomised Control Trials (RCT) and RWE, and meeting the evidence needs of healthcare decision-makers.

In 2023, GRI outlined its objectives and deliverables:

- Establish as a key forum for sharing understanding and prioritising critical opportunities and challenges in RWE.
- Undertake case studies, demonstration projects, and publish research to address scientific and operational uncertainties.
- Develop trusted resources and guidelines for best practices in RWE, filling existing gaps.
- Offer skill development training through partnerships with academic institutions (GetReal Academy).

A conference on 16th March in Utrecht underscored GRI's role as a vital European forum, with prominent representation from the rare disease community including speakers such as Bettina Ryll (Founder, Melanoma Patient Network Europe), Elizabeth Vroom (World Duchenne Organization), Hayley Chapman (Patient Focused Medicines Development), and EURORDIS.

Additionally, GRI contributed an article titled "Real-World Evidence to Inform Regulatory Decision Making: A Scoping Review" to the Journal of Clinical Pharmacology and Therapeutics, further establishing its commitment to advancing the use of RWE in regulatory frameworks.

VACCELERATE (January 2021-January 2024, Horizon 2020, Project Advisor)

VACCELERATE is a network facilitating COVID-19 vaccine trials across Europe, led by the University Hospital Cologne, Germany. It includes 29 national partners from 18 EU member states and five associated countries. Due to recruitment interruptions, its project duration extended by 12 months, introducing a new task on Clinical Trials Preparedness and response capacity. Ongoing trials focus on booster vaccinations in different age groups, with an added emphasis on long-term follow-up.

REMEDI4AL (September 2022-August 2027, Horizon Europe, Project Partner)

REMEDi4ALL seeks to support the development and implementation of repurposed medicines, addressing unmet medical needs, particularly in the rare disease (RD) field. As the WP lead on Patients & Users' Engagement, EURORDIS is pivotal in ensuring patient centricity throughout the drug repurposing lifecycle. Activities included developing processes for patient and user engagement, leading the Multi-stakeholder Meeting on drug repurposing for pancreatic cancer, and contributing to training and capacity building within the consortium.

EURORDIS organised the first REMEDi4ALL Multi-stakeholder Meeting “Drug Repurposing: An Attractive Strategy in Pancreatic Cancer Treatment” in 2023 and is currently working on a publication of the outcomes of this meeting.

In its first year, REMEDI4ALL, with EURORDIS’s significant input, particularly in WP3 on Training & Capacity Building, developed various internal training sessions and assessed training needs across the ecosystem. These efforts led to a detailed training landscape, laying the groundwork for a comprehensive training curriculum set to be defined in 2024.

Screen4Care (S4C, September 2021-September 2026, IMI 2, Project Partner)

Screen4Care proposes a novel method to expedite rare disease diagnoses through **genetic newborn screening and digital technologies.** EURORDIS is central to this initiative, leading the Patient Advisory Board (PAB) and influencing all project facets based on the rare disease community’s priorities. Efforts in 2023 focused on developing the ACT Panel, conducting surveys to gauge patient preferences on newborn screening (NBS), and hosting a workshop to discuss the actionability concept for NBS condition selection.

International Consortium for Rare Disease Research (IRDiRC)

The International Rare Diseases Research Consortium (IRDiRC) brings together national and international funding bodies, companies, patient advocacy organisations, and researchers to foster global collaboration in rare disease research. Its vision is for all individuals with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of medical attention.

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

EURORDIS has been a key member of IRDiRC since its inception in 2011, actively participating in the Consortium Assembly and contributing to numerous activities aimed at achieving IRDiRC goals. Additionally, EURORDIS staff are involved in the Therapies Scientific Committee, with further contributions from an Open Academy alumnus.

EURORDIS has led or been part of various task forces, including those focusing on Sustainable Economic Models in Repurposing and Drug Repurposing Guidebooks, contributing to publications and guidebooks essential for drug development. Ongoing efforts include participation in the Pluto Project on Disregarded Rare Diseases and the Newborn Screening Initiative, with involvement in upcoming publications and workshops.

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

In 2023, the Global Commission was reinvigorated by Takeda, Sanofi, and EURORDIS, with EURORDIS co-chairing alongside these companies. EURORDIS plays a significant role in the operational working group, contributing to content development and organising membership meetings, aiming to enhance diagnostic processes for children with rare diseases.

SOCIAL POLICY AND SERVICES 05 ^{05}

> NTEGRATED 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

ADVOCATE

ADVOCATING FOR INTEGRATED AND HOLISTIC CARE

EURORDIS continued its advocacy for holistic care, focusing on an integrated, lifelong, and person-centred approach as crucial for the future EU Action Plan on Rare Diseases. Furthermore, EURORDIS became a contracted partner in the upcoming Joint Action on the integration of ERNs into national healthcare systems (JARDIN), co-leading the design of Guidelines on Case Management for Rare Diseases. Additionally, EURORDIS promoted the good practices of RareResourceNet, rejoining its Board of Directors in May 2023.


“NOT BEDDED ON ROSES”
CEREBRAL CAVERNOUS MALFORMATION / GERMANY

ADVOCATING FOR QUALITY AND ADEQUATE SOCIAL RIGHTS

EURORDIS collaborated with the Social Platform and its members to engage in discussions on initiatives stemming from the European Pillar of Social Rights.

ADVOCATING TO IMPROVE ACCESS TO DISABILITY RIGHTS

In January 2023, EURORDIS was appointed as an observer member of the EU Disability Platform, supporting the implementation of the European Strategy for the Rights of Persons with Disabilities 2021–2030. Throughout the year, EURORDIS participated in the Platform’s plenary and subgroup meetings, advocating for the needs of the rare disease community.

EURORDIS contributed to consultations for the proposal to establish a European Disability Card, aimed at easing access to services for people with disabilities within the EU. After the European Commission’s Proposal for a Directive on the European Disability Card and Parking Card was published in September, EURORDIS continued advocacy efforts with the European Parliament, highlighting the eligibility of people with rare diseases and disabilities for the Card and calling for improvements in national disability assessment processes.

EURORDIS also engaged in key EU Disability events, including a seminar organised by the Swedish Presidency of the Council of the EU and the European Day of Persons with Disabilities, organised by the European Commission in partnership with the European Disability Forum.

ADVOCATING FOR THE MENTAL HEALTH AND WELLBEING OF PEOPLE LIVING WITH RARE DISEASES

To bolster its activities in mental health and wellbeing for those with rare diseases, EURORDIS launched a dedicated webpage offering information, factsheets, and webinars, alongside an online registration portal for the new Partnership Network.

EURORDIS joined Mental Health Europe (MHE) and collaborated on organising a European Parliament Policy Event titled "Addressing the Risks and Barriers Faced by People in Vulnerable Situations" on 28 November 2023.

A targeted literature review was completed by EURORDIS to map out existing mental health initiatives related to rare diseases in Europe, including within the European Reference Networks (ERNs). This summary was contributed to the Commission's call for evidence, aiding in the development of the new Communication on a Comprehensive Approach to Mental Health. Furthermore, EURORDIS submitted a poster presenting a concrete proposal for inclusion in the Communication.

EURORDIS also led various communications and engagement efforts to highlight the complex needs affecting the mental health and wellbeing of individuals with rare conditions.

EMPOWER

Relaunch the EURORDIS Social Policy Action Group (SPAG)

In November, the EURORDIS Board of Directors endorsed 20 volunteers from 12 European countries as the new Social Policy Action Group (SPAG). The SPAG, with a 3-year mandate starting in January 2024, aims to enhance access to holistic care, social, and human rights for people living with a rare disease and their families.

Launch of the EURORDIS Mental Health & Wellbeing Partnership Network

Preparatory webinars on the impact of rare conditions on mental health and dealing with trauma attracted over 120 participants. Based on these discussions, four factsheets were developed on mental health impacts and the intersectional needs related to rare diseases.

Following the call by the rare disease community, to look beyond the physiological symptoms of a rare disease and take concrete action to address the psychological impact associated with these complex conditions, EURORDIS has coordinated the establishment of **a new Mental Health & Wellbeing Partnership Network**. The network now includes over 100 members from 25 countries, with strong support from the rare disease community. The network connects with over 40 medical experts, psychologists, and researchers from various European Reference Networks and international centres.

EURORDIS is exploring collaboration models between ERN and the Partnership Network and the possibility of an ERN cross-working group on mental health. The inaugural meeting on World Mental Health Day celebrated solidarity with those affected by mental health issues, inviting members to become Mental Health Champions or join working groups focusing on population needs, research, quality of life, advocacy, and communication.

EURORDIS has formulated an action plan to guide and coordinate efforts under the new Partnership Network, supporting the mental health champions in representing the rare disease community in EU actions related to mental health.

PARTNER

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+) and within the Citizens, Equality, Rights and Values programme (CERV).

"IN BETWEEN"
HAPLOINSUFFICIENCY MEF2C / POLAND

STRATEGIC OBJECTIVE 03:

INCLUSIVE OF ALL RARE DISEASES, ALL REGIONS, "LEAVING NO ONE BEHIND"

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

All therapeutic areas, including genetic or non-genetic rare diseases, and rare cancers, with progress to be made with regard to rare infections and rare health hazards

All countries in geographical Europe prioritising Eastern and Southern Members of the EU, European Economic Area & EU Accessing Countries

All rare disease prevalence and incidence levels, particularly the ones affecting fewer than 1 in 1,000,000

CREDENTIALS

"MISHA WITH FRIENDS"
WILLIAMS SYNDROME / RUSSIA

ADVOCATE

RARE CANCERS

Publication of Updated EURORDIS Web Pages on Rare Cancers and RCAN:

In April 2023, EURORDIS updated its webpages on rare cancers, providing essential information such as main figures based on RareCareNet updates, details about the Rare Cancer Advocates Network (RCAN), acknowledgements of ePAGs representing rare cancer groups, partnerships, advocacy initiatives, and resources on rare cancers.

Activities with RCAN Members:

RCAN members prioritised raising the profile of rare cancer patients, fostering drug development and access, and integrating rare adult and paediatric cancers into National Cancer Plans (NCPs). EURORDIS supported RCAN members' involvement in the Rare Disease Day campaign and prepared tailored social media messages on rare cancers. Efforts to revise the EU General Pharmaceutical Legislation (GPL) were highlighted, with three calls organised around fostering drug development for rare diseases/cancers and addressing access inequalities.

National Cancer Plans (NCPs):

RCAN members identified the insufficient focus on rare cancers in NCPs, impacting the implementation of adequate healthcare pathways. A sub-working group within RCAN was formed to analyse NCPs in several EU Member States and the UK, aiming to enhance care for rare cancer patients by linking with National Rare Disease Plans, especially concerning access to orphan medicinal products.

Collaboration to Raise Awareness:

EURORDIS and RCAN members are collaborating with the ESMO Rare Cancer Working Group and the European Cancer Organisation (ECO) to elevate the challenges faced by rare cancer patients at the European level, including participation in conferences and responses to EU public consultations.

VERY RARE AND ULTRA-RARE DISEASES

EURORDIS has dedicated efforts to policy initiatives and solutions targeting the challenges faced by individuals with very rare and ultra-rare diseases. Through advocacy actions, such as contributing to the revision of the EU Regulation on Orphan Medicinal Products actions, such as contributing to the revision of the EU Regulation on Orphan Medicinal Products, and participation in various projects like SOLVE-RD (focused on undiagnosed diseases) and Screen4Care (aiming at early diagnosis and newborn screening), EURORDIS has actively addressed the needs of the rarest disease communities. Additionally, EURORDIS has been involved in activities within the EMA Committees, advocating for access to therapies for very rare diseases and advanced therapies, and supporting the provision of highly specialised services through European Reference Networks (ERNs) at the EU level.

EMPOWER AND PARTNER

Launch of the Rare Cancer Advocates Network (RCAN)

RCAN was inaugurated in January 2023, assembling rare cancer ePAGs from four ERNs related to rare cancers – PaedCan (paediatric cancers), EURACAN (rare adult solid cancers), EuroBloodNet (rare haematological diseases including rare adult blood cancers), GENTURIS (genetic tumour risk syndromes), alongside ePAGs in the "endocrine tumours" sub-network of ENDO-ERN. Dorica Dan, a EURORDIS Board member, Chair of the Romanian association for rare cancers, and the Romanian national alliance for rare diseases, also contributes to the network. Governed by the ePAGs Constitution and Rules of Procedure, RCAN started with 33 ePAGs, expanding to 34 by December 2023.

RCAN is distinct in uniting rare cancer patient advocates/ePAGs across all cancer types and ages at a national, European, and international level, fostering extensive outreach to patients and caregivers. This collaboration is further enhanced by connections with various societies, organisations, and networks such as WECAN, the Rare Cancers Working Group of ESMO, SIOPe, EHA PAC, and ECO PAC:

- WECAN – Workgroup of European Cancer Patient Advocacy Networks
- Rare Cancers Working Group of the European Society of Medical Oncology (ESMO)
- European Society for Paediatric Oncology (SIOPe)
- European Hematology Association Patients Advocacy Committee (EHA PAC)
- European Cancer Organisation Patients Advisory Committee (ECO PAC)

Four ePAGs, each aligned with a specific ERN covering a group of rare cancers, have played a crucial role in supporting EURORDIS's rare cancer initiatives. They have represented the rare cancer patient community in dialogues with DG SANTE and JRC officials, particularly following collective efforts to address the challenges and needs of the adult rare cancer patient community in the European Parliament's cancer report. Additionally, they offer guidance on refining EURORDIS's strategy on rare cancers.

Ukraine response programme

Throughout 2023, EURORDIS sustained its support programme for Ukrainians with rare diseases, initiated in March 2022. The organisation actively coordinated with patient organisations in Ukraine, providing direct grants to the National Alliance and other groups to aid Ukrainians with rare conditions.

In May 2023, the “Razem z Ukraina” project concluded, a year-long initiative in collaboration with 14 patient-focused non-profits. From June 2022 to May 2023, this project assisted 1,188 Ukrainians with rare diseases and their families by offering services such as family assistant consultations, medical equipment, psychological support, accommodation, and travel cost reimbursements for trips to and from Europe.

EURORDIS also backed the Rare Disease Hub, aimed at facilitating partnerships with European Reference Networks (ERNs) to deliver medical aid to Ukrainian families. Relocated to Kyiv at the National Children’s Hospital "Okhmatdyt" since December 2022, the Hub, co-founded and financially supported by EURORDIS until September 2023, handled over 138 applications from individuals with rare diseases, seeking treatments within Ukraine and the EU.

In advocacy, EURORDIS maintained efforts to highlight the unique difficulties encountered by Ukrainians with rare diseases to European, national, and Ukrainian officials, advocating for effective support measures.

PARTNER

Rare Disease Day 2023

Rare Disease Day, coordinated internationally by EURORDIS and nationally by alliances and patient organisations, is an awareness-raising event aimed at the general public and decision-makers about decision-makers about the impact of rare diseases on individuals’ lives. The 16th edition in 2023 saw over a thousand events globally in 112 countries, including first-time participants: Israel, Guinea-Bissau, and Tanzania. Events ranged from political conferences and medical symposia to community and family gatherings.

The campaign encouraged sharing of branded materials, including social media filters and virtual backgrounds, and introduced the #RareDiseaseDay challenge, inviting posts on themed topics throughout February. The global "Chain of Lights" saw landmarks, homes, and offices illuminated in solidarity with those living with rare diseases.

The day garnered global media attention, engaging politicians, researchers, medical professionals, and policymakers. The theme focused on equity, urging community engagement through the "Share your colours / Light Up For Rare" call to action. The Rare Disease Day website served as a central resource for information and event coordination worldwide.

Webinar Series – Capacity Building.

Two webinars were conducted to share experiences and assist in event preparation, covering topics like downloading awareness materials, engaging healthcare providers, and involving National Alliances more effectively in Rare Disease Day coordination. Outreach group meetings welcomed the three new national alliances to discuss communication strategies, benefiting from the expertise of skilled communication experts from national alliances.

New Rare Cancer Infographics.

In line with EURORDIS' strategy, two new social media cards were introduced to highlight rare cancers, with additional rare cancer information featured on the Rare Disease Day website.

CROSS-CUTTING PRIORITIES

TO SUPPORT OUR STRATEGIC OBJECTIVES

"FIGHTING KRABBE"
KRABBE DISEASE / USA

COMMUNICATION AND DISSEMINATION

"Rare on Air" Podcast:

In 2023, EURORDIS maintained its monthly podcast series, covering topics like gene therapies, newborn screening, and the value of new health technologies. A special series on European Reference Networks (ERNs) was also featured.

eNews:

EURORDIS's monthly eNews reached over 11,500 subscribers, with open rates between 24% and 36%. The newsletter's redesign improved click rates and user engagement.

Member News:

Now in its eighth year and available in six languages, this bi-monthly newsletter informs members about relevant news, events, and engagement opportunities. By December 2023, subscribers exceeded 2,700.

Website Improvements:

In 2023, EURORDIS significantly enhanced its website with new tools and features to improve accessibility and user engagement. Key updates include:

- Mental Health Focus Page: Introduced a section dedicated to the impact of rare diseases on mental health, offering expert research and articles.
- BeyondWords Integration: Implemented this feature to vocalise content, enhancing accessibility for users who benefit from auditory assistance.
- Projects and Partnerships Page: A redesigned page highlighting EURORDIS's involvement in rare disease research and advocacy, with improved search functionality and SEO.
- Newsroom and Multimedia Pages: Centralised press releases, news, blogs, testimonials, and updates, alongside multimedia content including podcasts and videos.
- New Web Components: Added components like Latest News, Events, Press Releases, Tabs, Carousels, and Stories for a dynamic online experience.
- Diagnosis Page in 7 Languages: Offers vital information on diagnosis in rare diseases, policy recommendations, and resources, including an updated section on Newborn Screening.
- Members' Resources Section: A new area for sharing resources, ideas, and advocacy materials, highlighting the collective impact of EURORDIS members.

Social Media Management

In 2023, EURORDIS significantly enhanced its social media strategy, focusing on expanding its audience and improving coverage of its activities across various platforms. This included promoting both public and invitation-only events and introducing new content types to boost engagement, particularly among young people and underserved groups like ultra-rare disease organisations and those from Eastern Europe. A dedicated social media management role was created to tailor content to the distinct audiences of each platform. Engagement rates showed notable improvements:

- **Twitter** increased from 1.72% in 2021 to 3.28%
- **LinkedIn** from 5.11% to 5.97%
- **Facebook** from 1.75% to 5.13%
- **Instagram** from 0.4% to 7.73%.

Reference Documents

The EURORDIS Activity Report 2022 and Work Programme 2023 were designed for accessibility and clarity, featuring straightforward language, clear headings, and ALT text for images. Digital formats were screen-reader friendly, and publications such as the Patient Partnership Framework for ERNs, and Championing the Rare were designed with legible fonts and high colour contrast for better readability.

EURORDIS also updated its infographics and strategy slides to enhance accessibility. Infographics were revised for better colour contrast, aligning with accessibility standards. Strategy slides improvements included adding ALT text to images, using legible fonts, and ensuring high text-background contrast. These changes were aimed at making content easily accessible to everyone, including those with visual, hearing, or other types of impairment.

Translations

In 2023, EURORDIS focused on expanding the accessibility of its content through targeted translation activities, though with some limitations compared to previous years. Key highlights of these translation efforts include:

Translation of priority pages:
A selection of EURORDIS's priority webpages was translated into six languages. This initiative aimed to make essential information more accessible to a broader audience across different linguistic groups, thereby enhancing the organisation's outreach and engagement with diverse communities within the rare disease sector.

Ukrainian translations:
In a significant effort to support Ukrainian people living with rare diseases, EURORDIS translated a crucial report into Ukrainian, ensuring that vital information was accessible in their native language.

PEOPLE (STAFF AND VOLUNTEERS)

02 07

EURORDIS Staff

In April 2023, a part-time Human Resources Director was appointed under a service agreement to carry out a three-way HR diagnosis:

- Twitter increased from 1.72% in 2021 to 3.28%
- 360° assessment of the Core Leadership Team
- Implementation of an annual Social Barometer with “Great Place to Work” over three years, assessing employee mental wellbeing and guiding staff towards the certification of a "Great Place to Work" via a People-Centric Approach.

In July 2023, the outcomes of the HR diagnosis were shared with the Board of Directors and all staff. This included a Human Resources Road Map outlining a people management system to be developed over 2024–2025 to foster a People-Centric Approach, Core Organisational Culture, Core Work Processes, and Measuring Performance, aligning HR processes with Strategic Objectives.

The position will transition to a permanent full-time contract from January 2024 to lead EURORDIS Human Resources and support the transition to a new CEO, with the recruitment process initiated in 2023 and effective in March 2024.

The number of full-time equivalents (FTE) was 51.18 in 2023, up from 48.7 in 2022. At year-end, the team comprised 56 staff members across seven countries:

France	Spain	Belgium	UK	Italy	Germany	Romania
32	10	08	02	02	01	01

No new positions were created in 2023. Two positions were discontinued:

- Rare Disease Day Senior Manager, Estelle Dubois, Paris
- Health & Social Science Project Senior Manager, Edith Gross, Paris

EURORDIS VOLUNTEERS

03 07

EURORDIS volunteers are experts on rare diseases. Most of them are either people living with a rare disease or their caregivers, usually family members. Those not directly affected by a rare disease are deeply involved in a rare disease patient organisation. The EURORDIS volunteers are committed to providing their expertise and time on specific areas on a voluntary basis. They greatly contribute to raising awareness of rare disease patients' needs and challenges to foster research on rare diseases, improve equal access to a timely, accurate diagnosis as well as adequate available treatments/medicines.

All volunteers adhere to the EURORDIS Charter of Volunteers, adopted on 8 May 2014 during the EURORDIS General Assembly in Berlin. This Charter outlines the values of EURORDIS, the commitments of the volunteers, and EURORDIS' obligations towards them.

In September 2023, after a year of preparation and discussions with the Core Leadership Team and the Board, the EURORDIS Board of Officers adopted an internal procedure for volunteer management to standardise processes across all teams.

EURORDIS volunteers are selected through a Call for Expression of Interest, based on strict criteria, including a commitment to rare disease advocacy, proficiency in English, and a substantial track record in advocacy. This rigorous selection ensures that EURORDIS consistently proposes qualified candidates for EMA and European Commission committees, often resulting in nominations by the European Commission.

In 2023, EURORDIS had 66 volunteer patient advocates involved in various groups or task forces, with some participating in multiple groups. Following the cessation of the RareConnect programme in mid-2023, there are no longer any volunteer moderators for RareConnect online patient communities.

EPAC/ADVOC (European Public Affairs Committee/Advocacy Committee)

EPAC, essential in EURORDIS' advocacy efforts, discusses pertinent issues affecting people with rare diseases and their families. Governed by specific Rules of Procedure, the committee included 24 members comprising volunteers and advocacy-involved staff as of late 2023. Following a Board decision in November 2023, EPAC/ADVOC was dissolved, transitioning to a new Advocacy 360 group from February 2024.

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

In 2023, 13 trained volunteers contributed to the DITA Task Force's initiatives, supporting EURORDIS representatives in EMA Scientific Committees and Working Parties.

TAG (Therapeutic Action Group)

TAG consists of 6 volunteers who represent patients on the European Medicines Agency's scientific committees, engaging significantly in monthly multi-day meetings.

HTA Task Force (Health Technology Assessment)

Comprising 8 volunteers and two coordinating staff, the HTA Task Force advises on HTA policies and shares insights on national assessments and the evolving European Cooperation on HTA.

SPAG (Social Policy Action Group)

Relaunched in 2023 with 20 volunteers from 12 countries, SPAG supports initiatives to enhance access to holistic care and social and human rights for those with rare diseases, starting its mandate in January 2024.

DAG (Digital and Data Advisory Group)

The 13-member DAG in 2023 advised on digital strategies and policies impacting EURORDIS and its constituents.

ePAGs – European Patient Advocacy Groups

Launched alongside the European Reference Networks (ERNs), ePAGs ensure the patient voice within ERN Boards and committees. A Steering Committee of two ePAG representatives for each of the 24 ERNs coordinates the efforts. In 2023, 17 members expressed continued interest in volunteering, supported by four staff members to enhance advocacy and raise ERN awareness.

GOVERNANCE

04[07]

EURORDIS Governance Bodies

The EURORDIS **Annual General Assembly** was held online on 17 May 2023. EURORDIS' full members voted on the Activity and Financial Reports 2022, and the Work Programme and Budget for 2023.

Members also voted on the vacant positions for the **Board of Directors (BoD)**. The following candidates were successfully re-elected to the BoD: Alain Cornet (Lupus Belgium); Dorica Dan (Romanian Prader Willi Association, Romania).

The Board also extended a warm welcome to new members:

- Graham Slater, EAT – Esophageal Atresia Global support groups, UK
- Anna Spinou, Hellenic Cystic Fibrosis Association, Greece
- Daniel de Vicente, FEDER — Rare Diseases Spain
- Tomasz Grybek, Foundation of Borys the Hero, Poland

Board members were elected for a full mandate of three years. One seat was open for a 2-year mandate (Graham Slater).

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 – Avril Daly, Ireland; Vice President – Dorica Dan, Romania and Simona Bellagambi, Italy; General Secretary – Geske Wehr, Germany; Deputy General Secretary – Anna Arellanesova, Czech Republic; Treasurer – Alain Cornet, Belgium.

The EURORDIS **Core Leadership Team (CLT)** prepares and executes on the strategies, ensures day-to-day decision-making, and includes the Chief Executive Officer (CEO), the Chief Financial Officer (CFO), the Chief Operating Officer (COO), the Public Affairs Director & Head of European Advocacy, the Human Resources Director and People Team Lead and the Governance Senior Manager. The EURORDIS Core Leadership Team (CLT) holds weekly meetings to address various strategic, management, and operational issues. These meetings aim to resolve challenges at this governance level, facilitating progress towards fulfilling EURORDIS' missions and activities.

EURORDIS By-laws

In 2023, parts of the by-laws were revised, and new terms of reference were established for the Mental Health & Wellbeing Partnership Network, along with revisions to the terms for the Council of National Alliances and the Social Policy Action Group (SPAG).

EURORDIS Key partnership with the AFM-Téléthon

EURORDIS and its co-founder and primary supporter, AFM-Téléthon, have worked on renewing their Memorandum of Understanding for 2024–2027. This agreement facilitates ongoing interactions and collaborative projects within the AFM-Téléthon ecosystem.

KEY 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 — The US Organization for Rare Disorders

JPA – The Japan Patients Association

RDI – Rare Diseases International

CORD – The Canadian Organization for Rare Disorders

RVA – Rare Voices Australia

EURORDIS also has partnerships with a number of learned societies, that are listed under the “External representation” webpage and detailed in our Work Programme 2024.

RESOURCE DEVELOPMENT & EURORDIS SUSTAINABILITY

05 07

In 2023, EURORDIS saw growth in engagement and revenue across its resource development channels:

- The EURORDIS Round Table of Companies convened for two key workshops:
    - Bringing Clinical Trials into the Future" in Brussels in February, with 146 attendees including 77 from 46 ERTC member companies.
    - "Overcoming Barriers to Rare Disease Diagnosis" in Barcelona in October, attended by 116 participants, including 67 from 39 ERTC member companies.
- Three webinars for corporate members included topics on EURORDIS strategy up to 2030, the impact of rare conditions on mental wellbeing, and involvement in Rare Disease Day.

- A total of 76 health sector corporations supported EURORDIS, including 10 new corporate donors.
- Fundraising from foundations saw significant growth, supporting programmes such as Rare Disease Week, a new Open Academy course on survey design, patient health data initiatives, and Rare Disease Day communications support grants and school toolkits.
- Both corporate and foundation supporters continued to sustain our Ukraine Emergency Response programme, as well as offering new support for the rare disease community affected by the earthquake in Turkey.

FINANCE & SUPPORT SERVICES

06[07]

Finance and support services' activities in 2023 included:

- Accounting and timely financial reporting, including detailed cash flow and risk analysis.
- Management of human resources activities, notably recruitment.
- Overseeing office support, including IT infrastructure, contact database, and office supplies.
- Handling legal and fiscal matters.
- The creation or updating of 45 EURORDIS procedures across accounting, HR, administrative, and IT sectors.

CONTRACT GRANTS

07[07]

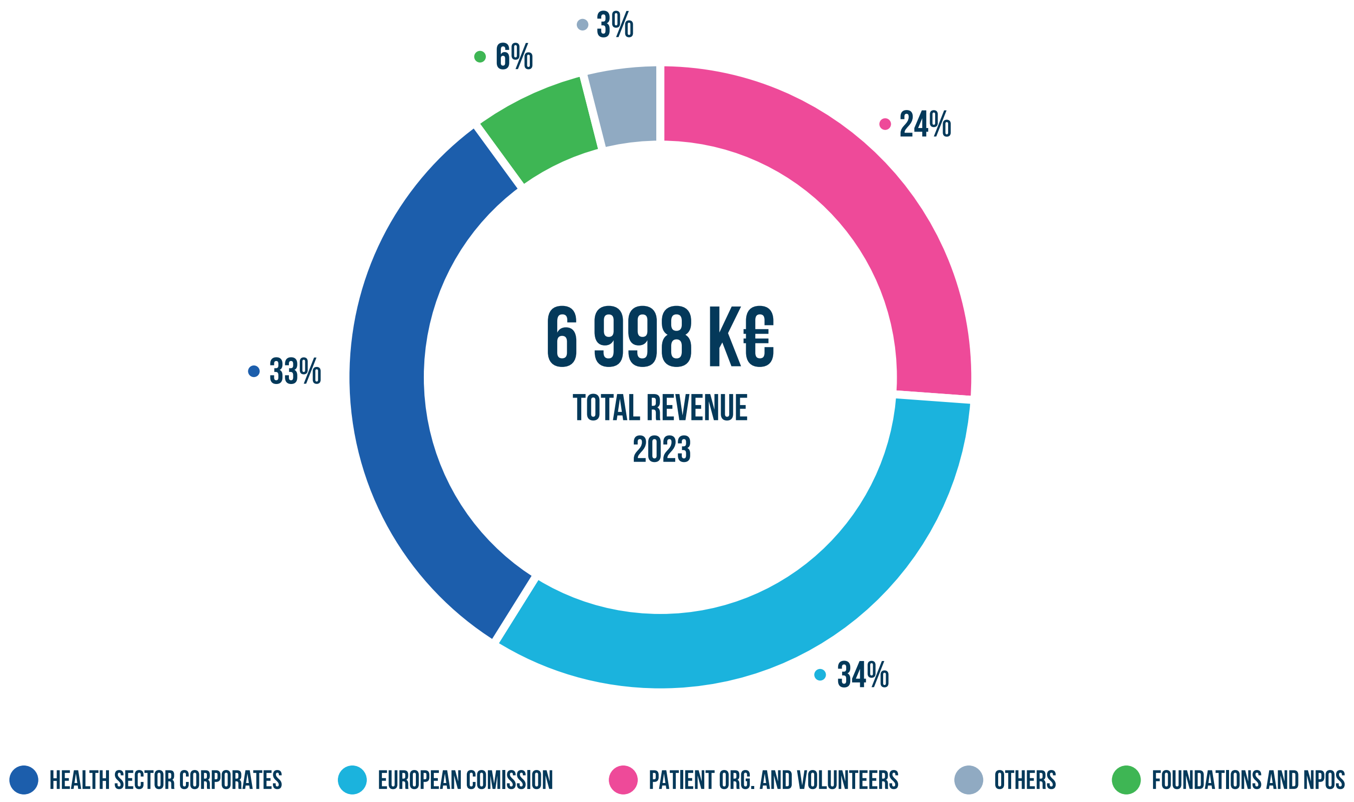
Ongoing:

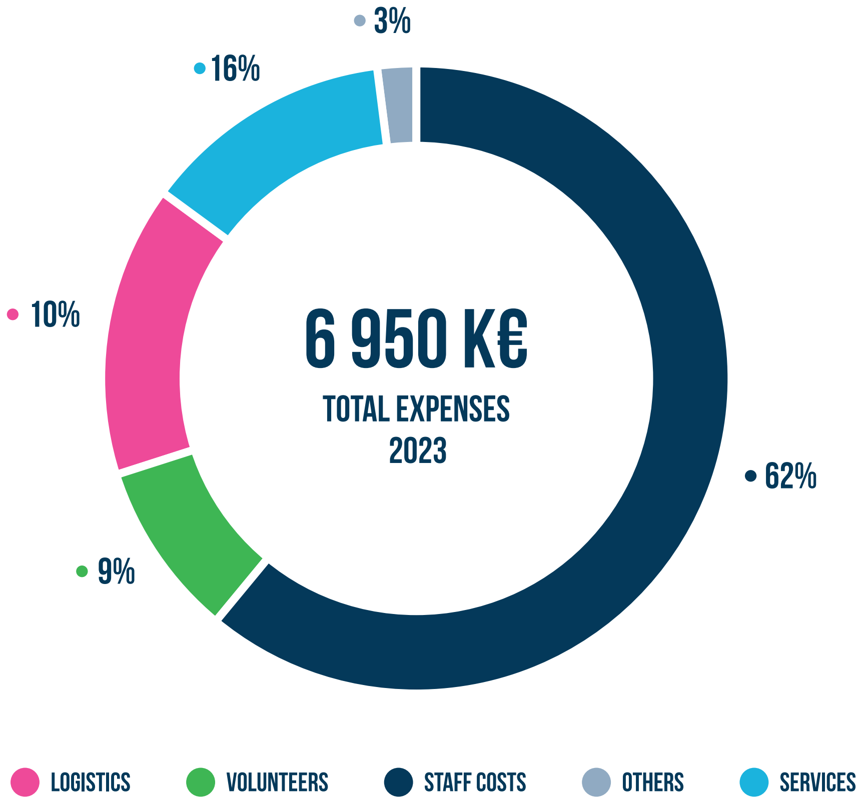
Advocacy and core activities, AFM-Téléthon, 2019–2023

Renewed:

Operating Grant for year 2023 (OG 2023), single beneficiary, DG SANTE, 12 months

REVENUES & EXPENSES 2023





“GET BREATHLESS FOR PULMONARY HYPERTENSION”
PULMONARY HYPERTENSION / CROATIA

BOARD OF DIRECTORS

MAY 2023 - MAY 2024

PRESIDENT

MS AVRIL DALY

Retina International

Ireland

DIRECTORS

MS ANNA ARELLANESOVA

Rare Diseases Czech Republic

Czechia

MR TERKEL ANDERSEN

Danish Haemophilia Society

Denmark

MR TOMASZ GRYBEK

Foundation of Borys the Hero

Poland

MS SIMONA BELLAGAMBI

UNIAMO – Rare Diseases Italy

Italy

MR ALAIN CORNET

Lupus Belgium

Belgium

MS DORICA DAN

Romanian Prader Willi Association

Romania

MR ALEXANDRE MEJAT

AFM - Téléthon

France

MS GESKE WEHR

Selbsthilfe Ichthyose eV

Germany

MS KIRSTEN JOHNSON

The Fragile X Society

United Kingdom

MS ANNA SPINOU

Hellenic Cystic Fibrosis Association

Greece

MR DANIEL DE VICENTE

FEDER

Spain

MS REBECCA TVEDT SKARBERG

Osteogenesis Imperfecta Federation Europe (OIFE)

Norway

MR GRAHAM SLATER

Esophageal Atresia Global support groups

United Kingdom

BOARD OF OFFICERS

MAY 2023 - MAY 2024

PRESIDENT

MS AVRIL DALY

Ireland

VICE-PRESIDENTS

MS DORICA DAN

Romania

MS SIMONA BELLAGAMBI

Italy

GENERAL SECRETARY

MS GESKE WEHR

Germany

DEPUTY GENERAL SECRETARY

MS ANNA ARELLANESOVA

Czechia

TREASURER

MR ALAIN CORNET

Belgium

EURORDIS MEMBERS 2023

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

Bulgaria Assocation Of People Suffering By Acromegaly In Bulgaria		Associate Member
Bulgaria Association of Tarlov Cyst patients in Bulgaria	http://tarlov-bg.eu/	Full Member
Bulgaria Bulgaria society of patients with pulmonary hypertension	http://www.bspph.net	Full Member
Bulgaria Bulgarian Association Wilson Disease	https://wilsonbg.org	Full Member
Bulgaria Bulgarian Cystic Fibrosis Association	http://lifewithcf.org/	Full Member

Bulgaria Bulgarian Huntington Association	http://huntington.bg	Associate Member
Bulgaria Bulgarian National Alliance of People with Rare Diseases	http://rare-bg.com/	Associate Member
Bulgaria Nas - National Association Sarcoidosis Bulgaria	http://sarcoidosisbulgaria.alle.bg/	Full Member
Bulgaria National Association For Child Support Congenital Hypothyroidism	http://www.abv.bg	Associate Member
Bulgaria National Association of Patients With Growth Hormone Deficiency	https://facebook.com/100069309093196/	Full Member
Bulgaria National Association of Patients with Mitochondrial Diseases in Bulgaria	http://www.mitobg.com/	Full Member
Bulgaria National Association of Syringomyelia		Full Member
Bulgaria National Gaucher Organization	http://gaucher-bg.org/	Associate Member
Bulgaria National Patients' Organisation	https://npo.bg/	Associate Member
Bulgaria PHA Bulgaria	http://www.phabulgaria.eu	Full Member
Bulgaria Retina Bulgaria	http://www.retinabulgaria.bg	Associate Member
Bulgaria 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 Cyprus Alliance For Rare Disorders	http://raredisorderscyprus.com/	Full Member
Cyprus Cyprus Association of Inherited Metabolic Diseases 'Aspida Zois'	http://www.aspidazois.com	Full Member
Cyprus Pancyprian Association For Rare Genetic Diseases "Unique Smiles"	http://www.monadikaxamogela.com	Full Member
Cyprus Thalassaemia International Federation	http://www.thalassaemia.org.cy	Full Member
Cyprus 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://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	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://filiere-mcgre.fr/le-parcours-patients/associations-de-malades/	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 Francaise 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://maladiesraresspm.simdif.com/	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

Georgia Georgian Foundation for Genetic and Rare Diseases		Full Member
Georgia 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-erkrankunger	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
Germany NCL-Gruppe Deutschland e.V.	http://www.ncl-deutschland.de	Full Member
Germany Nephie e.V. - Selbsthilfe nephrotisches Syndrom	http://www.nephie.de	Full Member
Germany Netzwerk Hypophysen- und Nebennierenerkrankungen e.V. - Network pituitary and adrenal disorders	http://www.glandula-online.de	Full Member
Germany Patienten- und Selbsthilfeorganisation für Kinder und Erwachsene mit Kranker Speiseröhree Mit Kranker Speiseröhre	https://keks.org/	Full Member
Germany Patientenverband Familiäre Amyloid Polyneuropathie	http://patientenverband-fap.de	Associate Member
Germany PCH-Familie e.V.	https://pch-familie.de/	Full Member
Germany Peutz-Jeghers-Germany e.V.	http://peutz-jeghers.eu	Full Member
Germany PKD Familiäre Zystenniere e. V.	http://www.pkdcure.de	Full Member
Germany Pro Retina Deutschland e.V.	http://www.pro-retina.de	Associate Member
Germany Pulmonale Hypertonie e.V.	http://www.phev.de	Full Member
Germany Sanfilippo Initiative e.V.	http://www.sanfilippoinitiative.org	Associate Member
Germany Selbsthilfe EPP e.V.	http://www.epp-deutschland.de	Full Member
Germany Selbsthilfe Ichthyose e.V.	http://www.ichthyose.de Full Member	
Germany Selbsthilfegruppe Ektodermale Dysplasie e.V.	http://www.ektodermale-dysplasie.de	Full Member
Germany Selbsthilfegruppe Für PXE-Erkrankte Deutschlands e.V.	http://www.pxe-shg.de	Full Member
Germany Selbsthilfegruppe Glykogenose Deutschland e.V.	http://www.glykogenose.de	Full Member
Germany Selbsthilfeorganisation für Menschen mit Anorektalfehlbildungen	http://www.soma-ev.de	Full Member
Germany Sklerodermie Selbsthilfe e.V.	https://www.sklerodermie-sh.de/	Full Member
Germany SMA Europe	http://www.sma-europe.eu	Full Member
Germany SSADH-Defizit e.V.	http://ssadh.wordpress.com	Associate Member
Germany Tom Wahlig Stiftung	http://www.hsp-info.de	Full Member
Verein Ahc18+	https://www.facebook.com/pages/	

	Association-ahc18/606879759444135	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	Asociate 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 h	ttp://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://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
Guatemala 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

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://www.malattierare.gov.it/associazioni/dettaglio/981	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 Musculare	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%ABmundjeveg	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
Lithuania 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

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
Mexico Proyecto Pide Un Deseo Mexico Iap	http://www.pideundeseo.org	Associate Member
Mexico 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
Morocco Sun'Hop		Associate Member

NEPAL

GBS/CIDP Foundation Nepal		Associate Member
Nepal Muscular Dystrophy Organization Nepal	https://www.facebook.com/ MuscularDystrophyOrganizatio	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.Nl	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	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 Organisations 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 - Associacao 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-686485228	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
Romania Asociatia Werdnig Hoffman Awh	https://www.facebook.com/werdnighoffman/	Full Member
Romania Autoimmune Diseases Patients Association	http://www.apaa.ro	Associate Member
Romania Charcot Marie Tooth Romania Association	http://www.asociatiacmt.ro	Associate Member
Romania DMD Care	https://dmdcare.org	Full Member
Romania Mastocytosis Support Association Romania	http://www.mastocitoza.ro	Associate Member
Romania Neuro Move CMT Association	https://neuromovecmt.ro/	Associate Member
Romania Romanian National Alliance For Rare Diseases	http://bolirareromania.ro	Full Member
Romania 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 Association	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
Associació 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/ A	ssociate 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
Associación 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	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 Sindrome 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

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 Membe

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://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	https://europeanlung.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://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	https://www.blueconemonochromacy.org/	Associate Membe
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

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

VENEZUELA, BOLIVARIAN REPUBLIC

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

ZIMBABWE

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

"THE CHILD'S WILL AND RESILIENCE"
MYOPATHY / FRANCE

PARTICIPATION
OF EURORDIS' REPRESENTATIVES IN PUBLIC EUROPEAN / INTERNATIONAL CONFERENCES & WORKSHOPS 2023

01. **Closing the funding gap. Rare Intelligence workshop from Ipsen Foundation, Paris**

EURORDIS Representative:	Date:
Roseline Favresse	24 January 2023

02. **Webinar: A New Era in Telemedicine: Opportunities, Challenges, and What Comes Next**

EURORDIS Representative:	Date:
Jelena Malinina	31 January 2023

03. **The European Health Data Space – A Game Changer?**

EURORDIS Representative:	Date:
Jelena Malinina	8 February 2023

04. **Global Conference on Advancing Social Pharmaceutical Innovation, Utrecht**

EURORDIS Representative:	Date:
Virginie Hivert	9-10 February 2023

05. **FOKUS Patient - Rare Cancers Seminar (organised by Ms Penilla Gunther, Founder of FOKUS Patient® and Member of the Cancer Mission Board)**

EURORDIS Representative:	Date:
Ariane Weinman	20 February 2023

06.

Rare Disease Moonshot Workshop, Brussels

EURORDIS Representative:	Date:
Roseline Favresse	**21 February 2023**

07.

DSL Tweetchat: How can patient registries improve treatment and care?

EURORDIS Representative:	Date:
Jelena Malinina	**21 February 2023**

08.

International UAE Rare Disease Society Congress

EURORDIS Representative:	Date:
Gulcin Gumus	**25-26 February 2023**

09.

Release of EUCOPE podcast with Simone Boselli

EURORDIS Representative:	Date:
Simone Boselli	**28 February 2023**

10.

Addressing the unmet medical needs of people living with a rare disease: What will it take?

EURORDIS Representative:	Date:
Jelena Malinina	**1 March 2023**

11.

Screen4Care annual meeting

EURORDIS Representatives:	EURORDIS (PAB) Representatives:	Date:
Gulcin Gumus **Edith Sky Gross**	**Simona Bellagambi** **Elizabeth Vroom**	**6-7 March 2023**

12.

ProMIS Winter School "Cohesion Policies and the Challenge of Regional Health Systems"

EURORDIS Representative:
Jelena Malinina

Date:
7-9 March 2023

13.

RE(ACT) Congress, Berlin

EURORDIS Representative:
Claudia Fuchs

Date:
15-18 March 2023

14.

Spain: EURORDIS receives an award from HM Queen of Spain

EURORDIS Representative:
Yann Le Cam

Date:
16 March 2023

15.

ESMO Sarcoma and Rare Cancers Annual Congress, Lugano, Switzerland

EURORDIS Representative:
Ariane Weinman

Date:
20-22 March 2023

16.

Rare Disease Innovation & Partnership Summit

EURORDIS Representative:
Edith Gross

Date:
21 March 2023

17.

Mental Health Stakeholder consultation meeting in Brussels

EURORDIS Representative:
Matt Bolz-Johnson

Date:
21 March 2023

18.

ERN ITHACA Webinar

EURORDIS Representative: Gulcin Gumus

Date: 21 March 2023

19.

DIA Euromeeting - #S0402-H: Will Europe Continue to Foster the Development of Orphan and Paediatric Medicines?

EURORDIS Representative: Virginie Hivert

Date: 22 March 2023

20.

DIA Euromeeting - #S0403-H: Expedited pathways, will Europe and UK be part of Innovation?

EURORDIS Representative: Virginie Hivert

Date: 23 March 2023

21.

DIA Euromeeting - #DMD10-H: Derisking Generation and Use Of Patient Evidence In Drug Development and Decision-Making

EURORDIS Representative: Virginie Hivert

Date: 24 March 2023

22.

HealthData@EU Pilot - External Advisory Board Kick-off

EURORDIS Representative: Jelena Malinina

Date: 27 March 2023

23.

Cell & Gene Meeting on the Med, BCN Panel Discussion: Hopes and Concerns: How Patients and Caregivers Approach ATMPs

EURORDIS Representative: Virginie Hivert

Date: 13 April 2023

24. Solve-RD Final Meeting, Prague

EURORDIS Representatives:
Simona Bellagambi
Roseline Favresse
Gulcin Gumus

Date:
23-24 April 2023

25. EU-PEARL final meeting: Paving the Way to Innovative and Patient-Centered Clinical Research, Brussels

EURORDIS Representative:
Virginie Hivert

Date:
25 April 2023

26. Undiagnosed day webinar

EURORDIS Representative:
Gulcin Gumus

Date:
28 April 2023

27. Patient Partnership webinar: Together4RD – Fostering a healthy collaboration between ERNs & Industry Partners

EURORDIS Representative:
Matt Bolz-Johnson

Date:
5 May 2023

28. HTx consortium meeting in Madrid

EURORDIS Representative:
François Houÿez

Date:
22 May 2023

29. European Parliament of Persons with Disabilities, Brussels

EURORDIS Representatives:
Dorica Dan
Raquel Castro

Date:
23 May 2023

30. More-Europa kick-off meeting

EURORDIS Representative:	Date:
François Houÿez	**30 May 2023**

31. MedTech Forum 2023: Health Data session

EURORDIS Representative:	Date:
Jelena Malinina	**31 May 2023**

32. General Assembly Meeting REMEDi4ALL, Amsterdam

EURORDIS Representatives:	Date:
Claudia Fuchs **Judit Baijet**	**June 2023**

33. GLOBAL SKIN Annual Meeting

EURORDIS Representative:	Date:
Matt Bolz-Johnson	**6 June 2023**

34. European Society of Human Genetics Annual Conference

EURORDIS Representative:	Date:
Gulcin Gumus	**10 June 2023**

35. c4c Annual Conference

EURORDIS Representatives:	Date:
Gulcin Gumus **Maria Cavaller**	**13-15 June 2023**

36. PGEU Annual Conference, EHDS panel

EURORDIS Representative:	Date:
Jelena Malinina	**13 June 2023**

37. Medicines for Europe Annual Conference, Malta

EURORDIS Representative: **Virginie Hivert**

Date: **14-16 June 2023**

38. HTA Cooperation Stakeholder Network

EURORDIS Representative: **François Houÿez**

Date: **14 June 2023**

39. EU Accessibility Event 2023 organised by IAAP-International Association of Accessibility Professional

EURORDIS Representative: **Raquel Castro**

Date: **14 June 2023**

40. Open Academy at ESEA Award ceremony I

EURORDIS Representatives: **Sharon Ashton-Sirot**
Marta Campabadal

Date: **15 June 2023**

41. RD Partnership meeting, Paris

EURORDIS Representative: **Roseline Favresse**

Date: **21-23 June 2023**

42. ACT-EU Stakeholder Meeting

EURORDIS Representative: **François Houÿez**

Date: **22 June 2023**

43.

Round Tables on “Barriers and drivers for joining the ERNs” in Bulgaria

EURORDIS Representative:	Date:
Matt Bolz-Johnson	22-23 June 2023

44.

EU4Health CSA event

EURORDIS Representative:	Date:
Julie Pernet	26 June 2023

45.

HTAi conference in Adelaide, Australia

EURORDIS Representative:	Date:
François Houÿez	26 June 2023

46.

Presidency Conference on Life Sciences

EURORDIS Representative:	Date:
Yann Le Cam	27 June 2023

47.

Swedish Presidency Seminar 'Union of Equality: Disability Rights and Strategies', Brussels

EURORDIS Representatives:	Date:
Rebecca Tvedt Skarberg Raquel Castro	27 June 2023

48.

POLITICO'S 'Overcoming the burden of rare diseases in Europe'

EURORDIS Representative:	Date:
Avril Daly	27 June 2023

49.

Patients & Consumers' Working Party, EMA

EURORDIS Representative:	Date:
Avril Daly	27 June 2023

50. European Expert Group on OD Incentives, EMA

EURORDIS Representatives:	Date:
Yann Le Cam Simone Boselli	29 June 2023

51. EJP RD Webinar "Replicated N-of-1 Randomized Controlled Trials for Rare Diseases

EURORDIS Representative:	Date:
Virginie Hivert	30 June 2023

52. European Joint Programme Policy and Governing Board meeting, Brussels

EURORDIS Representative:	Date:
Roseline Favresse	4-5 July 2023

53. WPC2023 in Glasgow

EURORDIS Representative:	Date:
François Houÿez	5 July 2023

54. EC ehealth stakeholder group meeting

EURORDIS Representative:	Date:
Jelena Malinina	5 July 2023

55. ERICA General Assembly

EURORDIS Representative:	Date:
Maria Cavaller	6-8 July 2023

56. Universités d’été de Pharmaceutiques, Versailles, France

EURORDIS Representative:	Date:
Virginie Hivert	6-7 July 2023

57.

Colloque TRANSFORM, Paris, France

EURORDIS Representative: **Virginie Hivert**

Date: **6 July 2023**

58.

ICAN & ISPI

EURORDIS Representative: **Maria Cavaller**

Date: **10-15 July 2023**

59.

ACT-EU workshop on Clinical Trials guidelines, EMA

EURORDIS Representative: **François Houÿez**

Date: **10-15 July 2023**

60.

SSIEM meeting

EURORDIS Representative: **Gulcin Gumus**

Date: **29 August 2023**

61.

ERN Rare Liver Annual Meeting

EURORDIS Representative: **Matt Bolz-Johnson**

Date: **1-2 September 2023**

62.

Retina International's Youth World Conference

EURORDIS Representative: **Jelena Malinina**

Date: **1 September 2023**

63.

ERN eUROGEN Annual Meeting

EURORDIS Representative: **Matt Bolz-Johnson**

Date: **11 September 2023**

64.

EJP RD General Assembly

EURORDIS Representative: **Roseline Favresse**

Date: **13 September 2023**

65. **Conect4children (c4c) cross-platform session: c4c Young Investigators Community (YIC) & young ESDPPP**

EURORDIS Representative:	Date:
Maria Cavaller	September 2023

66. **NORBS Caring for Rare Conference, Serbia**

EURORDIS Representatives:	Date:
Raquel Castro, Julien Delaye Dorica Dan	14-15 September 2023

67. **REMEDi4ALL 1st annual Sustainability Workshop, Barcelona**

EURORDIS Representative:	Date:
Judit Baijet	17 September 2023

68. **8th EC Advisory Body Meeting_ERN Guidelines**

EURORDIS Representative:	Date:
Matt Bolz-Johnson	18 September 2023

69. **c4c MSM on perinatal asphyxia**

EURORDIS Representative:	Date:
Maria Cavaller	18 September 2023

70. **RD Moonshot Steering Group, Brussels**

EURORDIS Representative:	Date:
Roseline Favresse	22 September 2023

71. **Reproductive Technologies: Screening and Diagnosis**

EURORDIS Representative:	Date:
Gulcin Gumus	September 2023

72.

EJPRD ISS Summer School on registries

EURORDIS Representative: **Gulcin Gumus**

Date: **25 September 2023**

73.

AEMPS meeting 'Shaping a European innovation Ecosystem'

EURORDIS Representative: **Maria Cavaller**

Date: **25 September 2023**

74.

European Health Forum Gastein, Austria

EURORDIS Representative: **Virginie Hivert**

Date: **26-29 September 2023**

75.

Data-Driven Drug Repurposing workshop, San Francisco

EURORDIS Representative: **Claudia Fuchs**

Date: **2 October 2023**

76.

'Rare 2023': Les rencontres des maladies rares, Paris

EURORDIS Representatives:
Roseline Favresse
Yann Le Cam

Date: **3 October 2023**

77.

Internet Governance Forum 2023

EURORDIS Representative: **Jelena Malinina**

Date: **8-12 October 2023**

78.

Screen4Care NBS Forum meeting

EURORDIS Representative: **Gulcin Gumus**

Date: **9 October 2023**

79.

European Social Network Seminar 'Social Services Leading Care in the Community', Barcelona

EURORDIS Representative: **Raquel Castro**

Date: **9-10 October 2023**

80.

ERN Research Training on 'Advances in Regenerative Medicine'

EURORDIS Representative: **François Houÿez**

Date: **13 October 2023**

81.

NORD Breakthrough Summit, DC

EURORDIS Representative: **Brian Howard**

Date: **13 October 2023**

82.

Early Access summit, London

EURORDIS Representative: **François Houÿez**

Date: **16 October 2023**

83.

SLRM EMA meeting, Madrid under ES Presidency

EURORDIS Representative: **Maria Cavaller**

Date: **16 October 2023**

84.

EFGCP Conference on Paediatrics, Amsterdam

EURORDIS Representative: **Virginie Hivert**

Date: **17 october 2023**

85.

Global Commission All members meeting

EURORDIS Representative: **Roseline Favresse**

Date: **17 october 2023**

86.

UDNI Annual Meeting

EURORDIS Representative: Gulcin Gumus

Date: 21-23 October 2023

87.

COST Association workshop in Brussels

EURORDIS Representative: Matt Bolz-Johnson

Date: 25 October 2023

88.

World Orphan Drug Congress, pre-session on Repurposing, Barcelona

EURORDIS Representative: Virginie Hivert

Date: 30 October 2023

89.

World Orphan Drug Congress Europe, Barcelona

EURORDIS Representatives:
Céline Schwob
Gulcin Gumus
Virginie Hivert
Maria Cavaller
Matt Bolz-Johnson
Roseline Favresse

Date: 30 October - 2 November 2023

90.

1st REMEDi4ALL Multi stakeholder Metting on Drug Repurposing in Pancreatic Cancer, Brussels

EURORDIS Representatives:
Claudia Fuchs
Judit Baijet
Virginie Hivert
Ariane Weinman

Date: November 2023

91.

Value Added Medicines conference, Brussels

EURORDIS Representative: Judit Baijet

Date: November 2023

92.

IRDiRC workshop on N=1 approaches, Barcelona

EURORDIS Representative:	Date:
Virginie Hivert	**2-3 November 2023**

93

US National Academies Meeting on Rare Diseases, online

EURORDIS Representative:	Date:
Virginie Hivert	**7 November 2023**

94.

State of Europe high-level roundtable

EURORDIS Representative:	Date:
Yann Le Cam	**8 November 2023**

95.

Ispor Conference

EURORDIS Representatives:	Date:
François Houÿez **Julien Delaye**	**12 November 2023**

96.

Conference on HTx and AI, Sofia, Bulgaria

EURORDIS Representative:	Date:
Julien Delaye	**23 November 2023**

97.

10th European Tay-Sachs and Sandhoff Family Conference, online

EURORDIS Representative:	Date:
Maria Cavaller	**28 November 2023**

98.

Addressing the Risks and Barriers Faced by People in Vulnerable Situations Call for the Comprehensive Approach to Mental Health be inclusive, leaving no one behind – EP Policy Event

EURORDIS Representative:	Date:
Yann Le Cam	**28 November 2023**

99. ## European Day of Persons with Disabilities Event, Brussels

EURORDIS Representatives:	Date:
Raquel Castro **Charissa Frank**	**30 November-1 December 2023**

100. ## The Hellenic Cystic Fibrosis Association Conference

EURORDIS Representative:	Date:
Avril Daly **Yann Le Cam**	**3 December 2023**

101. ## HMA/EMA Big Data Stakeholder Forum

EURORDIS Representative:	Date:
Maria Cavaller	**4 December 2023**

102. ## Patient Partnership Framework at ITHACA Annual meeting

EURORDIS Representative:	Date:
Andrea Cembrero Bonet	**7 December 2023**

103. ## European Health Summit

EURORDIS Representative:	Date:
Yann Le Cam	**7 December 2023**

104. ## Lighthouses and Mental Health at CNA-CEF, Brussels

EURORDIS Representative:	Date:
Ines Hernando **Matt Bolz-Johnson**	**12 December 2023**

105. ## WHO Tallinn Charter 15th Anniversary, Tallinn

EURORDIS Representative:	Date:
Matt Bolz-Johnson	**13 December 2023**

106. ## REMEDi4ALL presentation at the XVI foresight Training Course

EURORDIS Representative:	Date:
Claudia Fuchs	**18 December 2023**

JOURNAL PUBLICATIONS 2023

01. **Sustainable approaches for drug repurposing in rare diseases: recommendations from the IRDiRC Task Force, April 2023**

Lead author:
Virginie Hivert

Link:
https://www.oaepublish.com/articles/rdodj.2023.04

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02. **A Framework to Strengthen Health Systems for Gene Therapy, May 2023**

Co-Author:
Yann Le Cam

Link:
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10951444/

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03. **Patients, payers and developers of Orphan Medicinal Products: lessons learned from 10 years' multi-stakeholder dialogue on improving access in Europe via MoCA, June 2023**

Lead author:
Maria Cavaller

Co-authors include:
Yann Le Cam & Simone Boselli

Link:
https://ojrd.biomedcentral.com/articles/10.1186/s13023-023-02774-7

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04. **Advancing Use of Patient Evidence in Decision-Making**

Contributing author:
Virginie Hivert

Link:
https://globalforum.diaglobal.org/issue/june-2023/advancing-use-of-patient-evidence-in-decision-making/

05. How to START? Four pillars to optimally begin your orphan drug development, August 2023

Lead author:
Virginie Hivert

Link:
https://link.springer.com/article/10.1186/s13023-023-02845-9

06. Together4RD position statement on collaboration between European Reference Networks and industry, September 2023

Lead author:
Matt Bolz-Johnson, Ines Hernando

Link:
https://ojrd.biomedcentral.com/articles/10.1186/s13023-023-02853-9

07. Rare diseases' genetic newborn screening as the gateway to future genomic medicine: the Screen4Care EU-IMI project, October 2023

Lead author:
Edith Sky Gross

Link:
https://ojrd.biomedcentral.com/articles/10.1186/s13023-023-02916-x

08. Rare Disease Moonshot: Diagnostic Research Needs Recommendations, December 2023

Contributing author:
Roseline Favresse

Link:
https://www.rarediseasemoonshot.eu/wp-content/uploads/2023/12/DIAGNOSTIC-RESEARCH-NEEDS-RECOMMENDATIONS.pdf

09. **Paediatric Inflammatory Bowel Disease: A Multi-Stakeholder Perspective to Improve Development of Drugs for Children and Adolescents**

Contributing author: **Maria Cavaller**

Credentials: **J Crohns Colitis. 2023 Feb; 17(2): 249–258**

Link:
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC10024546/

10. **Treatabolome DB: linking gene and variants with treatments for rare diseases.**

Contributing author: **Gulcin Gumus**

Link:
https://hal.science/hal-04086225

11. **"Genetic newborn screening and digital technologies: A project protocol based on a dual approach to shorten the rare diseases diagnostic path in Europe."**

Contributing authors: **Gulcin Gumus and Edith Gross**

Link:
https://pubmed.ncbi.nlm.nih.gov/37992053/

12. **Genomic newborn screening: Are we entering a new era of screening?**

Contributing author: **Edith Gross**

Link:
https://pubmed.ncbi.nlm.nih.gov/37403863/

ACKNOWLEDGEMENTS

EURORDIS would like to thanks the following organisations and companies for their financial support in 2023

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.

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EUROPEAN COMMISSION
DG Health and Food Safety

- The Operating Grant for year 2023
- EUCAPA-European Capacity Building for Patients (Action Grant)

Co-funded by Health Programme of the European Union

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EUROPEAN COMMISSION
DG Research and Innovation

- The European Joint Programme on Rare Diseases (EJP RD)
- European Rare Disease Research Coordination and Support Action consortium (ERICA)
- Solving the Unsolved Rare Diseases (Solve-RD)
- The Next Generation Health Technology Assessment (HTx)
- More Effectively Using Registries to suppOrt PAtient-centered Regulatory and HTA decision-making (More-EUROPA)
- Remedi4All
- The Innovative Health Initiative (earlier IMI2) projects:
  - conect4children (c4c)
  - Screen4Care
  - FACILITATE – FrAmework for ClInicaL trIal participants' daTA reutilization for a fully Transparent and Ethical ecosystem

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

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

HEALTH SECTOR CORPORATES

EURORDIS appreciates the contributions received from health sector companies. Ensuring a sustained variety of funding is key to minimise potential conflicts of interest. EURORDIS had 76 different corporate donors in 2023.

These companies have supported EURORDIS through the EURORDIS Round Table of Companies[1], the EURORDIS Membership Meeting[2], the EURORDIS Black Pearl Awards[3], as well as International Initiatives such as Rare Disease Day[4], Rare Barometer[5] EURORDIS Open Academy[6], and multi-lingual communications.

Additional contributions include support of project development, the EURORDIS Ukraine Response programme[7], the Turkey Emergency Response 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

TAKEDA

01

HORIZON THERAPEUTICS

02

PFIZER

03

ROCHE

04

SANOFI

05

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[1] **EURORDIS Round Table of Companies:**
http://www.eurordis.org/content/ertc-members

[2] **EURORDIS Membership Meething:**
https://www.eurordis.org/eurordis-membership-meeting-emm-2023/

[3] **EURORDIS Black Pearl Awards:**
https://blackpearl.eurordis.org

[4] **Rare Desease Day:**
https://www.rarediseaseday.org

[5] **Rare Barometer:**
http://www.eurordis.org/voices

[6] **EURORDIS Open Academy:**
https://openacademy.eurordis.org

[7] **Ukraine Responce Programme:**
https://www.eurordis.org/our-priorities/eurordis-ukraine-response/

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

Adelphi ADELPHI REAL WORLD Leadership in Real World Evidence
aeglea
agios
ALEXION AstraZeneca Rare Disease
AliraHealth
Alnylam PHARMACEUTICALS
AMGEN
Amicus Therapeutics
AMRYT PHARMA
AMYLYX
argenx
astellas GENE THERAPIES
astria THERAPEUTICS
azafaros
BAYER
BeiGene
biocryst
Biogen
BIOMARIN
blueprint MEDICINES
Boehringer Ingelheim
bridgebio
Bristol Myers Squibb
camurus
Chiesi
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Cytokinetics
DENALI
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ferrer
GILEAD
GRIFOLS
GRIN THERAPEUTICS A Neurvati Neurosciences Company
illumina
Immedica pharma
Incyte
insmed
IONIS
IPSEN Innovation for patient care
IQVIA
janssen PHARMACEUTICAL COMPANIES OF Johnson&Johnson
Jazz Pharmaceuticals
Lundbeck
MERCK
Mereo BioPharma
moderna
MSD INVENTING FOR LIFE
NOVARTIS
novo nordisk
Orchard therapeutics
Orphalan Innovation for Orphan Diseases
Ovid Therapeutics
parexel
Passage Bio
Pharming
PTC THERAPEUTICS
RECORDATI RARE DISEASES GROUP
REGENXBIO
Sangamo THERAPEUTICS
Santen
SAREPTA THERAPEUTICS
Sciensus
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Boston Scientific Foundation Europe

Chan Zuckerberg Initiative

Everylife Foundation

Fondation Ipsen, under the aegis of Fondation de France

The Marigold Foundation

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 2023.

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)
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
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
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 level
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 RDConnect
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 byEC and by Member States, divided in work packages corresponding to specific activities.
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
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

EURORDIS & EUROPEAN REGULATORY NETWORK

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	People 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 (PACEERN) Consortium
PE	Patient Engagement
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.
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


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ACTIVITY REPORT

2023

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EURORDIS has received funding under an operating grant from the European Union's Health Programme (2021-2027) for this publication. Views and opinions expressed on this website are however those of the author(s) only and do not necessarily reflect those of the European Union or HaDEA. Neither the European Union nor the granting authority can be held responsible for them.

Funded by
the European Union