



# WORK PROGRAMME

2023

# EURORDIS WORK PROGRAMME 2023

Cover photo: "Springtime - When a smile lights up... even in difficult times."  
VACTERL and Total Bilateral Hemimelia  
Taken by Irene Ceneri, Italy

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# ACTION PLAN

2023



"Even with a serious illness, you can be a fashion model."  
Dima, living with Hunter syndrome  
Taken by Natalia Shugaeva, Russia





**A NEW EU POLICY  
FRAMEWORK**



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DISEASES**



**UKRAINE**

# 1. ADVOCATE

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



"Showing my colours in support of EPOF!"  
Fibrous dysplasia of bone  
Taken by Cecilia Lodi, Argentina

EURORDIS will support its strategic objectives through advocacy efforts and continue to promote rare diseases as a priority at national, European, and international levels. This includes:

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

- Exploring and consolidating political options with the EU institutions in 2023, to review and replace the overall EU rare disease strategy based on the **Rare2030** policy recommendations, as well as the calls for a **European Action Plan on Rare**

**Diseases** by the European Parliament (October 2021) and 22 Member States under the Czech EU Presidency (December 2022).

- Securing proposals impacting on rare diseases in the context of the **EU multi-annual financial framework 2021-2027** (e.g. the EU4Health Programme 2021-2027), including the sustainability of European Reference Networks, Orphanet, support for children and adult rare cancers under the EU Beating Cancer Plan; Horizon Europe 2021-2027, including the European Partnership for Rare Diseases and the future flagship European Clinical Research Network for Rare Diseases; new flagship



initiatives such as EU-level collaboration on newborn screening, European comprehensive health services for rare diseases, and an EU fund for the collection of real-world evidence.

- Involving the **European Network of National Alliances through its Council (CNA)** in advocacy activities, including through in-person meetings and monthly “hot topics” calls. We will share with the CNN medium-term advocacy plans to prepare upcoming engagement; providing tools and information for National Alliances to lead on Member States’ engagement in EU initiatives and legislation within the six strategic priority areas that are also part of the EURORDIS-CNA Common Goals and Mutual Commitments.
- Strengthening in 2023 the **European Network of Parliamentary Advocates for Rare Diseases** by engaging Members of the European Parliament (MEPs) in Rare Disease Week 2023 activities; collaborating with relevant MEPs to promote the asks of PLWRD in upcoming legislative files; and recruiting new MEPs and members of national parliaments.
- Coordinating the **Stakeholders Network for Rare Diseases established within the Health Policy Platform (HPP) of the European Commission**, as a dedicated forum for all rare disease stakeholders, identifying and implementing priority actions for 2023 and 2024.
- Promoting rare diseases as an international policy priority following the United Nations’ Political Declaration on Universal Health Coverage (2019) and the UN General Assembly Resolution on Addressing the Challenges of PLWRD and their Families (2021). Toward this end, we will continue to call for a **European Action Plan for Rare Diseases**; support the development and adoption of a WHO Resolution on Rare Diseases (desired adoption in 2024) in collaboration with Rare Diseases International (RDI) and the National Alliances in Europe; advocate for the integration of rare diseases into the next WHO Global Programme of Work; maintain a regular dialogue with WHO Europe regarding progressing a comprehensive rare disease strategy (desired outcome in 2025); co-organise an international, high-level policy event with RDI and the NGO Committee for Rare Diseases at the UN in New York

in September 2023; and, finally, chair and engage in the RDI Policy Committee, by contributing to the Committee’s identification and implementation of policy and advocacy actions.

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

- Reducing diagnostic delays for rare diseases and tackling undiagnosed diseases by calling for a harmonised European approach to newborn screening (NBS) based on our [11 Key Principles](#) and creating an EU-level expert working group to design this approach and collaborate on action between Member States.
- Using evidence and findings on the **needs and experiences of PLWRD from the EURORDIS Rare Barometer programme** to inform our contributions to policy-making and research. In 2023, the focus will be on disseminating the results of the **global survey on the Journey to Diagnosis for PLWRD** (13,300+ respondents worldwide, close to 10,500 in Europe) to a broad audience and the results of a new survey on NBS that will be carried out in 2023.

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**IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF “OPTIMISED DATA AND HEALTH DIGITAL TECHNOLOGIES FOR PEOPLE AND SOCIETAL BENEFITS”:**

- Advocating for a **European Health Data Space (EHDS)** to enable the secure sharing and use of data at the EU level in the context of the legislative proposal debated in 2023 in the European Parliament and the Council. We will promote the construction of an EHDS that optimises electronic health records, ensures the ethical use of data for research, policymaking, and treatment development, increases digital health literacy in the rare disease community and beyond, and encourages the formation of patient and public partnerships.

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF “INTEGRATED MEDICAL AND SOCIAL CARE WITH A HOLISTIC LIFELONG APPROACH AND INCLUSION IN SOCIETY GOAL OF 1/3 REDUCTION IN THE SOCIAL, PSYCHOLOGICAL AND ECONOMIC BURDEN”:

- Advocating for **integrated and holistic care** as a priority within the **European Action Plan for Rare Diseases**, the European Care Strategy, and the “Council Recommendation on access to affordable high-quality long-term care.” We will also contribute to the design of ERN initiatives on integrated health and social care through the Joint Action on ERN integration into national healthcare systems from 2023.
- Calling for the **rare disease community's access to quality and adequate social and employment rights** by contributing to consultations in 2023 on EU initiatives and legislative proposals resulting from the EU Pillar of Social Rights Action Plan.
- Supporting **improved access to disability rights by joining the EU Disability Platform** in 2023 to ensure that the disability-related needs of the rare disease community are addressed. We will also advocate for PLWRD to be provided with adequate levels of disability allowance, social protection, community services, and independent living arrangements in the Member States. We will provide inputs to the proposals for a European Disability Card for PLWRD.



Brussels Rare Disease Week, EURORDIS

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF “DEVELOPMENT AND AVAILABILITY, ACCESSIBILITY, AFFORDABILITY OF TREATMENTS, PARTICULARLY TRANSFORMATIVE OR CURATIVE THERAPIES GOAL OF 1000 NEW THERAPIES WITHIN 10 YEARS”:

- Advocating for the implementation of an **EU pharmaceutical regulatory and policy framework that addresses the challenges of PLWRDs**. This includes contributing to the revision of the **General Pharmaceutical Legislation, which includes revisions to the Regulation on Orphan Medicinal Products and Paediatric Medicines**, and responding to the draft legislation coming out in March 2023.
- Advocating for the strengthening of EU clinical research capabilities in 2023 in the context of the ACT-EU initiative. We will formulate new advocacy messages on drug repurposing, using the results from our work on the DITA Task Force, STAMP<sup>1</sup> and REMEDI4ALL<sup>2</sup>.
- Advocating for the implementation of the **Regulation on EU Cooperation on Health Technology Assessment** by contributing in 2023 to the development of the EU-netHTA methodological guidelines and to the EU HTA Cooperation Stakeholder Network. We will also follow European and national discussions on the implementation of the Regulation from 2025 and continue the dialogue on pricing policies and methodological guidelines for the economic evaluation of health technologies.
- Advocating to improve access to treatment for rare diseases by promoting the proposal of structured cooperation across European countries in pricing and reimbursement policies and the development of a European Fund to finance evidence generation post-marketing authorisation, which places particular emphasis on the rarest of diseases and advanced therapies. We will **engage with WHO Europe** to establish in 2023 the Novel Medicines

1 The STAMP project connects academic researchers with medicine regulatory agencies for early scientific advice on repurposing projects. This is instrumental to generate a robust data package to translate research into medicines access for patients.

2 REMEDI4ALL aims to establish Europe's leadership globally in the repurposing of medicines by creating a vibrant community of practice covering all relevant sectors and disciplines.



Platform - collaboration of all European countries on improving access to innovative therapies following the Oslo Medicines Initiative (OMI, 2020-2022).

- Advocating for the **harmonisation of Compassionate Use Programmes (CUP)** and/or a European fund for CUP, as they have proven to be one of the most effective approaches in bringing innovative medicines to patients and ensuring access to those most in need.

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

- Contributing to the **implementation of the cross-border healthcare legislative framework**. This will include: promoting a unified and integrated approach to monitoring and evaluating existing EU legislative measures enabling cross-border healthcare; calling for clear guidance on how to operationalise the existing legislative framework to improve access to innovative therapies.
- Shaping the concept with our members and stakeholders and advocating for an EU end-to-end approach to planning and delivering highly specialised healthcare services.
- Calling for the Mental Health EU Action Plan to be adopted in summer 2023 to integrating the needs of PLWRD and their families, recognising them as a particularly vulnerable population group.
- A new EURORDIS Mental Wellbeing (MW) Initiative is planned to be launched in 2023-24, with the overarching objective to promote the development of a ‘mentally healthy community’ that reduces the cumulative impact on mental health and wellbeing among people with rare diseases and their families. The MW Initiative will partner clinical experts and experts with lived experience to harness the existing evidence, innovations and best practices, and level up the capacities of rare disease advocates in mental health under a new EURORDIS Mental Wellbeing Partnership Network. This Network will build the capacities and resources necessary to



Brussels Rare Disease Week, EURORDIS

address the increased risk factors for poor mental health experienced by the rare disease population. EURORDIS will also leverage the opportunity of the new European Union’s Strategy on Mental Health to make visible the unmet mental health needs of people with rare diseases and their families and take affirmative action to address these needs.

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

- Continuing to increase the **visibility of rare cancers** and including them across our 2022-2023 advocacy activities.
- Focusing on policy initiatives and solutions in 2023 to address the specificities of **very rare and ultra-rare diseases**. This will be done through advocacy actions such as informing the revision of the EU Regulation on Orphan Medicinal Products and different projects such as: SOLVE-RD (undiagnosed diseases); Screen4Care (early diagnosis and newborn screening); various activities undertaken within the EMA Committees; proposing access to very rare diseases and advanced therapies; and proposing the provision of highly specialised services through the ERNs at the EU level.

## 2. EMPOWER

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

"Waterfall, Hugs, and  
Raspberries"  
Alex, living with  
STXBBP1  
Taken by Jessica  
Carr, USA



**EURORDIS will support its strategic objectives through the empowerment of its networks.**

**This includes:**

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

#### **BUILDING THE COMMUNITY, INFORMING, SUPPORTING, AND EMPOWERING OUR NETWORKS**

- Continuing to develop and engage the **Network of over 1000+ EURORDIS Members**, with a prioritisation in 2023-2024 of Eastern European, rare cancer, rare mental disorder, rare infectious disease, rare mitochondrial disease organisations, as well as those representing underrepresented therapeutic areas. We will also reach out to all European Patient Organisations at large, maintain and develop the EURORDIS contact database, and ensure interactions with members through the bi-monthly Member News.
- Coordinating and empowering the **unique network of 400+ volunteers and ePAGs organised by groups of interest or expertise**, supporting their work.
- Coordinating the **European Network of Rare Diseases National Alliances and its Council (CNA)** by organising a monthly online meeting on hot topics, including two in-person meetings of the CNA; following the adoption of the new EURORDIS' strategy, enhancing the strategic alignment of national alliances in 2023, involving National Alliances in common advocacy actions through monthly newsletters, regular emails and ad hoc group meetings, and empowering the network via regular bilateral meetings, peer-to-peer training, and sharing of best practices.
- Coordinating the **European Network of Disease-Specific European Federations and its Council of EF (CEF)** and providing continuous support to 20 of the weakest/younger rare disease European Federations by financing their network meetings and working more closely with them on key strategic items.
- Coordinating and developing the **European Network of Rare Disease Help Lines**

**(ENRDHL)** by identifying common interests, analysing the profile of callers and the purpose of their inquiries (Caller Profile Analysis), and exploring the use of Artificial Intelligence (AI) to engage helplines with social networks.

- Supporting the **safe closure of RareConnect** by providing adequate information to moderators, community members, and all partnering patient organisations.

#### **BUILDING CAPACITY FOR OUR NETWORKS**

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

- Organising **Brussels Rare Diseases Week (RDW) 2023** in the lead up to Rare Disease Day 2023 – **an in-person series of advocacy, capacity building, and networking events**, including meetings with relevant decision-makers, especially MEPs, and a conference at the European Parliament on a key legislative file. We will also ensure appropriate funding for the 2024 edition, prepare the advocacy programme, and develop an online training programme for **Rare Disease Week 2024** that takes into account the 2024 European Parliament elections.



Brussels Rare Disease Week, EURORDIS

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO CONTRIBUTE TO IMPROVING THE “DEVELOPMENT AND AVAILABILITY, ACCESSIBILITY, AND AFFORDABILITY OF TREATMENTS, PARTICULARLY TRANSFORMATIVE OR CURATIVE THERAPIES”, THE “GOAL OF 1000 NEW THERAPIES WITHIN 10 YEARS”, AND THE AMBITION OF ACHIEVING “RESEARCH AND KNOWLEDGE DEVELOPMENT THAT ARE INNOVATIVE AND LED BY PEOPLE’S NEEDS”:

- Delivering two **Schools in 2023 on “Medicines Research and Development” and “Scientific Innovation and Translational Research”** to empower patient representatives with the confidence and knowledge needed to bring their expertise to discussions on healthcare, research, and medicines development. The two schools will gather about 70 participants in Barcelona in the spring and will be followed by post-school training and alumni engagement.
- Supporting the programme development and delivery of the **European Joint Programme on Rare Diseases Leadership School** for about 60 participants in Gdansk in the autumn.
- Supporting patient engagement in therapeutic development through the continuous implementation and dissemination of materials produced in the context of the IMI PARADIGM<sup>3</sup>. We will also support patient engagement in therapeutic development by implementing new courses in 2023 within the Open Academy and developing in 2023 a framework for patient engagement in the context of the European Rare Disease Research Coordination and Support Action consortium (ERICA), which aims to improve the clinical research capacities of ERNs.
- Actively supporting patient engagement in European Medicines Agency (EMA) activities via EURORDIS’ representation on the EMA Management Board and various Committees, including the Patients and Consumers’ Working Party (PCWP) and the EMA Emergency Task Force. We will also:

EURORDIS staff,  
Barcelona



- **Support patient representatives** on the Committee for Orphan Medicinal Products (COMP), the Paediatric Committee (PDCO), the Committee for Advanced Therapies (CAT), and the Pharmacovigilance and Risk Assessment Committee (PRAC).
- Facilitate the EURORDIS Therapies Action Group (TAG), which gathers EURORDIS volunteers participating in the EMA Committees.
- Contribute to the CHMP Early Contact with Patient Organisations.
- Identify experts for Scientific Advisory Group Meetings, disseminate EMA Scientific Committee Surveys to relevant rare disease groups, and collaborate with the EMA to identify and support the participation of patient experts in Scientific Advice/Protocol Assistance procedures.
- Develop and disseminate monthly Therapeutic Reports.
- Supporting **patient involvement in HTA activities through the coordination of the EURORDIS HTA Task Force (HTA TF)** and the organisation of two face-to-face meetings. Toward this end, we will also:
  - Contribute to the implementation of the EU HTA Regulation (2021/2282).
  - Provide guidance on methods to involve patients in horizon scanning, joint scientific consultations, coping, and joint scientific assessment.
- Building the skills and knowledge of **patient advocates participating in HTAs**

<sup>3</sup> The PARADIGM project was a public-private Innovative Medicines Initiative (IMI) partnership active between 2018 and 2020. The project’s mission was to provide a unique framework that enables structured, effective, meaningful, ethical, innovative, and sustainable patient engagement and demonstrates the “return on the engagement” for all players.



with the EUCAPA – Health Technology Assessment training (EU4Health, January 2023-January 2025), ensuring that the EU HTA cooperation includes the patient perspective from the start. EURORDIS will lead the project, develop the training content, and be involved in the recruitment and selection of patients.

- Supporting **patient involvement in dialogue with payers through the Mechanism of Coordinated Access to Orphan Medicinal Products (MoCA)**.
- Supporting **patient involvement in developing quality information on medicines, through the coordination of the EURORDIS Drug Information, Transparency and Access Task Force (DITA TF)**. This will involve reflecting on how our members can be better informed about when they need to prepare for the evaluation of medicines and health technologies and how to work in parallel with the Patients and Consumers' Working Party.

IN LINE WITH OUR ORGANISATIONAL MODEL, EURORDIS – AS A NETWORK LEVERAGER – LEVERAGES A NETWORK OF MEMBER ORGANISATIONS, ADVOCATES, PARTNERS, AND STAKEHOLDERS TO ACHIEVE ITS STRATEGIC GOALS. IN 2023, WE WILL TAKE A STEP FORWARD TO STRENGTHEN OUR NETWORK LEVERAGE BY:

- Improving outreach to the network of **Open Academy alumni** through monthly newsletters, online meetups every two months, and structured communication. This will help facilitate their transition from trainees to active alumni, including follow-up training, post-school webinars, and online alumni masterclasses using a continuing education approach and peer-to-peer exchange.
- Developing the **Open Academy's e-Learning courses and digital platform** by updating the online modules for the School on Medicines Research and Development, developing whisper courses and nudge learning for the pre-training phase of the School on Scientific Innovation and Translational Research, and continuing to promote new e-learning courses. In particular, we will develop an online course about planning, designing, analysing, and disseminating surveys in cooperation with our Rare Barometer programme.

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "OPTIMISED DATA AND HEALTH DIGITAL TECHNOLOGIES FOR PEOPLE AND SOCIETAL BENEFIT":

- Promoting **digital health literacy**, including a new training in 2023 during the EURORDIS Membership Meeting on data safety and literacy. This training will teach simple ways to avoid privacy, security and well-being risks while using digital technologies. We will also conduct a EURORDIS Open Academy alumni masterclass on digital rights.
- Coordinating the **EURORDIS Digital Advisory Group (DAG)**, providing capacity-building sessions for DAG's members to be well informed and prepared to deliver their opinion in matters relating to digital health. Additionally, we will engage in the FACILITATE project and offer inputs to guide EURORDIS' policy and actions in relation to digital health aspects of clinical trials.

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

- Supporting **patient partnership in European Reference Networks (ERNs)** through:
  - Building skills and knowledge for the **network of European Patient Advocacy Group (ePAG)** advocates, creating opportunities for peer-to-peer learning.
  - Organising the annual all-ePAG meeting that will be organised back-to-back with the annual ePAG Steering Committee meeting in Barcelona.
  - Facilitating team building and shared leadership between patient representatives and clinicians within the ERNs, by contributing to the implementation of a harmonised governance approach to structure patient partnership.
- Engaging in 2023 with the ERN project managers and ePAG advocates to ensure

active participation of ePAG representatives in the five-year evaluation. Toward this end, we will:

- Explain how patients have been involved in the different activities and highlight areas of improvement in patient partnership.
- Develop new practical guides and toolkits to support patient involvement in the ERNs' activities and encourage the use of existing ones.
- Identify and promote good practices and stories on patient partnership in ERNs to develop communication resources.
- Facilitating – with special attention in 2023-2025 – the **collaboration between national alliances and ePAG advocates with regard to the integration of ERNs into national healthcare systems.**
- Supporting – with particular focus in 2023 – ERNs and ePAGs to manage the ERN application and decision-making process to onboard new ePAG advocates.

**IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF “INTEGRATED MEDICAL AND SOCIAL CARE WITH A HOLISTIC LIFELONG APPROACH AND INCLUSION IN SOCIETY GOAL OF 1/3 REDUCTION IN THE SOCIAL, PSYCHOLOGICAL AND ECONOMIC BURDEN”:**

- Relaunching the **EURORDIS Social Policy Action Group (SPAG) in 2023.** This will involve revising the Terms of Reference to consider the holistic and social priorities over the coming years, recruiting volunteers, and resuming regular meetings of the SPAG to actively contribute to relevant consultations and activities.
- Organising the **EURORDIS Membership Meeting 2023** in Stockholm (May 25/27), together with the Programme Committee around the theme of: **“Holistic approach to people's needs and full inclusion in society”** in line with our Strategic Objective to deliver on the goal of social inclusion. The EMM 2023 will gather up to 250 participants from our members to network and participate in capacity-building workshops, including on digital health.

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

- Continuing the **EURORDIS Newborn Screening Working Group (NBS-WG).** We will also continue our call for the adoption of our 11 Key Principles on NBS and maintain a dialogue in 2023 with EU institutions (DG Sante, DG Research, JRC) to design new, EU-funded instruments or projects with the aim to reduce inequalities across the EU.

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

- Continuing to provide direct assistance to ULWRD affected by the war in 2023. In particular, we aim to help them get access to healthcare services in Ukraine and Europe, as well as cover their basic needs in their host countries.
- Hosting displaced ULWRD with the help of local patient groups and partners in Ukraine and other European countries.
- Amplifying the voices of ULWRD.
- Better identifying the needs of ULWRD and advocating on their behalf where necessary.
- Collaborating with European, EU-national, and Ukrainian authorities, as well as humanitarian organisations to ensure that the needs of ULWRD both in Ukraine and abroad are covered through their response programmes.

**CAPACITY BUILDING, NETWORKING, AND AWARENESS RAISING  
ARE RECURRENT EURORDIS EVENTS THAT SUPPORT THE THREE  
EURORDIS STRATEGIC OBJECTIVES:**

- **Raising awareness at the European and international levels to achieve equity in social opportunities, healthcare, and access to diagnosis and therapies for people living with rare diseases (PLWRD).** To this end, the following actions will be taken:

- Coordinating the international **Rare Disease Day (RDD) 2023** campaign and planning RDD 2024 in over 100 countries. The campaign aims to raise awareness and works toward achieving equity in social opportunity, healthcare, and access to diagnosis and therapies for the 300 million people worldwide living with a rare disease.
- Focusing the campaign on basic calls to action around rare diseases.
- Extending the 2023 and 2024 campaigns throughout the year, with social media and patient stories in native languages, to increase the sense of belonging to the global rare disease community.
- Focusing on **young rare disease patients** and healthcare providers in collaboration with EURORDIS National Alliances.
- Co-creating and producing RDD campaign materials based on strategic review recommendations and a call to action.

- Translating media assets and webinars into different languages to spread the RDD message in more countries, especially the underrepresented ones.
- Expanding the movement of “the global chain of lights” in two ways while considering energy constraints and climate change.
- Taking advantage of the 2024 Paralympic Games to increase awareness.
- Organising the **Black Pearl Awards (BPA) 2023** as a hybrid event gathering up to 200 people in Brussels in February 2023 and up to 200 people online via an interactive platform. Participants will enjoy an awards ceremony, artistic performance, official Rare Disease Day animations, and keynote speeches. Building upon the 2023 experience, they will begin organising the 2024 edition, which will be a hybrid event with one on-site venue in Brussels or a multi-hub event.
- Preparing for the **European Conference on Rare Diseases and Orphan Products (ECRD) 2024** by:
  - Determining its format (in-person, online, or hybrid).
  - Selecting the Programme Committee.
  - Organising the conference, including the launch of a new conference website, a call for patient advocate fellowships to the ECRD, and a call for poster abstracts and registration.



# 3. PARTNER

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

"The dream does not die, it lives!"  
Desmoid Tumour  
Taken by Renan Santos, Brasil

EURORDIS will support its strategic objectives through partnerships within five horizontal strategic priority areas. This includes:



## RESEARCH POLICY AND ACTIVITIES

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

### TRANSLATIONAL RESEARCH

- Helping shape the **Rare Disease Partnership**<sup>4</sup> to ensure that the needs of PLWRD are reflected. Since November 2021, EURORDIS has been fully engaged in its co-design together with the main research infrastructure partners. The RD Partnership will consolidate and expand the European Joint Project (EJP) while creating the new European clinical research networks for rare diseases, in collaboration with the ERNs and the research infrastructures, as advocated by EURORDIS. In 2023, EURORDIS will be a key contributor to the European Commission’s Task Force established to draft the **Strategic Research and Innovative Agenda (SRIA)** that will define targeted impact, foreseen portfolio of activities, measurable expected outcomes,

<sup>4</sup> Rare Disease Partnership, a major research programme meant to start by late 2024.

resources, and milestones within a defined timeframe.

- Continuing to voice and incorporate the views of PLWRD in initiatives such as the **RD Moonshot**<sup>5</sup> (launched in 4Q2022) to enhance public-private partnerships with further engagement of private corporate partners in research.
- Participating in the **European Joint Programme on Rare Diseases**. This participation will involve:
  - Continuing to co-lead the EJP RD Pillar 3 Activities on **training and capacity building** and co-developing a 3rd EJP RD Massive Open Online Course (MOOC) in 2023 on “**Research data: ethics and regulatory challenges**.”
  - Continuing involvement in the mentoring and management of the first two EJP RD MOOCs.
  - Supporting the delivery in 2023 of the **next EJP RD paediatric patient training**.
  - Promoting and supporting patient organisations’ participation in the EJP RD calls for proposals.
  - Identifying and supporting patients as evaluators of EJP RD different calls for proposals.
  - Building further on the EJP RD PEN-REP (Patient Engagement in Biomedical Research Projects) principles by delivering in 2023 an executive version of the principles to be translated into different languages to help spread them among research communities in Europe.
  - Surveying further patients’ perceptions of their involvement in current and future research projects.
  - Contributing to the sustainability of the Orphan Drug Development Guidebook with updates, maintenance, and the development of contemporary or new content.

<sup>5</sup> The RD Moonshot is a coalition of partners from industry, research and patient organisations joining forces to accelerate scientific discovery and drug development in rare and paediatric diseases for which currently there is no therapeutic option.

“Marks on Snow”  
Cutis marmorata  
telangiectatica  
congenita (CMTC)  
Taken by Catriona  
Allen, Canada





"Best Friend's Ever"

Finja and Isabella, living with Spinal Muscular Atrophy  
Taken by Conny Wenk, Germany

- Continuing participation in the **European Rare Disease Research Coordination and Support Action (ERICA)** project up to February 2025. Toward this end, we will:
  - Contribute to the discussions and outputs on ERN registries' health data access and other data management aspects.
  - Scope the framework for patient engagement in clinical research.
  - Organise quarterly calls to share information and engage with the ePAG Research and Registries WG to obtain their feedback.

## UNDIAGNOSED DISEASES

- Participating and bringing the patient voice in the **EU-funded research project Solve-RD<sup>6</sup>** (Solve-RD, January 2018-June 2023, Horizon 2020). Toward this end, we will:
  - Lead the Work Package on Impact and Engagement.
  - Participate in the Steering Committee and in the Ethics Independent Advisory Board of the project.
  - Continue coordinating the Community Engagement Task Force and to ensure that the development of the activities within Solve-RD is patient-centred.
- Continuing to represent EU rare disease

<sup>6</sup> SOLVE-RD is a research project aiming to solve the unsolved rare disease by sophisticated omics approaches, genetic knowledge web (genes, genomic variants and phenotypes)

patients' perspectives and the priority of the undiagnosed community by:

- Participating in the **Undiagnosed Diseases Network International (UDNI)**.
- Coordinating the UDNI Patient Engagement Working Group in partnership with NORD and the Wilhelm Foundation.

## PAEDIATRIC CLINICAL TRIALS

- Continuously participating in **Conect-4Children** (C4C, May 2018 – April 2024, IMI 2): contributing to the external stakeholder management process of the pan-European Paediatric Clinical Research Network and the sustainability plan of this network; bringing EURORDIS' expertise in terms of patient engagement, e.g. drafting a White paper on improving parents and patient involvement; contributing to the work on data quality standards and to the Education and Training programme, including being a member of the Education Board.



## DATA AND DIGITAL HEALTH

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

- Ensuring a **coordinated and consistent approach to health data-linked issues within EURORDIS projects** and the development of an internal Q&A to facilitate better coordination.
- Participating in the **EU project FACILITATE<sup>7</sup>** (FACILITATE, January 2022-December 2025, IMI2). In 2023, EURORDIS will contribute to delivering:

<sup>7</sup> FACILITATE's main objective is the development of a new ethical, legal and regulatory framework to enable the return of clinical trial data to study participants and the health care professionals involved in their care. The secondary objective is to build a prototype process enabling the re-use of that data for future research needs.



- Ethical Standards and Guidelines by engaging all stakeholders.
- A report on the Stakeholder Engagement Plan. As part of this, the composition of the DAG+, a group composed of rare and non-rare patients and carers, led by EURORDIS, will be finalised. DAG+ is the patient expert group that will advise on all qualitative aspects regarding the development of the plan.
- An initial report on IMI DO-IT ICF template language updates for the processing of patient data in clinical trials, as well as for the results of such data.
- A Data management plan and a mid-term update.
- A literature review on patients' willingness to share their clinical trial data.
- Contributing to initiatives such as **Together4RD<sup>8</sup>** to leverage data to generate new knowledge and support product development.



## DEVELOPMENT AND ACCESS TO DIAGNOSTICS AND THERAPIES

IN LINE WITH OUR STRATEGIC OBJECTIVE 2 TO DELIVER ON PRIORITY AREAS AND TO MAKE CONTRIBUTIONS TO THE GOAL OF "DEVELOPMENT AND AVAILABILITY, ACCESSIBILITY, AFFORDABILITY OF TREATMENTS, PARTICULARLY TRANSFORMATIVE OR CURATIVE THERAPIES – GOAL OF 1000 NEW THERAPIES WITHIN 10 YEARS," WE ARE:

- Participating in the EU project **"More Effectively Using Registries to support Patient-centred Regulatory and HTA decision-making"** (More Europa, 2023-2028, Horizon Europe), which aims to develop, implement and establish evidentiary standards and methods to address

<sup>8</sup> Together4RD is a multi-stakeholder initiative aimed at supporting collaboration between European Reference Networks (ERNs) and industry in areas that will address the unmet medical need of 95% of rare diseases without a dedicated treatment.

the data and evidentiary needs of regulatory authorities and HTA bodies. The end goal is to progress the more efficient use of Real-World Evidence (RWE) for the development, registration and assessment of medicinal products in Europe.

- Continuing to participate in the EU project **"Next Generation Health Technology Assessment" (HTx,<sup>9</sup> January 2019-December 2023, Horizon 2020)** by:
  - Leading on the development of training materials (a toolbox) for patients and participants to the meetings of the other Work Packages.
  - Simplifying and disseminating project updates and information through communication and training materials.
- Participating in the **Get Real Institute<sup>10</sup>** as the co-founder and Board member working on reducing barriers to the use of secondary data source, bridging the gap between RCT and RWE, and addressing the evidence needs of downstream healthcare decision-makers.

<sup>9</sup> HTx aims to create a framework for the Next Generation Health Technology Assessment to support patient-centred, societally oriented, real-time decision-making on access to and reimbursement for health technologies throughout Europe.

<sup>10</sup> GetReal Institute builds on the success of two IMI projects: GetReal and The GetReal Initiative and brings together a wide variety of stakeholders to drive the sustainable development and adoption of tools, methods and best practices in the generation and use of RWE (real world evidence) for better health care decision-making.



"Lady in Blue"  
Osteogenesis Imperfecta  
Taken by Manasseh Nyirenda, Zambia

- Participating in the EU project **“VACCELERATE”**<sup>11</sup> (VACCELERATE, January 2021-January 2024, Horizon 2020) that works on Clinical Trial Site Capacity Building, Public Health Needs, Volunteer Registries, Booster Vaccination Doses in Elderly, Booster Vaccination Doses in Adults, and the vaccination regimen in children aged 12-16 years.
- Working to create more treatments for rare conditions via **drug repurposing through the “Remedi4ALL” project**<sup>12</sup> (REMEDI4ALL, September 2022 – August 2027, Horizon Europe). Toward this end, we will actively work with all stakeholders to ensure meaningful patient engagement throughout the entire drug development process and collaborate in the development of training materials such as the Annual Repurposing Academy and other virtual programmes.
- Representing EURORDIS in the **IRDiRC – International Rare Diseases Research Consortium** General Assembly and the Patient Advocacy Constituent Committee (PACC), as well as in the Therapies Sci-

<sup>11</sup> VACCELERATE is a clinical research network for the coordination and conduct of COVID-19 vaccine trials. The network is comprised of academic institutions from all over Europe: The consortium is led by the University Hospital Cologne, Germany, and currently includes 29 national partners in 18 EU-member states and 5 countries associated to the EU Horizon 2020 research programme.

<sup>12</sup> REMEDI4ALL aims to build a sustainable European Innovation Platform to enhance the repurposing of medicines. The project is expected to make a major leap forward in patient-centric drug repurposing, or finding new therapeutic options for existing drugs, in areas where there are high unmet medical needs, including the rare disease field.



“What it’s all about”  
Ford, living with CTNNB1  
Taken by Kristin Elliot, USA

entific Committee (TSC). Specifically in 2023, EURORDIS will:

- Contribute to the Task Force on Drug Repurposing Guidebook, which builds on the previous work carried out within IRDiRC on repurposing, as well as on the Orphan Drug Development Guidebook in order to provide guidance to developers undertaking repurposing approaches.
- Contribute to the Task Force on Disregarded Diseases (PLUTO project) for characterising specific commonalities among a large group of “disregarded” rare diseases, with the potential secondary aims to identify removable roadblocks to future research and development.
- Contribute to activities related to Newborn Screening in synergy with EURORDIS activities.
- Participate in the IRDiRC/RDI working group on Global Access to Rare Diseases Therapies, linking this group up to the New Medicines Platform of the WHO Europe.
- Follow-up on the work carried out within IRDiRC on Clinical Research Networks for Rare Diseases.
- Promoting the patient voice by chairing the **Screen4Care (S4C)**<sup>13</sup> **Patient Advisory Board (PAB)**, which consists of 15 patient representatives involved in the EURORDIS Newborn Screening Working Group, the Digital and Data Advisory Group, and ePAGs. The PAB provides strategic recommendations, guidance, and advice across all Screen4Care activities. This entails identifying patients’ preferences on existing symptom checkers and co-designing the S4C virtual platform.
- Working toward a definition in 2023 of ‘actionability’ in Newborn Screening terms. Toward this end, we will:
  - Finalise the **literature review on actionability within the Screen4Care (S4C, October 2021-September 2026, IMI JU) project.**
  - Launch a **Rare Barometer survey on**

<sup>13</sup> The aim of Screen4Care is to accelerate the diagnosis of rare diseases based on two central pillars: genetic newborn screening and digital technologies.



**Newborn Screening** to help frame criteria to define a list of actionable diseases to be included in the S4C ACT-Panel within the genetic Newborn Screening pilot studies.

- Continuing to engage in the **Global Commission to End the Diagnostic Odyssey for Children with Rare Diseases**, with a focus in 2023 on the results of the global survey on diagnosis, evaluation of the pilots on the diagnosis awareness campaign, and new policy initiatives.



## HEALTHCARE POLICY AND SERVICES

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

- Leading and co-designing, with a core group of pioneering ERNs, the H-CARE project, focusing in 2023 on the first phase of the PREM validation. This will involve finalising and publishing a scoping literature review to inform the definition of high-quality healthcare for rare diseases.
- Participating in the new Joint Action on integration of European Reference Networks into national health systems (JA, October 2023-2026, DG Sante). This will involve:
  - Teaming up and supporting patient organisations that will be involved in some of the activities of the Joint Action.
  - Maintaining our institutional relationship with the ERN coordinators, hospital managers, and Board of Member States representatives to contribute to the strategic planning of the ERNs for the next 5-year cycle.
  - Contributing in 2023 to the five-year

evaluation of ERNs to highlight successes but also help identify areas of improvement and measures to address shortcomings.



## SOCIAL POLICY AND SERVICES

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

- Engaging with the European Commission to provide input in 2023 to the **design of future funding instruments**, building on EURORDIS’ extensive involvement in the review of the Social Innovation (EaSI) programme. Toward this end, we will:
  - Identify and seize opportunities to develop project proposals aimed at addressing key holistic and social care priorities, notably within existing relevant funding instruments and calls for proposals, such as the European Social Fund Plus (ESF+).
  - Continue to seek opportunities to scale up the past **INNOVCare** project to support the implementation of further case management services and resource centre services.
  - Continue to support the work of the European Network of Resource Centres for Rare Diseases - RareResourceNet, as part of its Board of Directors.

# 4. CROSS-CUTTING PRIORITIES



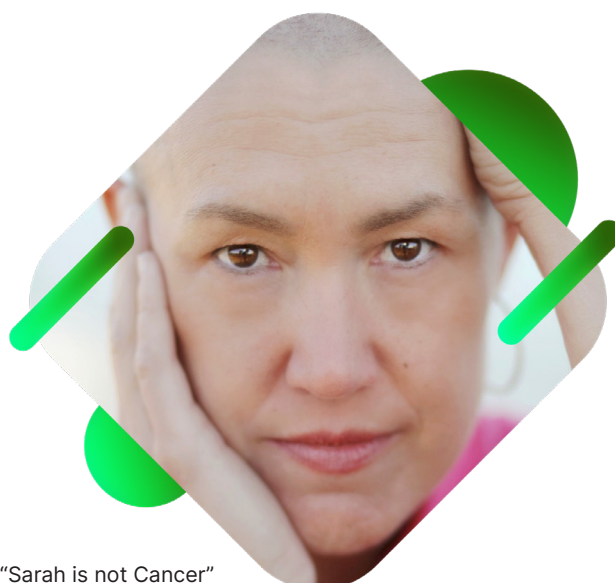
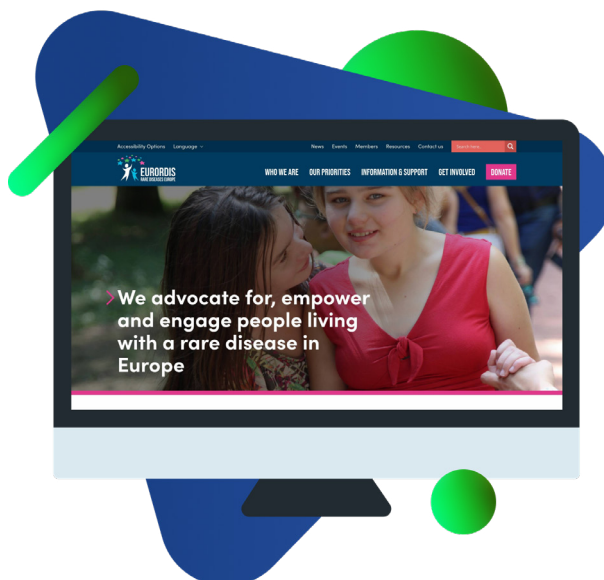
"Raising Rare"  
Blakely, living with  
Mucopolidosis type 2  
Taken by Lindsay  
Norman, USA



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

## COMMUNICATION AND DISSEMINATION

- Reviewing, updating, and implementing a **new Communication and Dissemination Strategy in 2023**, aligned with the new EURORDIS strategy for 2022-2023, leveraging our networks including by:
  - **Analysing current performance and finding new solutions** to ensure that the **new EURORDIS website** is user-friendly, intuitive, and easily accessible to all user groups.
  - Better engaging with EURORDIS' audience on **social media** by using interactive videos, animations, and infographics.
  - **Redesigning the EURORDIS monthly eNews newsletter.**
  - **Ensuring that scientific publications are easier to find** on the EURORDIS website by creating a dedicated space for them and effectively disseminating them.
- Focusing on the challenges linked to a growing number of advocacy files in 2023 while ensuring that **EURORDIS staff, Board members, and volunteers speak with one voice, drawing on the same brand identity, position papers, and talking points.** This will include updating the EURORDIS general presentation and preparing guidelines for when EURORDIS is represented externally.
- Innovating in 2023 through:
  - The launch of a **monthly EURORDIS podcast.**
  - The creation of a **Newsroom section on the website.**
  - Giving greater attention to **ultra-rare disease groups, Young Patient Advocates, and members in Eastern and South-Eastern Europe on social media.**



"Sarah is not Cancer"  
Adenoid Cystic Carcinoma  
Sarah McDonald, USA

## HUMAN RESOURCES (STAFF AND VOLUNTEERS)

- Enhancing team organisation that supports EURORDIS' strategy and operational needs with the appointment in 2023 of permanent positions (succession or new), including:
  - Human Resources Director (based in Paris, new position).
  - Office Assistant (based in Paris, new position).
  - European Public Affairs Senior Manager (based in Brussels, renewed position after disruption).
  - Rare Barometer Survey Project Manager (based in Paris, succession).
- Hiring a **Human Resources Director (new position) to lead a EURORDIS People Management System** (including staff and volunteers). This system will:
  - **Align our human resources to the new strategy.**
  - Recruit and retain talent, manage training and career development, develop HR policies, and ensure well-being at work.
  - Ensure that all HR operations are carried out smoothly and effectively.
- Supporting the development of a 360-degree view of the organisation for a part of the management team to enhance transversal work strategically, streamline operations, optimise time and fund allocation.
- Improving **EURORDIS' communication and acknowledgement of EURORDIS volunteers** on the EURORDIS website and Activity Report in 2023.
- Revising processes for effective and improved volunteer support and management.

## GOVERNANCE

- Disseminating the **revised EURORDIS long-term strategy for 2022-2030** and working toward strategic alignment with enhanced organisational capabilities and new management systems (people, workplace, information systems) to deliver on EURORDIS' strategic objectives. Toward this end, we will:
  - Conduct weekly Core Leadership Team meetings, monthly Operations Team meetings, and Advocacy and Strategy meetings to ensure the implementation of EURORDIS' strategy and action plan.
- Working toward a **multi-annual budget for 2023-2027** that implements our strategy and better manages uncertainties with funding sources (EU4Health Operating Grant).
- Renewing the **four-year multi-annual partnership with the AFM-Téléthon** that will provide a framework for common goals and financial support from 2023 to 2027.
- Managing **EURORDIS governance bodies** (General Assembly, Board of Directors, Board of Officers, Core Leadership Team) and conducting regular face-to-face and online meetings. This will involve:
  - Finalising the legal process on the revision of status.
  - Regularly reviewing and updating **EURORDIS by-laws**.
  - Enhancing governance processes and establishing two-way, structured communication between the Board and staff.
- Maintaining, reviewing, and developing **strategic partnerships (Memorandum of Understanding – MoU) with international patient organisations and other stakeholders**.



## RESOURCE DEVELOPMENT & SUSTAINABILITY

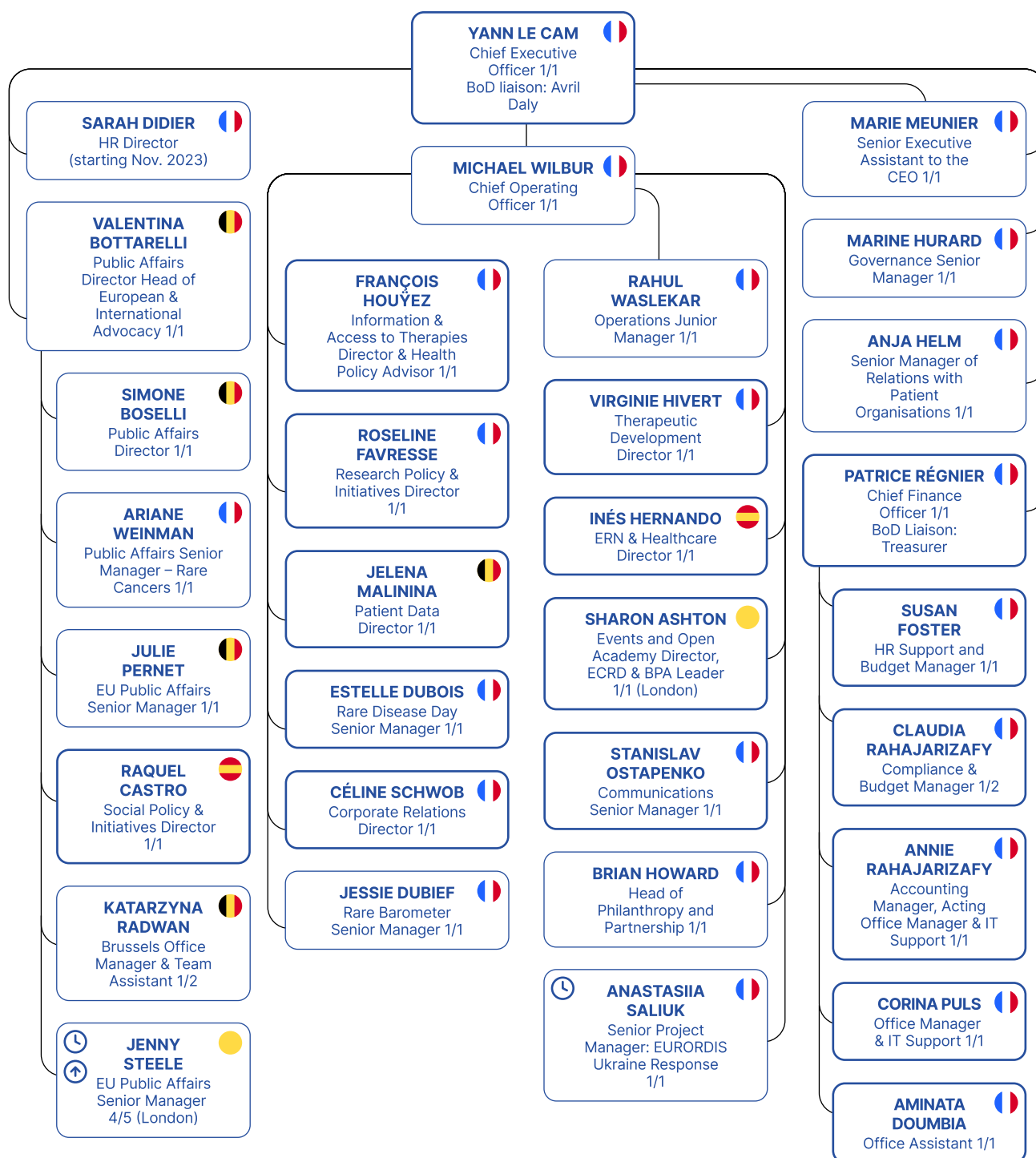
- Supporting the **EURORDIS Round Table of Companies (ERTC)** by engaging over 70 health companies in both bilateral and collective dialogue through regular webinars and two major face-to-face workshops focused on specific topics.
- Maintaining activity to support current contributions from the health sector within the confines of the EURORDIS Policy of Relationship with commercials.
- Seeking **new sources of funding** to implement our strategic objectives, including diversified sources from foundations and individual donors.
- Investing in **developing strategic projects based on innovative partnerships**.



"Enjoying life at Full speed"  
Jara, living with Dyskeratosis Congenita  
Taken by Uria Oficialdegui, Spain



# TEAM ORGANISATIONAL CHART EURORDIS JANUARY 2023



PARIS 

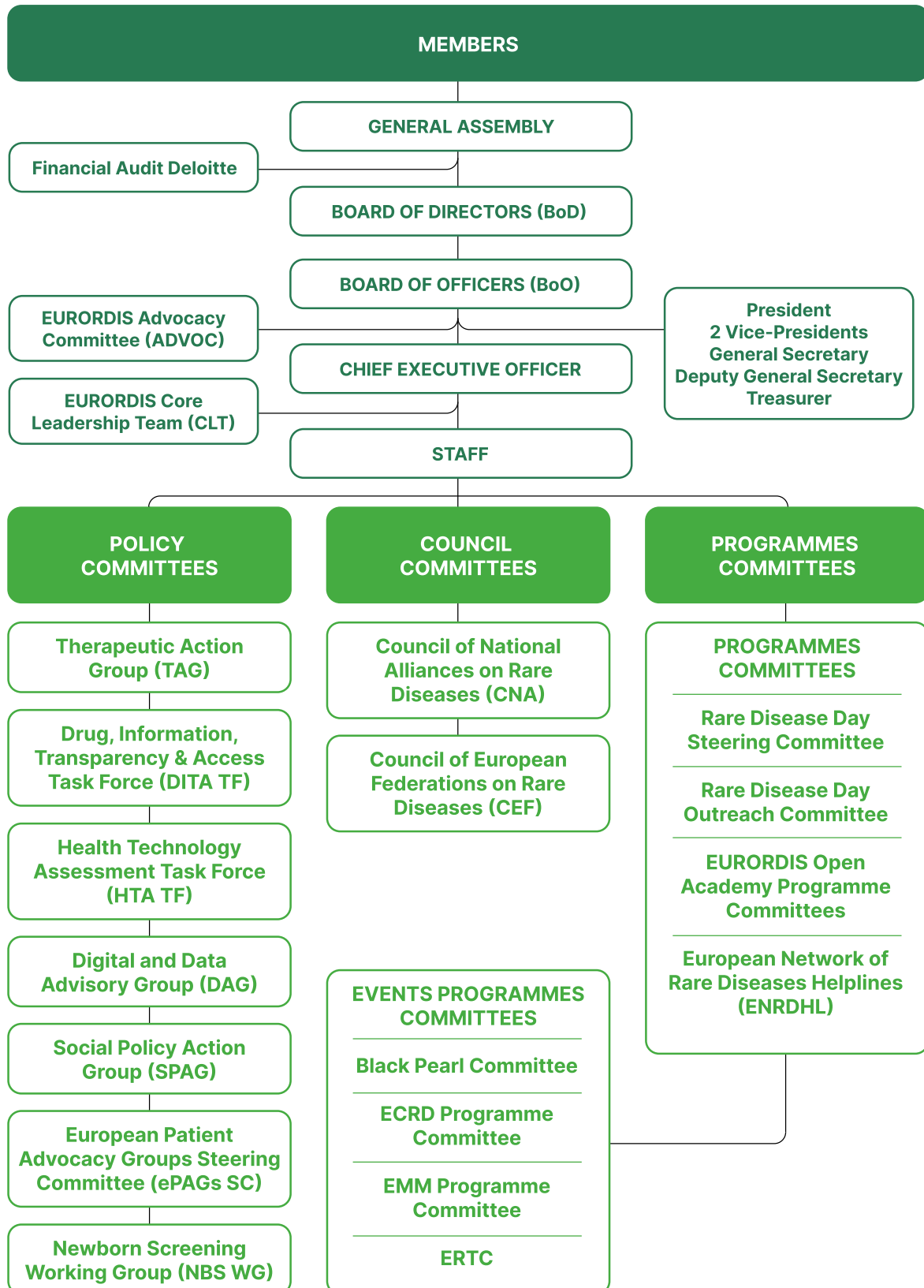
BRUSSELS 

BARCELONA 

OTHER 



# EURORDIS INTERNAL GOVERNANCE CHART 2023







"Get breathless for Pulmonary Hypertension"  
Juro, living with Pulmonary Hypertension  
Taken by Matej Djokić, Croatia



# EURORDIS EXTERNAL REPRESENTATION IN NETWORKS, ORGANISATIONS AND INSTITUTIONS IN 2023



## EUROPEAN NOT-FOR-PROFIT ORGANISATIONS & INITIATIVES

EPF: European Patients' Forum (founding member)

EU4Health Civil Society Alliance (founding member)

Rare Cancer Europe (founding member)

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

EDF: European Disability Forum

Social Platform - European Platform of European Social NGOs

RareResourceNet European Network of Resource Centres (Board member)

EFGCP: European Forum for Good Clinical Practice

Friends of Europe

FIPRA - International Policy Advisors

Rare Disease Platform in Paris (founding member)

Maladies Rares Info Service (French Helpline for RDs)

Get Real Institute (founding member)

ARRIGE - The Association for Responsible Research and Innovation in Genome Editing

RWE4DECISIONS

TOGETHER4RD

TRANSFORM

GO FAIR RD network

## PARTNERING WITH EUROPEAN NOT-FOR-PROFIT ORGANISATIONS & INITIATIVES

EFPIA European Federation of Pharmaceutical Industries and Associations Think Tank, Moonshot; and dialogue on Real World Evidence

European Expert Group on Orphan Drug Incentives (OD Expert Group)

## MEMBER OF EUROPEAN NETWORKS & PROJECTS

BBMRI Stakeholders Forum

HTx - Next Generation Health Technology Assessment

OpenMedicine

C4C - Conect4Children

CORBEL - MIUF

Decentralised Trial Project

SolveRD - Solving the Unsolved Rare Diseases

reCOVID consortium IMI2

EJP RD - European Joint Programme on RD

EUCAPA - European Capacity Building for Patients

MoreEuropa - More Effectively Using Registries to support Patient-centered Regulatory and HTA decision-making

REMEDI4ALL - The European Platform for Medicines Repurposing

Screen4Care - Shortening the path to RD diagnosis by using newborn genetic screening and digital technologies

VACCELERATE - pan-European backbone for the acceleration of phase 2 & 3

FACILITATE - Framework for Clinical Trial Participants' Data Reutilization for a Fully Transparent and Ethical Ecosystem

ERICA - European Rare Disease Research Coordination and Support Action

TEHDAS - Towards the European Health Data Space

LIVES

PROJECT STEERING COMMITTEES

- EuroCAB programme
- ERN-Hub for Ukraine
- RareConnect

EUROPABIO Patients Advisory Group

EUCOPE: European Confederation of Pharmaceutical Entrepreneurs

EUPATI-Spain (partnering on EuroCAB)

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

RD1: Rare Diseases International (founding member)

ORPHANET

IRDiRC: International Rare Disease Research Consortium (founding member)

UDNI - Undiagnosed Diseases Network International

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

NEWDIGS: New Drug Development ParadIGms

IAPO: International Alliance of Patients' Organizations

ICORD: International Conference on Rare Diseases and Orphan Drugs (founding member)

PFMD - Patient Focused Medicines Development Initiative

CIOMS - Council for International Organizations of Medical Sciences

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

## PARTNERSHIP WITH LEARNED SOCIETIES

European Hospital & Healthcare Federation (HOPE)

International Federation of Social Workers Europe (IFSW-Europe)

European Society of Human Genetics (ESHG)

International Society for Pharmacoeconomics and Outcomes Research (ISPOR)

European Connected Health Alliance - ECHAlliance

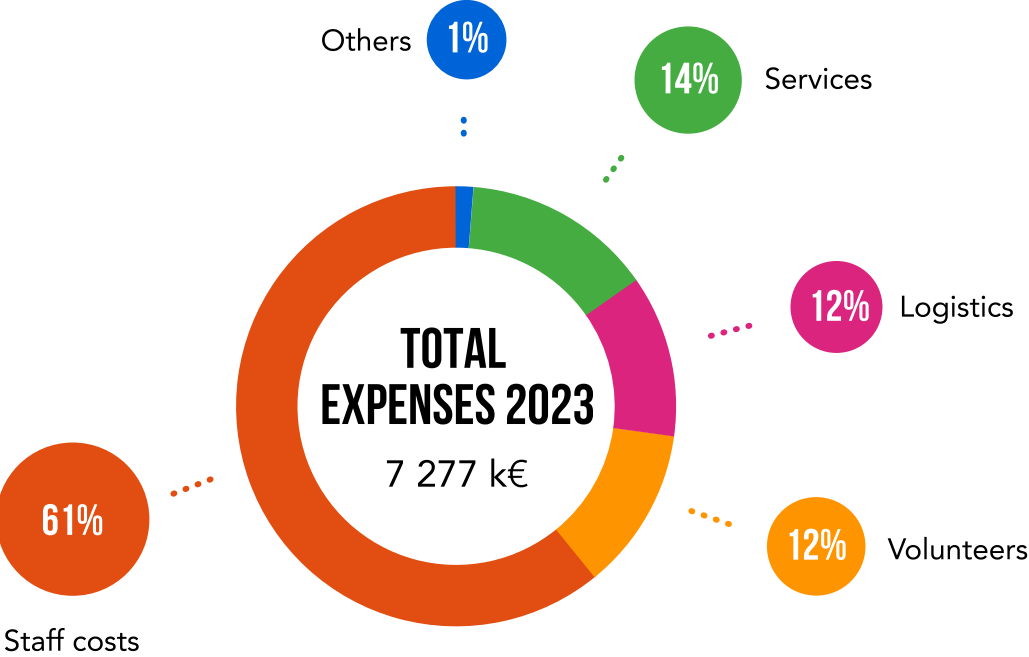
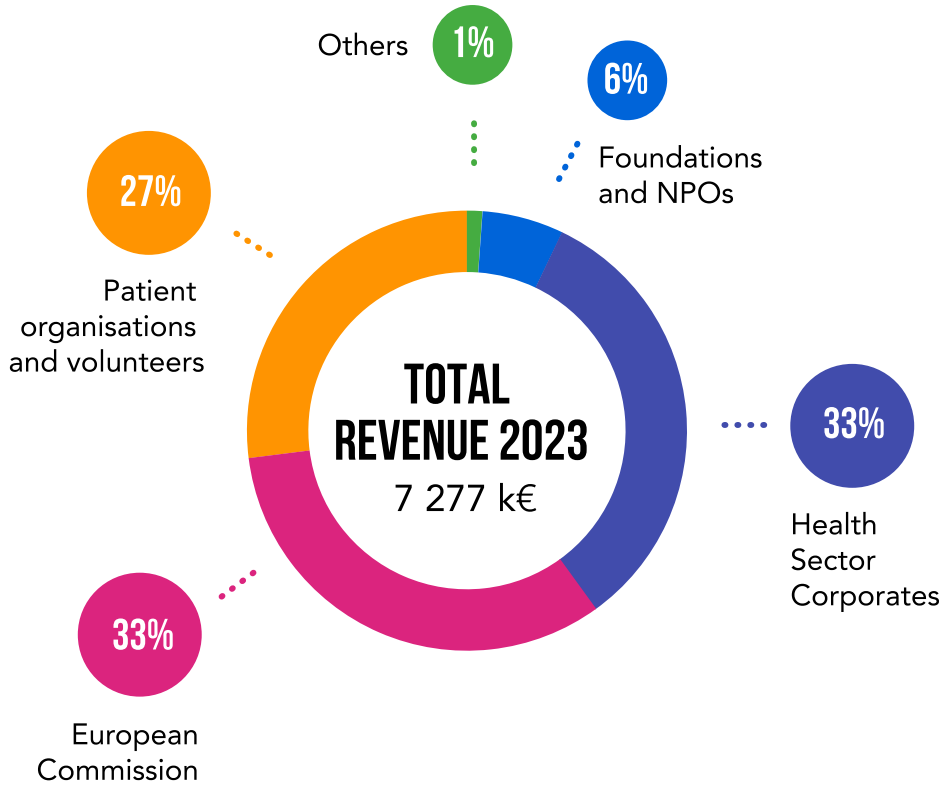
European Union of Medical Specialists (UEMS)

European Alliance for Personalised Medicine

European Association of Health Law



# EURORDIS REVENUE AND EXPENSES 2023





"Warrior Princess Abigail"  
Hyperoxaluria Type 1  
Taken by Lauren Carwardine, Canada



# ACRONYMS AND DEFINITION

## EURORDIS INTERNAL AND TASKFORCES

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

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

|  |  |
|--|--|
| <b>ERTC</b>                            | EURORDIS Round Table of Companies (with pharma & biotech developing Orphan Medicinal Products)   |
| <b>OA</b>                              | EURORDIS Open Academy  |
| <b>ePAG</b>                            | European Patient Advocacy Group  |
| <b>ECRD</b>                            | European Conference on Rare Diseases and Orphan Products   |
| <b>NGO Committee for Rare Diseases</b> | The NGO Committee for Rare Diseases engages at the UN level to elevate rare diseases to a priority within global public health.  |
| <b>RDD</b>                             | Rare Disease Day   |
| <b>RDI</b>                             | Rare Diseases International  |
| <b>EMM</b>                             | EURORDIS Membership Meeting  |
| <b>AGA</b>                             | Annual General Assembly  |
| <b>BPA</b>                             | Black Pearl Awards   |
| <b>RDW</b>                             | Rare Disease Week  |
| <b>RareConnect</b>                     | A safe, easy to use platform where rare disease patients, families and patient organizations can develop online communities and conversations across continents and languages.   |
| <b>Rare Barometer</b>                  | Rare Barometer is a community of people living with a rare disease who are willing to participate in EURORDIS-Rare Diseases Europe surveys and studies.  |
| <b>Rare 2030</b>                       | Rare 2030 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. |
| <b>BBMRI Stakeholders' Forum</b>       | Biobanking and Biomolecular Resources Research Infrastructure  |
| <b>ECRIN</b>                           | European Clinical Research Infrastructures Network   |
| <b>E-Rare</b>                          | Network of ten partners – public bodies, ministries and research management organisations – from eight countries, responsible for the development and management of national/ regional research programs on rare diseases  |



|                          |  |
|--------------------------|--|
| <b>EUnetHTA Forum</b>    | Support effective HTA collaboration in Europe that brings added value at the European, national and regional levels  |
| <b>EuroBioBank</b>       | European Network of DNA, cell and tissue banks for rare diseases – EU project 2003 – 2006 for which EURORDIS was administrative coordinator. It is now the biobank network of RD-Connect   |
| <b>EUROPLAN</b>          | Fostering National Plans in Europe (project ended in 2018)   |
| <b>EUPATI</b>            | Innovative Medicines Initiatives Joint Undertaking “Fostering Patient Awareness on Pharmaceutical Innovation”  |
| <b>EJA</b>               | "Joint Action on Rare Diseases of the EU Committee of Experts on Rare Diseases: Funded by EC and by Member States, divided in work packages corresponding to specific activities, e.g. continuity of Europlan (Work Package 4); developing guidelines for social services dedicated to RDs (Work Package 6)" |
| <b>FACILITATE</b>        | The project is focused on GDPR compliance in the context of clinical trials, and will look to find a way referencing previously completed trials to facilitate data sharing in a way that respects the needs and wishes of patients and adds value to all stakeholders.                                      |
| <b>Global Commission</b> | The Global Commission To End The Diagnostic Odyssey For Children With A Rare Disease is a group of multidisciplinary experts with the goal of achieving timely diagnosis for all rare diseases, especially those affecting children.   |
| <b>GCOF</b>              | Genetic Clinics of the Future: To map the opportunities and challenges that surround the clinical implementation of next generation sequencing technologies, Horizon 2020, 2015-2017   |
| <b>HTx Project</b>       | HTx is a project funded by the European Union which aims to take this to the next level.   |
| <b>InnovCare</b>         | Innovative Patient-Centred Approach for Social Care Provision to Complex Conditions, DG Employment and Social Innovation (EaSI), 2015-2018   |
| <b>IMI</b>               | Innovative Medicines Initiative  |
| <b>IRDiRC</b>            | International Rare Disease Research Consortium   |
| <b>PARADIGM</b>          | PARADIGM's mission is to provide a unique framework that enables structured, effective, meaningful, ethical, innovative, and sustainable patient engagement (PE).  |
| <b>SCOPE</b>             | The Strengthening Collaboration for Operating Pharmacovigilance in Europe (SCOPE) Joint Action   |
| <b>SCREEN4CARE</b>       | Its goal is to shorten the time to diagnosis and treatment for patients with a rare disease using a dual strategy based on NBS and AI tools.   |
| <b>SOLVE-RD</b>          | "Solve-RD - solving the unsolved rare diseases" is a research project funded by the European Commission for five years (2018-2022)   |
| <b>TREAT-NMD</b>         | Translational Research in Europe – Assessment and Treatment of Neuromuscular diseases  |
| <b>Web-RADR</b>          | Development of tools for patients and healthcare professionals to report suspected adverse drug reactions to national EU regulators, Innovative Medicines Initiative (IMI), 2014-2017  |
| <b>RD-Action</b>         | Joint Action to expand and consolidate the achievements of the former EUCERD JA, DG Sanco, 2015-2018   |
| <b>EJP RD</b>            | European Joint Programme for Rare Diseases   |

## EURORDIS & EUROPEAN REGULATORY NETWORK

|             |   |
|-------------|---|
| <b>CAT</b>  | Committee for Advanced Therapies  |
| <b>CHMP</b> | Committee for Human Medicinal Products                                  |
| <b>SAG</b>  | Scientific Advisory Group at the Committee for Human Medicinal Products |

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

### EUROPEAN COMMISSION

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

### NON GOVERNMENTAL PARTNERS

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

|                    |   |
|--------------------|---|
| <b>IAPO</b>        | International Alliance of Patients' Organizations   |
| <b>IFSW-Europe</b> | International Federation of Social Workers  |
| <b>Inserm</b>      | French National Institute for Health and Medical Research   |
| <b>ISPOR</b>       | International Society for Pharmacoeconomics and Outcomes Research   |
| <b>MRIS</b>        | Maladies Rares Info Services (French helpline for rare diseases)  |
| <b>NORD</b>        | National Organization for Rare Disorders (USA) - EURORDIS' counterpart in the US  |
| <b>RDI</b>         | Rare Diseases International   |
| <b>Orphanet</b>    | The online portal for rare diseases and orphan drugs: orpha.net   |
| <b>NORBS</b>       | The National Organisation for Rare Diseases Of Serbia   |
| <b>HUFERDIS</b>    | Hungarian Federation of People with Rare and Congenital Diseases  |
| <b>RADOIR</b>      | Rare Disease Foundation of Iran   |
| <b>San Pau</b>     | Fundació de Gestió Sanitària de l'Hospital de la Santa Creu i Sant Pau (Health Management Foundation of the Hospital de la Santa Creu & Sant Pau) |

## MISCELLANEOUS

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



|                         |   |
|-------------------------|---|
| <b>ADR</b>              | Adverse Drug Reaction   |
| <b>CUP</b>              | Compassionate Use Programme   |
| <b>ATMP</b>             | Advanced Therapy Medicinal Product  |
| <b>NBS</b>              | Newborn Screening   |
| <b>NGS</b>              | Next-Generation Sequencing  |
| <b>UN</b>               | United Nations  |
| <b>CoNGO</b>            | Conference of Non-Governmental Organizations in Consultative Relationship with the United Nations   |
| <b>#Resolution4Rare</b> | #Resolution4Rare is a campaign to support the call for a UN Resolution on Addressing the Challenges of Persons Living with a Rare Disease (PLWRD) and their Families. |



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