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Framing the European Rare Diseases field through a structured movement of patient organisations

Maria Cavaller-Bellaubi, Virginie Hivert, Roseline Favresse, Yann Le Cam

EURORDIS - Rare Diseases Europe, Paris 75014, France.

Correspondence to: Maria Cavaller-Bellaubi, Sant Antoni Maria Claret 167, Barcelona 08025, Spain. E-mail: maria.cavaller@eurordis.org

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Abstract

EURORDIS-Rare Diseases Europe is an alliance of patient organisations, working across countries, empowering patients with trainings, and generating evidence through surveys. It advocates for the needs of people living with rare diseases in Europe, influencing policy, legislation, research, healthcare services, social inclusion, medicines development and access. Since its creation, EURORDIS has maintained a global perspective. By nature of the rarity of these conditions, knowledge and expertise are scattered, so collaboration on a global level is required to have an understanding of disease progression. Thus, every rare disease patient group must be equipped to collaborate beyond its own borders. EURORDIS impacts global activities with a focus on raising awareness; empowering a global alliance; supporting international, national and regional initiatives; and recognising rare diseases within the international community while fostering collaboration among stakeholders. For these reasons, the organisation's impact extends beyond Europe, as demonstrated by its international partnerships. This perspective paper explores EURORDIS' achievements over two decades, examining its role in shaping policies and regulations, to demonstrate the vital importance of global collaboration in the rare disease field. Looking ahead, EURORDIS' Strategic Goals, to be achieved by 2030, align with the United Nations Sustainable Development Goals Agenda 2030 and advocate for a European Action Plan for Rare Diseases. This illustrates EURORDIS' continued commitment to transforming policies into tangible outcomes. With its ongoing dedication, collaborative efforts, and enduring impact on the rare disease community, EURORDIS remains a driving force for positive change in the lives of people living with rare diseases.

Keywords: Rare diseases, patient organisations, ecosystem, policy, framework, empowerment, advocacy, partnership

INTRODUCTION

EURORDIS is a non-governmental patient-driven alliance of 1,042 patient organisations in over 74 countries, covering more than 4,000 diseases. EURORDIS is a purpose- and value-driven organisation, multinational and multilingual by nature, with an impact in Europe and beyond. EURORDIS is an independent organisation with strong governance and balanced revenue sources[1].

Since its establishment in 1997, EURORDIS has been at the forefront of advocating for the rights and well-being of people living with rare diseases (PLWRD) globally. Building upon a powerful community by uniting PLWRD, their families, and support groups, EURORDIS has influenced research, healthcare, medicines development and access, and has simultaneously shaped policies at national, European, and global levels for more than two decades. EURORDIS has done so by promoting awareness, disseminating information, enabling networking, and sharing experiences to empower PLWRD, support groups and patient organisations[2]. EURORDIS, as a civil society organisation, brings together key stakeholders to engage with policy makers and competent authorities.

This perspective paper explores some of the main achievements of EURORDIS over the past two decades and examines how the organisation has contributed to impacting the lives of the 30 million people in Europe facing the challenges of living with rare diseases.

ADVOCACY - INFLUENCE FOR CHANGE

EURORDIS was created in 1997 by four funding members: the *AFM-Telethon, AIDES*, *Vaincre La Mucoviscidose*, and *La Ligue Nationale Contre le Cancer* with the initial goal to advocate for European Union (EU) laws to bring medicines to PLWRD[2].

Since then, EURORDIS has been a strong advocate for PLWRD at the EU level and beyond. Through its policy, advocacy efforts, and innovative solutions, the organisation has enabled change, advocating for rare diseases (RDs) to be recognised as a public health priority at the EU level, improving access to healthcare services, research funding, and development of orphan drugs. By creating a trustful and collaborative environment and dialogue with policymakers, regulators, researchers, developers, clinicians, payers, and other stakeholders, EURORDIS has successfully influenced the development of policies, regulations, and frameworks to address the challenges faced by PLWRD, improving the quality of life.

The founding members and EURORDIS have played a crucial role in shaping the EU Regulation on Orphan Medicinal Products (2000)[3] from contributing to reports, working closely with the National Organisation for Rare Disorders (NORD), the Food and Drug Administration (US FDA), leading patient advocates, industry associations in Europe, and the French EU Presidency and Minister of Social Affairs, Health and Cities, Simone Veil, in 1995, to promoting the key features and positive impact of the US Orphan Drug Act through media and conferences (French Senate, October 1996). The organisation went on to work with the European Commission on the legislative proposal (1997-1998) and with the European Parliament Rapporteur, Françoise Grossetête (1998-1999), on the amendments and eventual adoption.

As a direct result of EURORDIS' efforts, in 2008, the Commission issued a communication to promote RDs as a priority in the Member States through national plans and strategies. Together with the Council recommendation on addressing the challenges of PLWRD (2009), this led to new opportunities for policy development and investment in rare diseases. The organisation has also played a key role in shaping other legislation such as the Paediatric Regulation (2006)[4], the Cross-Border Healthcare Directive (2011)[5], and more recently, the Health Technology Assessment Regulation (2022)[6] and the European Health Data Space

(EHDS) Regulation (2024)[7], among others.

EURORDIS' way of working to promote and shape legislation, including national plans, is through consultation with its members. This includes: surveys of PLWRDs, structured multi-stakeholder dialogues such as EU-funded projects[8], the biennial European Conference on Rare Diseases (ECRD)[9], and the EURORDIS Round Table of Companies (ERTC)[10]; EURORDIS Position Papers[11] and numerous contributions to the Commission's fact-finding studies and public consultations. Upon the commencement of a new piece of legislation, EURORDIS engages in active dialogue with the Commission services drafting the legislative proposals, the most relevant Member States, and the members of the European Parliament who are rapporteurs for the legislation. After its adoption, EURORDIS plays a key role in the implementation of legislation, thanks to its European network of national alliances.

Most recently, with the revision of the General Pharmaceutical Legislation[12], EURORDIS aims to ensure that incentives are provided to keep enhancing research towards effective treatments for PLWRD.

The establishment of the 24 European Reference Networks (ERNs) in 2017 marked another significant milestone in EURORDIS' contributions to the RD field. These networks connect specialised healthcare centres across Europe, facilitating collaboration, knowledge, and expertise sharing. EURORDIS pushed and defined rules and guidelines for the creation, development, and functioning of the ERNs, while ensuring that they address the specific needs of PLWRD through cross-border healthcare[13].

EURORDIS advocacy style has evolved and matured over the years. Early on, EURORDIS initiated scientific publications to support its evidence-based advocacy[14]. In 2015, EURORDIS equipped itself to perform independent surveys through the Rare Barometer Programme. Rare Barometer runs surveys in 24 different languages through a panel of more than 20,000 PLWRD and/or their carers across the globe and conducts qualitative and quantitative studies[15]. The results, generated directly from the experiences of PLWRD, enable EURORDIS to deploy an evidence-based advocacy strategy geared towards person-centric policies [Figure 1].

EURORDIS places the empowerment of patients and capacity building of patients' organisations at the core of its activities. Through training programmes and educational resources, the EURORDIS Open Academy has equipped hundreds of patient advocates and researchers from more than 50 countries with the essential skills and knowledge to become effective patient representatives and change makers, as well as to act as full partners in research[16]. By strengthening the collective voice of PLWRD, EURORDIS enables greater patient engagement in research projects[17], Research & Development (R&D)[18], and participation in decision-making processes[19]. Some concrete examples include identifying patients, mentoring and supporting them to navigate and effectively contribute to regulatory processes within the European Medicines Agency (EMA)[20] and Health Technology Assessment bodies, as well as in early dialogues with developers and payers through the Mechanism of Coordinated Access (MoCA)[21].

EURORDIS also acts as a network leverager connecting advocates and stakeholders. The strategic approach of EURORDIS creates a critical mass voicing the common challenges across all RDs and European countries for the 30 million PLWRD. Over the years, EURORDIS has developed a set of activities involving the (at present) 36 European National Alliances[22] and the 80 European or International Federations[23] to foster cross-fertilisation amongst countries, member organisations, and disease areas, resulting in a pan-European movement of over 2,000 rare disease patient organisations.

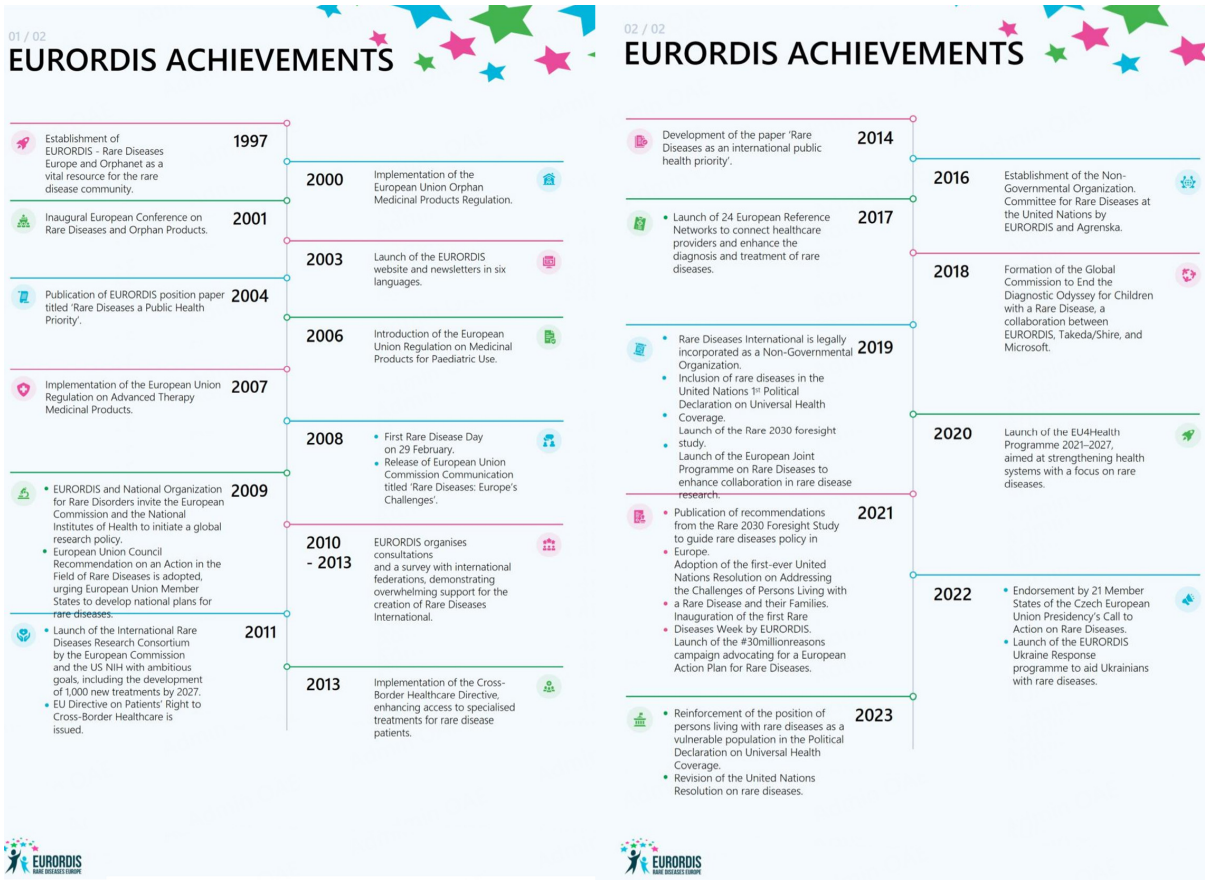

Figure 1. EURORDIS achievements.

Every two years, the EURORDIS Membership Meeting (EMM) provides an opportunity for all EURORDIS members to network with researchers, healthcare professionals, academia, and healthcare industry representatives to learn from each other and improve the quality of care of PLWRD[24].

Another impactful initiative is the Rare Disease Day campaign launched in 2008. This global awareness campaign takes place annually on the last day of February to raise public understanding of rare diseases, and advocate for better support and research for PLWRD. Through Rare Disease Day, EURORDIS successfully engages millions of people worldwide, with thousands of events organised every year in over 100 countries across the world. This global movement fosters a sense of unity among the rare disease community and provides a critical focal point through which patient organisations can capture the imagination of runway at the national and local levels for PLWRD[25].

Since 2017, EURORDIS has established the Network of Parliamentary Advocates for Rare Diseases. This initiative helps to elevate the patient perspective in new legislation and EU public policy[26].

PARTNERING WITH THE RD COMMUNITY

EURORDIS is instrumental in not only voicing patients' concerns and priorities but also supporting and engaging them as actors and equal partners. By establishing and facilitating networks with strategic partners and key stakeholders, the organisation has contributed to advances in diagnostic care pathways, research, holistic care, patient engagement, and therapy development, among other areas. The outcomes have been

invaluable in shaping research priorities, informing policymaking, and driving resources towards improving the lives of PLWRD[8].

In 2009, EURORDIS and NORD inspired the European Commission (EC) and National Institute of Health (NIH) to join forces with other public funders to promote research in rare diseases at the global level through the establishment of the International Rare Diseases Research Consortium (IRDiRC) in 2011[27]. IRDiRC is a global consortium aimed at advancing rare disease research by working together to build a multi-stakeholder collaborative approach to research funding and priorities, and to achieve targeted goals[28].

In 2013, Ågrenska Foundation and EURORDIS, together, thanks to their status as Non-Governmental Organisations (NGOs) and working relationship with the United Nations (UN), created the NGO Committee for Rare Diseases at the UN ECOSOC Committee of NGOs[29]. EURORDIS also supported the creation of Rare Diseases International (RDI), the global alliance of rare disease patient organisations worldwide. EURORDIS mobilised patient groups from around the world to support the initiative and provided financial and in-kind resources until 2023. Now, RDI is a full, independent, legitimate, and credible international NGO. EURORDIS-RDI's major joint accomplishments have been the inclusion of rare diseases in the UN political declaration of universal health coverage in 2019, which was reinforced in 2023. Additionally, the Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families was adopted in 2021[30].

REPOSITIONING EURORDIS FOR THE FUTURE: SCIENTIFIC ADVANCEMENTS, GROWING KNOWLEDGE AND TECHNOLOGIES

EURORDIS' more than 25 years of experience have positioned the organisation to lead the way for the future of the rare disease community, aligning with the UN Sustainable Development Goals Agenda 2030, One Sustainable Health Approach, and the future European Health Union. These initiatives promote an innovative, transformative European model of care that will be driven by the needs of PLWRD and a collective responsibility to prioritise solidarity and equity[31]. The Foresight Rare 2030 study enabled a multi-stakeholder consensus analysis of the main challenges and trends for the next 15 years. This study has confirmed the positioning of EURORDIS as a network leverager within an increasing and complex ecosystem and produced a set of recommendations[32]. Since these recommendations were published, EURORDIS and its members have been calling for a European Action Plan for Rare Diseases which would bridge the gaps between all actions and initiatives on rare diseases at the national and European levels[33].

Besides EURORDIS' long-term strategy, based on three complementary pillars - empowerment, partnering, and advocating - three strategic objectives have been defined. The first one is a new European policy framework to achieve measurable goals to prolong and improve the lives of PLWRD by 2030. The second goal is to deliver on six priority areas [Table 1]. The last one is to "leave no one behind" through the inclusion of all rare diseases with specific targeted actions (e.g., auto-immune rare diseases, rare infectious diseases, rare health hazards, ultra-rare diseases, the one in a million) in all regions of Europe, combining policy frameworks (e.g., Universal Health Coverage, UN Resolution, WHO Resolution, and WHO Work Programmes) with bottom-up empowerment of patient groups and clinicians, and local dialogue on national strategies[31].

PROMOTING AND SUPPORTING NEW FLAGSHIPS INITIATIVES: HOW THIS WILL BE TRANSLATED IN THE ECOSYSTEM?

With the upcoming changes to the General Pharmaceutical Legislation and other forthcoming initiatives,

Table 1. EURORDIS priority areas of work

Six priority areas:
1. Earlier, faster and more accurate diagnosis within 6 months
2. High-quality national and European healthcare pathways, including cross-border healthcare. The goal is to improve survival by 3 years on average over 10 years and to reduce the mortality of children under 5 years of age by one-third
3. Integrated medical and social care with a holistic life-long approach and inclusion in society by reducing the social, psychological, and economic burdens
4. Research and knowledge development that is innovative and led by the needs of people living with a rare disease
5. Optimised data and health digital technologies for the benefit of people living with a rare disease and society at large
6. Development and availability, accessibility, and affordability of treatments, particularly transformative or curative therapies

EURORDIS' next important milestone is to transform all the frameworks and current policies into opportunities of care and treatments for patients.

One of the latest initiatives is the Rare Diseases Moonshot, launched in 2022, aimed at accelerating research in "white spot" areas where no treatment is available, and where no research is being undertaken. This coalition of stakeholders to which EURORDIS contributes is a natural follow-up of previous joint endeavours aimed at building a trustful and collaborative research environment to speed up public-private partnerships for the ultimate benefit of patients[34].

EURORDIS is a full partner of the European Joint Programme on Rare Diseases (EJP RD programme). In 2019, EURORDIS constituted a group of patient representatives, funding agencies, and researchers in order to discuss patient partnerships in research and to develop a guide with best practices on how to co-create research, from the initial stages of biomedical research, together with patients and researchers. Those guidelines were the first guidelines in the biomedical rare diseases research area at the EU level and have helped address challenges in developing patient partnerships in research by providing information and resources to applicants trying to navigate the application process. Building upon the experience of the EJP RD[35], where the EU and Member States jointly co-fund activities, the European Rare Diseases Research Alliance (ERDERA) shall coordinate national, local and European research and innovation programmes from September 2024 onwards. ERDERA goes deeper in research cooperation, encompassing clinical research networks, a concept that EURORDIS has strongly pushed for many years, along with coordinating training activities and bringing patients' voices to research programmes, as has been done within EJPRD since 2019.

EURORDIS is also very active in scaling up the training opportunities provided to patients through the EURORDIS Open Academy and advocating for further development and progress in healthcare settings. For instance, since 2014, EURORDIS has been instrumental in developing the terms of reference for the ERNs, followed by the development of dedicated European Patient Advisory Groups (ePAGs)[36]. To further support ERNs' development and turn them into concrete opportunities for patients to receive treatment and/or to enrol in clinical research studies, EURORDIS is supporting the Together4RD initiative[37] which aims to alleviate the barriers to ERN-industry collaboration[38].

CONCLUSION

EURORDIS' achievements and contributions to the rare disease community are vast and far-reaching. EURORDIS has constantly redefined its unique role in the rare disease ecosystem. From its initiation as a network leverager engaging patient organisations, to its active involvement in shaping policies, fostering research, empowering patient organisations and individuals to be involved in decision making, EURORDIS

has played a pivotal role in driving positive change for PLWRD in Europe and beyond.

By 2030, in a new vibrant environment, EURORDIS will have catalysed multi-stakeholder collaborations and led impactful motivated networks of member organisations as well as new generations of advocates across diseases and borders. EURORDIS will continue to generate evidence, provide the views and opinions of PLWRD, and support the integration of the European Action Plan with national actions in key policy areas.

The organisation's dedication and collaborative efforts have had a profound impact on the lives of millions, making it a driving force for the rare disease community. Keeping a steady compass, clear vision, and strong values will ensure that EURORDIS keeps fulfilling its mission to improve the lives of people with rare diseases for the years to come.

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Drafting the table and the figure: Cavaller-Bellaubi M, Le Cam Y

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