CHAMPIONING THE RARE

Building the Engine of an Inclusive European Health Union

“RARE IS NOT RARE IF YOU SEE HOW MANY WE ARE. NO ONE DESERVES TO WALK THIS ROAD ALONE.”

ALEKSANDRA, THE NETHERLANDS

EURORDIS' priorities for the 2024 EU elections
Rare diseases affect over 30 million people, as well as their families and caregivers, across Europe. Since most rare diseases are chronic, progressive, degenerative, disabling and frequently life-threatening, they have a profound impact on the daily lives of people with a rare condition and their families. While this collective number is high, the number of people living with each of the 6,000 rare diseases is low and scattered across countries.

The most effective strategies to improve the lives of EU citizens living with rare diseases are therefore cross-border and EU-wide. At the time of writing, people living with a rare disease still have high unmet needs in a wide range of areas, across the globe and in Europe. There is still a lot to do to ensure equitable access to appropriate diagnosis, treatment, health, and social care, and to guarantee the full inclusion of people living with a rare disease in society.

In view of the upcoming European elections, EURORDIS - Rare Diseases Europe and its over 1,000 rare disease patient organisations, are calling for rare diseases to be a priority of the candidates to the European Parliament, the future European Commission, and all other relevant policymakers. We are calling on the EU to build a true European Health Union for rare diseases, including Treaty amendments to centralise decision-making and improve policymaking.

European policymakers have a rare opportunity to bring policy into step with scientific, technological, and societal shifts and to ultimately improve the lives of people living with a rare disease.

We believe that the following recommendations are key to building an inclusive European Health Union that leaves no-one behind.
1. European Policy Framework on Rare Diseases

2. Earlier, Faster, and More Accurate Diagnosis

3. Integrated National and European Healthcare Pathways

4. Timely Access to Affordable and Innovative Treatments

5. Integrated, Person-Centred, and Lifelong Holistic Care

6. Innovative and Needs-Led Research and Development

7. Optimised Data for Patient and Societal Benefit

8. Sustainable and Resilient Treatment Development

Our Priorities for EU Elections
Today, there is no comprehensive European framework or strategy to address the unmet needs facing the 30 million of Europeans living with a rare disease. The European Commission’s 2008 Communication on rare diseases, followed by the Council Recommendation a year later, have been cornerstone policies enabling many successes, including the spearheading of dedicated national plans across European countries. Yet, the policies of the past no longer keep pace with the needs of our community. We have new technologies and expectations that need to be reflected in policies from the EU and its Member States.

The European Union has regrettably failed to fully return to the question of how to improve the lives of people with rare diseases across a full breadth of policy areas. These past years, a solid political consensus in favour of a comprehensive European Action Plan for Rare Diseases has emerged, spanning successive EU Council Presidencies, EU Member States, and the European Parliament, who in their Resolution on the EU public health strategy in the post-COVID-19 era, called for such an action plan at the EU level. It is time for a fresh start in European collaboration on rare diseases.

THE EUROPEAN UNION SHOULD:

- Create a comprehensive and goal-based European Action Plan for Rare Diseases to address the existing high unmet needs of people living with a rare disease and bridge the gaps between different legislative pieces on data, research, treatment, healthcare, social protection and wellbeing, as well as national and EU initiatives. This Plan should set common objectives and measurable goals, as stated in the Rare 2030 Foresight study, to drive EU countries to:
  
  * Deliver a better and longer life for people living with a rare disease, with sub-targets such as reducing diagnostic delays to six months;

  * Ensure equal opportunities for people living with a rare disease, wherever they live, with sub-targets such as reducing by one-third the psychological, social, economic and environmental risk factors that increase the vulnerability of people living with a rare disease and their families;

  * Enable people living with a rare disease to benefit from innovation and resilient infrastructures, with sub-targets such as ensuring the approval of approximately 1,000 innovative (symptomatic or transformative) treatments for people living with a rare disease in the next ten years, irrespective of their status as orphan medicinal products;

  * Create a dedicated Drafting Group for a European Action Plan for Rare Diseases.
Because rare diseases are complex and patients are scattered across various countries, the search for an accurate diagnosis very often becomes a prolonged “odyssey” for people living with a rare disease. Combined with a lack of appropriate psychological support while awaiting diagnosis, this reality often leads to serious mental, financial and social distress for both the person with the condition and their family. Diagnosis is the first step in care pathways, hence the first step towards any improvements in health and wellbeing, which would highly benefit from greater collaboration amongst stakeholders at every level of the care process.

Furthermore, the heterogeneity of national capacities regarding screening and genetic testing and different approaches to the adoption of evolving technologies result in inequalities for patients and families across Europe. With the overarching goal for all people living with a rare disease to be diagnosed within six months of coming to medical attention [1], EU Member States and the European Union have a key role to play in co-designing adequate care pathways and in enhancing collaboration, gathering expertise, sharing best practices and supporting research.

**THE EUROPEAN UNION SHOULD:**

- Promote **equal access to rare disease diagnostics across Europe through an integrated approach**, ensuring screening and diagnostic opportunities for all;

- Sustain European networking of specialised healthcare providers, including ERNs for rare and complex diseases, and cross-ERN expert panels, guiding individuals from diagnosis to top-quality care and appropriate ERNs when needed;

- Support research and the equitable implementation of advanced diagnostic technologies across all stages of healthcare, including preconception, pregnancy, birth, and throughout life, including newborn screening and genome sequencing techniques; [2]

- Prioritise enhancing interoperability and the standardisation of data systems to **effectively support diagnostics**, particularly through transnational collaboration, in order to diagnose the most complex presentations, rare diseases, and unresolved cases;

- Acknowledge undiagnosed rare disease patients as a vulnerable population and establish a comprehensive European approach to individuals with currently undiagnosed conditions, guaranteeing access to optimal care and support irrespective of a diagnosis;

- Assist Member States in **informed and transparent decision-making for primary and secondary prevention** and promote **equitable access to well-structured newborn screening programmes across the EU**. This can be achieved through, for example, the establishment of an EU-level multistakeholder expert group;

- Help Member States establish training for primary and emergency care personnel to improve triage and patient referrals to expert centers.
Rare diseases are not only considered rare due to their low prevalence, but also the scarcity and geographic dispersion of healthcare experts who can treat them. Unfortunately, this has created a ‘geographic lottery’, in which some patients are ‘fortunate’ enough to live reasonably close to true experts in their conditions and may benefit from the fruits of their knowledge and experience. However, many patients may be limited in accessing the best existing care and treatment for their disease.

Numerous approaches and tools have been identified at the European level to eradicate such inequalities, which often exist between and within countries. For instance, the European Reference Networks (ERNs) have facilitated the dissemination, exchange and creation of knowledge and expertise across Europe. Enhanced pan-European, and indeed global, cooperation is called for to address the inequalities citizens face in accessing highly specialised healthcare and innovative therapies.

**THE EUROPEAN UNION SHOULD:**

- Sustain equitable and timely access to specialized healthcare for those with rare or complex conditions, integrating ERNs into cross-border and national health systems;
- Improve health outcomes for people with rare diseases by expanding evidence-based specialised healthcare, achieved through effective integration of ERNs into national health services using seamless connectivity mechanisms and structures;
- Secure sustainable EU budget investments to strengthen the capabilities of Centres of Expertise and enhance ERNs' competencies, management, and services. ERNs should receive support for training future rare disease experts, including clinical and surgical training, as well as education and training for local healthcare and therapy professionals;
- Define and implement specific solutions and European support for better cooperation and specialised healthcare delivery to meet the needs of individuals affected by extremely rare diseases (i.e. diseases impacting less than 500 people in the EU, as well as those requiring complex interventions with an annual procedure count below 500);
- Promote the strengthening of national person-centred and integrated healthcare systems, including by supporting research and innovation and the upskilling of the health workforce;
- Support and enable effective patient partnership and representation of people living with a rare disease in healthcare decision-making, including the ERNs.
The rare disease community still has vast unmet needs, and access to treatments and therapies was identified as the top priority for people living with a rare disease by 2030 [2]. The EU Regulations on Orphan Medicinal Products and Paediatric Medicines helped transform the lives of many people living with rare diseases and continues to be successful in fulfilling its primary purpose – attracting investment to the development of therapies for life-threatening or debilitating diseases for millions of people who today live without any, or without satisfactory, treatment options. Yet major difficulties in developing solutions for underserved areas and accessing approved treatments still exist. Whilst the number of approved therapies has increased dramatically, they have been overly concentrated in a certain selection of medical areas, whilst other rare conditions with lower prevalence have been neglected. Only 6% of all known rare diseases have an approved treatment option, and 69% of rare disease patients have received only symptomatic treatment for their rare condition, whilst unacceptable inequities between countries on time to accessibility of treatment still persist [3].

THE EUROPEAN UNION SHOULD:

- Evolve the incentives framework within the Pharmaceutical Package to target diseases lacking treatment options, prioritise early dialogue for evidence generation, boost European competitiveness, and ensure consistent sponsor rewards;

- Establish an R&D model focused on addressing the unmet needs of individuals with rare diseases, with compulsory involvement of rare disease patient advocates throughout the entire process, including research, development, regulatory procedures, and assessment and appraisal processes;

- Establish a comprehensive European pathway, from development to access, to balance innovation and affordability, achieve strategic autonomy in R&D, and minimise delays in patient access to treatments. This can be achieved through strategies like expanding early access or compassionate use programmes, ensuring legal clarity for cross-border access to approved treatments, and reviewing the directive on pricing transparency for medicinal products under national health insurance systems;

- Strengthen European collaboration among Member States in pricing and negotiation efforts, as voluntary cooperation mechanisms have gained momentum recently through multi-country negotiation platforms. These initiatives should be further consolidated and unified, while respecting existing treaty competencies, and can be reinforced through common purchasing initiatives, especially for orphan medicinal products;

- Define and implement specific policy and legislative measures to establish a European pathway to access complex therapies such as Advanced Therapy Medicinal Products, that can only be delivered in a few specialised centres throughout the EU.
People living with rare diseases often have specific care and independent living needs due to the very nature of their diseases, which are often serious, chronic, highly complex, degenerative, and associated with comorbidities. In addition, most people with a rare disease live with disabilities which can be visible or invisible. They often must go through long, complex and fragmented care pathways and are at risk of reduced income or unemployment, social isolation and discrimination, as the coordination of care tasks falls mostly on the family, and primarily on women.

Another major challenge for people living with a rare disease is obtaining an adequate disability assessment in their Member States – a situation often leading to exclusion from accessing social and independent living rights at national and European levels, including those deriving from the European Strategy for the Rights of Persons with Disabilities. These people are thus exposed to increased psychosocial, economic and environmental risk factors which affect their health and wellbeing. Consequently, people with rare diseases experience three times higher rates of depression than the general population.

**THE EUROPEAN UNION SHOULD:**

- **Acknowledge the specific complex and intersectional needs** of families with rare diseases, including holistic long-term care, and propose further initiatives to ensure their equal access to education, employment, and social protection, in alignment with the EU Charter of Fundamental Rights and the European Pillar of Social Rights;

- **Promote comprehensive, person-centred, lifelong care and support pathways** at the national level, involving rare disease representatives in policy design and implementation;

- **Ensure timely and equitable access to these care pathways**, including psychosocial support, social protection, and assistive services, throughout individuals’ lives;

- **Assist Member States in enhancing their disability assessment frameworks** and ensuring access to social, disability, and independent living rights for those with rare diseases;

- **Allocate Multiannual Financial Framework funds to support EU-wide networks and innovative projects for sharing best practices in rare disease care**;

- **Support Member States in providing training for healthcare providers** based on the expertise of rare disease specialists and patient organisations;

- **Recognise the mental health and emotional wellbeing needs** of rare disease individuals, in alignment with the WHO’s position recognising them as one of the vulnerable populations concerning EU policies on mental health;

- **Promote mental health and psychosocial support programmes for rare disease individuals and their families**, as per the UN General Assembly Resolution. [6]
Research leads to a better understanding of rare diseases, quicker and more accurate diagnoses, innovative treatments and cures, and better healthcare. It represents hope for the 30 million people living with a rare disease in Europe and their families. While improvements have been made in the past decades with dedicated public funding and coordinated actions at the EU level, rare disease research faces political and practical obstacles such as inadequate funding, small patient populations for clinical trials, and a lack of coordinated resources for patient registries. This translates into insufficient knowledge of these diseases and delays in both diagnosis and the development of much-needed treatments. The EU should continue to promote cross-border collaboration on rare disease research.

THE EUROPEAN UNION SHOULD:

- Support European cross-sectoral collaboration in rare disease research, particularly under the Horizon Europe’s Partnership for Rare Diseases;

- Invest in pre-competitive infrastructures to advance patients’ needs-led research, boosting the research capability of ERNs, including to conduct clinical research and trials;

- Optimise the use of limited resources through strategic investments and incentivising all stages of research to achieve excellence in basic, transnational, clinical, and social research;

- Develop and fund a robust European regulatory science agenda with particular attention to the specificities of rare diseases, emerging technologies, advanced therapies and innovations in the diagnosis field and to ultra-rare and disregarded conditions which lack therapeutic options;

- Promote and facilitate patient engagement in rare disease research;

- Enhance long-term multinational public-private research partnerships and sustain international research collaboration through IRDiRC (International Rare Diseases Research Consortium) and other international consortiums.
Sharing health data to advance scientific research and improve clinical practice is of particular importance in the field of rare diseases, where knowledge and expertise are limited, patient populations are geographically dispersed, and health data scattered. For people living with a rare disease, data equates to hope: it is an opportunity to access specialist healthcare in other European countries, the path to an earlier diagnosis, or the chance to understand more about a little-understood disease through research. In practice, sharing data across countries is hampered by divergent national rules and the lack or inoperability of electronic health records. The European Health Data Space Regulation, as proposed by the European Commission, is an unprecedented opportunity for Europe to unlock the potential of health data in Europe and ultimately improve the lives of people living with a rare disease through enhanced and safe access to data, as well as through advancing diagnosis, treatment, care and research. However, there is a clear need to find the right balance between ensuring that patient data is safe and secure and allowing these data to be made available to boost knowledge on rare diseases.

THE EUROPEAN UNION SHOULD:

• Support the harmonisation, optimisation and interoperability of electronic health records to ensure safe and timely sharing of health data across countries, thereby facilitating cross-border healthcare;

• Ensure the ethical use of secondary health data. Secondary use should enable health providers, research organisations and regulatory bodies to have access to health data for the purposes of research, innovation, policymaking, educational activities, patient safety, regulatory activities, or personalised healthcare. Access to cross-border health data should become possible within a trusted governance framework based on clear rules and standards, with guidance from the ERNs;

• Increase digital health literacy. The rare disease community is often faced with decisions on highly sensitive data categories, such as genetic data and cross-border uses of data through established ERNs. It is therefore essential that the new system of data sharing has educational programmes for citizens and healthcare professionals to ensure choices are well-informed;

• Encourage patient and public partnership in policy and programme design and evaluation to understand what people living with a rare disease expect from rare disease research and data sharing, while making sure their wishes and needs are embedded within research and healthcare delivery design. The new data sharing governance should reflect patients’ needs – both in terms of developing robust standards ensuring secure, ethical and responsible data sharing, and allowing health data to be seamlessly shared across borders to benefit all people living with a rare disease.
The development of treatments and therapies as well as resource-intensive provision of care, require a wide range of resource commitments, including commitments of organizational, financial, natural, energy, and even animal and human biological resources. Given the global constraints posed by increasing demands on environmental resources, supply chain limitations, and socioeconomic changes, it is crucial that the EU pays attention to these challenges. There is a pressing need for the EU to shift towards a more integrated and resilient approach that effectively balances and optimises the health and wellbeing of individuals, animals, and the environment throughout the entire lifecycle of research, development, and access to treatments and therapies, all the way to the provision of health or social care. An adaptive and balanced approach will foster a deeper understanding of current and future co-benefits, risks, trade-offs, and opportunities, ultimately advancing equitable and holistic health solutions for people living with rare diseases.

**THE EUROPEAN UNION SHOULD:**

- Facilitate the development of digital tools, telemedicine, and more mobile *healthcare*, which aim to make clinical trials more easily accessible and convenient for trial participants, whilst maintaining the protection and transparent use of patients’ health data;

- Implement the 3R principles in regulatory medicine testing [7], which relate to replacing animals with non-animal methods when feasible, minimise the number of animals needed for valid results, and refine practices to enhance animal welfare and reduce stress. This will ensure the optimal use of animals while maintaining scientific validity;

- Establish a comprehensive initiative to promote collaboration within the *pharmaceutical industry for enhanced environmental sustainability*. This should involve implementing green chemistry to reduce environmentally hazardous by-products, minimising packaging waste, and optimising efficiency in transport and logistical processes;

- Establish a comprehensive framework to develop resilient and innovative treatments for *individuals with rare diseases*. This framework should enable agile adaptation to changing circumstances, such as environmental or socioeconomic risks, while preserving the continuity and integrity of planned research.
“COLOURS BREATHING”
BY SUPRATIM DAS
“DALMATIAN(S)”
BY LUCÍA LAMATA
REFERENCES

[1] Rare2030 Recommendations, Goal 2: https://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf

[2] RARE 2030 FORESIGHT STUDY, June 2021

[3] Rare Barometer 2019, Results unpublished but available upon request


Scan to read the summary of our priorities

EURORDIS—Rare Diseases Europe is a unique, non-profit alliance of over 1,000 rare disease patient organisations from 74 countries that work together to improve the lives of over 300 million people living with a rare disease globally.

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