



#ACTRARE2024

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 - 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 other relevant policymakers.

As we approach the European Parliament elections, 30 million people living with a rare disease in Europe call for a world where they can have longer and better lives and achieve their full potential, in a society that values their well-being and leaves no one behind.

We believe that the following recommendations are key to building an inclusive European Health Union that leaves no-one behind:

**EUROPEAN POLICY
FRAMEWORK ON RARE
DISEASES**

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**EARLIER, FASTER,
AND MORE ACCURATE
DIAGNOSIS**

02

**INTEGRATED NATIONAL
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**TIMELY ACCESS TO
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EUROPEAN POLICY FRAMEWORK ON RARE DISEASES

No comprehensive framework or strategy currently addresses the challenges of the rare disease community, and policies of the past no longer keep pace with the needs of people living with a rare disease. Building on a solid political consensus [1], we call on the European Union to **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 to break the silos of different legislative pieces on data, research, treatment, healthcare, social protection and well-being, as well as national and EU initiatives.

[1] See for instance MEPs' call for an EU Action Plan for RD and 21 Member States endorsing Czech EU Council Presidency's Call to Action on rare diseases.



EARLIER, FASTER, AND MORE ACCURATE DIAGNOSIS

Due to the complexity and rarity of these diseases, the search for accurate diagnoses often becomes a "**diagnostic odyssey**", causing significant mental, financial, and social hardships. Differences in national capacities and approaches to diagnosis create inequalities across Europe. We urge the European Union to **coordinate a unified approach**, promoting **collaboration among specialised healthcare providers, sharing best practices among Member States** to guarantee **earlier, faster and more accurate rare disease diagnoses across Europe**. Additionally, **EU-wide approaches to newborn screening** and support for the best **research and advanced diagnostic technologies** is crucial to providing all children with equal opportunities across Europe.



INTEGRATED NATIONAL AND EUROPEAN HEALTHCARE PATHWAYS

Rare diseases pose unique challenges not only due to their low prevalence but also because of the scarcity and dispersion of healthcare experts. This has created a **"geographic lottery"** limiting many patients' access to optimal care. To address these inequalities within and between countries, we call on the EU to ensure **timely, equal access to highly specialised healthcare** for individuals with rare or complex conditions. This can be achieved through the **integration of the European Reference Networks (ERNs)** into both cross-border and national healthcare systems. We also urge the EU to define and implement specific solutions and to provide support for better cooperation and specialised healthcare delivery, particularly for **ultra-rare diseases** impacting fewer than 500 people in the EU.



TIMELY ACCESS TO AFFORDABLE AND INNOVATIVE TREATMENTS

Currently, only 6% of rare diseases have approved treatments, with 69% of rare disease patients receiving only symptomatic care. [2] Despite the OMP Regulation's success, accessing approved treatments remains challenging. In the creation of a European pathway from development to access, we urge the EU to systematically engage with patients and their representatives at all stages of the research and development and regulatory processes. The EU should establish a **robust framework**, focusing on patients' unmet needs, **promoting innovation and access to transformative affordable treatments**, including through early access, compassionate use, early dialogues, and enhanced pricing and reimbursement cooperation, backed by unified European purchasing methods.

[2] Rare Barometer 2019, EURORDIS



INTEGRATED, PERSON-CENTERED, AND LIFELONG HOLISTIC CARE

People with rare diseases have unique challenges, including obtaining appropriate disability assessments and facing a higher-risk of mental health issues. To address these issues, **the European Union should recognise the lifelong care needs of people living with a rare disease and their families and assist Member States in developing comprehensive care path-ways**, including well-trained healthcare and social service providers. Additionally, the EU should **support Member States in sharing good practices to improve national disability assessment frameworks**, to ensure that there are **no gaps in the integration of complex diseases causing disabilities**, and to **recognise the rare disease community as a vulnerable population** with specific mental health needs in its policies on the matter.



OPTIMISED DATA FOR PATIENT AND SOCIETAL BENEFIT

Sharing health data is vital for advancing rare disease research, given the dispersed knowledge, expertise, and patient populations. The **European Health Data Space Regulation** presents an opportunity to unlock the potential of health data in Europe for the benefit of rare disease patients. The EU must **balance data security with enabling data access for new therapies**. We urge the EU to **support harmonising and optimising electronic health records for secure cross-border data exchange**, along with establishing a **trusted governance framework that involves ERNs for ethical data utilisation**. **Collaborating with patient representatives** is crucial for understanding patients' expectations in research and data-sharing during policy development and evaluation.



SUSTAINABLE AND RESILIENT TREATMENT DEVELOPMENT

The development of treatments and therapies, along with resource-intensive care provision, involves a wide range of commitments, including organisational, financial, natural, energy, and even biological resources. The EU must adopt **a more integrated and resilient approach that balances and optimises the health and wellbeing of individuals, animals, and the environment** across the entire research, development, and access lifecycle of treatments and care. We urge the European Union to **facilitate the development of digital tools, telemedicine, and mobile health-care** to enhance accessibility and convenience for clinical trial participants. Implementing **3R principles in regulatory medicine testing**, ensuring reduced and rethought animal use while maintaining scientific validity, is essential.



INNOVATIVE AND NEEDS-LED RESEARCH AND DEVELOPMENT

Research enhances our understanding of rare diseases, leading to quicker diagnoses, innovative treatments, and improved health-care. Challenges like inadequate funding, small patient populations for trials, and uncoordinated patient registries persist, resulting in delays in diagnosis and treatment development. The **EU should continue fostering long-term cross-border collaboration in rare disease research**, with a focus on regulatory science tailored to rare diseases, leveraging technology, and advanced therapies. Specifically, we urge the EU to **bolster the research capacity of ERNs, support cross-sector partnerships** in rare disease research under **Horizon Europe**, ensure **continuity for the European Joint Programme for Rare Diseases**, and facilitate patient engagement in research.



Rare diseases affect over 30 million people in Europe, impacting their families and caregivers. These conditions are often chronic, progressive, degenerative, and life-threatening, significantly affecting daily life. While the collective number is substantial, individuals with each of the 6,000 rare diseases are few and dispersed across countries.

1 IN 2,000

is the EU's threshold for classifying a rare disease, affecting that number or less of citizens

30 MILLION

people live with a rare disease in 48 countries in Europe

6,000+

is the estimated number of distinct rare diseases

72%

of rare diseases are of genetic origin

To improve the lives of EU citizens with rare diseases, effective strategies must be cross-border and EU-wide. Currently, unmet needs persist for people with rare diseases in various areas globally and in Europe. Much work remains to ensure equitable access to diagnosis, treatment, healthcare, and social support while promoting the full inclusion of people with rare diseases in society.

We need renewed European action for rare diseases to rebuild the momentum around national plans and strategies, and everything within them. Nobody can do it alone for rare diseases.

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VICTORIA
PATIENT ADVOCATE



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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.

Scan the QR-code to read the detailed version of our priorities or visit our website: www.eurordis.org

