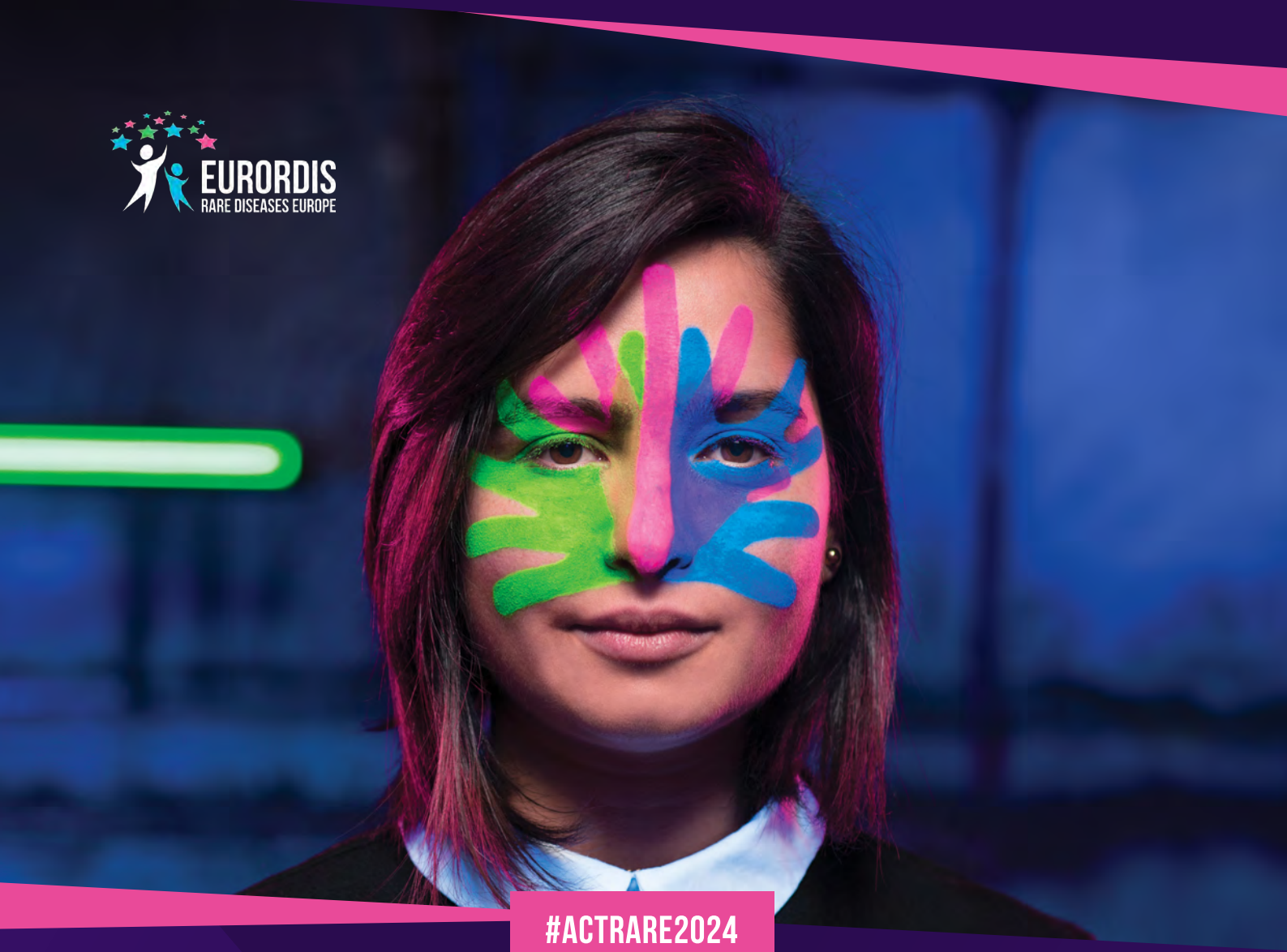
EURORDIS
RARE DISEASES EUROPE



#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

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.

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

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.

"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."

VICTORIA, PATIENT ADVOCATE

OUR PRIORITIES FOR EU ELECTIONS

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EUROPEAN POLICY FRAMEWORK ON RARE DISEASES

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, 21 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.**

EARLIER, FASTER, AND MORE ACCURATE DIAGNOSIS

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 well-being, 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 diagnostics** for rare diseases across Europe, ensuring comprehensive screening and diagnostic options for all;
- Strengthen **European networks of specialised healthcare providers, including ERNs** for rare diseases, and cross-ERN expert panels, guiding patients from diagnosis to high-quality care;
- Support research and equitable implementation of advanced **diagnostic technologies at all healthcare stages**, including **newborn screening and genome sequencing**; [2]
- Prioritise enhancing **data system interoperability and standardisation** for effective diagnostics, especially through transnational collaboration for complex and rare diseases;
- Recognise **undiagnosed rare disease patients** as a vulnerable group and **establish a comprehensive European approach** for optimal care, regardless of diagnosis;
- Assist Member States in decision-making for prevention, promoting **equitable newborn screening programmes across the EU**, facilitated by 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.

INTEGRATED NATIONAL AND EUROPEAN HEALTHCARE PATHWAYS

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.**

TIMELY ACCESS TO AFFORDABLE AND INNOVATIVE TREATMENTS

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** in the Pharmaceutical Package to focus on diseases lacking treatments, encourage early dialogue for evidence generation, enhance European competitiveness, and ensure consistent rewards for sponsors;
- **Develop an R&D model centred on the unmet needs of rare disease patients**, mandating patient advocate involvement in all stages, including research, development, regulatory processes, and assessment;
- **Create a comprehensive European pathway from development to access, balancing innovation and affordability**, aiming for strategic autonomy in R&D, and reducing delays in patient access to treatments. Strategies could include expanding early access programmes, clarifying legal aspects of cross-border treatment access, and revising the directive on pricing transparency for medicinal products under national health systems;
- **Strengthen European collaboration in pricing and negotiations,** building on the momentum of multi-country negotiation platforms and voluntary cooperation mechanisms. These should be further unified and may include common purchasing initiatives, particularly 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.

INTEGRATED, PERSON-CENTRED, AND LIFELONG HOLISTIC CARE

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 complex, intersectional needs of families with rare diseases,** including comprehensive long-term care, and propose initiatives for equal access to education, employment, and social protection, aligning 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;
- Help Member States enhance their **disability assessment frameworks** and ensure access to social, disability, and independent living rights for those with rare diseases;
- Allocate Multi-annual 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 and promote the **mental health and wellbeing needs** of people with rare diseases in EU mental health policies, in line with the WHO's recognition of them as a vulnerable population and as per UN General Assembly Resolution [6].

INNOVATIVE AND NEEDS-LED RESEARCH AND DEVELOPMENT

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.

OPTIMISED DATA FOR PATIENT AND SOCIETAL BENEFIT

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 specialised 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:

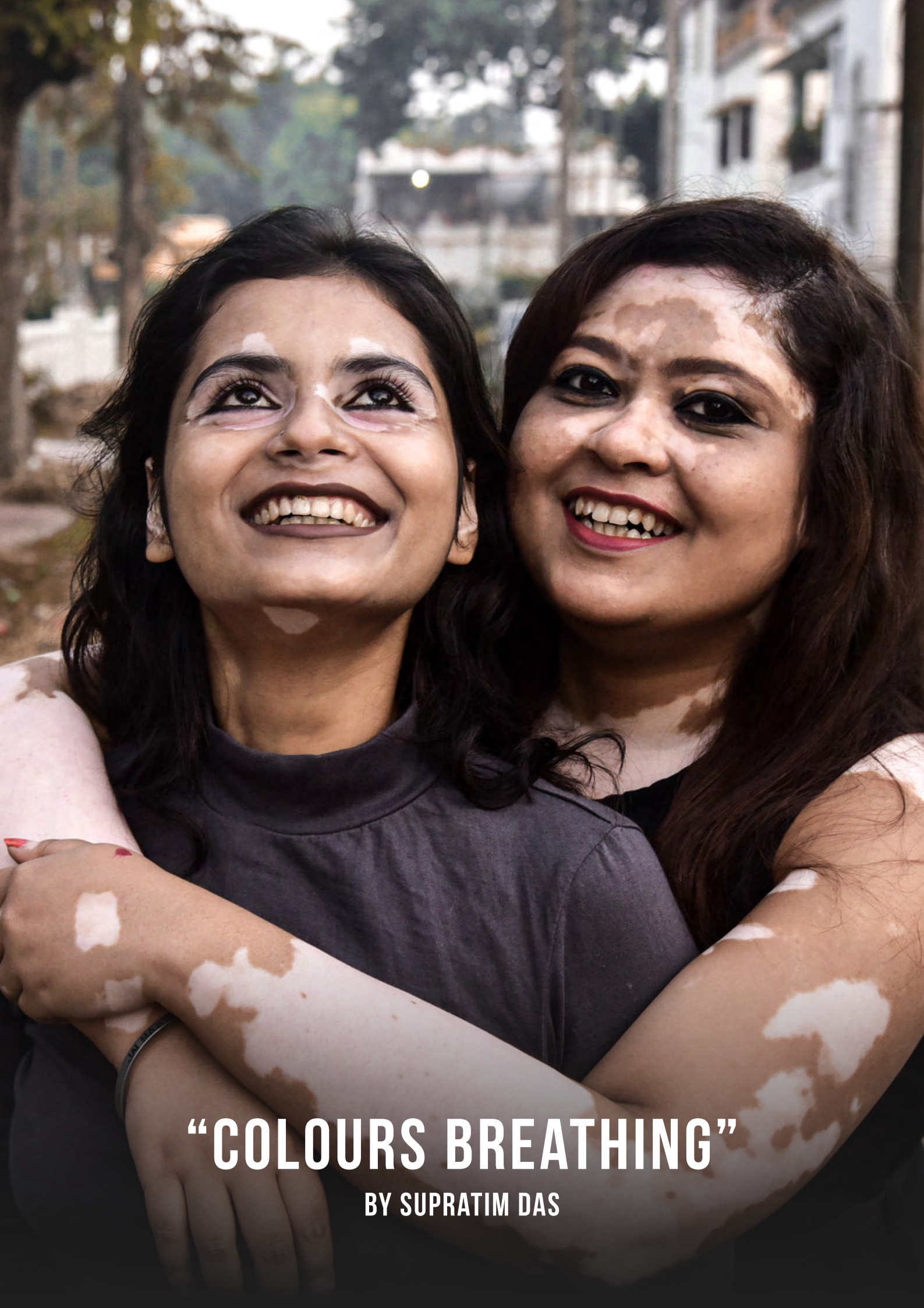
- **Support the harmonisation, optimisation and interoperability of electronic health records** to ensure safe and timely sharing of health data across countries, thereby facilitating cross-border healthcare;
- **Ensure the ethical use of secondary health data.** Secondary use should enable health providers, 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.

SUSTAINABLE AND RESILIENT TREATMENT DEVELOPMENT

The development of treatments and therapies as well as resource-intensive provision of care, require a wide range of resource commitments, including commitments of organisational, 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.

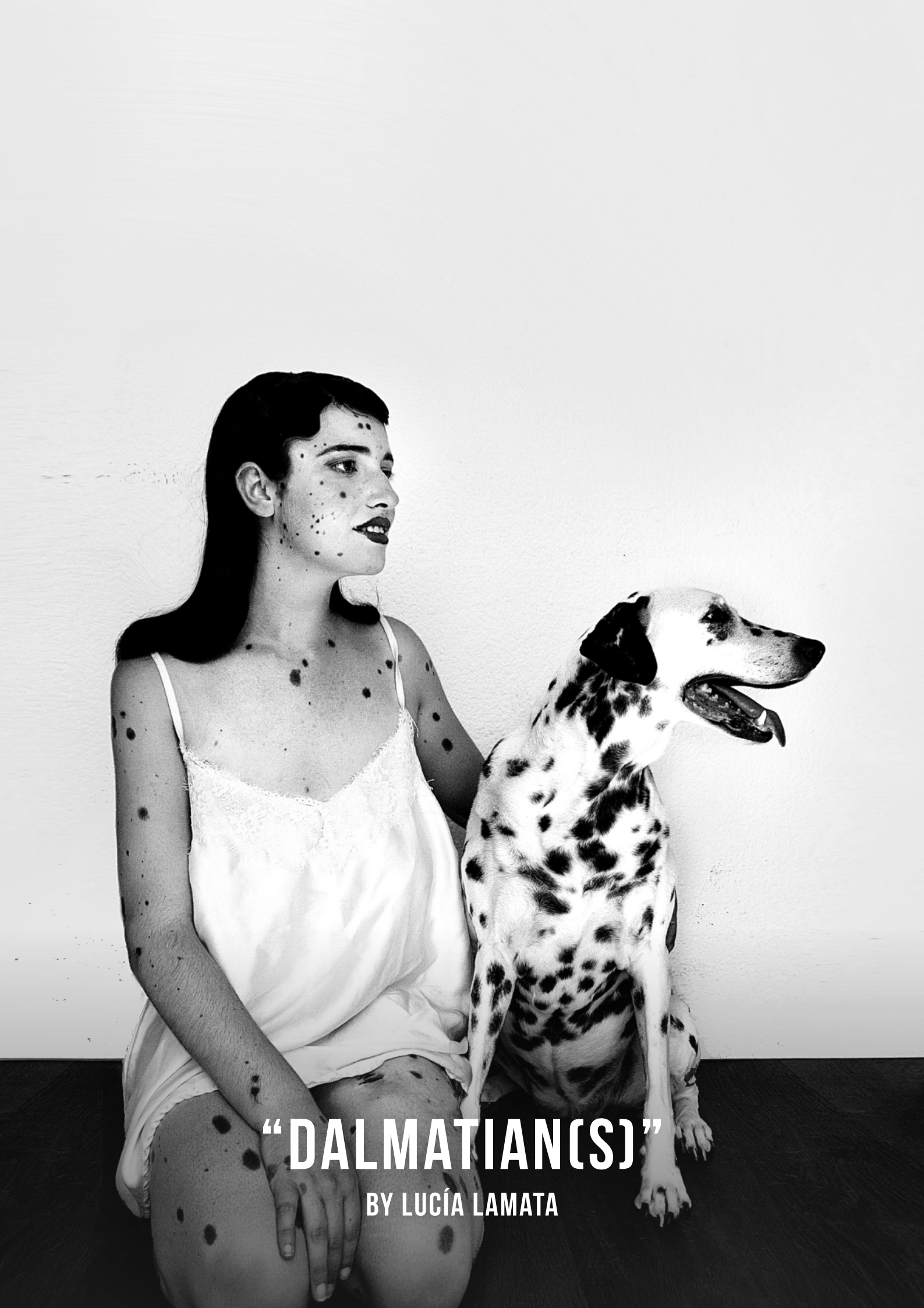
THE EUROPEAN UNION SHOULD:

- **Facilitate the development of digital tools, telemedicine, and more mobile health care**, 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 effeciency 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

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Scan the QR-code to open the link

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[4] Directive 2011/24/EU of the European Parliament and of the Council of 9 March 2011 on the application of patients' rights in cross-border healthcare and Regulation (EC) No 883/2004 of the European Parliament and of the Council of 29 April 2004 on the coordination of social security systems (for the part on the provisions of access to cross border care)

[5] https://www.rarediseasesinternational.org/wp-content/uploads/2022/01/Final-UN-Text-UN-Resolution-on-Persons-Living-with-a-Rare- Disease-and-their-Families.pdf

Scan the QR-code to open the link

[6] https://ec.europa.eu/health/scientific_committees/opinions_layman/en/non-human-primates/glossary/tuv/three-rs-principle.htm

Scan the QR-code to open the link

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Building the Engine of an Inclusive European Health Union

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**

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