A working proposal:
Europe's Action Plan for Rare Diseases

November 2021

This document is put forward by EURORDIS-Rare Diseases Europe as part of a proposal on how to update the European Union's rare disease strategy taking stock of achievements, lessons learned, persistent challenges and resulting recommendations of the Rare 2030 Foresight Study.

It calls for a **European Action Plan for Rare Diseases**, a comprehensive policy framework, to connect all policies and initiatives affecting people living with a rare disease at European and national levels.

Rare diseases are expected to feature on the agenda of the upcoming Trio of EU Council Presidencies, held by France, Czech Republic and Sweden between January 2022 and July 2023. For the first time in December 2021, the United Nations General Assembly is expected to adopt a Resolution on Addressing the Challenges of Persons Living With a Rare Disease.

With support from Members of the European Parliament, Member States and EU Citizens as part of the campaign #30millionreasons for European Action on rare diseases, this is a crucial moment to ensure Europe acts now to leave no one behind.

Find out more: eurordis.org/30millionreasons

1. A new European Plan proposed to address the public policy priority of rare diseases

A disease is defined as rare in Europe when it affects less than 1 in 2000. Yet with more than 6000 rare diseases**, a community of 30 million people are affected across Europe**, even more when considering their families and carers. More than 70% of rare diseases have a genetic origin and are often chronic, heavily disabling and life-threatening. Despite progress in recent decades, a high level of unmet needs remains and rare diseases continue to be accompanied by a scarcity of knowledge and expertise, limited access to diagnosis and holistic care, and a general lack of public awareness.

In the spirit of the **Better Regulation agenda and in line with the lifecycle of European and national policies for people living with rare diseases across Europe**, Europe's Action Plan for Rare Diseases proposes to turn the European Commission's exhaustive review of its strategy on rare diseases, as proposed by a recent Court of Auditors report, into a proposal of concrete actions with uniquely high community added value to address the challenges faced by people living with a rare disease in Europe. It should **consolidate on-going activities, contribute to the initiatives put forth by the European Commission to ensure a strong European Health Union** and align its measurable targets with relevant UN Sustainable Development Goals – particularly the commitment to leave no one behind by 2030.

The proposed Action Plan for Rare Diseases would set out actions to support, coordinate or supplement the Member States' efforts at every stage of the journey of a person living with a rare disease.

A renewed focus on rare diseases as a public health and public policy priority will ensure that scientific, technological and therapeutic advances result in the greatest impact for the 30 million people living with a rare disease in Europe – a vulnerable population challenged by premature death, significant inequalities in health and well-being, and a lack of access to effective treatments. It would do so by creating a comprehensive and cohesive plan across European Commission's policy areas and programmes, integrating EU and national level actions and ultimately creating the ecosystem required to address the unmet needs and persisting inequalities across Europe.

The proposed Action Plan is based on a wide consultation with stakeholder groups, including European and national policymakers - the concluding recommendation from the Rare 2030 Foresight Study which applied a robust foresight methodology to draw upon the inputs from over 250 European and international experts, a panel of Europe's Young Citizens and thousands of people living with a rare disease. By spearheading innovation-led research, technological and therapeutic development, the Action Plan can now make a significant contribution to Europe's leadership in highly innovative treatments, thus laying the foundation of an innovation-based and competitive economy, for the broader prosperity of citizens in Europe.

2. Building on the success of past, ongoing and upcoming actions in the field of rare diseases and beyond

Building on 20 years of successful EU Regulations on Orphan Medicinal Products, the Paediatric Use of Medicines and Advanced Therapies and the Directive on Cross-border Healthcare, and over two decades of public investments in collaboration and research, the proposal for Europe's Action Plan for Rare Diseases provides the policy framework required to capitalise on and further develop the tremendous scientific and therapeutic advancements to date, to bring together research and care powered by digital

technologies and data analytics, to define healthcare pathways to improve the patient experience, and to promote an integrated medical and social care throughout the journey of a person living with a rare disease.

It would provide Europe and its citizens with an ambitious and cohesive strategy to guarantee that progress made in Member States since the 2009 Council Recommendation "on an action in the field of rare diseases" is not lost and that Europe remains a leader on the international stage.

3. Rare diseases: an exemplary area demonstrating EU added value

As part of a larger assessment of the current rare disease strategy to decide whether it needs to be updated, adapted or replaced by 2023 (as per the European Court of Auditors' Special Report n°7/ 2019), the Rare 2030 Foresight Study - initiated by the European Parliament and co-funded by the European Commission- concludes that renewed European action on rare diseases is required now to:

> **Address the remaining unmet needs and inequities** all along the patient journey in accessing a diagnosis, treatments and care, leaving people living with a rare disease marginalised in society;
>
> **Keep pace with new technologies, new values and new expectations** of Europe's citizens and give a new focus to national rare disease plans and strategies;
>
> **Sustain the European Commission's strategic approach** in addressing a distinctive domain of high European added-value and bring together existing and upcoming actions, across countries, across sectors and policy areas, and across the rare disease pathway, where the EU can add the most value under one interconnected framework.

Europe's Action Plan for Rare Diseases would address these gaps while incorporating conclusions of parallel evaluations and revisions of legislation on Orphan Medicinal Products Regulation, Paediatric Use of Medicines and pharmaceutical legislation; Cross-Border Healthcare Directive, including European Reference Networks.

It would also align and build on the EU's major ongoing, upcoming, and new policies and initiatives, such as the European Health Data Space, the Research Partnership on Rare Diseases, the Action Plan of the European Pillar of Social Rights and the European Strategy for the Rights of Persons with Disabilities.

As an initiative under the European Health Union supporting President von der Leyen's vision for a Union of equality, tolerance and social fairness, Europe's Action Plan for Rare Diseases would provide a road map for all European countries to respond to these unmet needs and ensure that inequalities in addressing the challenges faced by people living with a rare disease are not exacerbated by their country of residence.

"I CAN ASSURE THAT RARE DISEASE POLICY WILL REMAIN AN IMPORTANT FOCUS FOR THE COMMISSION AND THAT PATIENTS' VOICES WILL CONTINUE TO INFORM OUR APPROACH. CURRENT ACHIEVEMENTS NEED TO BE CONSOLIDATED AND FURTHER IMPROVED."

STELLA KYRIAKIDES, COMMISSIONER FOR HEALTH AND FOOD SAFETY, SPEAKING AT THE RARE 2030 FINAL POLICY CONFERENCE (23 FEBRUARY 2021)

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4. A goal-oriented approach

Unique to this new generation of rare disease policy, the proposed Action Plan would set three ambitious goals and a number of sub-targets in tackling Europe's challenges in rare diseases, that would translate to tangible results that contribute to several of the UN Sustainable Development Goals (SDGs):

1. Ensuring healthy lives and promoting well-being for all people living with a rare disease, with sub-targets such as reducing diagnostic delays to six months

SDG 3 - GOOD HEALTH AND WELL-BEING: 3.2, 3.4, 3.8, 3.b

2. Reducing inequalities for people living with a rare disease, with sub-targets such as reducing psychological, social and economic vulnerability of people living with a rare disease and their families by one-third

SDG 10 – REDUCED INEQUALITIES: 10.2, 10.3

3. Building resilient infrastructure, promoting inclusive and sustainable industry and fostering innovation for people living with rare diseases, with sub-targets such as ensuring the approval of approximately 1000 innovative (symptomatic or transformative) treatments for people living with a rare disease, irrespective of their status as orphan medicinal products

SDG 9 – INDUSTRY, INNOVATION AND INFRASTRUCTURE: 9.2, 9.5

Long-term plans and strategies for rare diseases the Action Plan would require the partnership of a large number of stakeholders across countries, disease areas and sectors, due to the complexity of the diseases (contributing to *SDG 17.6, 17.16, 17.17, 17.18*). By achieving genuine partnership with patients in policy making, clinical practice, or research and development Europe's Action Plan would foster partnerships between governments, the private sector and civil society.

No one country, institution or sector can address the challenges of rare diseases alone. By collaborating, coordinating research and sharing its results across sectors (public, private and civil society), institutions and countries – particularly in less developed countries - the rare disease research community could boost the role of science technology and innovation in the achievement of the 2030 Sustainable Development Goals.

5. An ambitious approach structured around key areas of action

Europe's Action Plan on Rare Diseases should set the common ambition for a strong European Health Union through **6 key areas of action, 2 cross cutting themes and 1 special focus**. These should all be composed of supporting actions generating model solutions to important challenges faced by people living with a rare disease and society as a whole.

- **6 proposed key areas of action would** drive change through **innovative European collaboration** and **improve care pathways for patients and their families**:

1. Innovative and Patient Needs-led Research
2. Making the EU a Global Leader in Rare Disease Innovation
3. Accessible, Affordable and Sustainable Transformative Medicines
4. Earlier, Faster and More Accurate Diagnosis
5. Delivery of High-Quality, Highly Specialised Healthcare Services
6. Delivery of Integrated and Person-Centred Care and Full Inclusion in Society

The Action Plan would specifically address the bottlenecks encountered along the journey of a person living with a rare disease, and be supported by actions spanning across policy areas from employment, education, social policy and equality, through research, digital, industry, internal market, cohesion policy, international cooperation and development, and health.

- **2 cross-cutting areas would help catalyse change**:

a) making rare diseases a European added value use case in digital technologies and data analytics;

b) a whole-system and whole-government partnership to achieve the goals

To support these actions and achieve the critical mass of data, knowledge and attention the people living with rare diseases deserve, the proposal for Europe's Action Plan for Rare Diseases is designed to support an ecosystem of digital technologies and data analytics making rare disease an exemplary use case of the European Health Data Space (EHDS) with an integrated and strategic European approach for the capture, use, and reuse of all data sources on rare diseases. The solutions put forward by the rare disease community to maximise data collection and sharing will be able to serve as an example for many other areas of health and research across Europe.

The Action Plan would also be instrumental in integrating the European and national levels of action with strong cooperation between EU Member States steered by common expected outcomes. To this aim, the Commission should adopt a proposal for a new Council Recommendation in the field of rare diseases, which would renew the call to Member States to adopt or refresh national plans and strategies on rare diseases in European countries.

The proposed Action Plan recognises that many of these actions can only be fully achieved with international collaboration beyond Europe's borders. Europe's Action Plan for Rare Diseases would provide an opportunity to **implement the recently adopted** **UN Resolution to Address the Challenges of Persons Living with Rare Diseases**, with Europe leading the world by example in an area of public health where EU common values are a cornerstone for overcoming the remaining challenges faced by people living with a rare disease.

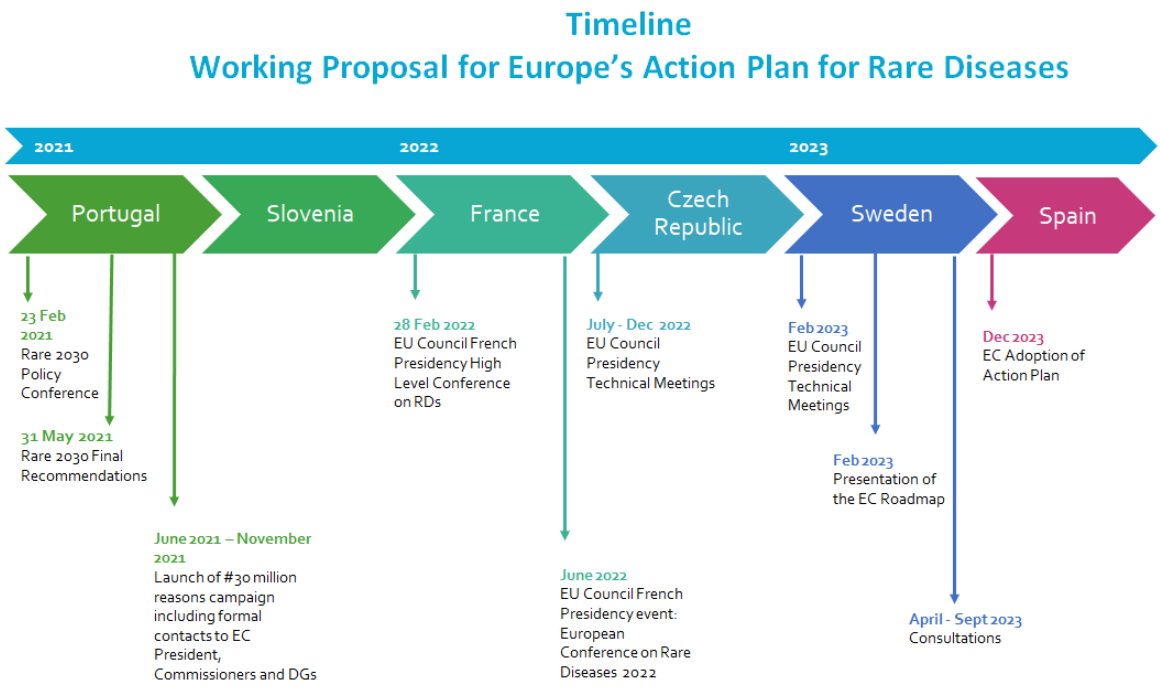
- **1 special focus: the rarest of Europe's citizens require international and cross-cutting collaboration**

Europe's Action Plan would also consider the special focus required of the rarest of diseases affecting less than one person in 1 million for which often even less progress has been made and a targeted approach is required. Targeted approaches to diagnosis, care pathways and research and development of treatments for these extremely rare diseases are considered in the proposal for Europe's Action Plan for Rare Disease, based on piloted programmes, such as a programme for "the undiagnosed"; European Centres or Rare Diseases an EU Fund for post-marketing approval evidence generation for therapies for rare diseases with very low prevalence. These are solutions for Europe's undiagnosed citizens, infrastructures and collaboration pooling even rarer knowledge and expertise on care and special incentives to ensure that research and development leaves no disease rare behind - no matter how rare.

6. Proposed timeline

Europe's Action Plan for Rare Diseases responds to calls from the European Parliament and Member States as well as social partners, other stakeholders and most importantly EU citizens. A new European policy framework on rare diseases was the concluding recommendation from the Rare 2030 Foresight Study, initiated by the European Parliament and funded by the European Commission, concluded in February 2021. Rare diseases are now expected to feature on the agenda of the upcoming Trio of EU Council Presidencies, held by France, Czech Republic and Sweden between January 2022 and July 2023.

A possible timeline and a working proposal are illustrated below:



For more information about Europe's Action Plan for Rare Diseases and how you can support this proposal please contact Anna Kole, Director of Public Health Policy at Anna.Kole@eurordis.org**.**

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of 962 rare disease patient organisations from 73 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe.