

Rare2030

Foresight in Rare Disease Policy



RECOMMENDATIONS FROM THE RARE 2030 FORESIGHT STUDY

THE FUTURE OF
RARE DISEASES
STARTS TODAY



The health of 30 million people living with a rare disease in Europe should not be left to luck or chance. The Rare 2030 foresight study prepares a better future for people living with a rare disease in Europe by gathering the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations.

Since the adoption of the Council Recommendation on European Action in the field of Rare Diseases in 2009, the European Union has fostered tremendous progress to improve the lives of people living with rare diseases. Rare2030 will guide a reflection on rare disease policy in Europe through the next ten years and beyond.

PARTNERS



Bone disorders (ERN BOND)



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FOREWORD

I first worked with the rare disease community in Europe on joining the Commission's Health DG in 2004.

Rarely, before or since, have I encountered the Rare Disease community's special blend of knowledge and objectivity, passion and maturity. It was a privilege and a pleasure to be in touch with the work in those still early days, and I have been very happy to offer some small contribution to the major foresight and strategy exercise that is Rare 2030.

It is the rigour and breadth of the underlying process of the Rare 2030 effort that has produced the high quality vision set out in these pages.

Here you will find a set of ideas whose time has come, as Europe seeks to revalidate its way of living the health journey, making it fit for the post-COVID 21st century. This is just in time for implementation, as the work of Horizon Europe and the post-pandemic health reset get underway.

If I could offer one personal suggestion to colour the findings here described, it is to frame the Rare 2030 prescriptions as lessons from rare disease for the good of all.

Rare disease challenges are the challenges facing society more widely. If we can deliver on this vision, it will be to the benefit not only of the millions of rare disease sufferers, their families, employers and communities. It will also bring in its wake crucial improvements in healthcare beyond the rare disease area.

I say this because, for example, the undiagnosed can be even more numerous beyond the defined field of rare disease. In all cases, rare or not, an undiagnosed patient not only faces a long search for diagnosis, with no real relief

for symptoms meanwhile, but faces the stigma of health professional doubts and dismissal and frequently experience, even today, a failure to listen and to hear the patient. This trial lasts for as long as the symptoms elude satisfactory explanation and then treatment. All who have struggled, in our overloaded systems, to secure the full focus that their diagnosis depends on, will gain from the breakthroughs proposed here.

In the same way, rare diseases are not alone these days in facing the huge challenge of defining a sustainable and feasible access and funding model for potentially curative treatments that appear "too" costly to some. Here, as in the search for timely diagnosis, we must find a positive answer if we are to be true to the principle of unalloyed solidarity. And solidarity is surely intrinsic to the European Way of Life that we promote these days even beyond our borders. Personally, I am encouraged by work underway in Europe to define ways to spread high costs over the decades of good life that a cure or significant treatment can uniquely offer.


The community of those living with rare disease and its consequences, those helping as carers and those searching for new cures, all deserve our admiration. Admiration is not enough. But the real shifts in policy suggested here are in no way special pleading: the changes set out in these pages can unlock society-wide public good.

ROBERT MADELIN

Director General for Health 2004-2010
Rare 2030 Research Advisor
Chairman of FIRPA International

An open letter

FROM THE **Rare2030** YOUNG CITIZENS

 ear Members of the European Parliament, dear European Commissioners, dear present policy makers and thought leaders,

We are the Rare 2030 Young Citizens, representing a new generation of advocates, patients and actors in the rare disease community. We come from diverse backgrounds, different countries and have different experiences of rare diseases – as patients, young parents, doctors, students, and junior policy advisors. Nevertheless we have all come to the same conclusions about what needs to happen for a better tomorrow for the 30 million people living with a rare disease in Europe, where no one is left behind.

While the European Union has fostered tremendous progress in the past decade to improve the life of people living with a rare disease in Europe, it is undeniable that the world we live in today is different than the one we knew back then. We are a generation in a new context, with new challenges but also new opportunities. It has taken a generation to get where we are. Now it's time to listen to what we have to say about the future for Europe to indeed be a place where no one is left behind.

In ten years' time, we will be living with a rare disease. In ten years, we may also be parents of a child living with a rare disease. By 2030, we want to live in an inclusive society that recognises all our needs and challenges. We want to live in a society that acknowledges that our conditions are more than collections of physiological symptoms but rather require a holistic approach to be fully understood and, more importantly, addressed. We want to have access to the right diagnosis without having to go through the all too common diagnostic odyssey and to be given the appropriate treatment wherever we live and whatever our socio-economic status may be.

In ten years' time, we will be doctors, nurses, psychologists. By 2030, we want to work and collaborate with colleagues in our respective countries and throughout Europe to whom rare diseases are not an abstract notion or mystery but a surmountable challenge. We want – as doctors, nurses, health and social care providers of any kind – to know that collaboration and help beyond national borders exists to take the best care possible of people living with a rare disease. We hope that we – and the generation of health care providers that will follow ours – will benefit from a solid education on rare diseases and on the means we have at our disposal to appropriately recognize, diagnose and treat them. It is time for us to become the best version of our professions to make sure that our patients can live the healthiest and happiest life they can.

In ten years' time, we will be policy makers. By 2030, we hope we will not have to advocate anymore for the need to have a European policy framework that encompasses all the important aspects of rare diseases and addresses the needs of the rare disease community as a whole. We want Europe to be an example of fairness, equity and inclusiveness in all areas having an impact on the lives of people living with a rare disease. We want policy makers to have understood by 2030 that rare diseases **are** a public health priority that highly benefits from cross-border collaboration, in terms of diagnosis, care and research, to indeed guarantee that we are a society that truly leaves no one behind.

In ten years' time, we will be sitting where you are today and, we hope, as part of a stronger rare disease community all together. In the next ten years we want to be the generation of change.

As the leaders of today you can make a meaningful difference, to drive change for our generation, and the generations of rare disease patients, carers, doctors and policy makers to come.

Dear Members of the European Parliament, dear European Commissioners, dear present policy makers and thought leaders, please don't let the health and well being of 30 million people living with a rare disease in Europe be left to luck or chance. Consider our words in your policies changes today.

EXECUTIVE SUMMARY

This document presents the conclusions of the Rare 2030 Foresight Study, initiated by the European Parliament and co-funded by the European Commission Pilot Project and Preparatory Actions Programme.

The field of rare diseases has witnessed tremendous strides in the last decades. But significant unmet needs for people living with a rare disease in Europe remain and current policies are no longer fit for purpose.

Over the past two years, over 200 experts from across the rare disease field and thousands of patients have come together through the Rare 2030 Foresight Study to make an important decision:

What future do we want for people living with a rare disease in Europe?

Presented with four possible scenarios, the rare disease community came up with eight recommendations of how to achieve the preferred future “Investment for Social Justice”. This is defined by: needs-led innovation, collective responsibility and outcomes-based goals. Only by implementing new policies fit-for-purpose for this decade can we actively design this preferred future and prepare for the shortcomings of other possible scenarios.

The recommendations of the Rare 2030 Foresight Study set out how to make these needed changes happen in a strategic and concerted way - not by replacing or replicating other assessment efforts, but rather serving as a unique reference for an updated European policy framework for rare diseases that will, in turn, guide a new generation of national plans and strategies with measurable goals.

RARE DISEASES: EUROPEAN ADDED VALUE, COLLABORATION AND INNOVATION

Rare diseases¹ are a heterogeneous group of largely incurable, often complex conditions. Although individually characterised by low prevalence, the sheer number of rare diseases results in a directly-affected community of 30 million people across Europe alone. Rare diseases are typically accompanied by a scarcity of knowledge and expertise. These characteristics single out rare diseases as a major area of very high community added value, demanding collaboration in terms of knowledge, data, and research.

With such a large portion of the population affected by rare diseases, and the severity of their condition, improving the whole rare disease pathway offers a significant opportunity to contribute to the United Nations’ Sustainable Development Goals which aspire to a better and more sustainable future for all. In particular, the goal of Universal Health Coverage will be met when all people living with a rare disease can access high quality healthcare with financial protection.

Out of necessity rare diseases have become pioneers in tomorrow’s transformations and creative solutions for research and healthcare. The community continually brings forward innovative concepts and models created at the margins of the healthcare system but with benefits for all.

1 - In this paper the term rare diseases also includes rare cancers

WHY DO WE NEED A NEW GENERATION OF RARE DISEASE POLICIES?

Rare diseases were nearly invisible in our health and social care systems in previous decades. Today, they are recognised as a public health and human rights priority in large part due to a policy framework largely defined by the Commission Communication on '[Rare Diseases: Europe's Challenges](#)' (2008) and the Council Recommendation of 8 June 2009 on "[An action in the field of rare diseases](#)" setting the road map for a number of legislative acts and policies such as the EU Regulations on Orphan Medicinal Products, the Paediatric Use of Medicines and Advanced Therapies and the Directive on Cross-border Healthcare.

Yet for the 30 million people in Europe living with a rare disease, huge challenges remain: a diagnostic odyssey, lacking treatments, decreased quality of life and isolation, lack of access to the highest available quality of care, invisibility in healthcare and research systems.

The policies of the past no longer keep pace with the needs of our community. We have new technologies and new expectations that need to be reflected in policies from the European Union and its Member States. More strategic and collective decision making processes need to be put in place in order to truly address these needs.

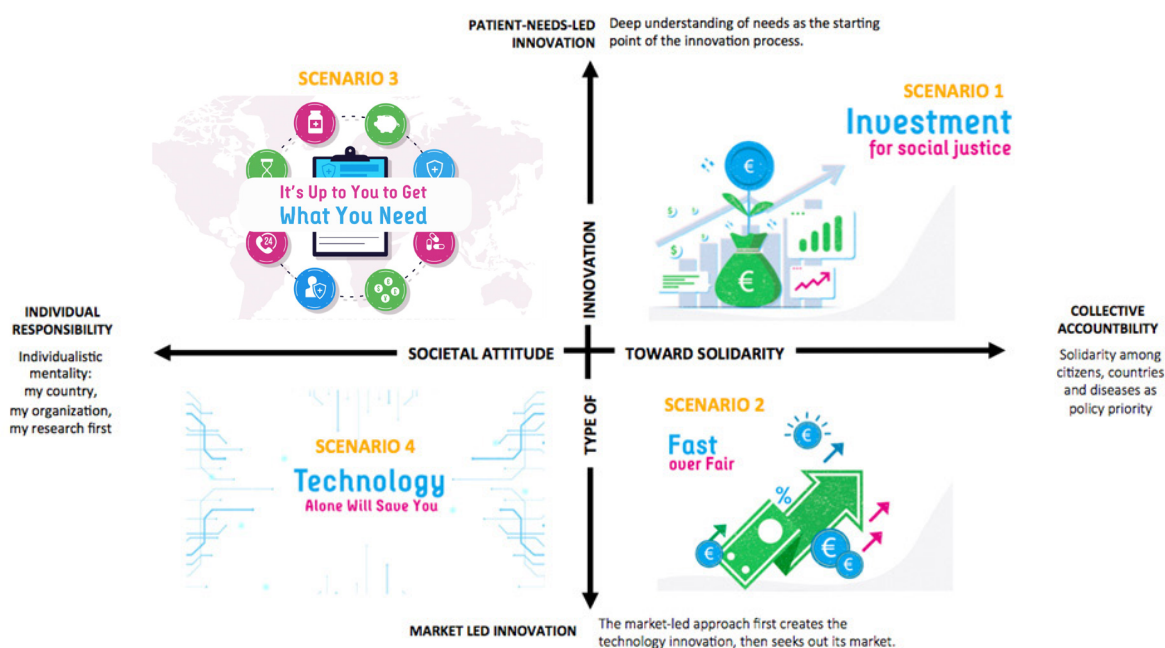
THE RARE 2030 FORESIGHT STUDY: A BETTER FUTURE BY DESIGN

Health needs and solutions change so quickly that we need methods to make decisions in an agile way. The foresight approach allows opinion leaders to consider several possible future scenarios and identify the steps needed to actively reach the preferred future.

Supported by EU bodies, the Rare 2030 Foresight Study called upon Europe's most dedicated doctors, companies, researchers, young citizens, advocates, patients, payers and policy makers. Together they comprise expert panels of over 200 key thought leaders and thousands of people living with a rare disease who have all participated in this iterative, participatory and inclusive study between January 2019 and March 2021.

THE PREFERRED SCENARIO 2030: "INVESTMENT FOR SOCIAL JUSTICE"

The Rare 2030 Foresight Study provided us with 12 trends clustered around changing values and technology that will affect rare disease policy over the next decade. In combining different progressions of the trends, we established four possible scenarios for 2030.



During the 2020 European Conference on Rare Diseases, the rare disease community has identified Scenario 1 as most preferred, Scenario 2 as most likely and Scenario 4 as the least preferred.

The preferred scenario “Investment for Social Justice” is defined by:

- + **Innovation** driven by patient’s unmet needs rather than financial gain
- + **High collective responsibility** recognising societies’ role in paying special attention to this vulnerable population
- + **Outcome-based goals** (earlier faster and more accurate diagnosis; an integrated and person-centred approach to care; improved availability, accessibility, affordability and sustainability of medicinal products)

Only by proposing new policies fit-for-purpose for this decade will we actively reach this preferred scenario and prepare for the shortcomings of other possible scenarios which may include distrust in supranational efforts to manage health and innovation led only by market rewards.

FROM THE PREFERRED SCENARIO TO A POLICY FRAMEWORK

To reach the preferred scenario “Investment for Social Justice” we need a new policy framework for rare diseases as a roadmap to get there. This should build on existing policy and legislative pillars that have shaped the field of rare diseases to date. The recommendations for a new policy framework are designed to work in step with ongoing European strategies and programmes in pharmaceutical development², data³, social rights⁴, research⁵, public health⁶, cancer⁷ and an overall more resilient European Health Union.

This new generation of rare disease policies move the rare disease community’s goals from mechanisms and infrastructure needed to address challenges to setting measurable outcomes for people living with a rare disease for the next decade:

- + Earlier, faster and more accurate diagnosis
- + A holistic approach to the person’s needs and improved access to high quality integrated medical and social care
- + Holistic approach to care and research, centred on the patient
- + Improved availability, accessibility, affordability and sustainability of medicinal products

Six approaches are needed to achieve these goals:

- + **A human rights approach:** Drawing on the principles and overarching values of universality, access to good quality care, equity and solidarity to guarantee the same universal human rights for people living with a rare disease, as endorsed in European and international treaties.
- + **A multi-stakeholder approach:** Bringing all players to the table to form an efficient and agile eco-system for rare diseases.
- + **A partnerships approach:** Partnering with patients, between public and private sectors, and between countries to share knowledge.
- + **An integrated approach between regional, national and European levels:** Ensuring all players are moving towards measurable outcomes.
- + **A technological approach:** Leveraging innovative technology across the pathways, such as Whole Genome Sequencing in diagnostics, digital assets such as telemedicine and Artificial Intelligence, in clinical trials design and cell and gene therapies.
- + **A holistic approach to care and research:** Centred on the patient and the point of care, powered by digital technology and data analytics, requiring standards, interoperability and adapted capabilities for the healthcare providers.

2 - Pharmaceutical Strategy for Europe, https://ec.europa.eu/health/human-use/strategy_en

3 - European Data Strategy, https://ec.europa.eu/info/strategy/priorities-2019-2024/europe-fit-digital-age/european-data-strategy_en

4 - European Pillar of Social Rights, https://ec.europa.eu/info/strategy/priorities-2019-2024/economy-works-people/deeper-and-fairer-economic-and-monetary-union/european-pillar-social-rights_en

5 - Horizon Europe - Rare Disease Research and Innovation Partnership, https://ec.europa.eu/info/sites/info/files/research_and_innovation/strategy_on_research_and_innovation/documents/ec_rtd_orientations-he-strategic-plan_122019.pdf

6 - EU4Health Programme, https://ec.europa.eu/health/funding/eu4health_en, Directive on patients’ rights in cross-border healthcare, https://ec.europa.eu/health/cross_border_care/overview_en

7 - Europe’s beating cancer plan, <https://ec.europa.eu/info/law/better-regulation/have-your-say/initiatives/12154-Europe-s-Beating-Cancer-Plan>

A WINDOW OF OPPORTUNITY FOR RARE DISEASE POLICY

We have a rare opportunity to bring the policy in step with the scientific, technological and societal shifts. Alongside wider legislative areas under review, the three EU Institutions that together drive the policy agenda for their 27 Member States recognise the need for action:

- + The recently published EU Court of Auditors Report on the implementation of Directive on Cross-border Healthcare provides an additional impetus for an updated framework for rare diseases by 2023. The report recommends
- + that by 2023, "the Commission should: (a) assess the results of the rare disease strategy (including the role of the European Reference Networks) and decide whether this strategy needs to be updated, adapted or replaced".
- + The European Parliament in its Resolution on the EU public health strategy in the post COVID-19 era⁸ calls for an action plan for rare diseases at the EU level.
- + France, the Czech Republic and Sweden have recognised their upcoming presidencies of the EU Council as a platform of discussion to bring rare diseases forward as a priority in this same 2022/23 time frame.



"Europe has built a strong ecosystem for rare diseases and the Rare 2030 Foresight Study is an opportunity for continued multi-stakeholder dialogue to prepare for the future. Rare 2030 provides impetus to the European Parliament, the Council of Ministers and the Commission to design ambitious plans for how we get there together - not as accidental progress in one field or another but as an active design to get the kind of future we all want."

Terkel Andersen
President of EURORDIS-Rare Diseases Europe
Rare 2030 Research Advisor

The Rare 2030 recommendations represent the first step in providing Europe's institutions with clear instructions on how to ensure a better future in the decades to come.

But whether this is implemented is not just in the hands of policy makers.

The vision of rare disease patients and experts will only be realised with a collective voice – from every corner of Europe, from every disease group and every discipline - showing how we can improve the lives of people living with rare diseases.

8 - 2020/2691(RSP) Resolution on the EU's public health strategy post-COVID-19

THE RARE 2030 RECOMMENDATIONS: A ROADMAP FOR RARE DISEASE POLICY

The future of people living with a rare disease cannot be left to luck or chance. To play a positive role in future, we call upon Europe's institutions and Member States to deliver on our eight recommendations for a new generation of rare disease policies by 2030.

RARE 2030 RECOMMENDATION

1



LONG-TERM, INTEGRATED EUROPEAN AND NATIONAL PLANS AND STRATEGIES

A European policy framework for rare diseases defined by societal responsibility, equity and driven by the needs of people living with a rare disease should guide the implementation of consistent national plans and strategies, secure major investments at both the European level and by governments that are fairly shared across Europe in order to pool scarce resources, share expertise and information, scale-up good practices and provide access to timely and accurate diagnosis and the highest available quality of treatment and care for people living with a rare disease, no matter where they live in Europe. Both EU and national policies are supported by measurable outcomes that are monitored and assessed by a multistakeholder body on a regular basis.

RARE 2030 RECOMMENDATION

2



EARLIER, FASTER, MORE ACCURATE DIAGNOSIS

The time to diagnosis should be shortened - whilst avoiding erroneous and subsequent negative consequences - which should be achieved by better use and accessibility of currently effective and available diagnostic testing technologies, best practices and programmes. New technologies and innovative approaches must be driven by patient-needs and applied rapidly and strategically. Inequalities in access to diagnosis and ensuing care must be eradicated through the harmonisation of standards and programmes across Europe (and beyond). A particular focus is necessary for patients with undiagnosed rare diseases, which demands greater and more strategic global collaboration via data-sharing and diagnostic platforms and infrastructures.

RARE 2030 RECOMMENDATION

3



ACCESS TO HIGH QUALITY HEALTHCARE

Political, financial, operational and technical support at European, national and regional levels should be provided to establish a mature highly specialized healthcare ecosystem that, in collaboration with patient organizations and all relevant stakeholders, leaves no person living with a rare disease with uncertainty regarding their diagnosis, care and treatment.

RARE 2030 RECOMMENDATION

4



INTEGRATED AND PERSON-CENTRED CARE

Implement EU-wide and national actions by all stakeholders that guarantee the integration of people living with a rare disease in societies and economies, enabling them to live life to their full potential, by implementing innovative solutions and approaches to integrated and person-centred care along the full lifespan of people living with a rare disease.

EU-wide and national actions must be undertaken by all stakeholders to guarantee equal opportunities and access to the labour market, active support for employment, fair working conditions, social protection and inclusion and integrated and person-centred, long-term care for people living with a rare disease and their families.

RARE 2030 RECOMMENDATION

5



PARTNERSHIP WITH PATIENTS

An overall culture, reflected in policies and funding, that encourage the meaningful participation, engagement, involvement and leadership of people living with a rare disease in the research, care and development of diagnostic tools, treatments and innovative solutions to improve the health and social status, healthcare delivery, autonomy, quality of life and well-being of people living with a rare disease in Europe. Patient partnership should be encouraged in both the public and private sectors and people living with a rare disease and their representatives may often serve as a partnering link between the two.

RARE 2030 RECOMMENDATION

6



INNOVATIVE AND NEEDS-LED RESEARCH AND DEVELOPMENT

Maintain basic, clinical, social and translational research on rare diseases as a priority by increasing the funds for competitive and pre-competitive research, establishing greater incentives in more neglected areas (or in areas of high unmet needs), and supporting infrastructures required to expedite discovery and knowledge acquisition. Research in public health, social sciences, healthcare organisation, health economics and health policy research must also be promoted, to ensure that research outputs are applied for the benefit of people living with a rare disease.

RARE 2030 RECOMMENDATION

7



OPTIMISING DATA FOR PATIENT AND SOCIETAL BENEFIT

All European data sources of relevance to addressing the challenges faced by people living with a rare disease should be federated in a continuum encompassing epidemiological, healthcare, research, quality of life and treatment-related data, and should be linked at the global level where possible. Sharing of data for care and research should be optimised across infrastructures and countries, relying upon commonly adopted codification systems (Orphanet nomenclature), harmonised standards and interoperability requirements. Cohesive data ecosystems should be developed at national level, linking seamlessly via Findable, Accessible, Interoperable and Reusable (FAIR) data approaches to an integrated European ecosystem, positioned within the European Health Data Space and centred on robust European Reference Networks (ERNs), the European Platform on Rare Disease Registration, and other key infrastructures. Legal and ethical guidelines and regulations should incentivise practices that best lead to addressing these challenges while respecting international, national and regional laws and conventions – particularly the preferences and privacy of people living with a rare disease and their families.

RARE 2030 RECOMMENDATION

8



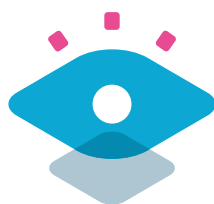
AVAILABLE, ACCESSIBLE AND AFFORDABLE TREATMENTS

Establish streamlined regulatory, pricing and reimbursement policies. These policies should encourage a continuum of evidence generation along the full life cycle of a product or technology as well as the patient journey from diagnosis to treatment access. A European ecosystem able to attract investment in areas of unmet need, foster innovation, and address the challenges of healthcare system sustainability.



THE FUTURE OF RARE DISEASES STARTS TODAY

[RARE2030.EU/RECOMMENDATIONS](https://rare2030.eu/recommendations)



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Foresight in Rare Disease Policy