

Rare 2030

Foresight in Rare Disease Policy

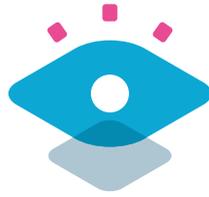


RECOMMENDATIONS FROM THE RARE 2030 FORESIGHT STUDY

THE FUTURE
OF RARE DISEASES
STARTS TODAY

FEBRUARY 2021

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



Rare2030

Foresight in Rare Disease Policy

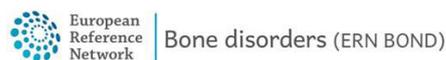
The health of 30 million people living with a rare disease in Europe should not be left to luck or chance. The Rare 2030 foresight study prepares a better future for people living with a rare disease in Europe by gathering the input of a large group of patients, practitioners and key opinion leaders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



FONDAZIONE



The Rare2030 project is co-funded by the European Union Pilot Projects and Preparatory Actions Programme (2014-2020). This leaflet is part of the pilot project PP-1-2-2018-Rare 2030. The content represents the views of the author only and is his/her sole responsibility; it cannot be considered to reflect the views of the European Commission or any other body of the European Union.

This document was prepared as part of the Rare 2030 Foresight Study, under the leadership of EURORDIS-Rare Diseases Europe and with input from all partners. The document should be referenced as follows: Kole, A., Hedley V., et al. (2021) Recommendations from the Rare 2030 Foresight Study: The future of rare diseases starts today.

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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 and
Consumer Policy 2004-2010
Rare 2030 Research Advisor
Chairman of FIPRA International

An open letter

FROM THE **Rare2030** YOUNG CITIZENS

Dear 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.

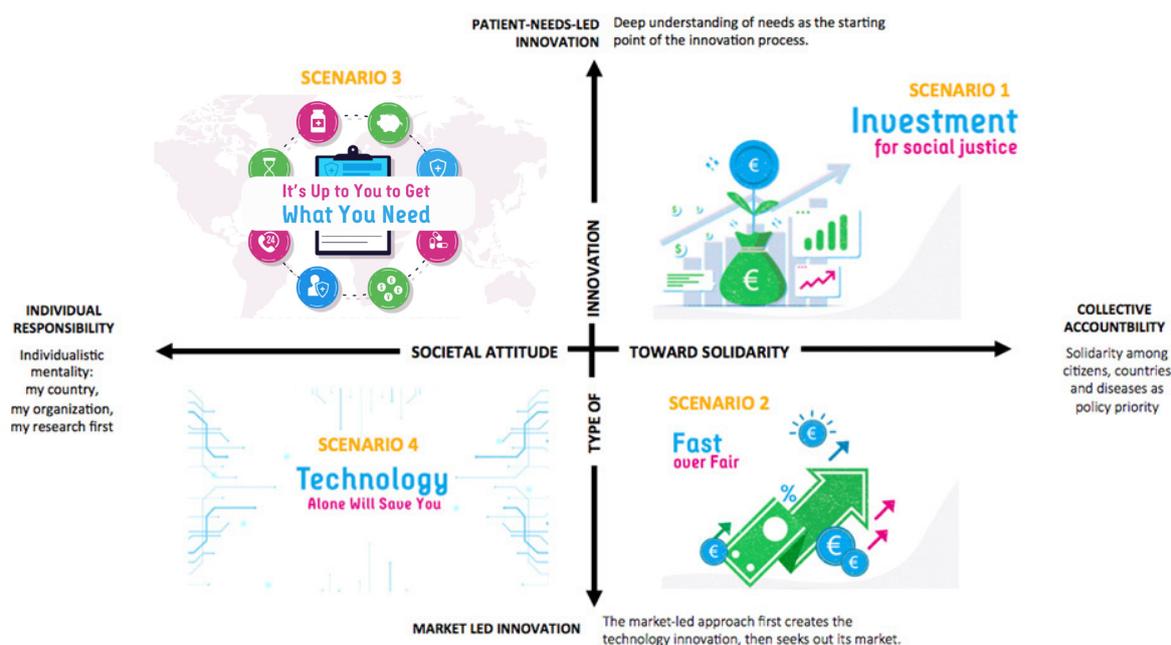
THE RARE 2030 FORESIGHT STUDY: A BETTER FUTURE BY DESIGN

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.

INTRODUCTION

TO THE RARE 2030 RECOMMENDATIONS

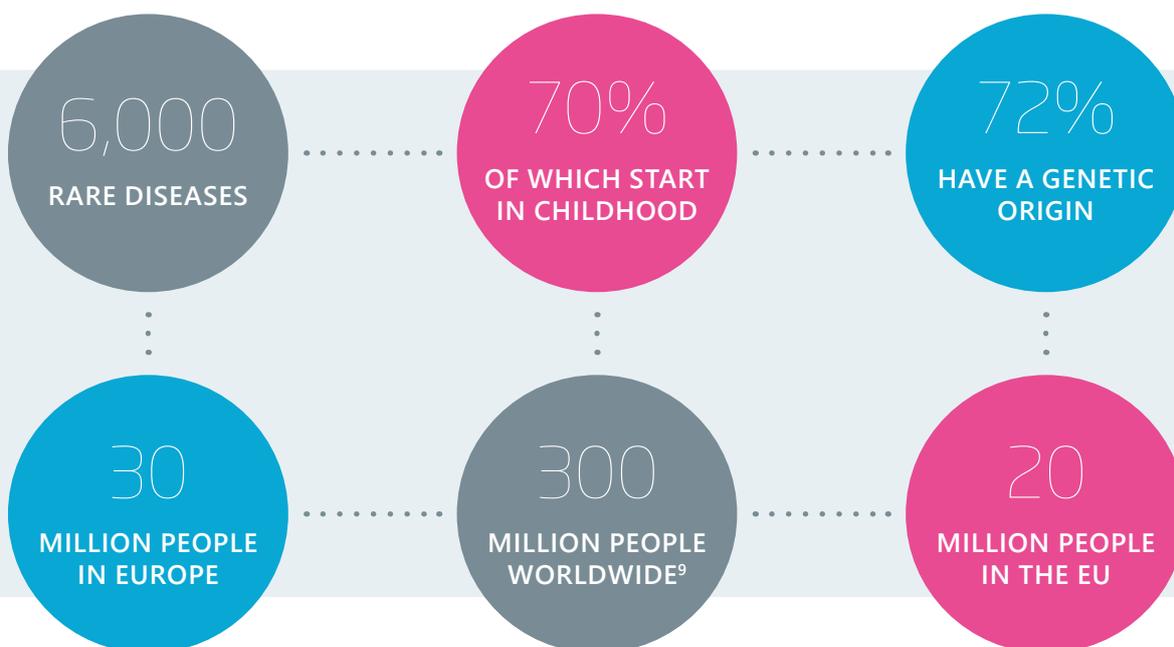
RARE 2030 FORESIGHT STUDY: A BETTER FUTURE BY DESIGN

Supported by EU bodies, the Rare 2030 Foresight Study is the opportunity to bring strong policy recommendations - stemming from those closest to rare diseases through their own experiences and work - that take account of both current and future trends in rare diseases to the European Parliament and the European Commission in February 2021.

With the participation of over 200 experts from across the rare disease field and beyond, the Rare 2030 Foresight Study concludes with eight recommendations that set a roadmap for a generation of new rare disease policies that will lead to a better future for people with rare diseases.

In implementing these recommendations the European Union, its Member States and beyond have an opportunity to prepare for a better tomorrow for people living with a rare disease.

RARE DISEASES: A MODEL OF EUROPEAN ADDED VALUE, INTERNATIONAL COLLABORATION AND INNOVATIVE SOLUTIONS



9 - Nguengang Wakap, S., Lambert, D.M., Olry, A. et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet* 28, 165–173 (2020). <https://doi.org/10.1038/s41431-019-0508-0>

In Europe, rare diseases¹⁰ are defined as any disease affecting fewer than 5 people in 10,000 in the EU. Despite their heterogeneity, they are mostly disabling and degenerative conditions, most of them with no cure. Knowledge and expertise on most rare diseases is disparate across disciplines and geographic borders. The result is that people living with a rare disease very seldom have access to the most timely, accurate diagnosis and quality care and treatments where they live in Europe. As a result of their

impact on European citizens coupled with the common challenges that rare diseases present across borders, disciplines and disease areas - it is clear that this public health priority requires a concerted European and multi-stakeholder response. This approach, given the number of people affected, also allows Europe to move closer towards the United Nations' Sustainable Development Goals (SDGs), which we recognise in our recommendations.

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. For more than 10 years, civil society and patient organisations have pushed to have rare diseases included in European health and research programmes, acknowledging the strong EU added value with regards to diseases rarity and other related challenges. Over time, these efforts have considerably encouraged and stimulated progress to improve the lives of people living with a rare disease at both national and European levels.

Today, they are recognised as a public health and human rights priority in large part due to a policy framework largely defined by the Council Recommendation of 8 June 2009 on an action in the field of rare diseases setting the road map for a number of regulations and soft legislations such as the EU Pharmaceutical Regulations on Orphan Medicinal Products, the Paediatric Use of Medicines and Advanced Therapies and the Directive on Cross-border Healthcare. With this political recognition have come investments, transformative treatments, new knowledge and hope.

Yet for the 30 million people in Europe living with a rare disease, unmet needs and inequalities still remain. Healthcare is a rapidly changing landscape that requires actors to be prepared for what is coming next. Should it be in terms of science, technology, social care, politics, financing or institutions, the healthcare sector is in constant move and, adding up to already existing challenges and opportunities, constantly transforms the way rare diseases are seen and taken care of.

With the end of the European Commission Expert Group on Rare Diseases and EU Joint Actions on Rare Diseases (EUCERD Joint Action, RD-ACTION) in 2018, a new European Parliament, European Commission and Council in 2019, and the recent developments in several healthcare-related domains (health technology assessments, use of big data and new technologies, pharmaceutical regulations, etc.), the need to foresee today what could happen in the upcoming decades and to be prepared accordingly is of primary importance for the rare disease community as a whole, hence the foresight approach of this study.

USING FORESIGHT TO DEVELOP SCENARIOS AND POLICY RECOMMENDATIONS

Foresight helps to prepare for the future. This exercise is extensively used across policy development in spheres such as energy, public transport and geo-politics. They are more rarely used in health.

By analysing current and future trends and applying them to various scenarios, foresight expands our thinking and uncovers inevitable or near-inevitable futures. This allows challenge to

conventional wisdom and enables the design of strategic pathways.

It also is a process of co-production. Starting from the same evidence based, people from across the field and beyond can decide, together, on the recommendations. Actors are all on the same page, and can therefore move in the same direction.

The COVID-19 pandemic has shown that foresight in health, and particularly for the most vulnerable of Europe's citizens, is proving instrumental in preparing resilient and strong health and social sectors.

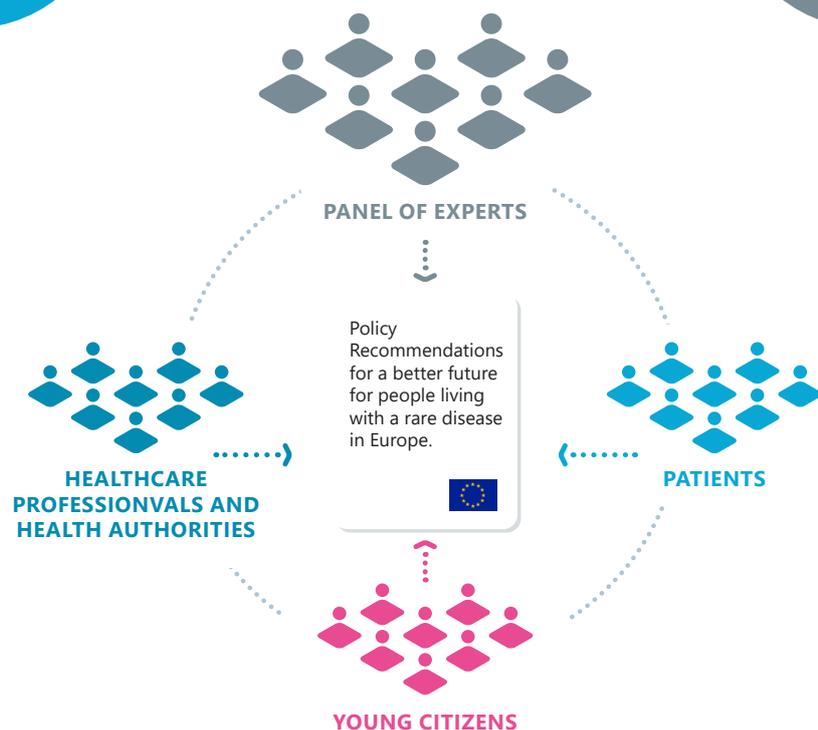
10 - In this paper the term rare diseases also includes rare cancers

HOW THE RARE 2030 FORESIGHT STUDY WORKED

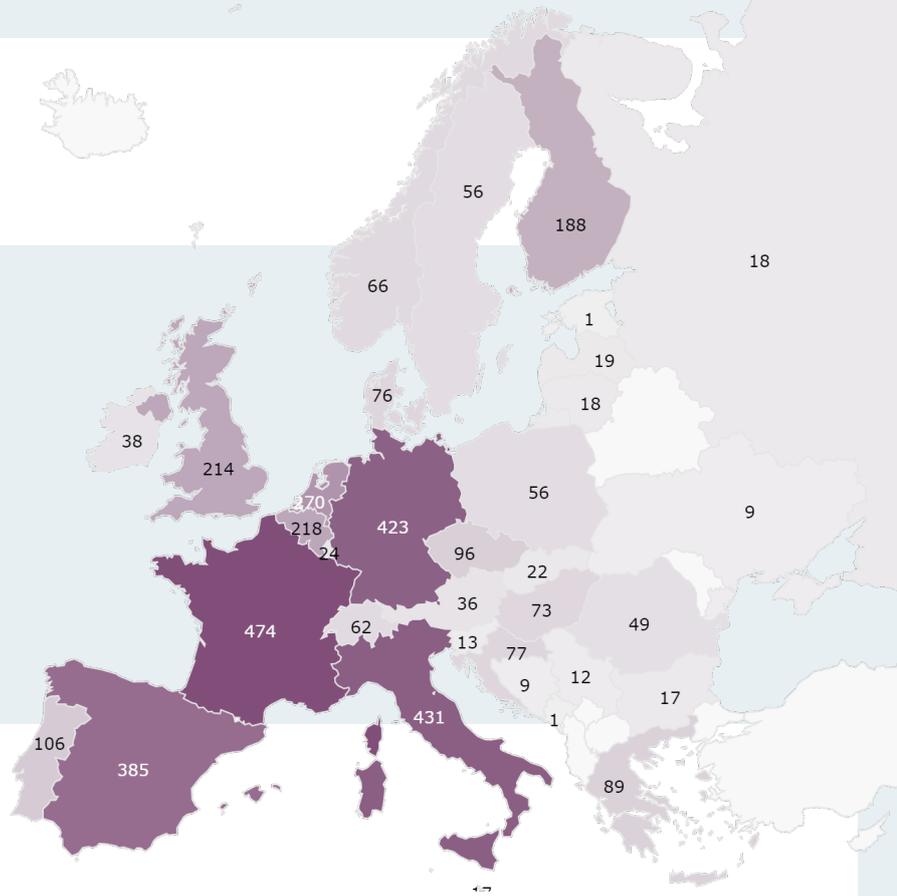
To prepare a better future for people living with a rare disease the Rare 2030 Foresight Study called upon Europe's most dedicated doctors, companies, researchers, young citizens, advocates, patients, payers and policy makers.

Together they comprised several expert panels of over 250 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.

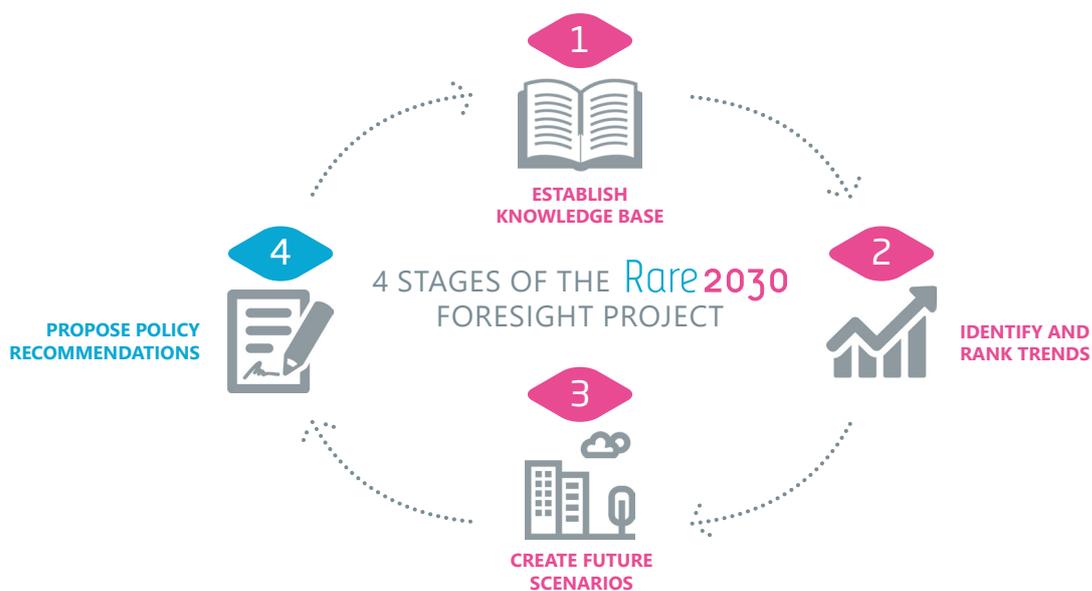
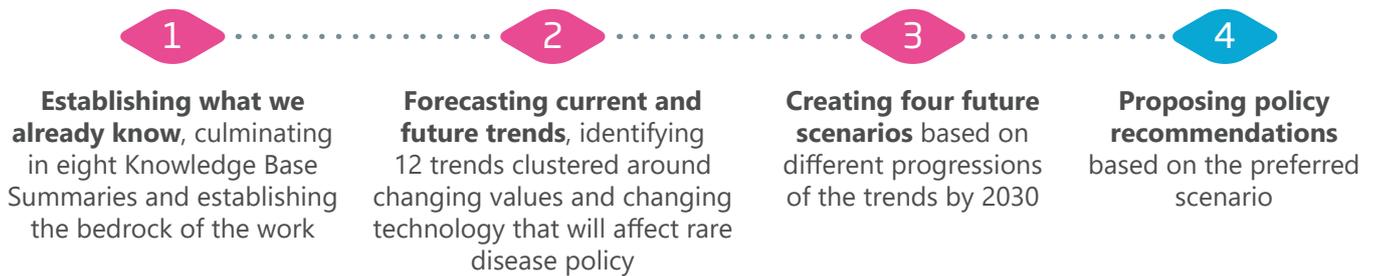
THE RARE 2030 FORESIGHT STUDY IN NUMBERS



TO ENSURE THAT RECOMMENDATIONS LEAD TO MEANINGFUL OUTCOMES, A SURVEY GATHERED THE OPINIONS OF OVER 3663 PEOPLE LIVING WITH A RARE DISEASE IN EUROPE THROUGH THE EURORDIS RARE BAROMETER PROGRAMME.



The Rare 2030 Foresight Study included three initial steps of reflection:



THE RARE 2030 TIMELINE



18 MAY 2019
Knowledge Base
Workshop – EURORDIS
Membership Meeting

JULY - AUGUST 2019
16 Topic Specific
Panel of Experts
Teleconferences

7 NOV 2019
Panel of Experts
Workshop - Validation
of Trends and Building
of Scenario Space

NOV/DEC 2020
4 Regional Conferences aligned
with upcoming EU presidencies

28 OCT 2020
Online Young Citizen
Conference Fall Debate

26 OCT 2020
European Reference Network
Plenary Event

SEPT - OCT 2020
4 Backcasting Workshops on
European Reference Networks

JULY - AUGUST 2020
16 Topic Specific Panel of
Experts Teleconferences

7 - 8 JULY 2020
Young Citizen
Conference

14 - 15 MAY 2020
The Rare Disease
Patient Journey in
2030 – European
Conference on
Rare Diseases and
Orphan Drugs

23 FEBRUARY 2021
Final
Rare 2030
Conference

2019

2020

2021

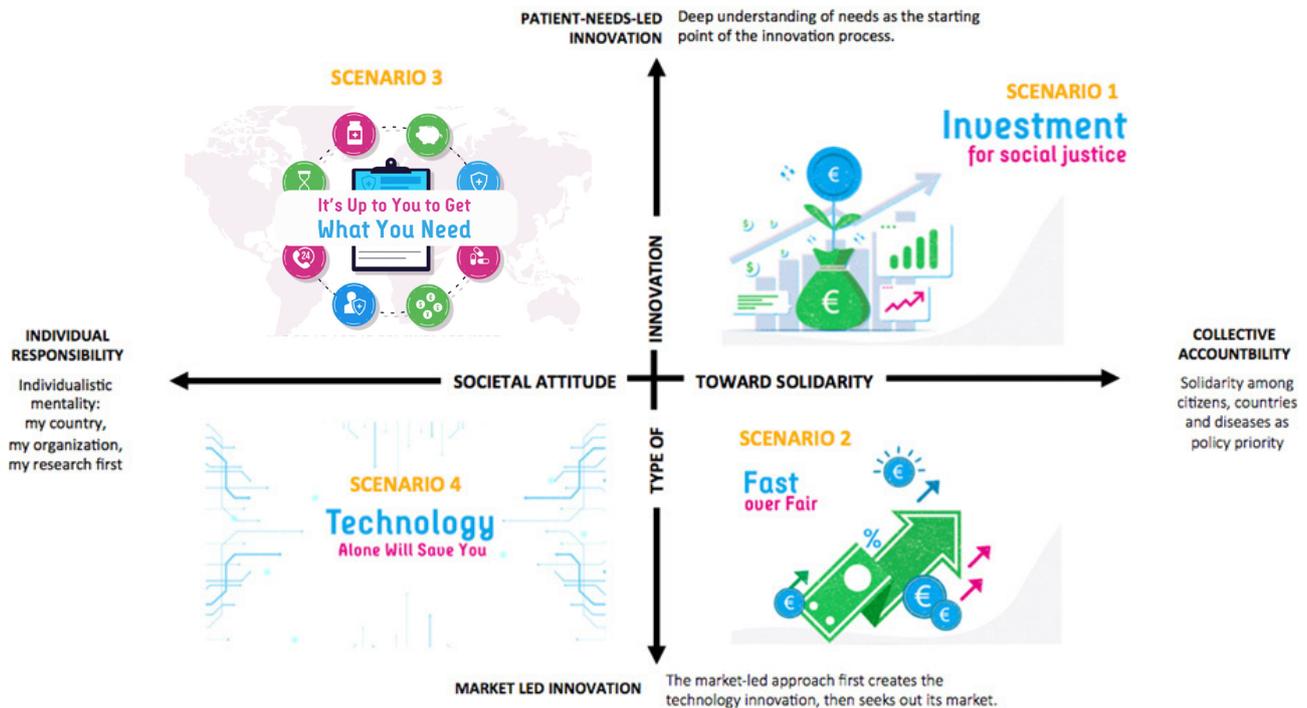
TWELVE RARE 2030 TRENDS

Through a series of consultations held during meetings in 2019 (the EURORDIS Membership Meeting, the Panel of Experts Workshop, the EURORDIS Council of National Alliances and Council of European Federation meeting), the Rare2030 Panel of Experts summarised and validated 12 trends that have been identified as instrumental to shaping the future of rare disease policy.

	OVERARCHING TRENDS IN RARE DISEASES	RELEVANT BROADER TRENDS IN HEALTH AND HEALTHCARE	TYPE OF TREND
1	Rise of pan-European multi-stakeholder networks to advance diagnostics, treatment and care for rare diseases	Multi-stakeholders governance	POLITICAL
2	Strains on the health care budget and the emergence of new care delivery models	New healthcare delivery models	ECONOMIC
3	Greater variation in access to treatments and care resulting in more inequality across Europe for people with rare diseases	Access to medical products	ECONOMIC
4	Demographic change of RD patients introducing new challenges	Ageing population in a changing family structure	SOCIO-CULTURAL
5	Threats to solidarity equity, and the prioritization of rare diseases	Increase inequality and threats to solidarity	SOCIAL AND ECONOMIC
6	Increasingly empowered rare disease patient and the patient advocacy evolution	Advocacy evolution and patient empowerment	SOCIO-CULTURAL AND POLITICAL
7	Rise in innovation-oriented, multi-stakeholder, needs-led (patient-led) research	Innovation in Healthcare Research	SOCIO-CULTURAL AND POLITICAL
8	Facilitation of knowledge exchange and local care delivery through digital health	Digitization of healthcare	TECHNOLOGICAL
9	Increased potential for large sets of standardised and interoperable data	Big Data	TECHNOLOGICAL
10	Rise in the use of AI for diagnostics, treatment and care, opening-up the potential of 'big data'	Big Data and Artificial Intelligence	TECHNOLOGICAL
11	New technologies and advanced therapeutics	Innovation in Medical knowledge	TECHNOLOGICAL
12	Application of Whole Genome Sequencing from the research to the clinical sphere	Genomics	TECHNOLOGICAL, ETHICAL AND LEGAL

TWO CLUSTERS OF TRENDS EMERGED AND THEIR PROGRESSIONS CREATED FOUR POSSIBLE FUTURE SCENARIOS VARYING IN DEGREES OF **SOLIDARITY** AND **DRIVERS OF INNOVATION**.

THE RARE 2030 SCENARIOS



THE RARE DISEASE COMMUNITY HAS IDENTIFIED SCENARIO 1 AS MOST PREFERRED, SCENARIO 2 AS MOST LIKELY AND SCENARIO 4 AS THE LEAST PREFERRED.

SCENARIO 1

If we prioritize societal responsibility, equity and the collaborative policy frameworks to achieve them, we will end up in the **Investments for Social Justice** world where major investments have been made by governments and are fairly shared across Europe ensuring health and well-being of all European citizens, and equity for vulnerable populations including those living with a rare disease. The European Union's increased legislative power in areas of health and social welfare fosters access to care, treatment, education, employment, leisure, psychological support and reduces the risk of being left behind no matter where you live in Europe. Healthcare systems are led by an understanding of a holistic view of a patient's needs, including the curative but also the preventative, rehabilitative and palliative. This understanding is

based on comprehensive health data collection seamlessly shared across borders in a system also designed to integrate data from consumer and profit driven companies. Multistakeholder initiatives prioritize investments in rare disease research that responds to patient needs. These efforts cover as many diseases as possible. The resulting innovations are regulated and assessed at the European level and with greater transparency, accountability, cost-effectiveness and consideration for the patient experience. With the right public and private partnerships, the field of rare diseases could play a pioneering role in promoting such a deep positive change in European health and social policy for all citizens.

SCENARIO 2

If we continue as we are, we'll end up in the **Fast Over Fair** world where stakeholders collaborate but only when they share the same interests. Due to significant private investment in research and development for rare diseases, many breakthrough technologies are available for diseases that are well understood, but very rare and complex diseases are left behind. Patient organizations, health care professionals and other stakeholders working across disease areas and multiple countries are key players. They help identify the gaps and disparities and advocate for better healthcare, treatments and research by considering the patient experience.

New IT solutions gathering large amounts of health-related data provide great potential to advance care and treatment. However, major bottlenecks exist due to a lack of clear and simple rules and incentives to share information, especially between the public and private sectors. Health remains the responsibility of each country. Driven by budget constraints, services are prioritized and although new technologies may come quickly, access to them is slow and limited. The challenges in driving fast AND fair innovation are likely to create frustration as well as an increased distrust in science, health care, and the systems that govern them.

SCENARIO 3

If distrust in supranational efforts to manage health continues, we may find ourselves in the **It's Up To You To Get What You Need** world with national two-tiered health care systems. In these systems, basic care is provided by the government and a second tier of care exists for those who can pay for better quality or faster access. In this scenario, the health and social welfare systems are well integrated and patients may get the holistic care they need, but it will highly depend on the country in which they

live. Given that no country has sufficient numbers of patients to conduct adequate research, innovation is hampered in this scenario due to a lack of investment in multinational collaboration, data collection and sharing platforms. The innovation that does reach the market for rare diseases is extremely expensive and only accessible for the richest countries and wealthiest patients, eventually making the market too small to attract investment.

SCENARIO 4

Following the principles of a free market economy we might find ourselves in the **Technology Alone Will Save You** scenario. Here, the government doesn't have a leading role, and private companies are the ones managing the health of people living with a rare disease. Health care systems are private, insurance-based, market-led and profit driven. Many innovations such as health applications are available to people living with a rare disease enabling them to be responsible for their own health. Patient organization support may even be largely replaced by technologies that allow patients to manage their own needs. Several pri-

vate companies have created fast and accurate diagnostic options but only for those who can pay for it. Thanks to the investments of health data companies, artificial intelligence and other cutting-edge science have led to breakthroughs for some of the most complex and rarest diseases, but many still remain without diagnostic and treatment options. Genomics has also developed dramatically and is regularly used by doctors and individuals. When commercially exploitable, the data is gathered in a collective way to find information that is helpful to patients. Patients and consumers may even be financially rewarded for their participation.

The four scenarios left the rare disease community with a choice on which direction they wanted to move in for a better future.

Their choice: a future based on **“INVESTMENT FOR SOCIAL JUSTICE”**, defined by patient-need driven innovation; high collective responsibility and performance based goals. This is driven by approaches prioritising human rights, multi-stakeholder and patient partnerships, technology and the holistic view to care and research. Importantly, this highlights the key principles justifying a coordinated European, and even an international approach, to address the challenges of rare diseases.

FROM THE PREFERRED SCENARIO TO A POLICY FRAMEWORK

To get there, we need new policy recommendations. This final step, consolidating the reflection through extensive participation at regional, national and European level, has resulted in the final recommendations, defined by the whole community, for a new generation of European policies for rare diseases by the year 2022.

For the first time these recommendations set goals for measurable outcomes for people living with a rare disease, not just establishing the infrastructure of how to get there, but where we need to go.

The Rare 2030 recommendations reflect new values, innovative technologies and approaches to address continued challenges and a new political climate that recognises rare diseases at the highest political levels¹¹. This makes for a mature ecosystem for a new generation of European policies for rare diseases that will guide Member States in their plans and strategies for the next 10 years and ultimately improve the health and well-being of a significant but vulnerable portion of Europe’s citizens.

11 - UN Member States include rare diseases in political declaration on universal health coverage
<https://www.un.org/pga/73/wp-content/uploads/sites/53/2019/09/UHC-HLM-silence-procedure.pdf>

WHO WAS CONSULTED IN THE RARE 2030 FORESIGHT STUDY

A CENTRAL CONSULTATIVE BODY: THE RARE 2030 PANEL OF EXPERTS

As the Rare2030 project seeks to identify future policy priorities and needs across the field of diagnostics, treatment, care and social support for people with rare diseases it was important to establish a similarly broad Panel of Experts (PoE), uniting a range of stakeholders with knowledge and expertise across different aspects of the field. This group of experts comprised one of the main consultative bodies of the project. The

PoE involved 200 experts from 38 countries¹². To stimulate meaningful discussion on the status quo, needs, trends and drivers of change in rare disease policy, the Panel was sub-divided into 8 distinct -though interconnected- working groups across the 8 recommendations in this report. Through a series of teleconferences and workshops these experts were consulted on the four stages of the foresight process.

AN ACADEMY OF ADVISORS: THE RARE 2030 RESEARCH ADVISORY BOARD



Ruediger Krech
WHO



Ruxandra Draghia-Akli
Johnson & Johnson
(formerly DG Research)



Robert Madelin
FIPRA (formerly DG
Sanco, DG Connect)



Milan Macek
Natl. Coord. Centre RD,
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Orphenet



Natacha Assopardi-Muscat
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Kate Bushby
Newcastle University
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Rare Diseases)



Tanja Bratan
Fraunhofer Institute



Simon Kos
Next Practice
(formerly Microsoft)



Didier Schmitt
European Space Agency
(formerly EC on Foresight)



Terkel Andersen
EURORDIS - Rare
Diseases Europe

The Rare 2030 Research Advisory Board represents one panel of key opinion leaders involved in the Rare 2030 Foresight Study. It is composed of high level experts in the field of rare diseases,

in health and research policy in general, emerging technologies and foresight studies - complementing the governance of the project.

¹² - Full membership list of the Rare 2030 Panel of Experts - <https://www.rare2030.eu/panel-of-experts/>

A PLAN FOR EUROPE SUPPORTED BY ITS MEMBER STATES: RARE 2030 REGIONAL CONFERENCES

Four Rare 2030 Regional Conferences were organised to provide key stakeholders at the national level consider how the trends identified by Rare 2030 Panel of Experts apply to the regional context, to debate which scenarios should be focused on or favored and to reflect on how policies may help shape a sustainable future for people living with a rare disease in the country of interest. Attendees included: representatives of National Alliances, Orphanet, Ministries of Health, local healthcare professionals involved in ERN, Young Citizens and other stakeholders considered as relevant based on the specific national context. To do so, participants of the

Regional Conferences were asked to review and adapt key trends at regional level, to identify explorative future scenarios taking into account country/region specific needs and relevant stakeholders' experience/expertise, and ultimately to identify key recommendations at the national and European levels.

- + **Croatia** – November 24 online
- + **France** (including participants from Belgium and Luxembourg) – December 3 online
- + **Spain** – December 4 online
- + **Italy** – December 1 and 9 online

A PLAN FIT FOR THE NEXT GENERATION: RARE 2030'S YOUNG CITIZENS

If policies are to be fit for the next decade they need to take into account the views and ambition of Europe's Young Citizens. The Rare 2030 Foresight study brought together 23 Young Citizens from across Europe to give the voice of the next generation: those currently training to be the doctors, researchers, health advocates, policy makers who will be following the policies in their work, and those young people already living with, or caring for someone with a rare disease.

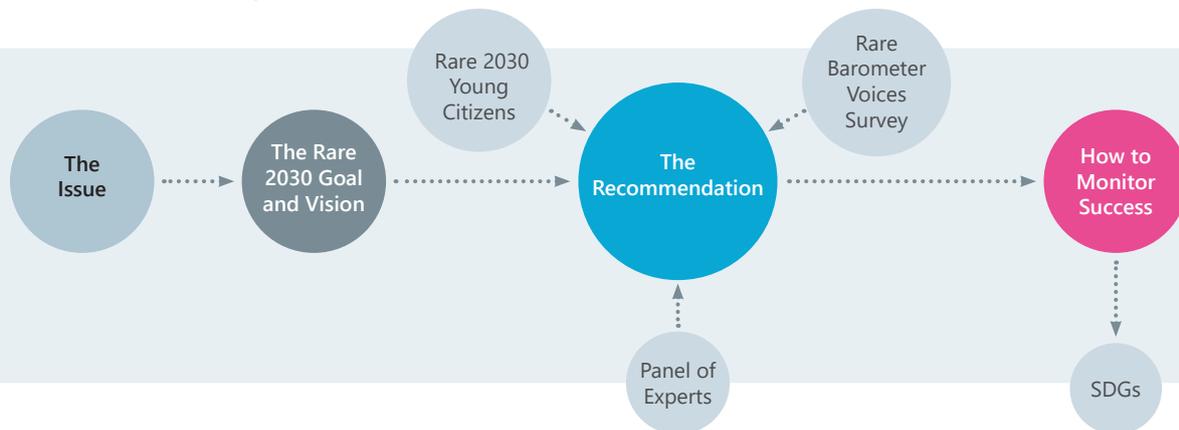
Based on the future they want to live in, they put forward their recommendations driven by the following four values

- + **Collaboration**
With a move towards more nationalistic policies across Europe, Young Citizens want to stress the need and value of collaboration in rare diseases, across borders, sectors and disease areas
- + **Innovation**
To make real strides forward, Young Citizens want the community to make more of data and technological advances, and drive innovation in research
- + **Person-centered care**
Looking beyond the physiological symptoms of a rare disease was an absolute priority for Young Citizens, with a focus on integrated support for mental health
- + **Education and training**
For a cross-cutting understanding of what it means to live with a rare disease, and for a greater understanding by all healthcare professionals.



HOW TO USE THIS DOCUMENT

The remaining chapters of this report detail each of the eight recommendations coming out of this foresight study. Each chapter is structured around a central recommendation which is the consolidation of inputs from all stakeholders involved in the two-year process and includes the following elements for each topic :



- + **The Issue** - this text defines an unmet challenge faced by people living with a rare disease in Europe.
- + **The Rare 2030 Goal and Vision** - where possible this goal describes what measurable changes we can strive for by 2030 to meet this challenge. Even when measurable goals are difficult to define, an aspirational vision describes what the rare disease community hopes to achieve as a future scenario by 2030.
- + **The Recommendation and How to Achieve This?** - Each recommendation summarises the highest level recommendations described in the How to Achieve This section. Both consolidated with consultation via the following resources:
 - The outcomes of the European Conference for Rare Diseases 2030 debates (Rare 2030 DELIVERABLE 5.2),
 - 32 teleconferences with Rare 2030 Panel of Expert topic groups (Rare 2030 Deliverable 6.2)
 - Recommendations from the Rare 2030 Young Citizens Conference and Debate
 - The Rare Barometer Voices Rare 2030 Survey
 - Existing frameworks or recommendations recognised by the rare disease community
- + **What Do People Living with a Rare Disease Think?** - Each figure displayed in this section comes from the surveys of the EURORDIS Rare Barometer programme. Those surveys were carried out in Europe between 2016 and 2021. In these surveys, people living with a rare di-

sease reported their views on a range of issues, such as participation in research, access to treatments, social care needs, the impact of COVID-19 on their lives and their hopes for 2030. You can read more about these surveys here:

- [Juggling Care and daily life: the balancing act of the rare disease community.](#)
- [Rare disease patients' participation in research.](#)
- [Share and protect our health data: Rare disease patients' preferences on data sharing and protection.](#)
- Rare disease patients' experience of treatments (report not available yet).
- [Improve our Experience of Healthcare! Patients' and carers' experience of medical care for their rare diseases.](#)
- [Rare disease patients' experience of COVID-19](#)
- Rare Disease patients' opinion on the future of rare diseases: Rare2030 foresight study
- To find out more about rare barometer surveys: eurordis.org/voices
- + **Monitoring Success** - where possible the Rare 2030 consortium identified indicators of Success that will be further developed and monitored as part of the Sustainability Plan of the project.
- + **Supporting the Sustainable Development Goals** - Rare 2030 Recommendations and aspiring to reach Rare 2030 goals are matched with UN Sustainable Development Goals to show how they contribute.



LONG-TERM, INTEGRATED EUROPEAN AND NATIONAL PLANS AND STRATEGIES



Rare diseases are a heterogeneous group of largely incurable, often multisystemic 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. Such collaboration, whether at the local, national or European and international level, requires clear policies to coordinate efforts in a strategic and concerted manner.



VISION OF THE RARE DISEASE COMMUNITY

Innovation to address unmet needs of 30 million people living with a rare disease in Europe as well as societal responsibility, equity and collaboration are prioritised in a European policy framework for rare diseases to achieve a future defined by 'Investments for Social Justice'. This policy framework guides the implementation of national plans and national strategies for rare diseases all designed to achieve the same measurable objectives. This policy framework complements existing legislations¹³ and brings together a concerted strategy across research, digital, healthcare, social global cooperation. It encourages major investments in the field of rare diseases at both the European and national levels which are fairly shared across Europe. The rare disease community envisions a future in which robust, action-oriented national

plans and strategies are strategically evaluated and implemented in order to meet the medical, holistic and research-related needs of people living with a rare disease. All stakeholder groups, including national and European policy makers, are able to share experiences and good practices via a suitable pan-European forum for rare disease policies, enabling the translation of innovative and effective approaches at the national level. Robust indicators addressing the myriad aspects of rare disease diagnostics, treatment, care, research and holistic wellbeing are agreed and collected across national jurisdictions under a comprehensive monitoring and quality improvement approach, to support meaningful and measurable progress for people with rare diseases.

13 - e.g. Pharmaceutical Strategy for Europe, European Data Strategy, European Pillar of Social Rights, Horizon Europe - Rare Disease Research and Innovation Partnership, Cross-border Healthcare, Transparency



RARE 2030 **GOAL**

1



A new EU policy framework for rare disease - including rare cancers, rare infections and rare poisonings - guaranteeing that rare diseases remain a public health priority through concerted European actions and guiding the implementation of long-term national plans and policies across all countries in Europe



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.

Specifically:

- + this framework should be aligned with other European and national strategies (e.g. cancer, data, research, access, social rights)
- + it should support a harmonisation of the definition of rare diseases and rare cancers in European and national plans, strategies and policies
- + this European policy framework should be aligned with the international objectives established in Sustainable Development Goals, Universal Healthcare and other UN system policies relevant to rare diseases.
- + a new focus should be placed on EU level monitoring of rare disease diagnostics, treatment, care, research, and holistic wellbeing, with countries encouraged to collect and pool such data to publically and transparently illuminate the status quo and enable benchmarking
- + European and national plans and strategies should be sustained on a long-term basis, with adequate funding, and should be monitored by the appropriate authorities and key opinion leaders in the field
- + a renewed focus should be placed on the state of the art of current national plans and strategies and the adoption of renewed national plans and strategies
- + a suitable forum should be created or designated to advance multistakeholder policy-oriented debate on rare diseases, enabling the identification of good practices and support for implementation to suit national realities



HOW TO ACHIEVE THIS?

AT THE EUROPEAN LEVEL

- + Greater pan-European - indeed sometimes global - collaboration is essential to address the health, research, economic, and holistic challenges posed by rare diseases, which know no borders and cannot be met by any single nation alone; in particular, the role of the European Union in the sphere of health should be augmented, through eventual adoption of a new charter
 - + A new Council Recommendation on an action in the field of rare diseases should be elaborated and adopted, as part of the new legislative and policy framework, following the overall assessment of the implementation of the current one, as demanded by the Council Conclusion of 16 June 2017 on Encouraging Member States-driven Voluntary Cooperation between Health Systems and the [2019 Special Report of the European Court of Auditors on implementation of Directive 2011/24/EU](#).
 - This new Council Recommendation should take into account specific recommendations pertaining to rare cancers (the rare diseases of oncology) set out in the [Rare Cancer Agenda 2030](#) (JARC, 2016-2019)
 - + Application of the EU Open Method of Coordination to the rare disease field should be explored, along with the potential to add rare diseases to the [agenda of the European Semester](#) ;
 - + Lessons must be learned from the COVID-19 pandemic, in terms of establishing the impact on already vulnerable rare disease populations and ensuring global, European, national, regional and local efforts to redress the damage and promote equality across all sectors, whilst capitalising on the positive momentum the crisis has created in terms of rapidly and efficiently streamlining procedures, research collaborations, clinical trials and regulatory activity.
- + Consensus indicators should be developed at European level to monitor rare disease diagnostics, treatment, care, research, and holistic wellbeing, with countries encouraged to collect and pool such data to publically and transparently illuminate the status quo and enable benchmarking:
 - This could be achieved through the [EU Open Method of Coordination](#) and/or the [Resource on the State of the Art of Rare Diseases Activities in Europe](#), and could build upon the [EUCERD Recommendations on Core Indicators for National Plans and Strategies](#).
 - Such indicators should illuminate cross-country collaborations as well as national-level activities, and should serve the purpose of identifying good practices which might be expanded or replicated elsewhere
 - + To align the rare disease field with the growing trend for outcomes-based medicine, the incorporation of rare diseases (including rare cancers) to the '[State of Health in the EU](#)' and to OECD activities concerning patient-centred outcomes, should be explored
 - + A dedicated multi-stakeholder body -with participation from all national competent authorities, along with European Reference Network coordinators, patient advocates, Industry, researchers, and independent experts should be established, with a remit to identify and assess best practices, and to review existing - and elaborate new - policies and recommendations on any subject under the 'rare diseases' remit.
 - This could be a new body, building on the new Rare Diseases Stakeholder Network under the EU Health Policy Platform, or perhaps be a subgroup under the Steering Group on Health Promotion, Disease Prevention and Management of Non-Communicable Diseases.



- This body should operate in collaboration with the Board of Member States of European Reference Networks on any relevant issues, and with the Policy Board under the European Joint Programme for Rare Diseases Research for research-related matters
- This body should promote specific opportunities and avenues for collaboration and crosstalk between countries linked by geography, size, language, or other relevant considerations, to enable a tailored approach to tackling the challenges posed by rare diseases

AT THE NATIONAL LEVEL:

The elaboration, implementation, evaluation and renewal of robust and effective national plans and strategies for rare diseases must once again be embraced as a key policy priority.

- + The European Union shall consider an updated request to Member States in connection with national plans and strategies for rare diseases, structured within the frameworks of the health and social systems
- + The aforementioned EU-level multistakeholder group tasked with overseeing policy challenges and opportunities for the full breadth of rare disease/rare cancer issues should ensure a key focus on revitalising the national plans and strategies agenda
- + Support should be provided from the European level in terms of updated KPIs for national plans/strategies and the identification and dissemination of good practices and solutions to shared challenges
- + National plans and strategies should be robustly evaluated and – in the case of time-bound policies – renewed or replaced by national authorities in a timely and transparent manner. National authorities should ensure intersectoral collaboration in the elaboration, evaluation and implementation of national frameworks for rare diseases/rare cancers, encompassing also social and holistic actions alongside the medical and research angles
- + National authorities should dedicate designated funding to implement the national plans and their constituent activities (which should include SMART objectives, wherever possible)
- + The integration of rare cancers (both in adults and paediatric cancers) in national cancer control plans should be fostered, with relevant synergies with national rare disease plans
- + National authorities should avoid subsuming 'rare diseases' into broader health strategies which reduce addressing their specificities and their strategic prioritisation and; however, where relevant strategies exist (for instance for genomics or cancer) appropriate links to the rare disease field should be ensured
- + National authorities should consider the applicability of rare diseases to the [UN Sustainable Development Goals](#) and [Universal Health Coverage](#) debates and incorporate this to their strategic agendas
- + Countries should create a Mirror Group on rare disease research, to interact with the European Joint Programme on Rare Diseases Policy Board on research matters, and integrate this to their national plans and strategies for rare diseases/rare cancers
- + Each renewed national cancer control plan should include relevant and specific measures for both paediatric cancers and rare cancers in adults, addressing the issues of research and access to adequate care, in synergy with national plans or strategies for rare diseases where relevant.
- + By 2025, all countries should have a 'live' national plan or strategy for rare diseases, with a dedicated multistakeholder oversight body and an annual budget separate from the wider health and social system



WHAT DO THE RARE 2030 YOUNG CITIZENS RECOMMEND?

ON COLLABORATION...

Young Citizens define collaboration across countries and sectors as a key component for the success of future policies. With the overarching goal to guarantee equality for the rare disease community in all relevant fields, Young Citizens recommend:

- + **Establishing a European Rare Disease Agency** that gathers expertise and resources into one single body responsible for all aspects of rare diseases, encompassing funding, social support, market access, data management, disease/patient registries, interoperability and use of best practice
- + **Developing a more cohesive and transparent reimbursement process**, through a European common system that would pay healthcare professionals for the time they spend with foreign patients. This would simplify the reimbursement process, There is currently a not sufficiently clear understanding of these procedures, which is currently unclear, as it varies from country to country and remains the responsibility of Member States
- + **Establishing a European reward system for treatment and care abroad**, to foster cross-border healthcare while ensuring transparency and legitimacy.. As Young Citizens believe a common European funding system has potential to bridge the gap between patients' needs and national resources to ensure care for all
- + **Developing a legal framework for data ownership and protection** that takes account of the opportunities and challenges new technologies bring
- + **Improving articulation between public and private sectors to clarify their roles** in guaranteeing optimal collaboration to positively impact access to care through common efforts



ON PERSON- CENTRED CARE...

The role of a political and strategic framework goes beyond concerns over practical and economic aspects of rare disease. With rare disease being often misunderstood and stigmatised, Young Citizens believe that a new framework should address these issues and thus call for:

- + A **framework that address discrimination, misunderstanding and stigma surrounding** rare diseases through training, awareness campaigns and new laws and policies making states more accountable for discrimination and stigmatisation
- + A framework that addresses **mental health issues and social care challenges** and provides funding support to hospitals for person-centred care rather than solely on healthcare

ON INNOVATION...

Young Citizens believe innovation should be a key component of national and European plans and strategies. Diagnosis in particular was identified as an area that could greatly benefit from innovative techniques. Young Citizens recommend:

- + **Developing more company-oriented incentives for diagnosis**, which could in turn stimulate research aiming at re-
- ducing the well too known 'Diagnostic Odyssey' through European structural and funding support



HOW TO MONITOR SUCCESS

- ✓ Develop consensus indicators for monitoring rare disease diagnostics, treatment, care, research, and holistic wellbeing, and the existence and implementation of national plans and strategies for rare diseases.
- ✓ Monitor the national and cross-border level data systematically collected. This could be achieved through the EU Open Method of Coordination and/or the Resource on the State of the Art of Rare Diseases Activities in Europe, and could build upon the EUCERD Recommendations on Core Indicators for National Plans and Strategies.
- ✓ Health, disability and socioeconomic statistics related to rare diseases are monitored by [Eurostat](#)



" I would like for ALL rare diseases the recognition that collectively rare diseases are common and that they are a health & human rights issues and not to be seen as only 'disabilities' [...] I would like to see more acceptance of genetic differences and more knowledge and awareness of the fact that not all disabilities are visible. Some [rare diseases] are life-limiting, some need orphan drugs/therapies and others require good treatment & management and, ultimately, we all want the same thing - timely access to health & social care and access to educational opportunities."

Rare Disease Patient



SUPPORTING THE SUSTAINABLE DEVELOPMENT GOALS BY 2030

3 GOOD HEALTH AND WELL-BEING



IMPROVING HEALTH AND WELL-BEING

Strong EU and national plans and strategies centralise objectives linked to improved health and well-being for people living with a rare disease.

4 QUALITY EDUCATION



INCREASING THE QUALITY OF EDUCATION

EU and national plans and strategies should include the building of capacities of people living with a rare disease to better advocate for their needs and raising awareness amongst healthcare professionals on the challenges faced by people living with a rare disease. As such we improved learning opportunities for people living with a rare disease or working in the field.

10 REDUCED INEQUALITIES



REDUCING INEQUALITIES

Monitoring of EU national plans and strategies provides a platform for benchmarking where inequalities within and among countries for people living with a rare disease can be identified and subsequently targeted for reduction.

17 PARTNERSHIPS FOR THE GOALS



INCREASE PARTNERSHIPS

By definition strategic and long-term plans and strategies for RDs imply the partnership of a large number of stakeholders across countries, disease areas and sectors.



REFERENCES

- + 2019 Special Report of the European Court of Auditors on implementation of Directive 2011/24/EU
<https://op.europa.eu/webpub/eca/special-reports/cross-border-health-care-7-2019/en/>

- + Rare Cancer Agenda 2030
https://www.jointactionrarecancers.eu/attachments/article/265/Rare_Cancer_Agenda_2030.pdf

- + Agenda of the European Semester
https://ec.europa.eu/info/business-economy-euro/economic-and-fiscal-policy-coordination/eu-economic-governance-monitoring-prevention-correction/european-semester_en

- + EU Open Method of Coordination
<https://www.eumonitor.eu/9353000/1/j9vvik7m1c3gyxp/vh7eg8ym3xy5 2>

- + Resource on the State of the Art of Rare Diseases Activities in Europe
<http://www.rd-action.eu/wp-content/uploads/2018/09/Final-Overview-Report-State-of-the-Art-2018-version.pdf>

- + EUCERD Recommendations on Core Indicators for National Plans and Strategies
<https://www.radico.fr/images/documents-utiles/europe/EUCERD-Recommendations-Indicators-adopted.pdf>

- + State of Health in the EU
https://ec.europa.eu/health/state/summary_en

- + UN Sustainable Development Goals
<https://sdgs.un.org/goals>

- + Universal Health Coverage
https://www.who.int/health-topics/universal-health-coverage#tab=tab_1

- + Eurostat
<https://ec.europa.eu/eurostat/web/health/overview>

- + «Rare Cancer Agenda 2030: Ten Recommendations from the EU Joint Action on Rare Cancers», 2019
<https://www.eurordis.org/publication/rare-cancer-agenda-2030-ten-recommendations-eu-joint-action-rare-cancers>

- + «Strengthening Europe in the Fight Against Cancer: Going further, faster», study for the ENVI Committee, European Cancer Organisation, 2020
[https://www.europarl.europa.eu/RegData/etudes/STUD/2020/642388/IPOL_STU\(2020\)642388_EN.pdf](https://www.europarl.europa.eu/RegData/etudes/STUD/2020/642388/IPOL_STU(2020)642388_EN.pdf)

- + Prades, et al. «Priorities on rare cancers' policy in National Cancer Control Plans (NCCPs): A review conducted within the framework of EU-JARC Joint-Action», Journal of Cancer Policy, 2020
<https://www.sciencedirect.com/science/article/abs/pii/S221353832030014X>



EARLIER, FASTER,
MORE ACCURATE
DIAGNOSIS



EARLIER, FASTER, MORE ACCURATE DIAGNOSIS

The search for an accurate diagnosis very often remains a diagnostic 'odyssey', for many reasons; for instance, the sheer number of conditions under the heading of 'rare diseases'; the scarcity (by definition) of patients with any single condition and the corresponding scarcity of experts acquainted with each condition; the tendency for rare diseases to manifest as complex, multisystemic conditions; the fact that

existing technologies are not fully utilised or implemented; to name a few. The lack of a diagnosis (or perhaps of an accurate diagnosis) can have far-reaching consequences for patients and their families. Heterogeneity of national capacities regarding genetic testing and different approaches to implementing evolving technologies can impact access to diagnosis, resulting in inequalities for patients and families across Europe.



VISION OF THE RARE DISEASE COMMUNITY

The rare community recognises that diagnosis is the first step towards any improvements in health and well-being and envisions a future where families across Europe no longer have to endure a diagnostic odyssey to obtain this critical information. Whether it be access to a treatment or the ability to plan for their future, by 2030 all people living with a rare disease will receive the attention they need as quickly as

possible - and this begins with a timely and accurate diagnosis. The community strongly supports the better application of current diagnostic testing and screening approaches (which includes low-cost but highly effective solutions such as raising awareness) as well as the use of new technologies to overcome current barriers in diagnosis equally across all countries.





RARE 2030 **GOAL**

2



All people living with a rare disease known in the medical literature will be diagnosed within six months of coming to medical attention.

All people will have access to the most effective diagnostic technologies, best practices and programmes (including screening) without discrimination and regardless of where they live in Europe. All currently undiagnosable individuals will enter a European and globally coordinated diagnostic and research pipeline.



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.

Specifically, to achieve earlier, faster, and more accurate diagnosis, the following actions should be supported:

- + Promote equality in access to diagnostic opportunities for people living with a rare disease or suspected rare disease, no matter where they live
- + Enable patients to navigate health systems with ease, following the most appropriate and direct route to obtaining a diagnosis, to connect with others in similar situations, and learn how to best manage their disease and participate in research in a safe and timely manner.
- + Co-design - with healthcare professionals and patients - care pathways to most efficiently guide people living with a rare disease from diagnosis to highest quality care and, where possible and appropriate, to the most relevant European Reference Network
- + Improve diagnostic expertise by fostering European and global networking of highly specialised healthcare providers and by ensuring greater interoperability and standardisation of data able to support diagnostics whilst ensuring particular transnational collaboration to diagnose the most complex presentations, rarest diseases and undiagnosed cases.
- + Ensure an integrated, international approach to patients with currently undiagnosable conditions, ensuring the absence of diagnosis does not preclude access to the best possible care and support
- + Raise awareness on rare diseases in medical curricula and amongst all primary/ front-line health and social care professionals and specialists
- + Foster broad and equitable implementation of next generation sequencing and other emerging new technologies into national healthcare systems, to facilitate and speed up access to diagnosis.



HOW TO ACHIEVE THIS?

BETTER USE AND ACCESSIBILITY OF EXISTING SOLUTIONS, WITHIN A MORE STRATEGIC AND COORDINATED DIAGNOSTICS ECOSYSTEM

Obtaining a timely and accurate diagnosis is a human right, whether there is an available medical treatment or not. The following steps should be pursued to better apply existing tools, best practices and programmes

AT THE EUROPEAN AND GLOBAL LEVELS

- + A clear, systematic and European-wide (indeed sometimes global) approach to rare disease diagnostics must be ensured, founded upon the ability to guide patients towards centres of expertise or equivalent, access transnational diagnostics platforms, and capture - and systematically manage - data on patients for whom a diagnosis is not forthcoming
- + Continued support must be ensured for multinational and multistakeholder research linking omics data, clinical data and biomaterials with well-defined patient cohorts and applying them in the clinic, building on the work of existing initiatives such as the European Joint Programme on Rare Diseases and Solve-RD
- + Existing and future best practice guidelines to support the diagnosis of rare diseases (such as decision trees and patient pathways) should be visible and findable at the European level (via European Reference Networks

ERNs and Orphanet) and should be adopted and implemented to a greater degree at the national level

- + The Orphanet services pertaining to diagnostics (resources concerning the definition and inventorying of diseases, and the database on expert clinical centres and laboratories) should increasingly be co-created and co-curated together with ERNs, and should be sustained by European action
- + Funding bodies in Europe and all other world regions should target diagnostics for subpopulations, indigenous people, and other culturally and linguistically diverse populations in a culturally safe manner (including populations in developing nations): this will support the genetic and phenotypic characterisation of rare disease populations to enlarge patient cohorts and advance knowledge and understanding.
- + Appropriate and targeted funding should be dedicated at EU and national levels to foster research into aetiology of rare diseases with no evident underlying genetic causes
- + Research should be fostered at European level to elucidate the determinants of the heterogeneity across EU Member States in terms of diagnostic performance
- + Research should be fostered at the European level (inline with the [Commission Expert Group on Rare Disease Recommendations on Cross-Border Genetic Testing](#)) to conduct a cross-border health economics assessment of diagnostic and screening technologies, comparing costs and benefits relative to those currently incurred under the diagnostic 'odyssey'



"My disease is extremely rare as there are only nine people living with this disease today, me included. I would like to see the creation of a worldwide network of patients (patient database) as a prerequisite to launch clinical trials and to improve research and diagnosis of my disease."

Rare Disease Patient



AT THE NATIONAL LEVEL

- + Countries should define clear national strategies to support RD diagnostics and should support professionals involved in diagnostics -and through them, patients- in their national territory to access specialised diagnostic platforms; in particular, to utilise genome-phenome platforms and similar tools suited to rare disease diagnostics, especially those recommended by IRDiRC now and in the future for diagnostic purposes
- + Countries should strive to meet the IRDiRC goal stating 'patients with a suspected diagnosable RD should receive an accurate diagnosis within 1 year of coming to specialist medical attention' and indeed should treat this as a minimum, reduced to 6 months or less in the case of conditions for which a preventive strategy demands neonatal or infant diagnosis.
- + Countries should fully implement the provisions within the [Commission Expert Group on Rare Disease Recommendations on Cross-Border Genetic Testing](#)
- + Countries should adopt, and provide the means (financially and organisationally) to actually implement EU level best practice recommendations on diagnosis and screening
- + Countries should ensure an available and appropriately trained workforce to address rare disease diagnostics in the clinics of the future
- + Countries must ensure that genomic and rare disease diagnostics services promote cultural awareness of all populations, including indigenous populations and other culturally and linguistically diverse populations, approaching diagnoses with sensitivity and ensuring appropriate coordinated and integrated care

European and national authorities must take action to reduce the inequalities stemming from the existing heterogeneity in national approaches to screening and prevention of rare disease, and collaborate to support more informed and transparent decision-making for primary and secondary prevention

- + The proposed activities highlighted in the [EUCERD Opinion on Newborn Screening](#) should be revisited and implemented through a European-level body or programme; new solutions proposed in [EURORDIS Key Principles for Newborn Screening](#) should be considered
- + Countries should work collaboratively to share best practices and HTA data concerning newborn screening programmes
- + The cost-effectiveness of newborn screening should be calculated and set against the costs of the diagnostic odyssey and costs to the health and social systems in the absence of an accurate diagnosis
- + An EU level recommendation on NBS should be created, addressing the following: the potential of genome sequencing for newborn screening; the need for screening panel expansion to be based on scientific advancement and health technology assessment; the recognition that screening is not just a test, but rather a process which requires adequate communication with families and the public; adequate training for healthcare professionals; and more
- + A greater focus on preconceptional prevention and care for rare diseases is required, encompassing primary prevention and screening, the need for improved communication with affected persons and family members, a greater emphasis on professional awareness and alertness, the need to ensure regular disease follow-up, and more. Regarding primary prevention, the [Recommendations of EUROCAT and EUROPLAN on Primary Prevention](#) should be revisited



LINKING BETTER DIAGNOSTICS TO CARE PATHWAYS

National and European authorities must place particular strategic emphasis on reducing the diagnostic 'odyssey' from primary care to specialised diagnostic support, by establishing and ensuring care pathways to

most efficiently accompany people living with a rare disease from diagnosis to highest quality care and where possible to the European Reference Network (ERN) covering the disease.

“

It is difficult to get the diagnosis. [...] We need to share all the information about this disease – how it is manifested, how it progresses, all the experience the patients and doctors have”

Rare Disease Patient

- + Individuals with suspected diagnoses must be referred to the most relevant specialist centres/ centres of expertise / coordination hub at the earliest opportunity: the precise role which ERNs could play in facilitating a diagnosis for rare disease patients lacking one should be clarified and better implemented at the national level.
 - + Governments should identify and implement optimal methods to share electronic health records (which include rare disease-specific data elements) across borders, in order to increase diagnostic efficiency
 - + Countries should prioritise the raising of awareness of rare diseases in primary care (essential for effective triage), focusing on the following:
 - Healthcare professionals must be encouraged to refer more readily when unsure of a patient's pathology– they should be encouraged to 'know what they do not know'
 - Countries/health systems should explore and invest in symptom-checking suspicion prompt tools in primary care settings, designed to raise 'red flags' and guide doctors towards specialised therapeutic centres to streamline the diagnostic odyssey
 - As triage to identify a possible area of specialism for referral rests upon awareness in primary and secondary care of how and where to access such expertise, national referral pathways to tertiary centres of expertise (or a catch-all coordination hub/centre for rare diseases) must be elucidated and made publicly available
 - Mandatory medical training for all health-care professionals should include education on rare diseases, including the following concepts: the specificities common to all rare diseases, and subsequent challenges; the main sources of information on rare diseases; the ways in which national and cross-border systems for rare diseases have been set-up; the unique value of research; and the considerations for diagnosis, treatment and care.
 - Online training and accredited courses in rare diseases should be made available to primary care workers
- ## STRATEGIC COLLABORATION TO ADDRESS THE NEEDS OF UNDIAGNOSED PATIENTS
- + Countries should implement the [International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients](#)
 - + Countries should build knowledge on existing undiagnosed rare disease patient populations – research should be conducted to establish number of undiagnosed patients, and socio-economic impact including impact on patients' and families' quality of life and ability to access health and social care



EARLIER, FASTER, MORE ACCURATE DIAGNOSIS

- + Undiagnosed patients should be properly coded in health information systems, by annotation of electronic health records with specific codes to ensure traceability and enable appropriate action from healthcare providers: the recommendations of initiatives including RD-CODE should guide and structure this activity
- + Whilst patients are awaiting a confirmed diagnosis for a suspected rare disease, access to appropriate health and social services should nonetheless be ensured:
 - Cross-country explorations are needed, to assess the feasibility of a temporary diagnosis based upon clustering of phenotype and symptoms: specific codes should be added to electronic health records, and diagnosis assertions metadata should be added to ORPHAcodes, based upon the recommendations of [RD-CODE](#) and other relevant initiatives
 - European guidance on genetic counselling following a diagnosis of a rare disease -or failure to find a diagnosis in a suspected rare disease patient- should be elaborated and implemented in all countries.
 - European countries should agree on a strategy for sharing core case details and samples - leveraging existing biobanking infrastructures - for unsolved patient cases, to ensure support for their unique needs in the absence of a diagnosis, and to practically and systematically ensure repeated testing as knowledge advances.
- + Ensure European support to best research and implement modern diagnostic technologies and advances equally across countries
 - at preconception, using novel techniques such as pre-implantation genetic diagnosis
 - during pregnancy, such as maternal blood test, ultrasound or chorionic villus sampling
 - at birth, considering the potential of genome sequencing in newborn screening programmes
 - later in life, taking into account artificial intelligence and genome sequencing techniques in the expansion of screening programmes
- + Robust data should be collected and analyzed on diagnostic utility, clinical utility, and cost-effectiveness while evaluating the impact of new technologies

APPLICATION OF NEW TECHNOLOGIES

AT THE EUROPEAN LEVEL

- + Continued support must be ensured for multinational and multistakeholder research linking omics data, clinical data and biomaterials with well-defined patient cohorts and applying them in the clinic, building on the work of existing initiatives such as the European Joint Programme on Rare Diseases and Solve-RD

AT THE NATIONAL LEVEL

- + Facilitate and expand access to scientific advancements such as next generation sequencing techniques, imaging, artificial intelligence and other digital solutions by applying them in a clinical setting in accordance with recommendations from consortia such as [Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease](#).
- + A wider range of agreed ontologies should be deployed in health and research data capture systems, to support diagnostics and facilitate extraction and mining of information - from real-world data particularly - to evaluate the impact and facilitate reimbursement of new technologies
- + Countries should ensure an available and appropriately trained workforce to address rare diseases diagnostics in the clinics of the future
- + Countries should ensure a greater investment in and development of clinical (phenotypic) interfaces, to complement investment in the genomic and screening aspects of diagnostics
- + Countries should ensure appropriate funding to support the translation of pilot projects on new diagnostic technologies into value-based solutions in the clinic



WHAT DO PEOPLE LIVING WITH A RARE DISEASE THINK?

The following figures come from several Rare Barometer surveys carried out in Europe between 2016 and 2021. In these surveys, people living with a rare disease told us their views on a range of issues, such as participation in research, access to treatments, social care needs, the impact of COVID-19 on their lives and their hopes for 2030. Find out more: eurordis.org/voices

GET A FASTER DIAGNOSIS IF DISORDERS ARE KNOWN IN THE MEDICAL LITERATURE

97% of people living with a rare disease agree to share their health data in order to foster research. This dataflow (well analysed, securely stored and constantly updated) is central to build a robust and up-to-date medical literature and to coordinate diagnosis techniques across the globe.

ACCESS TO MORE EFFECTIVE DIAGNOSIS STRATEGIES: NEW-BORN SCREENINGS

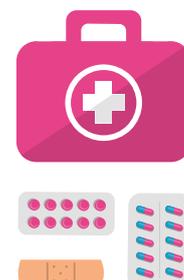
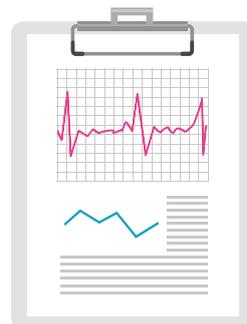
94% of respondents support the diagnosis of rare conditions at child's birth, pushing for implementing new-born screening across Europe.

People living with a rare disease also favour the use of new technologies to diagnose rare conditions before birth: either during pregnancy (87%) and around the time of conception (80%).

GET THE BEST DIAGNOSIS, REGARDLESS OF WHERE ONES LIVE, THANKS TO THE EUROPEAN REFERENCE NETWORK

+ The centers of expertise in rare diseases put in place in Europe (European Reference Network) are already showing results in significantly reducing the time between the first sought medical advice and the confirmed diagnosis of a rare or complex disease: it currently takes 2.8 years in ERNs to diagnose while it takes 4.1 years elsewhere in Europe.

+ ERNs also perform more genetic testing than non-ERN HCPS to either get or confirm a rare/complex disease diagnosis.





EARLIER, FASTER, MORE ACCURATE DIAGNOSIS



WHAT DO THE RARE 2030 YOUNG CITIZENS RECOMMEND?

ON COLLABORATION...

The diagnosis of rare diseases is often a long and difficult process. While society tends to see care as a synonym for treatment, young citizens see diagnosis as a currently under-investigated step of the care pathways, which would highly benefit from greater collaboration amongst stakeholders at every level of the care process. They thus recommend:

- + **Supporting those with 'undiagnosed' rare disease**, where collaboration across sectors is key to provide those with 'hard to diagnose rare disease' with more innovative diagnosis tools, professionalism, understanding and support.
- + **Solidifying pathways from first symptoms to care**, possibly involving ERNs more than they currently are in diagnosis and in raising awareness on rare disease diagnosis.

ON PERSON-CENTRED CARE...

Diagnosis is a challenging step for people living with a rare disease, which often lack psychological support and accompaniment. Young Citizens think that current efforts do not sufficiently take account of aspects that go beyond the physiological side of diagnosis and therefore recommend:

- + **Bringing support to people living with a rare disease before and at the time of diagnosis**, in the form of psychological support and other initiatives aimed at accompanying patients from symptoms onset to diagnosis and beyond.

ON INNOVATION...

Innovation is at the centre and basis of potential progresses in diagnosis. Young Citizens consider that future policies should be aimed at fostering this crucial step of the care pathways, gathering all relevant stakeholders to guarantee that the most innovative techniques are the main subject of collaboration and investment. They recommend:

- + **Improving existing newborn screening programs**, with more research to highlight the benefit of expanding the scope of rare diseases included in national screening programs while harmonising their scope across Europe
- + **Increasing investments in diagnostic solutions**, mostly with regards to linking disease/patient registries to diagnosis instead of treatment alone. Such efforts would require action and investments both at the European (harmonising and linking data) and national levels (collecting and sharing data)
- + **Employing telemedicine and remote care to support improved diagnosis**, as the COVID-19 crisis has demonstrated the enormous potential for telemedicine in delivering healthcare and that further developments of remote care would allow patients to have appointments and tests for diagnostic purposes without having to travel unnecessarily



HOW TO MONITOR SUCCESS

- ✓ Monitor numbers - and diagnoses of - existing and newly diagnosed patients attending ERN HCPs, through the ERN Continuous Monitoring and Quality Improvement System (ERN CMQS)
- ✓ Metrics collected within the scope of EU-wide monitoring for myriad aspects of rare diseases, relating to diagnosis of people with a rare disease in each country (tracking of the status quo for undiagnosed patients), including the implementation of new diagnostic technologies and best practices in the clinic
- ✓ Regularly monitor the experience and expectations of people living with a rare disease and their carers on the extent to which diagnostic needs are addressed
- ✓ Interrogate the ecosystem of integrated and federated data from across the research and care continuum, to objectively monitor patient journey from first symptoms to accessing the highest available quality of care





EARLIER, FASTER, MORE ACCURATE DIAGNOSIS



SUPPORTING THE SUSTAINABLE DEVELOPMENT GOALS BY 2030

3 GOOD HEALTH AND WELL-BEING



IMPROVING HEALTH AND WELL-BEING

Addressing the diagnostic needs of people living with a rare disease not only directly helps achieve better health and well-being of a large percentage of Europe's citizens by more quickly accessing the highest quality of research and care, but also allows parents, carers and their families best plan and prepare all other aspects of their lives including reproductive decisions and psychological and social well-being.

5 GENDER EQUALITY



ACHIEVING GENDER EQUALITY

Women living with or caring for someone living with a rare disease are disproportionately affected. As patients and caretakers they face more bottlenecks in reaching a correct and timely diagnosis because their concerns, including symptoms at the time of diagnosis, are taken less seriously.

9 INDUSTRY, INNOVATION AND INFRASTRUCTURE



BUILDING RESILIENT INFRASTRUCTURE, PROMOTE INCLUSIVE AND SUSTAINABLE INDUSTRIALISATION AND FOSTER INNOVATION

Increasing research for rare diseases both in terms of disease mechanisms but also development of diagnostics, telemedicine tools and data sharing technologies contributes to goals to enhance scientific research, upgrade the technological ability of many sectors across all countries, increase the scientific research workforce and increase access to information and technology all part of the 2030 SDGs.



REFERENCES

- + Commission Expert Group on Rare Disease Recommendations on Cross-Border Genetic Testing
https://ec.europa.eu/health/sites/health/files/rare_diseases/docs/2015_recommendation_crossbordergenetictesting_en.pdf

- + EUCERD Opinion on Newborn Screening
<https://isns-neoscreening.org/wp-content/uploads/2016/06/Expert-opinion-document-on-NBS-FINAL.pdf>

- + EURORDIS Key Principles for Newborn Screening
<https://www.eurordis.org/newbornscreening>

- + Recommendations of EUROCAT and EUROPLAN on Primary Prevention
<https://eu-rd-platform.jrc.ec.europa.eu/sites/default/files/EUROCAT-EUROPLAN-Primary-Preventions-Reccomendations.pdf>

- + International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients
https://www.udninternational.org/documenti/schede/international_joint_recommendations.PDF

- + RD-CODE - Standard procedure and guide for the coding with Orphacodes
http://www.rd-action.eu/wp-content/uploads/2017/05/D5.2_Standard-procedure-and-guide_final.pdf

- + Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease
<https://www.globalrarediseasecommission.com/>

- + Rare Barometer Survey: Improve our Experience of Healthcare! Patients' and carers' experience of medical care for their rare diseases
https://download2.eurordis.org/rbv/HCARE/HCARE_FS_long.pdf

- + Rare Barometer Survey: Rare Disease patients' opinion on the future of rare diseases: Rare 2030 foresight study (report available soon)
<https://www.eurordis.org/voices>



ACCESS TO HIGH QUALITY HEALTHCARE



Rare diseases are not only considered rare due to their low prevalence but also the scarcity and geographic dispersion of health care experts that can treat them. Unfortunately this has created a 'geographical lottery', in which patients 'fortunate' enough to live reasonably close to true experts in their conditions might hope to benefit from the fruits of their knowledge and experience (accrued through a concentration of patient

cases across the years). However, many patients may be limited in accessing the best existing care and treatment on their disease. Numerous approaches and tools have been identified at European level, to try to eradicate such inequalities which can often exist within countries, as well as between countries and above all for people living with a rare disease as compared to those who do not.



VISION OF THE RARE DISEASE COMMUNITY

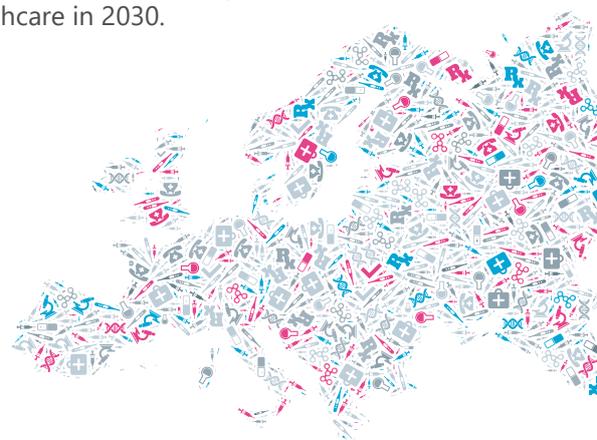
The rare disease community envisions a future in which:

- + patients are empowered to navigate healthcare systems and are able to have access in 3 'clicks' to the right information, to remote care services and to specialist advice based on global knowledge.
- + frontline clinical services are equipped with 'actionable intelligence' powered by digital healthcare pathways that provide timely access to evidence and patients' data, allowing them to combine evidence-based medicine and personalised care.
- + national health systems capacities are further strengthened, under an EU health system for rare diseases, which offers better coordination of healthcare services, invests more resources, and has greater competencies to plan and deliver a more effective response to rare diseases as a serious cross-border public health priority.

With increased solidarity, EU Member States will overcome healthcare fragmentation and inequalities in access, provide an integrated whole-system response and be stronger to protect, prevent and meet the evolving needs faced by people living with a rare disease.

Such an EU 'whole-system' response to rare diseases would provide clear lines of sight from frontline services to expert centres at both a national and European level, enabling the expertise to travel and - where needed - the centralization of care, under leading expert centres recognised as 'rare disease lighthouses'.

Ultimately, the rare disease community envisages that these actions can reduce health inequalities in several fundamental ways: by facilitating earlier diagnosis; ensuring access to affordable, evidence-based care, treatment, and therapies; accelerating clinical trials; developing new guidelines on treatments and care; and supporting the lifelong education of a healthcare workforce sensitive to and specialised in the needs of people with complex and rare diseases. Each of these is necessary to optimise access to healthcare in 2030.





RARE 2030 **GOAL**

3



All citizens can exercise their right to access a timely diagnosis, high quality essential healthcare, and safe, effective and affordable medicines and treatment, as close to home as possible or else have easy access to physical or remote cross-border healthcare, without unnecessary delay, under an EU “whole-system” approach for rare diseases.



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.

Specifically, there is a need for:

- + Greater solidarity and EU competencies in health, to facilitate the centralization of decision-making where needed, better policy and strategic planning as well as pooling of resources and expertise for highly specialized healthcare via the European Reference Networks (ERNs), under an EU health system for rare diseases.
- + Sustainable and proportionate investment from national and EU budgets into strengthening Centres of Expertise capacities and enhancing ERNs competencies and services to better protect people living with a rare disease. Investment by both Member States and the EU is critical for ERNs to become a core component of national healthcare systems.
- + Scaling up the capabilities and strengthening EU healthcare systems, leveraging network-based health data, experience and knowledge, powered by digital tools; where Centres of Expertise act as a trusted universal source and curators of global knowledge and integrate it to daily clinical practice.
- + An interoperable infrastructure and a fully-fledged data strategy for rare diseases to collect and exploit the full value and potential of health-related data in alignment with and contributing to the European Data Space and the European data strategy. EU healthcare systems must leverage the technological advancements that enable health data sharing for primary and secondary purposes, with appropriate safeguards, to develop digital healthcare pathways and accelerate the development and uptake of treatment options for rare diseases and facilitate European-wide clinical research.



HOW TO ACHIEVE THIS?

Given the breadth and depth of the topic 'access to healthcare', several categories of recommendations are required, directed towards different actors:

- + **a)** Recommendations for a more strategic and directive European role in enabling access to high quality healthcare for rare and complex diseases
- + **b)** Operational recommendations to lead Europe towards optimal European Reference Networks (ERNs) of the future
- + **c)** Recommendations at the national level, to optimise access to care at national level and support more seamless cross-border care for all patients requiring it

A) RECOMMENDATIONS FOR A MORE STRATEGIC AND DIRECTIVE EUROPEAN ROLE IN ENABLING ACCESS TO HIGH QUALITY HEALTHCARE FOR RARE AND COMPLEX DISEASES

- + Greater pan-European, and indeed global, collaboration is called for to address the health inequalities citizens face in accessing highly specialised healthcare in the EU, in particular the EU should develop a Health Framework for Rare Diseases, that formalising MS collaboration in strategic and workforce planning and decision making to develop and manage a European highly specialised healthcare system, where ERNs are the operational arm for delivery.
- + Pan-European policy should be elaborated to ensure the centralisation of care and expertise, for the rarer diseases, to organise care pathways, based on prevalence and incidence levels, and to commission services on an optimal population size to ensure safe and sustainable services are accessible for all.
- + Shared resources should be developed for commissioning and contracting designated European Centres of Expertise, for rarer diseases and highly specialised interventions that affect an annual national caseload of <250 to be accessed by all affected individuals.

B) OPERATIONAL RECOMMENDATIONS TO LEAD EUROPE TOWARDS OPTIMAL ERNS OF THE FUTURE

- + The strategy of future ERNs must be targeted towards all rare disease patients in Europe, and not only those attending ERN HCPs or 'affiliated' centres: ERN operations (from guidelines to data collection, knowledge generation to research) should always target this wider population, wherever possible
- + A common EU agency should be created/ adapted to enable ERNs to operate more flexibly and effectively, and receive funding from a range of sources (including 'external' sources such as industry and private donors, with an appropriate governance for public-private partnerships)
- As many believe ERNs should ideally each be legal entities, they should – as an interim solution, at least - be nested within such an organisation, or a foundation, to provide a mechanism for ERNs to easily receive funds.
- + ERNs need a long-term funding framework which should consider ALL possible sources of funding: such a framework needs to be defined urgently and should include a definition of all central functionalities and policies to support ERNs' financial management and governance
- + The realistic costs of network coordination, relative to the activities of the ERNs, should be established, and coordination funding provided on these grounds – a core coordination budget, available to all ERNs, should be supplemented by an additional variable budget, based on size, scale, coverage and activities
- + A special category of association or collaboration or affiliation should be created to allow formal collaboration and recognition of centres from countries outside of the EU Member States /EEA; clear rules on shared activities (what is and is not permitted) should be created.



- + ERNs and the BoMS should embrace a strategic mission of promoting more integrated care, encompassing integration of different medical specialities, but also of paramedical and social actors, in line with the [EUCERD Recommendations on Rare Disease European Reference Networks](#) and the [Commission Expert Group Recommendations to support the incorporation of rare diseases to social policies and services](#)
- + The European Commission should support Member States and EEA countries to implement the actions outlined in the [ERN BoMS Statement on Integration of ERNs into national health systems](#), specifically by funding national multistakeholder workshops, with patient organisations, clinical leads and national authorities, to facilitate discussions and actions on integration into each of the national health systems.
- + ERNs should develop strategies to minimise disparity between European regions in access to high quality healthcare: disease-related metrics should be agreed and monitored
- + A robust focus on continuous monitoring of ERNs is required, to demonstrate their impact: the EU-monitored indicators should be supplemented with more nationally-relevant and disease-relevant indicators
- + ERNs should develop clear and transparent rules for patient engagement, adequately supporting the involvement of patient organisations and their representatives in the different ERN activities, and should fairly compensate patient representatives for expenses and expertise.
- + ERNs must be supported to educate and train the future experts in rare diseases, in terms of clinical training, surgical training, and also training in holistic care and wider wellbeing
- + ERNs should be supported to review and expand their disease-specific membership criteria, in collaboration with patients and professional associations, with an emphasis on the necessary multidisciplinary expertise: in this way, EU countries (perhaps even the global RD community) could make use of robust criteria by which to define expertise in given disease areas
- + A cross-ERN working group on integrated and holistic care should be established as soon as possible, in partnership with RareResourceNet (the European Network of Resource Centres for Rare Diseases), as a gateway to build joint guidance on collaborative approaches for the provision of integrated and holistic care to people living with a rare disease: dedicated funding should be made available for broad stakeholder meetings and activities to advance this goal
- + ERNs should gather and create, in collaboration with patient organisations, resources which could support rare disease patients in receiving more integrated and more personalised care in their local environment: such resources should translate to heterogeneous care and social settings, by focusing on clarifying and explaining the (often poorly-understood) needs of patients with complex conditions, and adaptations/approaches which could help
- + A dedicated study/project should be funded, to support countries in developing their Electronic Health Records and virtual care delivery services to best address the specificities of rare diseases and highly specialised healthcare, and promote interoperability: this could aim at wider national deployment of the CPMS or a system compatible with it, as the basis for virtual care provision for complex rare disease cases nationally (whilst ensuring that any move towards more virtual care must be proportionate, to avoid further marginalization of a vulnerable population).
- + The Clinical Patient Management System (CPMS) should be fully compatible with any referring HCP systems, enabling automatic and two-way cross-talk with Electronic Health Records, to populate and update records post case referral: CPMS data should be fully searchable, and cases accompanied by an appropriate PPRL (privacy preserving record linkage) solution
- + An efficient project/tender should be funded to establish a pricing model to reimburse expert time spent on CPMS case review and propose options for payment (e.g. a quid pro quo system, a straightforward billing of another Member State/EEA country (perhaps with a differential GDP-based pricing scheme), a reduction in the workload of ERN HCP clinicians in lieu of payment for CPMS reviews, etc)
- + The Social Security Regulation and/or Cross-Border Healthcare Directive should be amended to allow for payment of time spent on cross-border virtual consultations performed through the CPMS, following a systematic national referral process.



- + Countries should consider automatically authorise requests for treatments or therapies if deemed beneficial by an ERN panel through

CPMS: the ERNs' expertise should hold more weight than national expert bodies who make such decisions at present



"I was searching for specialists/experts from other countries to understand my condition. Ideally they are around and you can meet them from time to time. But I prefer to talk with knowledgeable experts via the phone compared to meeting a doctor/specialist who doesn't understand my condition."

Rare Disease Patient

C) RECOMMENDATIONS FOR NATIONAL AUTHORITIES, TO IMPROVE ACCESS TO CARE AT THE NATIONAL LEVEL AND CREATE AN ENVIRONMENT SUPPORTING SEAMLESS CROSS-BORDER CARE FOR ALL PATIENTS AS REQUIRED

IN TERMS OF OPTIMISING NATIONAL ECOSYSTEMS BROADLY:

- + The elaboration, implementation, evaluation and renewal of robust and effective national plans and strategies for rare diseases must once again be embraced as a key policy priority at national and European levels
 - The European Union should consider an updated request to Member States in connection with national plans and strategies for rare diseases structured within the frameworks of the health and social systems
 - The aforementioned multistakeholder group tasked with overseeing policy challenges and opportunities for the full breadth of rare disease issues should ensure a key focus on revitalising the national plans and strategies agenda
 - Support should be provided from the European level in terms of updated KPIs for national plans/strategies and the identification and dissemination of good practices and solutions to shared challenges

- National plans and strategies should be robustly evaluated and – in the case of time-bound policies – renewed or replaced by national authorities in a timely and transparent manner
- National authorities should ensure inter-sectoral collaboration in the elaboration, evaluation and implementation of national frameworks for rare diseases, encompassing also social and holistic actions alongside the medical and research angles
- National authorities should dedicate designated funding to implement the national plans and their constituent activities (which should include SMART objectives, wherever possible)
- National authorities should avoid subsuming 'rare diseases' into broader health strategies which reduce their strategic prioritisation; however, where relevant strategies exist (for instance for genomics or cancer) appropriate links to the rare disease field should be ensured
- National authorities should consider the applicability of rare diseases to the UN SDGs and Universal Health Coverage debates and incorporate this to their strategic agendas
- By 2025, all countries should have a 'live' national plan or strategy for rare diseases, with a dedicated multistakeholder oversight body and an annual budget separate from the wider health and social system
- National authorities should carefully assess medical education and training materials designed to support professionals in providing an optimal standard of care to



people with rare diseases; where appropriate, resources generated at European level, representing best practice, should be recommended for national use (including educational resources emerging from the ERNs and Multidisciplinary Joint Committee of Rare and Undiagnosed Disease (MJC RUD) at the European Union of Medical Specialists)

IN TERMS OF GREATER SUPPORT TO -AND INTEGRATION OF- ERNS:

- + Renewing or updating national plans and strategies for rare diseases should remain a key priority for all countries - all such documents should stipulate the strategy to engage bidirectionally with ERNs: support in this task should be provided by a group/body with a remit to encompass all rare disease topics, beyond ERNs alone
 - + Countries should be encouraged to revisit and update their national designation of Centres of Expertise (CEs) for rare diseases and strengthen the organisation of national rare disease and specialised care networks - this should then translate to a more strategic engagement of national CEs with ERNs, via a limited number of full member HCPs.
 - + The concept of a Centre of Expertise for rare diseases should be revisited/affirmed at national level: countries should ensure they designate all such centres in a comprehensive and transparent way, and make the result of such a mapping and designation publicly available, demonstrating how ERN HCPs and 'affiliated' centres fit within wider national networks (where applicable). The [EUCERD Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases](#) remain a robust resource here and countries should aim to meet this, to support a baseline comparability in quality criteria
 - + National competent authorities should define national referral pathways for rare disease (or suspected rare disease) patients or those requiring a concentration of expertise, addressing transition from paediatric to adult care and containing clear guidance on how and when to seek referral to an ERN; ERNs should then compile and publish these pathways, explaining the process in each country and producing -with their patient advocates- patient-friendly information and advice on accessing specialist advice under an ERN.
- + All Member States and EEA countries should identify and publicise a clear process to facilitate the referral of patients for ERN care: this might include endorsing one centre as a 'National Coordination Hub' (or, if a federated system, endorsing a centre in each region, or in several strategically-selected regions) to manage referrals and function as gateways to accessing the specialist advice of the ERNs collectively - any such centre should work in partnership with the national patient community, and build relationships with national professional societies and research leads
 - + The European Commission should provide coordination funding to coordinating HCPs but Member States /EEA countries should provide funding to each national HCP/affiliated member within their national territory (providing they meet performance and impact indicators) to support their engagement in ERN activities
 - + Hospitals must strengthen support for the participation of their clinicians and other professionals in ERNs
 - + Each Member State and EEA country should define a mechanism, centred upon Orphanet, for instance, to disseminate and utilise the knowledge and evidence generated by the ERNs, to impact across the wider health and social systems; in particular, clinical practice guidelines/clinical decision support tools generated or endorsed by an ERN should be fully applied in all Member States and EEA countries, and national committees and structures dedicated to rare disease or specialised healthcare should include some level of national ERN HCP representation

RECOMMENDATIONS ON THE ROLE OF ERNS IN DATA GATHERING, RESEARCH AND INNOVATION

ERNs must be provided with the financial, technical, political and operational support required to collect and use findable, accessible, interoperable and reusable data as a means to support the accelerated development and uptake of treatment options for rare diseases and integrate European-wide clinical research and care settings. **Specific recommendations have been included throughout this report in respective sections pertaining to research and data.**



WHAT DO PEOPLE LIVING WITH A RARE DISEASE THINK?

The following figures come from several Rare Barometer surveys carried out in Europe between 2016 and 2021. In these surveys, people living with a rare disease told us their views on a range of issues, such as participation in research, access to treatments, social care needs, the impact of COVID-19 on their lives and their hopes for 2030. Find out more: eurordis.org/voices

REMOTE CONSULTATIONS WILL HELP ACCESSING THE HIGHEST POSSIBLE QUALITY OF HEALTHCARE

39% of People living with a rare disease are willing to use remote health care to access higher quality of care.

79% would be happy to use remote consultation if it enables to gather different doctors on the same consultation also known as multi-disciplinary care (for instance reunite a GP and a specialist on the call).

76% would also use remote consultations if it gives them access to doctors who live abroad but are specialised in their rare disease.

PEOPLE LIVING WITH A RARE DISEASE ARE MORE WILLING TO TRAVEL TO ANOTHER COUNTRY TO RECEIVE CARE OR TREATMENT THAN THE GENERAL POPULATION

9% of the people living with rare disease would not be willing to travel to another country to receive medical treatment; they were 46 % in the general EU population in 2014. In particular:

84% (vs 71% in the general population) of People living with a rare disease would be willing to travel in order to receive treatment that is not available in their country.

45% (vs 38% in the general population) of People living with a rare disease would be willing to travel in another country to receive treatment from a renown specialist.

NATIONAL COMPETENT AUTHORITIES NEED TO ADDRESS PATIENT'S RESERVATIONS ABOUT GETTING TREATED ABROAD

48% of respondents said they are not willing to travel to another country because they do not have enough information about the availability and quality of medical treatments abroad.

44% have no information on patient safety and quality of care abroad.

43% of respondents would have issues understanding the language.

43% are not aware of their rights in case things would go wrong.



WHAT DO THE RARE 2030 YOUNG CITIZENS RECOMMEND?

ON COLLABORATION...

Equal access to care throughout Europe vastly depends on collaboration across countries and sectors. A multidisciplinary approach going beyond national borders is thus a critical condition for effective initiatives and policies, mostly regarding the role of ERNs. Young Citizens recommend:

- + **Clarifying and reframing the role of ERNs** through better communication on how an individual person living with a rare disease could adequately benefit from these networks. Currently there seems to be a misunderstanding – and therefore a missed opportunity – of ERNs' benefits and structure as well as on the way one can interact with them
- + **Establishing a European reward system for care and treatment abroad**, fostering cross-border healthcare and facilitation of authorisation process to seek treatment beyond national borders. Young Citizens believe developing a common European funding scheme for rare disease treatment at the EU level could bridge the gap between patients' needs and national resources to ensure care for all. This mechanism could also improve social support to patients while seeking care abroad.
- + **Improving telemedicine practices**, as fostering efforts and further developing telemedicine practices would allow patients to have remote appointments without having to travel and benefit from telemedicine for care



HOW TO MONITOR SUCCESS

- ✓ With respect to ERNs, the ERN Continuous Monitoring and Quality Improvement System (ERN CMQS) will be utilised
- ✓ The planned revision of the Cross-Border Healthcare Directive should stipulate how to monitor the fulfillment of citizens' rights to access a timely diagnosis, high quality essential healthcare, and safe, effective and affordable medicines and treatment



"Before getting taken on by [Centre of Expertise] as a rare disease patient, I had over twenty years of poor care. But [Centre of Expertise] now looks after me incredibly well."

Rare Disease Patient





SUPPORTING THE SUSTAINABLE DEVELOPMENT GOALS BY 2030

3 GOOD HEALTH AND WELL-BEING



IMPROVING HEALTH AND WELL-BEING

Addressing the holistic needs of people living with a rare disease not only directly facilitates earlier diagnosis, access to clinical trials, access to treatments, new guidelines on treatments and care and supporting the lifelong education of a healthcare workforce sensitive to and specialised in the needs of people with complex and rare diseases their health and well-being are improved and the goal to achieve universal health coverage, including financial risk protection, access to quality essential health-care services and access to safe, effective, quality and affordable essential medicines and vaccines for all is supported.

4 QUALITY EDUCATION



INCREASING THE QUALITY OF EDUCATION

By raising awareness of rare diseases in medical curricula across Europe the quality of medical education is improved overall. Investments in continuous training of healthcare professionals within the rare diseases field will sustain knowledge gathered. Encouraging training of healthcare professionals outside the field of rare diseases will promote a culture of awareness about rare diseases that will spread throughout the chain of care of people living with a rare disease and their families.

10 REDUCED INEQUALITIES



REDUCING INEQUALITIES

People living with a rare disease are currently a marginalised and often invisible population, with little information available about their diseases and very few treatment options. They often suffer inequality in accessing health care services and treatment, and in the prices they have to pay, due to their social status or their country of origin. By ensuring that treatment and consideration of people living with a rare disease is equal across Europe, we contribute to the reduction of inequality.



REFERENCES

- + EUCERD Recommendations on Rare Disease European Reference Networks
<https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-9-30>

- + EUCERD Recommendations to support the incorporation of rare diseases to social policies and services
https://ec.europa.eu/health/sites/health/files/rare_diseases/docs/recommendations_socialservices_policies_en.pdf

- + ERN BoMS Statement on Integration of ERNs into national health systems
https://ec.europa.eu/health/sites/health/files/ern/docs/integration_healthcaresystems_annex_en.pdf

- + EUCERD Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases
https://www.eurordis.org/sites/default/files/publications/factsheet_Centres_Expertise.pdf

- + EURORDIS-Rare Diseases Europe - Recommendations to achieve a mature ERN system in 2030
<https://www.eurordis.org/maturevisionern>



INTEGRATED AND PERSON-CENTRED CARE



INTEGRATED AND PERSON-CENTRED CARE

Today, the 30 million Europeans living with a rare disease and their family members (often the main carers) remain a marginalised and largely invisible population, with little information about their diseases and their rights, few treatments, and a high level of psychological, social and economic vulnerability. The need for an integrated, social and holistic approach to person-centred care is particularly important for people living with a rare disease, for which only approximately

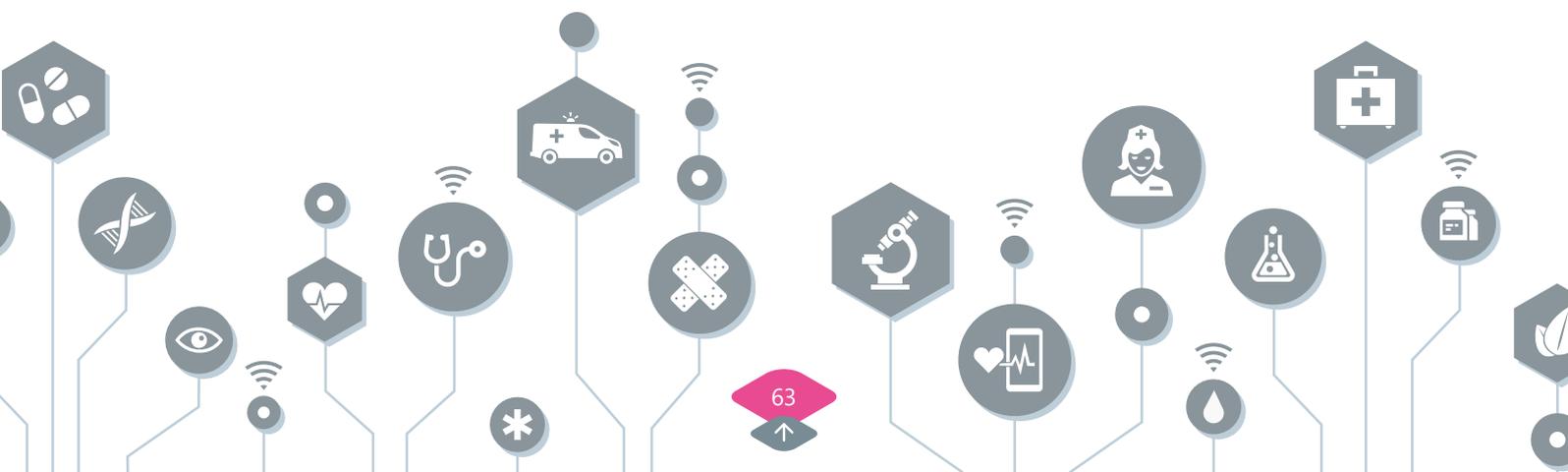
6% of conditions have a dedicated therapy of any kind. The rare disease community recognises that despite tremendous progress, even in 2030 curative or stabilising treatments will exist only for a relatively small number of all identified rare diseases and hence only for a minority of all people living with a rare disease. Therefore, for the vast majority, the greatest gains in quality of life will result from social determinants as often as clinical determinants.



VISION OF THE RARE DISEASE COMMUNITY

By 2030 the community envisages a paradigm shift in the design of current health and social systems to better address and integrate the holistic needs of people with rare diseases - including the curative but also the preventative, rehabilitative and palliative - aiming at social inclusion and ensuring that no European citizens are left behind in reaching their full potential

regardless of where they live. Caring for people living with a rare disease will mean recognising the complexity of the conditions, the associated impairments and barriers to social inclusion of the person and addressing the multi-faceted impacts of the condition on the families and carers.





RARE 2030 **GOAL**

4



Reduce the level of psychological, social and economic vulnerability of people with a rare disease and their families by one third.

Rare diseases seriously impact everyday life

7 in 10 patients & carers

reduced or stopped professional activity due to their or their family member's rare disease.



8 in 10 patients & carers

have difficulties completing daily tasks (household chores, preparing meals, shopping etc.)



2/3 of carers

spend more than 2 hours a day on disease-related tasks.



3 times more people

living with a rare disease and carers report being unhappy and depressed than the general population*

* Rare Barometer Voices sample compared to International Social Survey Programme, 2011



RARE 2030 RECOMMENDATION

4

INTEGRATED AND PERSON-CENTRED CARE

Implement EU-wide and national actions by all stakeholders that guarantee 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

Specifically:

- + Ensure the principles set by **existing conventions** including the The [Universal Declaration of Human Rights](#); The [United Nations Convention on the Rights of Persons with Disabilities](#); The [European Charter of Fundamental Rights](#) are effectively achieved
- + Implementing the principles in the **European Pillar of Social Rights** recognising the synergies but also the specificities of people living with a rare disease
- + Ensure that the [Directive \(EU\) 2019/1158 on work-life balance](#) for parents and carers of 20 June 2019 is implemented in EU Member States
- + Foster a **multi-sector approach** from research, to diagnosis, access to treatment, health care and social care, and long-term care, at both national and European levels, putting the person at the centre
- + Safeguard the **continuity of important frameworks** and Europe-wide platforms essential for the development and sharing of good practices in rare disease social and health care, including the European Reference Networks and the [European Network of Resource Centres for Rare Diseases](#).
- + Support the **design of health and social systems** which address the holistic needs of people with rare diseases at policy, programme and service delivery levels: recognising the profession of case management, entitling access to social workers and adequate social support, guaranteeing the "right to be forgotten" for financial, social, and professional contexts
- + Ensuring the recognition and adequate compensation of the disabilities experienced by people living with a rare disease



HOW TO ACHIEVE THIS?

All people with a rare disease should be supported to participate fully and equally in society, labour, education and leisure, without discrimination and in fulfilment of their basic human rights but also in recognition of the benefits to society at large.

Policies, programmes and services to address the social and person-centred needs of people living with a rare disease should synergise with global, European, national, regional and /or local disability programmes or strategies, to build solidarity in areas of commonality, whilst maintaining a focus on the features which demark rare diseases for special attention .

People living with a rare disease, their representatives and families should contribute directly and indirectly to the design of such policies, programmes and services to ensure they are person-centred care and prioritised in terms of

true needs. The disproportionate impact of rare diseases on women carers, in particular, must be ameliorated through appropriate policies across multiple domains including social well-being, employment, diagnostics, reproductive choices.

INTEGRATED, PERSON-CENTRED AND LONG-TERM CARE

Policies and practices must be designed and implemented to ensure a local, regional, national and European focus on identifying and addressing the social and holistic needs of people with a rare disease: more integrated care (both in terms of integration across medical disciplines but also bridging the medical and social spheres) should be provided for rare and complex diseases

“

“Since I was followed by the [Centre of Expertise], my quality of life has improved considerably, as has my mental approach to my pathology and life. [...] It allows me to lead a full life, as if I did not suffer from any disease. Without the support of the specialists and their staff, this would not have been possible.”

Rare Disease Patient

AT THE EUROPEAN LEVEL:

- + A common definition and indicators concerning person-centred care - bridging health and social domains- for people with rare diseases should be defined at European level, through a future Council Recommendation or similar, in view of the specificities and knowledge-gap associated with rare diseases, which necessitate a European approach
- + Financial and structural support should be allocated to ensure the sustainability of relevant Europe-wide platforms including the European Reference Networks (ERNs), the European Network of Resource Centres for Rare Diseases and Orphanet. These platforms gather and share essential knowledge and good practices that support countries to

effectively address both the health and the social needs of people living with a rare disease. An enabling environment should thus be created to integrate these initiatives with national health and welfare systems.

- + The European Commission should support a dedicated initiative or body to collect and review concrete good practices for ensuring an integrated and holistic approach to care for rare diseases, and to assess the impact of different approaches and interventions in a structured and systematic manner
- + European Reference Networks and the Board of Member States should embrace a strategic mission of promoting more integrated care, encompassing integration of different medical specialities, but also of paramedical and





social actors, in line with the [EUCERD Recommendations on Rare Disease European Reference Networks](#) and the [Commission Expert Group for Rare Diseases Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies](#)

- + A cross-ERN working group on integrated and person-centred care should be established (by 2025) in partnership with European Resource Centres for Rare Diseases, as a gateway to build joint guidance on collaborative approaches for the provision of integrated and person-centred care to people living with a rare disease: dedicated funding should be made available for broad stakeholder meetings and activities to advance this goal
- + ERNs should gather and create - in collaboration with patient organisations - resources and data which could support rare disease patients in receiving more and better adapted integrated and more personalised care in their local environment: such resources should translate to heterogeneous care and social settings, by focusing on clarifying and explaining the (often poorly-understood) needs of patients with complex conditions, and adaptations/approaches which could help patients; making use of digital tools where needed and helpful
- + The European Commission - in collaboration with the European Network of Resource Centres for Rare Diseases - should raise particular awareness of the need for cross-border collaboration between rare diseases resource centres and other centres for complex diseases and disabilities, promoting identification and sharing of best practices and exchange of knowledge
- + The European Commission should increase funding opportunities to assess the true impact (clinical, social, personal, and financial) of rare diseases through collaborative research as referenced in Recommendation 10 of the [Commission Expert Group for Rare Diseases Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies](#)
- + The European Commission should support proof of concept studies to demonstrate how preventative, integrated care can result not only in better quality of life and grant people living with a rare disease the right to the standards of health and social care they are

entitled but also in economic savings; sharing this evidence base widely

- + The European Commission should support research to assess and publicise the respective levels of functioning and disability associated with rare diseases, through a publically-available database accessible for all (for instance through expansion of the Orphanet Disability Project or similar)

FOR THE NATIONAL LEVEL:

- + Countries should invest as needed to fully implement EU level standards, infrastructures and tools including the existing consensus recommendations and resources concerned with the social and holistic needs of people with rare diseases, namely the following:
 - The [Commission Expert Group for Rare Diseases Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies](#)
 - [Recommendations of the INNOVCare project](#)
 - The recommendations within the [EURORDIS Position Paper 'Achieving Holistic Person-Centred Care to Leave No One Behind'](#) (which were built upon past European projects including the EUCERD Joint Action and RD-ACTION)
- + In view of the fact that national plans/strategies for rare diseases should structure activities within health and social systems, bodies intended to implement, evaluate, and/or renew these plans/strategies should involve representatives from both Ministries of Health, Ministries of Social Affairs, Welfare, Labour or equivalent and Ministries of Education, to support a multidisciplinary perspective.
- + National plans/strategies for rare diseases should provide dedicated funds to encourage the bridging of health and social care and enable holistic wellbeing (encompassing also educational -including transitional- and employment opportunities) along with other incentives to encourage coordinated care across-sectors (with a particular focus on the opportunities offered through eHealth, cancer plans and data strategies)
- + Countries should set aside more resources to cover or reimburse the costs of non-pharmacological therapies including preventative, rehabilitative and palliative care (when sup-



ported by best practice guidelines in peer-reviewed literature)

- + Countries should ensure that the concept of a centre of expertise within the national territory (including ERN HCPs) is as aligned as possible with the EUCERD Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases, including also the requirements to ensure multidisciplinary and to collaborate with paramedical, social, and educational actors
- + Countries should ensure that all centres of expertise for rare diseases (including ERN HCPs) in the national territory include a patient care coordinator or case manager role – these positions should be accompanied by an official

career pathway, in terms of qualifications, (continuous) training and salary, and should support patients in accessing the health and social care they need, closer to their home: the INNOVCARE training resources can be instructive here

- + Countries should ensure robust networking between centres of expertise for rare diseases and Resource Centres for Rare Diseases addressing disabilities: such networking may need to take place on a cross-border basis, in the absence of domestic centres, in which case national authorities should facilitate the sharing of best practices and exchange of knowledge

“

“People believe I am making up symptoms, contributing to my poor mental health and making me able to cope less”

Rare Disease Patient

EQUAL OPPORTUNITIES AND ACCESS TO THE LABOUR MARKET

- + All EU and national level legislation must guarantee that there is no form of discrimination based on health or disability status. The [Horizontal Equal Treatment Directive](#) should be swiftly adopted and any discrimination on all grounds covered in the Article 21 of the [European Charter of Fundamental Rights](#), and in all fields, should be tackled.
- + All legislative proposals and recommendations deriving from the European Pillar of Social Rights must take into account the specific needs of people living with a rare disease, their carers and others with complex diseases/disabilities.
- + The [‘Social Scoreboard’](#) should introduce clear indicators that reflect the reality on the ground and monitoring tools to support effective policy changes.

ACTIVE SUPPORT TO EMPLOYMENT

- + Access to high quality education must be guaranteed to all people with rare diseases and complex conditions. When necessary, adapted schooling should be accessible and delivered in a way that supports all individuals to reach their maximum potential.
- + Tailor-made assistance to improve employment or self-employment for people living with a rare disease, such as career counselling to explore fulfilling professional avenues, is needed.
- + All EU and national level legislation must guarantee that there is no form of discrimination based on health or disability status, concerning all forms of employment, including recruitment, hiring, employment, career advancement and safe and healthy working conditions. The Employment Equality Directive must be fully implemented with targeted support via EU funds, appropriate legislative frameworks, and exchange of practices to support the labour market integration of groups in disadvantaged situations as part of active labour market policies.



- + Access to social protection measures, pension rights and care support must be guaranteed for people living with a rare disease, their carers and others with complex conditions when leaving the labour market or having to work part-time due to the disease.
- + Measures to ensure people living with a rare disease and with disability who wish to study and/or to be active as volunteers for civil society organisations, are in no way deprived from their rights, including disability and retirement benefits.

FOR FAIR WORKING CONDITIONS

- + European countries, via the implementation of the Work-Life Balance Directive and other means, must ensure that people with complex conditions/disabilities and their carers have the right to specific mechanisms that support their access and retention in the labour market:
 - Flexible work arrangements, such as flexible working hours and remote work;
 - Reasonable leave of absence due to their health/disability condition or caring responsibilities;
 - Reasonable accommodation in the workplace.
- + The European Commission should provide MS with the necessary support to ensure the full implementation of Directive (EU) 2019/1158 on work-life balance for all parents and carers of people living with a rare disease who need those provisions.
- + The European Commission should provide guidelines for MS on how to ensure reasonable accommodation for people living with a rare disease in the workplace, in line with Article 5 of Directive 2000/78/EC. The guidelines should encourage MS to entitle people living with a rare disease with adequate leave of absence and flexible work arrangements, in line with the provisions offered to parents and carers within the Directive (EU) 2019/1158 on work-life balance.

SOCIAL PROTECTION

- + The future EU Child Guarantee must fully integrate the challenges of children in most

vulnerable situations, as is the case of children living with a rare disease, or suffering from a cancer or surviving a childhood cancer, guiding EU and national policy frameworks and financial resources to ensure adequate resources for childcare and early intervention services;

- + EU MS must implement specific mechanisms to guarantee coordination between national policy sectors within a multidisciplinary approach, engaging health, social, work, education and research Ministries. Inter-Ministerial working groups and shared budgets between Ministries should be implemented;
- + The future European Strategy on the Rights of Persons with Disabilities must provide guidance to Member States on disability assessment procedures to ensure persons with all types of disability, including persons with rare conditions or multiple impairments, are not overlooked and are provided with adequate levels of disability allowance, social protection schemes, community-services and independent living arrangements;
- + A Convention on the Rights of Persons with Disabilities (CRPD) Unit should be established within the European Commission, placed in the EU Directorate General for Justice and Consumers, under the supervision of the EU Commissioner for Equality. This unit would be responsible for the implementation of the UN Convention on the Rights of Persons with Disabilities and for coordinating the work of all disability focal points in EU institutions;
- + EU MS must guarantee that all people living with a rare disease and their carers are entitled to access a social worker and adequate social protection and social inclusion provisions, adapted to their individual needs and to the cost of living;
- + The “right to be forgotten” should be enacted in national legislation: medical information relating to rare diseases or cancers should not be collected or held by insurance organisations for longer than ten years following the end of treatment (five years in the case of paediatric patients)
- + Patient organisations also provide specific support, information and counsel to patients, their families and carers along the patients’ journey. Their social action should be supported by European and national authorities.



WHAT DO PEOPLE LIVING WITH A RARE DISEASE THINK?

The following figures come from several Rare Barometer surveys carried out in Europe between 2016 and 2021. In these surveys, people living with a rare disease told us their views on a range of issues, such as participation in research, access to treatments, social care needs, the impact of COVID-19 on their lives and their hopes for 2030. Find out more: eurordis.org/voices

LIVING WITH A RARE DISEASE INCREASES THE VULNERABILITY AT A PSYCHOLOGICAL LEVEL

37% of the respondents declared that they often feel unhappy and depressed, compare to 11% of the general population.* (Comparison with the general population: ISSP, International Social Survey Programme, 2011)

58% think possible that, within 10 years, they could get the psychological and emotional support they are lacking today.

24% need, by 2030, more access to meditation and other related techniques to better manage their symptoms.

THE SOCIAL VULNERABILITY OF PEOPLE LIVING WITH A RARE DISEASE IS VERY HIGH

52% declared that their rare disease has a severe impact on everyday life (e.g. capacity to carry out daily tasks, motor and sensorial functioning, personal care). The majority of people living with a rare disease need to visit different health, social and local support services in a short space of time, and find that hard to manage.

Remote consultations is seen as a great opportunity (79%) to do multidisciplinary consultations that will ease the coordination between the different healthcare providers.

42% of people with rare diseases spend more than 2 hours a day on care-related tasks. This time burden falls heavily on women, often the main carers. Remote consultations can play a role as "saving time" will be, according to rare disease patients and carers, the main reason for them to use remote consultations within 10 years.

LIVING WITH A RARE DISEASE REDUCES JOB OPPORTUNITIES AND JEOPARDISES PEOPLE'S FINANCES

76% of the respondents declare that the fact they are affected by a rare disease has limited their professional choices

67% also declare that the disease has limited them in being promoted. Which has repercussions on their finances. Hence 12% would favour remote consultations, within the next 10 years, to save money.

13% have decided to look after their child or children full time and had to make the decision of leaving or significantly reduce employment.

19% declared that living with a rare disease prevented them from studying according to their wishes, therefore impacting their career.





WHAT DO THE RARE 2030 YOUNG CITIZENS RECOMMEND?

ON PERSON-CENTRED CARE...

Living with a rare disease and what this entails remains a mystery to a large part of society, potentially leading to stigma and discrimination. Taking account of aspects such as mental health, sexual health or idiopathic disease/symptoms is a key component to include in future policies and frameworks. Young Citizens recommend:

- + **Reframing what it means to be a person living with a rare disease**, therefore taking account of domains going beyond clinical care and treatments and adopting a more holistic perspective in rare diseases policies and societal understanding
- + **Better addressing psychological and social challenges for people living with a rare disease**, including increased funding in hospitals and other healthcare settings to integrate a truly holistic approach to care, justified both by legislation and policies in the private and public sectors – with a particular focus on discrimination and stigmatisation – while making member states more accountable for their implementation

ON EDUCATION AND TRAINING...

Education and awareness raising on disability and the holistic needs of people living with a rare disease is currently not sufficiently emphasised nor addressed in society. Young Citizens call for:

- + **Enhanced education around rare diseases and their challenges** to both professionals and society as a whole, including awareness on rights to cross-border healthcare, increased development of training on discrimination, as well as well as fostering already existing trainings in all appropriate settings (hospitals, social sector, schools, etc.)

ON INNOVATION...

The importance of research to demonstrate the importance of and improve holistic care should be acknowledged and increasingly supported. People living with a rare disease should be involved in designing social research to best address priorities. Young Citizens therefore recommend:

- + **Improving assessment methods**, thus involving patients who have first hand-experience in reporting outcomes and experiences (PROMs and PREMs) to identify unmet needs and prioritise research areas that would truly make the difference for those who would benefit from upcoming research



HOW TO MONITOR SUCCESS

- ✓ Evaluate the implementation of relevant European and international legislation with increased accountability
- ✓ Monitor the use of European Commission Structural Reform Support Service (SRSS) and other common funds to support implementation of relevant legislation and policies
- ✓ Monitor the shift of policies, programmes and services from one of pure clinical disease evaluation towards the assessment of the person-centred, holistic experience
- ✓ Conduct cross-sector analysis (healthcare-economic-social) of the impact of policies, programmes and services through patient-centred studies
- ✓ Regularly monitor the experience and expectations of people living with a rare disease and their carers on the issue of integrated and person-centred care and those working in the field of psychological and social policy, programme planning and service delivery

“

“The appointments should be multidisciplinary and allowing for the various specialists to see the patient on the same day and the same place. Besides facilitating communication and the definition of adequate treatment it would also avoid that the patient has to travel to different appointments and places, being absent from work and spending a lot in travel.”

Rare Disease Patient



SUPPORTING THE SUSTAINABLE DEVELOPMENT GOALS BY 2030

1 NO POVERTY



REDUCING POVERTY

By addressing the economic vulnerability of people with a rare disease and their families we contribute to eradicating poverty amongst Europe's citizens.

3 GOOD HEALTH AND WELL-BEING



IMPROVING HEALTH AND WELL-BEING

Addressing the holistic needs of people living with a rare disease not only directly helps achieve better health and well-being of a large percentage of Europe's citizens, but also improves the psychological and social well-being of carers and families.

4 QUALITY EDUCATION



INCREASING THE QUALITY OF EDUCATION

As 70% of rare diseases affect children, many people with rare diseases face difficulties in attending school due to the frequency of medical appointments, inaccessibility of facilities, and non-adapted teaching methods. By spreading knowledge on rare diseases to teachers, educators and the next generation, we promote a culture of acceptance and a world where all children have a chance to learn.

5 GENDER EQUALITY



ACHIEVING GENDER EQUALITY

Women living with or caring for someone living with a rare disease are disproportionately affected. As patients, their symptoms are taken less seriously than men. They also more often assume the role of primary carer of a person living with a rare disease in their family and thus experience greater interruptions to their professional life, greater levels of stress and physical and emotional strain.

8 DECENT WORK AND ECONOMIC GROWTH



DECENT WORK AND ECONOMIC GROWTH

By integrating people living with a rare disease in societies and economies decent work and economic growth can be improved for a significant portion of Europe's population.



10 REDUCED INEQUALITIES



REDUCING INEQUALITIES

People living with a rare disease are currently a marginalised and invisible population, with little information available about their diseases and very few treatment options. They often suffer inequality in accessing health care services and treatment, and in the prices they have to pay, due to their social status or their country of origin. By ensuring that treatment and consideration of people living with a rare disease is equal across Europe, we contribute to the reduction of inequality.

REFERENCES

- + The Universal Declaration of Human Rights
<https://www.un.org/en/universal-declaration-human-rights/>

- + The United Nations Convention on the Rights of Persons with Disabilities
<https://www.un.org/development/desa/disabilities/convention-on-the-rights-of-persons-with-disabilities.html>

- + The European Charter of Fundamental Rights
https://ec.europa.eu/info/aid-development-cooperation-fundamental-rights/your-rights-eu/eu-charter-fundamental-rights_en

- + 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

- + Directive (EU) 2019/1158 on work-life balance
<https://eur-lex.europa.eu/legal-content/EN/TXT/?uri=celex%3A32019L1158>

- + EUCERD Recommendations on Rare Disease European Reference Networks
https://ec.europa.eu/health/sites/health/files/ern/docs/eucerd_rd_ern_en_0.pdf

- + The Horizontal Equal Treatment Directive
<https://eur-lex.europa.eu/legal-content/en/TXT/?uri=CELEX%3A52008PC0426>

- + Council Directive 2000/78/EC of 27 November 2000 establishing a general framework for equal treatment in employment and occupation
<https://eur-lex.europa.eu/legal-content/EN/TXT/?uri=celex%3A32000L0078>



PARTNERSHIPS WITH PATIENTS



It is now well recognised and demonstrated in the field of rare diseases that people living with a disease and their carers are experts on the diseases that affect them and have a valuable contribution to make to shaping meaningful RD research, policies and services. 'Patient partnership' can be defined as a mutual relationship between all these stakeholders including patients where input from

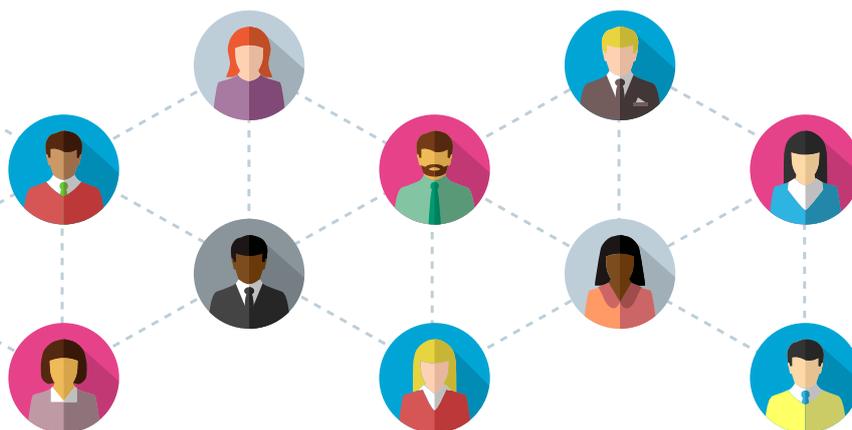
people living with a rare disease or caring for someone with a rare disease routinely and formally informs policy reflections and decisions. Patient partnership implies going beyond empowerment and engagement but considering people living with a rare disease and their advocates as equal partners and actors in policy and programme design and evaluation.



VISION OF THE RARE DISEASE COMMUNITY

The rare disease community recognises that, out of necessity, people living with a rare disease are often the most motivated stakeholders to make progress on their disease when not only the number of patients living with the disease is low but also knowledge, expertise and funding available are lacking to do more. The rare disease community also recognises that working on both the problems and solutions for rare diseases as a partnership between professionals and the communities they serve, better address actual patient-needs and ultimately leads to better outcomes for all. People living with a rare disease are part of collective and responsible

solutions to address their needs and their empowerment creates value for all. The knowledge and experience of people living with a rare disease are valuable assets in shaping future actions and policies and supporting others facing the same challenges. The rare disease community envisions a future in which people living with a rare disease and their representatives can support progress by partnering at the appropriate level of action or policy making and being supported with the relevant information, adequate training and capacity building to achieve this as equal partners.





RARE 2030 **GOAL**

5



All people living with a rare disease and their representatives wishing to improve policies and actions in rare diseases should find their role in an ecosystem which fosters and rewards meaningful patient partnerships, exemplified by an ethos of co-creation that includes but goes beyond mere involvement, engagement, and empowerment people living with a rare disease and their representatives



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 with rare diseases and their representatives may often serve as a partnering link between the two.

Specifically the following actions that foster or enforce the participation of people living with a rare disease and their representatives should be supported at the European, national, regional and local levels:

- + Patient partnerships in research including collection of patient reported outcomes (PROMs) and patient reported experiences (PREMs) to improve disease knowledge and evidence supportive of accelerating development of high quality treatments
- + Communication – each health and social care or research initiative should be responsible and adequately supported to inform and update participants. Health and social care professionals should engage patients as equal partners in the decision regarding their care.
- + Patient partnership and meaningful involvement in multi-stakeholder initiatives, projects and dialogues
- + Support of national and European umbrella and disease specific patient groups to build the capacity through training and empower people living with a rare disease and their representative to support existing initiatives or lead their own, where appropriate
- + Support for new European or global umbrella patient communities, groups or organisations for the rarest diseases



HOW TO ACHIEVE THIS?

AN ECOSYSTEM FOSTERING MEANINGFUL PATIENT PARTNERSHIPS

All stakeholders involved in rare disease diagnostics, prevention, treatment, research, care, and holistic support should contribute to the creation of an ecosystem which fosters and rewards meaningful patient partnerships, exemplified by an ethos of co-creation and exceeding mere involvement, engagement, and even empowerment.

- + Policy makers, physicians, researchers and all other stakeholder groups should place equal value on the work and contributions of rare disease patients and carers, acknowledging the unique services and insights they provide
- + More meaningful and equitable patient partnerships must become the gold standard in all health-related activities, not only in research but in all relevant domains ranging from care delivery to policy making:
 - Concrete indicators should be developed at European -and where relevant, global-level, to measure the success of patient partnerships in the respective activities; in the case of research, these should build on the outputs of the [PARADIGM IMI 2 project](#)
 - Stakeholders should appreciate the absence of a strict 'one size fits all' model for rare disease patient partnerships, and be prepared to adapt approaches as necessary
 - Robust and concrete examples of meaningful patient partnerships in each domain should be disseminated globally
- + National competent authorities should ensure meaningful patient partnerships in the

elaboration, implementation, monitoring and evaluation, updating of national plans and strategies and other relevant policies for rare diseases, or measures for childhood cancers and rare cancers in adults in national cancer plans, and in all activities stipulated therein

- + Policy makers, physicians, researchers, patients, and all other stakeholders should recognise that robust and equitable patient partnerships cannot exist when services, time and expertise are bestowed without remuneration; consequently, they should ensure a fair and transparent system of financial support and compensation (which will simultaneously broaden representation by removing the current de facto requirement for independent financial means)
- + Policy makers, physicians, researchers and all other stakeholders should give particular thought to accessibility when building patient partnerships in rare diseases, considering not only barriers such as language, but also accessibility requirements for those with learning disabilities, hearing and/or sight impairments, etc.
- + The ability of people living with a rare disease (and their carers) to fulfill essential advocacy roles and build patient partnerships is hampered by the disproportionate challenges they face in all walks of life, from psychosocial difficulties to financial, educational and employment-related barriers; therefore, the provision of adequate holistic support for rare disease patients and carers that encompasses social care and adequate social policy measures, should be a priority for national competent authorities seeking to support an ecosystem in which patient partnerships can thrive

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“I would like a professionalisation of patient associations with start-up funds to give oxygen to those who want to employ staff to work better”

Rare Disease Patient



STRATEGIC, COHESIVE AND SUSTAINED SUPPORT

European and national authorities should provide strategic, cohesive and sustained support to perfect and scale-up robust training activities and programmes supporting patient partnerships in the rare disease field

- + European-level training to understand how to form effective patient partnerships in the rare disease domain should be further elaborated and scaled-up, comprising bespoke elements for patients/families/carers, on the one hand, and for researchers and health-related professionals on the other, with opportunities for joint stakeholder training
- + European-level training in 'core skills' for rare disease patients/families/carers should be further elaborated and scaled-up, with an emphasis on building confidence and fostering strategic, diplomatic, and decision-making competences for those aspiring to leadership roles
- + Patient organisations should ensure particular emphasis on engaging and building capacity in the next generation of young patient advocates
- + National authorities should consider endorsing and utilising the training courses and materials provided by groups/initiatives such as the [European Joint Programme for Rare Diseases](#), [EURORDIS Open Academy](#), and [EUPATI](#) - which espouse best practices for rare disease patient partnerships as agreed at the European level - by supporting their implementation and facilitate their access at national level, in the national language
- + Although it is essential to provide specialised training on how to build and sustain meaningful patient partnerships in the rare disease and highly specialised care field, the value of patient partnerships should nonetheless be emphasised by relevant national authorities in more generic training and education programmes for care, research, and policy-related professions at large.

ERNS TO EMBODY ROBUST PATIENT PARTNERSHIPS

The potential for ERNs to embody robust patient partnerships, internally and indirectly by spreading good practices and resources, should be fully realised:

- + European Reference Networks (ERNs) should develop clear and transparent rules for patient engagement, adequately supporting the involvement of patient organisations and their representatives in the different ERN activities and fairly compensate patient representatives
- + Within ERNs, opportunities must be created for patients (not only those attending ERN healthcare centres) to foster robust data partnerships, determine governance, and contribute/extract data to or from appropriate registries, care records and other relevant data sources
- + ERNs should be supported to review and expand their disease-specific membership criteria, in partnership with patients and professional associations, with an emphasis on the necessary multidisciplinary expertise: in this way, EU countries (perhaps even the global RD community) could make use of robust criteria by which to define expertise in given disease areas
- + ERNs should gather and create, in partnership with patient organisations, resources which could support rare disease patients in receiving more integrated and more personalised care in their local environment: such resources should translate to heterogeneous care and social settings, by focusing on clarifying and explaining the (often poorly-understood) needs of patients with complex conditions, and adaptations/approaches which could help
- + ERNs should encourage the development of - and adherence to - codes of conduct and Terms of Reference for ePAGs and patients representing wider communities in ERN-related roles.
- + ERNs should help to promote a culture of shared decision-making in the patient-physician relationship, encouraging professionals in ERN healthcare providers and 'affiliated' centres to discuss all options with regard to the treatments and approaches available, empowering patients to make more informed decisions in partnership with their care team.



TOOLS, RESOURCES AND GOOD PRACTICES

Tools, resources and good practices to develop robust and equitable patient partnerships in rare disease research should be elaborated and implemented

- + The principles of the [EJP RD Short guide on patient partnerships in rare diseases research projects](#) should be noted and implemented by researchers and research funders
- + The [Patient Engagement Toolbox created by the PARADIGM project](#) should be implemented by researchers and research funders, to assess patient capability, avoid conflicts of interest, support patients in managing competing interests, assess fair market value of patient services, amongst other benefits.
- + The [Guiding Principles on Reasonable Agreements between Patient Advocates and Pharmaceutical Companies](#) should be more widely used. This multi-stakeholder initiative aims to make legal agreements between both parties easier and more acceptable while providing adequate protection and rules for both sides. Patient partnerships should span the full research and development pipeline, including the preclinical stage: researchers and research funders must support patients and patient representatives to shape the research agenda, identifying research priorities and knowledge gaps and contributing to call texts, in addition to partnering in research once underway; the Joint Transnational Calls of the European Joint Programme for Rare Diseases exemplify good practices in building patient partnerships in rare diseases research, and can be viewed as a good starting model for improvement
- + Patients and researchers should ensure two-way communication and collaboration to improve patient partnerships in rare disease research: researchers should involve patients in activities such as organisation of conferences, publications and seminars (to help educate and inform other researchers about the added value of patient partnerships), and patient organisations should involve researchers in family days, conferences, newsletters and other appropriate activities
- + To complement training activities designed to build capacity and confidence for patients wishing to participate in research, specialised training should be provided to researchers, funded by research bodies and initiatives, to demonstrate how to engage rare disease patients in the full cycle of research activities and build mutually beneficial partnerships
- + By 2030, following incremental demonstration of the added-value of patient partnerships in the rare disease field, patient organisations should routinely and systematically be considered and included as full partners in any basic, preclinical, clinical, translational, or social research
- + Targets for patient partnerships in research activities and research events should be agreed, based on agreed metrics (building on the outputs of the PARADIGM IMI 2 project)
- + Patient partnerships should be considered a fundamental cornerstone of translational research for rare diseases: evidence of robust patient partnerships should be available as part of the Marketing Authorisation process for all orphan medicinal products
- + The achievements and resources of initiatives such as EFPIA and the EURORDIS Community Advisory Board (CAB) programme, aimed at developing policy principles and codes of conduct to guide Company-Patient interactions, should be sustained
- + Industry, researchers and patient organisations should explore a transparent working model (or models) to provide baseline financial support for rare disease patient organisations to organise and achieve their core goals whilst remaining independent and maintaining credibility



WHAT DO PEOPLE LIVING WITH A RARE DISEASE THINK?

The following figures come from several Rare Barometer surveys carried out in Europe between 2016 and 2021. In these surveys, people living with a rare disease told us their views on a range of issues, such as participation in research, access to treatments, social care needs, the impact of COVID-19 on their lives and their hopes for 2030. Find out more: eurordis.org/voices



PATIENT REPRESENTATIVES WISH TO BE MORE INVOLVED IN THE RESEARCH ECOSYSTEM

9 in 10 are willing to:

- + Help researchers recruit patients for clinical trials and research projects.
- + Disseminate information about research projects.
- + Review research proposals to ensure its feasibility and relevance from a patient's perspective.
- + Being actively involved in research projects as official partners and co-investigators.



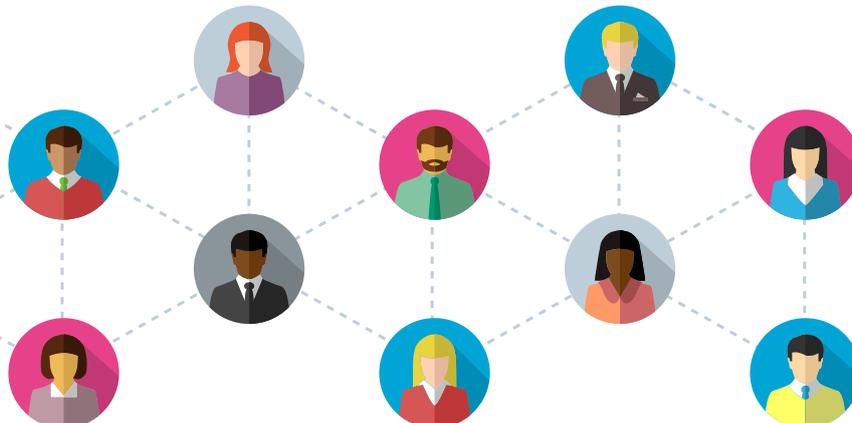
THE MAJORITY OF PATIENT REPRESENTATIVES WANT TO GO BEYOND AND LEAD THEIR OWN PROJECTS

7 in 10 are willing to:

- + Raise funds for research projects in their disease.
- + Lead their own research project.

To be fully involved in rare disease research, patient organisations need to be supported:

- + Financially (8 in 10).
- + With human resources (7 in 10).
- + In terms of Knowledge or abilities (6 in 10).





WHAT DO THE RARE 2030 YOUNG CITIZENS RECOMMEND?

ON COLLABORATION...

The role of patients and 'expert patients' is increasingly being seen as an opportunity to identify current gaps in research and care. Their expertise and experience are currently under- or misused while their potential benefits are broadly acknowledged by both public and private sectors. Young Citizens thus recommend:

- + **Promoting partnership between actors**, mostly between patient organisations and academic institutions through programs to discover and promote new tests and technologies
- + **Stimulating networking to increase awareness while creating/validating/promoting the role of 'expert patients'**, to clearly demonstrate the benefits of collaboration between doctors, industry and patients to increase knowledge and expertise amongst all stakeholders. Such collaborations could be emphasised and regulated through policies to define ethical and legal guidelines ensuring no conflict of interest and stipulating terms of collaboration
- + **Establishing bi-directional specialised department for partnership in both companies and patient organisations**, alongside the definition and justification of the 'expert patient' to guarantee and encourage optimal articulation between public and private sectors

ON EDUCATION AND TRAINING...

Achieving effective collaboration between stakeholders and patients is key but not a given. To become a truly effective tool and approach, education and training are required to guarantee optimal outcomes and process. Young Citizens recommend:

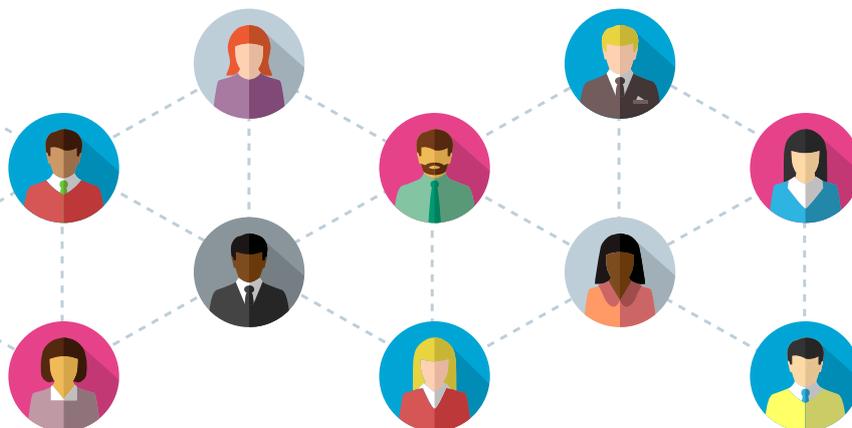
- + **Enhancing education at a younger age**, putting the stress on rare disease in general both at societal level and in academic curriculum, acknowledging the importance of holistic research and care to stimulate understanding and willingness to work collectively.
- + **Developing trainings and educational programs for all stakeholders**, specifically addressing cultural differences and barriers which could hamper collaboration amongst stakeholders





HOW TO MONITOR SUCCESS

- ✓ Monitor the participation of patients in healthcare, research and drug development initiatives at the European and national levels and in public and private sectors
- ✓ Further review of the impact of patient partnership on healthcare, research and development outcomes and describe remaining barriers and facilitators to realising effective partnerships
- ✓ Monitor the percentage of patients or patient representatives with voting rights in decision making committees in legislative regulatory and policy making bodies
- ✓ Monitor inclusion of patient reported outcomes and experiences in standardised national/regional regulatory procedures





SUPPORTING THE SUSTAINABLE DEVELOPMENT GOALS BY 2030

17 PARTNERSHIPS FOR THE GOALS



INCREASE PARTNERSHIPS

As complex diseases, rare diseases require partnership across all sectors and people and their representatives are often best placed to bring together all stakeholders around a common goal that ultimately leads to progress. By achieving genuine partnership with patients in policy making, clinical practice, or research and development the partnerships between governments, the private sector and civil society required to meet this Sustainable Development Goal are also achieved.



REFERENCES

- + PARADIGM IMI 2 project
<https://imi-paradigm.eu/petoolbox/>

- + Patient Engagement Toolbox created by the PARADIGM project
<https://imi-paradigm.eu/>

- + European Joint Programme on Rare Diseases - (EJP RD)
<https://www.ejprarediseases.org/>

- + EJP RD Short guide on patient partnerships in rare diseases research projects
<https://www.ejprarediseases.org/wp-content/uploads/2020/08/SHORT-GUIDE-ON-PATIENT-PARTNERSHIPS-IN-RARE-DISEASE-RESEARCH-PROJECTS.pdf>

- + EURORDIS Open Academy
<https://openacademy.eurordis.org/>

- + European Patients' Academy on Therapeutic Innovation (EUPATI)
<https://eupati.eu/>

- + Guiding Principles on Reasonable Agreements between Patient Advocates and Pharmaceutical Companies
https://wecanadvocate.eu/wp-content/uploads/2019/03/Guiding-Principles_final-document6.2_clean.pdf



INNOVATIVE AND NEEDS-LED RESEARCH AND DEVELOPMENT



Individual researchers and consortia have been studying rare diseases for several decades. However, the rare disease research community has historically been very fragmented mainly due to the large heterogeneity of rare diseases despite common barriers – most particularly the low numbers of people living with each disease, limited funding for research in each disease and a lack of concerted ef-

forts to organise research and thus reach the requisite threshold for progress. Due to the multisystemic nature of many rare diseases a coordination between basic, clinical, translational and social research is key including the study of the impact of policies and programmes on the populations they are meant to serve through public health and health economic studies.



VISION OF THE RARE DISEASE COMMUNITY

The rare disease community envisages a dramatic change in the research and development ecosystem in order for quicker and more pervasive progress to be made for all people living with a rare disease. The community aspires that by 2030 knowledge and resources will be more plentiful, and currently unmet needs will be better addressed by efficient investments and alignment of strategies in rare disease research.

Overarching policy making consortia will foster innovative solutions for ecosystem changes, by advancing needs-led research which will result in demonstrable improvements. Real world evidence of research impact will be readily available - facilitated by a mature European data ecosystem - illustrating how the fruits of rare disease research have delivered concrete benefits.





RARE 2030 **GOAL**

6



People living with any rare disease will benefit from faster development and safer use of health and well-being innovations in terms of curative or symptomatic treatments, improved organisation and management of care and holistic support.



RARE 2030
RECOMMENDATION

6

INNOVATIVE AND NEEDS-LED RESEARCH AND DEVELOPMENT

Basic, clinical, social and translational research on rare diseases should be maintained 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.

Specifically:

- + European and national stakeholders must optimise the use of limited resources through strategic investments and incentivising the sharing of data and other research assets, particularly through the upcoming Horizon Europe Partnership on Rare Diseases
- + European and national stakeholders need to ensure greater incentives for all stages of research, to optimise competitiveness and excellence in basic, clinical, translational and social research
- + European, national and international stakeholders must invest in pre-competitive infrastructures to advance needs-led research, fostering the research capability of European Reference Networks and building bridges with Clinical Research Networks for rare diseases
- + Long-term multinational public-private research partnerships should be enhanced
- + International research collaboration through IRDIRC and other international consortiums should be promoted and sustained



HOW TO ACHIEVE THIS?

GREATER PRIORITISATION AND STRATEGIC SUPPORT FOR RARE DISEASE RESEARCH

AT THE EUROPEAN AND GLOBAL LEVEL:

- + European cross-sectoral partnerships in rare disease research should be sustained, particularly under Horizon Europe, to ensure continuity for the European Joint Programme for Rare Diseases.
 - + More streamlined collaborations should be ensured between European Commission Directorate Generals (particularly RTD, CNECT and SANTE) as regards rare disease research, to reduce bureaucracy and continue to strategically align rare disease funding programmes, avoiding duplication
 - + Strategic approaches employed in rare disease research should be considered a model and/or use case for broader health and research domains
 - + Resources should be designated to foster research and development in very rare and disregarded conditions which lack therapeutic options: the benefits of new incentives for this group of diseases (ideally with a global reach) should be explored.
 - + Funding bodies world-wide should develop globally-reaching research opportunities for the rarer diseases, with dedicated resources
 - + More investments, prioritization and incentives should be ensured for basic and clinical research in areas where these are lacking - research funders must address the significant gap in basic research and discovery science for rare diseases, and simultaneously build more bridges to translate innovative and promising research from bench to the clinic and back.
 - + The merits of designating a European body to identify and elucidate the unmet needs of disregarded rare disease groups should be explored, as part of the mission to address the research and therapy development gaps for all conditions
- + European and global research programmes and funding bodies should ensure better accountability and coordination of current funding to minimise waste and avoid duplication of efforts
 - + A robust regulatory science agenda (building on the existing EMA agenda) should be developed and financially supported at European level, with particular attention to the specificities of rare diseases, emerging technologies and advanced therapies.
 - + Research pertaining to communities with natural synergies to rare diseases must be conducted collaboratively: in particular, cross-talk and collaboration in the paediatric sphere must be ensured between European Joint Programme for Rare Diseases and Conect4Children, and all relevant future organisations
 - + Initiatives and grants supporting trans-national research collaborations for rare diseases must continue to strengthen incentives for the newer EU Member States (EU 13)
 - + Investments into public private partnerships operating in the pre-competitive space should be increased, with greater coordination and collaboration between funding sources and across sectors, and with particular attention to tech-intensive and other advanced approaches
 - + Repurposing of therapies for rare diseases should be supported at the transnational level, as a strategic priority

IN ADDITION, THE FOLLOWING RECOMMENDATIONS ARE PROPOSED TO ENSURE GREATER STRATEGIC SUPPORT AND PRIORITISATION FOR RARE DISEASE RESEARCH AT THE NATIONAL LEVEL:

- + Countries should take all necessary steps to meet the [International Rare Disease Research Consortium \(IRDIRC\)](#) Goals and implement the recommendations issued from IRDiRC Task Forces.



- + National authorities should ensure that national plans and strategies for rare diseases as well as national cancer control plans for rare cancers – which should be evaluated and renewed, if time-bound - include specific goals and plans to facilitate research, and should address the following:
 - Future plans and strategies should highlight the services available to researchers in-country through the ESFRI (European Strategic Forum of Research Infrastructures) Infrastructures and through the help-desk of the European Joint Programme for Rare Disease Research, and should provide guidance on how to access these respective services
 - Dedicated funding and/or a plan of tax incentives should ideally be stipulated in the plan/strategy, to facilitate rare disease/rare cancer research either in-country or on a trans-national basis (or both), and should be proportionate to the size and situation of the country
 - National Mirror Boards for rare disease research (that also include rare cancers) should be created, to ensure a bidirectional dialogue with the European Joint Programme for Rare Disease Research and future European Partnerships

“

“Initially I participated [in research] because there were no treatment options for me and I nearly died several times. I started as a child and my parents saw the clinical trials as a way to keep me alive. It worked, I’m still alive!”

Rare Disease Patient

ACCELERATING EXCELLENT SCIENCE IN THE RARE DISEASE DOMAIN, TO MAXIMISE COMPETITIVENESS

Specific support for research that will expedite the discovery of rare disease mechanisms into direct benefits for people living with a rare disease is required.

- + Research funders should support researchers to gain access to existing national, European, and global-level resources, infrastructures and networks, to ensure future research takes note of acknowledged best practices and avoids reinventing wheels: these should include facilitating access to rare disease-relevant services available via the European Strategic Forum of Research Infrastructures (ESFRI) and through the help-desk of the European Joint Programme for Rare Disease Research
- + Research funders should insist upon greater reproducibility of data, for all stages of rare disease research, and should follow leading publications by increasingly assessing the robustness of strategies for data management, interoperability, reproducibility and sharing/linkage when evaluating proposals
- + There should be a requirement to share data (at a minimum metadata) from publicly-funded research, once complete, to inform and streamline future research: patient organisations and Industry should be encouraged to act similarly, whilst respecting intellectual property rights
- + Researchers should be encouraged and incentivised to publish data from ‘failed’ basic or clinical research; companies must publish data from ‘failed’ clinical research, to inform future research
- + Research funders, regulators, and academic/scientific organisations must adopt and promote a new paradigm as regards incentives



and rewards for research into rare diseases: an open and collaborative approach must be incentivised, favouring the publication of research results in a manner that enables discovery rights to the researcher whilst enabling access to the research data as promptly as possible (stepping away from esteem indicators based solely on competitive publications)

- + Greater investments are required, to transform -omics investigations into improved diagnostics, care and treatment knowledge
- + FAIR data stewardship should be available to support individual research projects or clinical trials in preparing relevant data from the outset, for potential secondary use in future - the costs for this should be included in the initial funding proposal, to ensure all results are FAIR-compliant.
- + All public and private stakeholders involved in the therapy development cycle should consider the [IRDIRC Orphan Drug Development Guidebook](#) and utilise the materials and recommendations therein when approaching academic, patient-led, and industrial drug development
- + Facilitate developers in continuing the development of orphan medicines abandoned by other entities for commercial reasons.
- + Embed, in the regulatory landscape, proven approaches to utilizing shared platforms and innovative trial methodologies capable of targeting multiple rare diseases at once and developing therapies for multiple conditions
- + The applicability of AI to enhance myriad types of rare disease research should be ascertained through dedicated projects
- + Investments in all areas of innovation should be guided by large observational research utilising real world evidence, to demonstrate real-world impact of research outputs (including therapies) for patients
- + Initiatives should complement the creation of expert resources to improve rare disease research by providing more accessible, direct, stakeholder-specific training opportunities; in particular, research funders and research bodies should invest in training and mentoring of junior scientists, to facilitate their familiarisation with the rare disease research pipeline and R&D processes
- + Member States should ensure that all clinical trials ethics applications are assessed within

stipulated timelines, to accelerate study start-up, which is essential for research into medical conditions which are severely debilitating and/or life threatening and for which therapeutic options are limited or non-existent.

- + Competent authorities should harmonize the requirements in terms of pre-clinical data and documentary packages for cross-national clinical trials

LINKING CLINICAL CARE TO RESEARCH - OPTIMISING CAPACITY OF EUROPEAN REFERENCE NETWORKS

ERNs must be supported by Member States/EEA countries and at a centralised research support structure at the European Level to fulfil their potential, as key components of a coordinated research ecosystem performing high quality collaborative clinical research that complies with the expected standards required by regulatory and Health Technology Assessment (HTA) bodies

- + European Reference Networks should receive earmarked -and adequate- funding through European programmes to conduct clinical research and trials (involving centres inside or outside of the Networks, as required) and to research neglected topics including rehabilitative, holistic and social research
- + European Reference Networks should be specifically and adequately funded to develop and conduct natural history (and where possible accompanying biomarker) studies, a minimum of 5 every 2 years, to build the knowledge base and capacity for clinical research in disregarded diseases/areas lacking research
- + The Coordination and Support Action funded by the H2020 programme to support the creation of Clinical Research Networks (covering 4 domains: clinical research (including PCOMs); data management; engagement and dissemination; and administrative support) should be supplemented by additional funding to deploy core services to become fully operational by 2025
- + Collaboration between the European Reference Networks and global entities, such as the NIH Clinical Research Networks, should be supported



- + European Reference Networks must collaborate with their relevant scientific and learned societies, in discussing research priorities, in order to build synergies around activities pertaining to rare diseases
- + European Reference Networks' potential to positively impact the development and use of medical devices for rare diseases should be explored
- + European Reference Networks should receive funding to employ research-oriented staff to complement their clinical experts, particularly for HCPs in countries where research capacity-building is most needed
- + Clear rules are required that enable European Reference Networks to collaborate with industry across a range of pre-agreed activities, clarified and tested through pilots, using shared SOPs to accelerate research and build mutually-agreeable public private partnerships: a central business develop-

ment/tech transfer office could promote, coordinate and supervise European Reference Networks interactions and agreements with industrial partners

PLACING PATIENTS AT THE CENTRE OF CLINICAL RESEARCH

Placing people living with and caring for someone with a rare disease at the centre of clinical research, drug development, and evaluation is increasingly recognized as paramount to fully understanding a disease and to identifying meaningful endpoints. Their knowledge, contribution, empowerment, and participation are crucial to increasing the efficiency of such efforts. **Specific recommendations on partnering with advocacy organisations and people living with and caring for someone with a rare disease are elaborated in the section on Patient Partnerships of this document.**

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“I haven't participated in research because I've found it all too far away.”

Rare Disease Patient

SOCIALLY-ORIENTED RESEARCH INTO RARE DISEASES

Both cross-border and national foci are required to prioritise and advance socially-oriented research into rare diseases:

- + The European Commission should increase funding opportunities to assess the true impact (clinical, social, personal, and financial) of rare diseases through collaborative research
- + The European Commission should support proof of concept studies to demonstrate how preventative, integrated care can result not only in better quality of life but also in economic savings
- + The European Commission should support research to assess and publicise the respective levels of functioning and disability associated with rare diseases, through a publically-available database accessible for all (for instance through expansion of the Orphanet Disability Project or similar)

Additional Recommendations to improve integrated and person-centred care for people with rare diseases can be found in the chapter on Integrated and Person-Centered Care.

BRIDGING THE RESEARCH AND DEVELOPMENT DIVIDE FOR RARE DISEASES

Concrete actions and strategic directions are required in order to optimise the regulatory pathway for would-be therapies and devices for people with rare diseases. Therapy development, which is a cornerstone of rare disease research, should take place within a cohesive, multistakeholder ecosystem. Recommendations to steer the European rare disease community in this direction are elucidated in the chapter on Available, Accessible and Affordable Treatments.



WHAT DO PEOPLE LIVING WITH A RARE DISEASE THINK?

The following figures come from several Rare Barometer surveys carried out in Europe between 2016 and 2021. In these surveys, people living with a rare disease told us their views on a range of issues, such as participation in research, access to treatments, social care needs, the impact of COVID-19 on their lives and their hopes for 2030. Find out more: eurordis.org/voices



IT IS URGENT TO INCREASE RESEARCH TO FASTER THE DEVELOPMENT OF INNOVATIONS

- + **18%** of people with rare diseases have participated in research in their life to develop treatment and therapies. This lack of research is mainly due, according to patients and carers, to the lack of public and private funding and to the small patient population.
- + Despite all that, research on treatments and therapies remains the top priority for people living with a rare disease as they believe it is the main factor to improve their care and ease their symptoms.
- + People living with a rare disease count on research because they believe there is hope for them to: have their rare disease stabilised within 10 years (53%), to better manage the symptoms (50%) or to cure their disease (19%) within 10 years.



CARE MANAGEMENT AND HOLISTIC SUPPORT ARE HIGHLY VALUED BY PEOPLE WITH RARE DISEASES

- + Patients' unmet needs are not limited to treatments and therapies. The actual clinical experience (coordination between healthcare professionals, the access to specialists...), along with the social recognition of the disease and the psychological impact are highly valued by patients and carers. Yet, only 15% have participated in research on quality of life.
- + People have hopes and realistically expect to better manage the psychological and emotional aspects of the disease (58%) within 10 years or that they will not be discriminated against (33%) by 2030.





WHAT DO THE RARE 2030 YOUNG CITIZENS RECOMMEND?

ON COLLABORATION...

The importance of collaboration in research is crucial as expertise and means tend to be scattered throughout Europe. All relevant actors, from patients to the industry, have the potential to greatly contribute to future research and Young Citizens therefore recommend:

- + **Improving cross-country collaboration** to gather means and knowledge beyond national borders and fostering the sharing of best practices
- + **Creating and validating the role of “expert patients”** to officialise their role and job across sectors contributing to rare diseases, which would comply with ethical and legal guidelines to ensure no conflict of interest and be formally acknowledged and paid.
- + **Improving articulation between public and private sectors** to guarantee optimal collaboration between actors

ON INNOVATION...

Innovation is at the centre and basis of potential progresses in research. Young citizens consider that future policies should be aimed at gathering all relevant stakeholders to guarantee that the most innovative techniques are the main subject of collaboration and investment. They recommend:

- + **Establishing patient-centricity as the standard model of research and development in Europe** to ensure patients' needs are indeed the drive for future innovations
- + **Improving assessment methods** to put the stress on identifying gaps that remain in terms of person-centred care and enhancing the use of innovative tools that effectively highlight where these gaps currently are and thus driving research



HOW TO MONITOR SUCCESS

- ✓ Continue to inventory and monitor research activities in the field of rare diseases through Orphanet
- ✓ Ensure rare disease research is specifically embedded in the monitoring of EU policies and activities in European research as conducted by the European Commission's Joint Research Centre
- ✓ Support additional European and global committees (such as the NGO Committee for Rare Diseases and the International Rare Disease Research Consortium) in their potential to monitor rare disease research, identify unmet needs, align research strategies and priorities and develop roadmaps.
- ✓ Implement monitoring systems of public funding for R&D on orphan treatments and its impact on marketing authorization and availability to patients to better define the future research agenda. IRDiRC has become the forum where that strategic research agenda could be defined.





SUPPORTING THE SUSTAINABLE DEVELOPMENT GOALS BY 2030

3 GOOD HEALTH AND WELL-BEING



IMPROVING HEALTH AND WELL-BEING

Increased research and development of the highest quality of care and treatments for people with rare diseases helps improve the health and wellbeing of this vulnerable population. Strategic investment into research today can also help the generations of tomorrow as many rare diseases are inherited. Researching rare diseases can also lead to the discovery of strategies and solutions for more common diseases that may have a close connection from the biological, sociological, public health or other disciplinary point of view.

9 INDUSTRY, INNOVATION AND INFRASTRUCTURE



BUILD RESILIENT RESEARCH INFRASTRUCTURES AND FOSTER INNOVATION

By supporting networks, platforms and repositories of data, knowledge, tools and activities in rare disease research (e.g. biobanks, registries, clinical research infrastructures, bioinformatics networks, etc.) information and technologies are made available to all benefit the needs of all stakeholders in the rare disease community also building resilient infrastructures and fostering innovation in broader areas of health and research.

17 PARTNERSHIPS FOR THE GOALS



INCREASE PARTNERSHIPS

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 in a multis-takeholder approach the rare disease research community can boost the role of science technology and innovation in the achievement of the 2030 sustainable development goals.



REFERENCES

- + European Joint Programme for Rare Diseases
<https://www.ejprarediseases.org/>

- + International Rare Disease Research Consortium (IRDIRC) Goals
<https://irdirc.org/about-us/vision-goals/>

- + IRDIRC Orphan Drug Development Guidebook
<https://irdirc.org/orphan-drug-development-guidebook-materials/>



OPTIMISING DATA FOR PATIENT AND SOCIETAL BENEFIT



RARE 2030 **GOAL**

7



All data associated with rare diseases is properly coded using the Orphanet nomenclature, including all people with rare diseases that have encountered a centre of expertise having this code as part of their electronic health records

All data of relevance to rare diseases (including quality of life, health economics, public health, natural history, genomics) is federated within national systems, and is Findable, Accessible, Interoperable and Reusable (FAIR compliant)

The majority of people living with a rare disease have their data integrated to a federated European (and where possible, global) health and research data ecosystem



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 with rare diseases and their families.

Specifically, the semantic, technical, legal, financial and political frameworks at the European and national levels and the knowledge, skills and political will to use and re-use existing data and collect new (including real-world data) should be enhanced in order to:

- + advance diagnostic capability, research and development of effective treatments and health and social care solutions
- + monitor the public health impact of rare diseases
- + ensure that any investments in rare diseases respond to unmet medical needs (those driven by the needs of people living with a rare disease all along their patient journey and in recognition of the life cycle of development of care solutions, treatments and technologies)
- + encourage multistakeholder dialogue and collaboration to test and evaluate newly available solutions and values around data collection and use (e.g. the applicability of big data, artificial intelligence, the commercialization of patient data and data donation)
- + integrate education around data collection, use and sharing to build understanding and encourage a data-sharing friendly culture on all levels and develop relevant skills across all stakeholders
- + facilitate data collection and use at the international level by promoting adoption of internationally recognised best practices and standards





HOW TO ACHIEVE THIS?

INTEGRATED AND STRATEGIC EUROPEAN DATA FRAMEWORK

An integrated and strategic European framework for the capture, use, and reuse of data of relevance to rare diseases must be elaborated and adopted, to unlock the potential of rare disease data for health-related and research purposes (which in rare diseases are often intertwined)

- + The rare disease field must become a central component of the European Health Data Space, as well as research-oriented initiatives such as the EU Open Science Cloud, to support and accelerate **FAIR** -compliant data-sharing
 - + National authorities should - with support from the European level - implement integrated electronic health record (EHR) systems capable of capturing data on rare disease patients at each healthcare encounter, utilizing the Orphanet nomenclature (ORPHAcodes) to ensure visibility of patients within national health and social systems, thus building a robust and accurate longitudinal care record
 - + Optimal strategies for mining unstructured or differently-structured data (for instance built upon different syntactic and semantic standards) should be identified, to make best use of the myriad of data sources available to theoretically inform health and research for rare diseases
 - + The role of the biopharmaceutical industry in an overarching rare disease data framework must be established, as part of an ecosystem involving the European Commission, Member States/EEA authorities, ERNs, patients, the EMA, and all other relevant actors, to ensure ethical and effective public private partnerships centred around data
 - + Privacy Preserving Record Linkages or other solutions to federate and link rare disease data in line with GDPR should be agreed with the support of legal, IT and technical experts, and thence be promoted by European and national authorities to support the use of such solutions
 - + Workable governance frameworks and guidance should be elaborated to ensure that data remain able to support rare disease health and research goals under the GDPR.
- + Consensus should be developed at the European - and ideally global - level, to identify and agree the most appropriate standards and ontologies for all types of data, addressing not only diagnoses and phenotypes but also treatments, quality of life, and more: these standards should be henceforth used for public and private data generated at source, including clinical (health and social sector) and research level (including registries and data repositories) data
 - + European and national authorities should promote the implementation of FAIR (Findable, Accessible, Interoperable and Reusable) data principles, particularly for rare disease data: they should:
 - Provide incentives which favour data sharing or at least shareability, with a standard requirement to share data from publicly-funded research - such as placebo data and data from failed trials - to inform and streamline future research: companies should be encouraged to act similarly, whilst respecting IPR
 - Financially support the GO-FAIR (Findable, Accessible, Interoperable, Reusable) Implementation Network for Rare Diseases, or equivalent body, to provide strategic community advice on FAIRification of any and all types of data of relevance to rare diseases
 - Invest in training expert data stewards able to advise stakeholders in the national territory on FAIR-compliant data management and to support individual research projects or clinical trials in preparing relevant data from the outset, for potential secondary use in future, capitalizing on the experienced achieved by the European Joint Programme for Rare Diseases
 - + All disease and specialist communities - centred on or in collaboration with the ERNs - should be encouraged and supported to develop meaningful datasets and data dictionaries, for health and research purposes, based upon suitable international nomenclatures for particular types of data, and these must be made publically available to support reuse of assets and greater data interoperability on a global scale



- + Specific projects/funding should be initiated, involving patients, healthcare professionals, researchers and regulatory authorities, to strategically define and agree patient-centred outcome measures for rare disease and specialised care communities, using the hierarchies of the ERNs as a basis
- + Sustainable funding must be secured to ensure continued improvement and curation of the Orphanet nomenclature and associated cross-harmonisation of terminology, in line with the EUCERD Recommendations on Ways to Improve Codification for Rare Diseases in Health Information Systems

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“I agree to share my data if the rare disease community gets benefit from it”

Rare Disease Patient

MULTI-STAKEHOLDER DIALOGUE FOR RARE DISEASES REGISTRIES AND REPOSITORIES

A renewed multistakeholder dialogue is required, at the regional, national, European and global levels, to ensure a more strategic approach to the creation and connectivity of rare diseases registries and data repositories, at all levels.

- + The [EUCERD Recommendations on Rare Disease Patient Registration](#) remain robust and very valuable: they should be promoted by national authorities and all rare disease registries should strive to implement them
- + An appropriate forum should be created/designated at the European level to ensure multidisciplinary and strategic ‘oversight’ of the topic of rare disease registration, in its broadest sense, and should be open to all stakeholders (including ERN representatives, European Platform on Rare Diseases Registration, European Commission, national policy-makers and/or national rare disease registry owners, patients, regulators, and Companies: this forum should:
 - advance discussions on the optimal ways to develop or orientate existing and/or future national and regional registries for (all) rare diseases, with flexibility to support working groups between countries facing similar challenges in view of size, geography, or other relevant characteristics
 - be supported to clarify the different types of rare disease registries, the possible functions and added-value each can bring, and the kind of data collection or access is required for particular purposes, to support a more strategic future for rare disease registration in Europe
- support the sharing of best practices, to propose solutions for future national plans and strategies for rare diseases to advance or initiative rare disease registration, considering the wider European or global context as appropriate
- consider mechanisms to incorporate direct patient-reported data to registries, and enable access to a patient’s own data
- propose and assess mechanisms to make EHRs and registries/other data collections interoperable, to foster the reuse of health data for secondary purposes.
- + The European Platform on Rare Disease Registration should provide guidance and assistance to current or prospective registries in Europe (whether established by ERNs or not) which register with the Directory of Registries, supporting them to contribute to and share in the broader registry data ecosystem in collaboration with the European Joint Programme for Rare Diseases
- + A dedicated body, building on the achievements of the European Platform on Rare Disease Registration and the European Joint Programme for Rare Diseases, should be able to advise any (current or prospective) registry owner/curator as to what the GDPR means in reality for registries and data collection/sharing.





- + The EMA should provide more strategic scientific advice at early stages to companies developing therapies in the same space, directing them toward existing disease registries wherever possible (via collaboration with the ERDRI Directory of Registries), and providing impartial support for public private partnerships for rare diseases which meet the needs of all actors involved
- + Decisions on new and renewed European funding for rare disease registries should be made following cross-DG, EMA and ERDRI input and advice from a dedicated EU-level forum on rare disease registration, as above, to enhance strategic alignment and reduce duplication

ERNS TO CONSOLIDATE RESEARCH DATA ECOSYSTEM

The unique potential of European Reference Networks to consolidate and streamline a European health and research data ecosystem for rare diseases and highly specialised healthcare must be realized through tangible actions:

- + The future ERN health data strategy must be anchored to the wider European health data and IT ecosystem, driven by a concerted policy action of all the relevant DGs and aligned with national health data strategies from the majority of MS/EEA countries; in this context, ERNs should help to shape the future Health Code of Conduct for secondary use of data (addressing the need to make GDPR research-friendly)
- + The future ERN data strategy must be targeted towards all rare disease patients in Europe, and not only those attending ERN HCPs: opportunities must be created for patients to foster robust data partnerships, determine governance, and contribute/extract data to or from appropriate registries, care records and other relevant data sources.
- + ERNs should be financially supported to co-create (together with the European Joint Programme for Rare Diseases) a comprehensive data strategy and implementation plan by 2023, envisaging the necessary activities across 6 action lines: architecture - cloud computing services and IT support for registries and other databases; data collection protocols; data curation services; data management tools (services and tools to search, access and share data, tools to manage own data); data analytics tools and services; and a data governance framework.
- + Data from hospital EHR (electronic health record) systems should be interoperable with the ERNs' Clinical Patient Management System (CPMS), and with ERNs' new epidemiological registries, allowing minimal data entry and maximum automation (with accompanying quality assurance) - all such systems should be aligned with the European Health Data Space.
- + Disease-specific registries (where positively evaluated by ERNs based on rigorous criteria OR created anew by ERNs in future) should be interoperable with the new ERN registries and any robust national RD registries: these should all be connected (sustainably) to ERDRI and the European Joint Programme for Rare Diseases Virtual Platform to provide a fully functioning registration ecosystem
- + ERNs should sit at the centre of all future efforts to refine and evolve all ontologies and standards for data collection and utilisation into a common data model, including efforts to facilitate extraction and mining from real-world data



WHAT DO PEOPLE LIVING WITH A RARE DISEASE THINK?

The following figures come from several Rare Barometer surveys carried out in Europe between 2016 and 2021. In these surveys, people living with a rare disease told us their views on a range of issues, such as participation in research, access to treatments, social care needs, the impact of COVID-19 on their lives and their hopes for 2030. Find out more: eurordis.org/voices

SHARE RELEVANT AND ELIGIBLE DATA

97% of people living with a rare disease are willing to share their health data to foster research on their own disease.

95% are also willing to share their data to improve research on diseases other than theirs.

80% want to keep control over their data to avoid their information being shared with 3rd parties without their consent or being used in a different context from the one they disclosed or being victims of discrimination. This is a direct call to develop a legal framework for data ownership and protection.

SUPPORT ACCURATE ASSESSMENTS OF PATIENT'S NEEDS

96% of patient's representatives agree that patient organisations should participate in reviewing research proposals to ensure feasibility and relevance of the study from a patient's perspective.

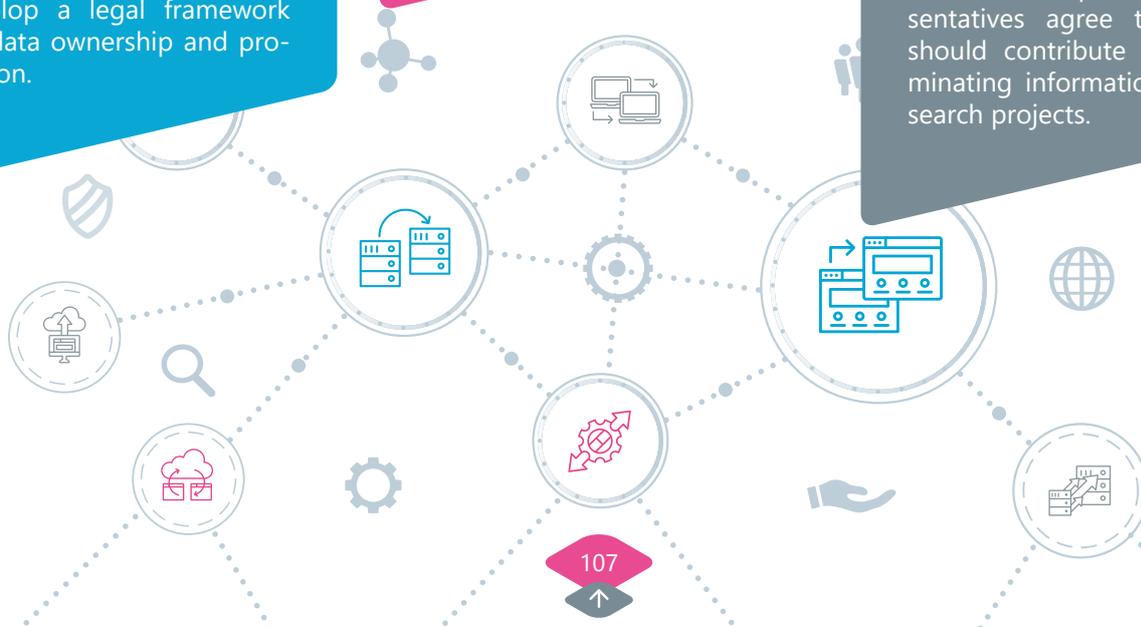
Patient's top 3 most unmet needs that need to be filled in 2030 are: finding treatments and therapies, a better coordination between all healthcare professionals, having access to specialists in their disease.

MEASURE THE IMPACT OF CURRENT SOLUTIONS IN RARE DISEASE PEOPLE'S LIVES

37% only of people living with a rare disease have participated in medical research which aims at understanding rare disease treatment effects, improvements in patient's quality of life, efficacy of medical devices, etc.

95% of patient representatives believe they have a key role to play in helping to recruit patients to participate in clinical research.

99% of patient representatives agree that they should contribute to disseminating information on research projects.





WHAT DO THE RARE 2030 YOUNG CITIZENS RECOMMEND?

ON COLLABORATION...

As knowledge on rare diseases are limited as compared to other disease areas, Young Citizens strongly support increased collaboration on data-related topics to make better use of data and resources. They recommend:

- + **Improving cross-country collaboration**, to guarantee not only the sharing of best practices throughout Europe but also for the collection, storage and use of data in a unified interoperable way that would greatly benefit research
- + **Developing a legal framework for data ownership and protection**, as this constantly evolving and increasingly sophisticated technological era brings challenges related to data safety and ownership, alongside opportunities for better use of data in health and well-being solutions

ON INNOVATION...

A better collection and use of data across Europe and sectors could also have great benefits in expanding research horizons, especially when putting the patient at the very centre of research. Young Citizens recommend:

- + **Establishing patient-centricity as the standard model of research and development in Europe**, to ensure that existing and upcoming data and knowledge are best used in the interest of those it benefits

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“We are only 6 families in [my country] affected by this. If we don't offer our database, I think it's impossible for someone to help us, to know much about us”

Rare Disease Patient





HOW TO MONITOR SUCCESS

- ✓ European Platform on Rare Diseases Registration
- ✓ European Reference Networks
- ✓ Orphanet database and reports (biobanks, infrastructures, registries, research projects etc.)
- ✓ State of the Art of Rare Diseases Resources
- ✓ EU Health Data Space



SUPPORTING THE SUSTAINABLE DEVELOPMENT GOALS BY 2030

17 PARTNERSHIPS FOR THE GOALS



INCREASE PARTNERSHIPS

As complex diseases, rare diseases require multidisciplinary care and thus multiple sources of data to better understand and address them from the clinical, psychosocial and economic point of view. To reach the critical mass of data required to best understand and develop solutions for people with rare diseases, data must be integrated from a variety of sources across countries, institutions and sectors.



REFERENCES

- + FAIR Initiative
<https://www.go-fair.org/>

- + EUCERD Recommendations on Ways to Improve Codification for Rare Diseases in Health Information Systems
https://ec.europa.eu/health/sites/health/files/rare_diseases/docs/recommendation_coding_cer-grd_en.pdf

- + EUCERD Recommendations on Rare Disease Patient Registration
http://www.eucerd.eu/?page_id=13

- + Rare Barometer Survey - Share and protect our health data: Rare disease patients' preferences on data sharing and protection.
http://download2.eurordis.org.s3.amazonaws.com/rbv/dataprotectionsharing/EN_RB_Data%20Survey%20Infographics_Complete.pdf

- + Rare Barometer Survey - Rare disease patients' participation in research.
http://download2.eurordis.org.s3.amazonaws.com/rbv/2018_02_12_rdd-research-survey-analysis.pdf



AVAILABLE, ACCESSIBLE AND AFFORDABLE TREATMENTS



In today's world, science and technology offer an unprecedented chance to address the unmet medical needs of people living with a rare disease. This potential is currently not translated into actual health benefits for the large majority of people living with a rare disease

due to issues concerning availability, accessibility and affordability of treatments. Coordinated, more strategic policies are required to address these shortcomings and inequalities and ultimately improve the health outcomes and quality of life of people living with a rare disease.

VISION OF THE RARE DISEASE COMMUNITY

The rare disease community aspires to a research, development and delivery ecosystem for rare disease therapies in Europe in which efforts at the local, regional, national and international levels remain concerted for success. This ecosystem must be co-designed by both public and private sectors. Although the community strives to remain united to overcome its common challenges, tailored approaches to care, research and treatment may be required for diseases which have historically been neglected

and/or for whom curative treatments have not yet materialised.

No matter what the disease area, the rare disease community will work towards the faster uptake of new technologies and the decision making processes will be adapted in order to make treatments available, accessible and affordable for all rare disease patients, no matter where they live.





RARE 2030 **GOAL**



More and better quality curative, stabilising, palliative, assistive, rehabilitative and preventative technologies and therapies are available, accessible AND affordable for all people living with a rare disease in Europe

Europe is a world leader in the development of rare disease therapies with a competitive regulatory ecosystem and a more robust pharma and biotech manufacturing presence, leading to greater investments in research and product development, with accompanying improvements in patient access and health monitoring

*1000 new therapies should be available by 2030,
in line with the [IRDiRC vision](#)*

- + Treatments should be approved in the EU for 500 different rare diseases and for 50% of the overall population of people living with a rare disease.
- + These new treatments and technologies should focus on unmet needs with two goals:
 - Curative, transformative or stabilising and symptomatic treatments for 200 of the 400 most frequent rare diseases covering over 90% of the population living with a rare disease;
 - Curative or transformative treatments for at least 100 rare diseases from the group affecting less than one in 100,000

Therapies should be 3 to 5 times more affordable than currently available treatments



RARE 2030
RECOMMENDATION



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.

Specifically, policies should enforce:

- + early-stage multistakeholder identification of unmet needs and subsequent priorities and investments
- + a threshold of eligibility: including prevalence of no more than 5/10,000 individuals (an incidence for rare cancers of less than 6 per 100 000 per year) and avoids artificial breakdown of common diseases into rare subsets
- + a graduated system of incentives, rewarding earliest dialogue and favouring areas with no therapeutic options (currently disregarded diseases)
- + a strengthened mandate for the European Medicines Agency (EMA) Committee for Orphan Medicinal Products encompassing early dialogue, designation, ongoing scientific advice and protocol assistance, scientific qualifications (in particular of registries and patient-centred outcome measures (PCOM)), risk-benefit assessment, and post-marketing requirements
- + a functional and efficient EU Health Technology Assessment (HTA) Framework to support the assessment of effectiveness and relative effectiveness (and, in the interim, incentivisation of joint EMA/HTA assessment at the European level and uptake at the national level)
- + a continuum of comparative evidence generation throughout the product life cycle and patient journey, enabled via multi-purpose disease registries and all other relevant data sources
- + a European Table of Pricing and Negotiation enabling European collaboration between Member States
- + an EU-Fund to co-finance the generation of evidence across EU Member States and reduce uncertainties during the first years following approval, for advanced therapies for the rarest diseases (affecting less than 1/100 000.)



HOW TO ACHIEVE THIS?

REVISING CURRENT REGULATORY FRAMEWORKS ON ORPHAN MEDICINAL PRODUCTS

In general:

Pan-European – and ideally global – action is necessary, to improve a currently unsustainable and inequitable status quo regarding the availability and accessibility of orphan medicinal products

- + Existing resources to address the current challenges should be implemented, by national and European-level authorities, including the [EUCERD Recommendations on the CA-VOMP Information Flow](#) and the recommendations in [Breaking the Access Deadlock](#)
- + [IRDiRC Recognized Resources](#) and recommendations to improve accessibility and availability should be utilised at national and regional level, to encourage cross-country and ideally global action necessary for a paradigm shift
- + EU Member States and all relevant EU authorities should revisit the actions defined in the [Commission Communication](#) and [Council Recommendation](#) concerning orphan medicinal product accessibility and availability, where these have not been addressed
- + The positive aspects of the current EU regulatory framework governing pharmaceuticals for orphan diseases should be sustained, to continue to incentivise investments, whilst simultaneously increasing the robustness and transparency of the ecosystem
- + Regulators and competent authorities must ensure that adaptive pathways and rapid access mechanisms continue to bring medicines to patients who need them, providing essential safety and efficacy considerations are met.
- + Voluntary and early dialogue between stakeholders and countries to collaborate on coordinated access should be continued via initiatives such as the [Mechanism of Coordinated Access to Orphan Medicinal Products \(MoCA\)](#).

- + Orphan Medicinal Product developers should consider both the individual value of products for patients but also the wider societal value, weighing the burden of lack of treatment against investments
- + Orphan medicinal products should be developed, launched, and monitored within a continuum of comparative evidence generation spanning the whole product lifecycle and patient journey, enabled by a more strategic and standards-based approach to data sharing and federation centred on multi-purpose disease registries and all other relevant data sources: the application of agreed standards to increase the FAIR-ness of relevant data sources (including adequate codification of health records, to support a health innovation ecosystem) will be essential

To improve availability:

- + National, European and global authorities should ensure that the development of future therapies for rare diseases is not hampered by an increasing Industry focus on distinct subgroups and mutations of otherwise common conditions under the growing trend for precision medicine
- + Therapy developers - whether from the private, public or civil society sectors - should be encouraged to utilise expert rare disease resources and guidance to optimise the development and launch of orphan medicinal products (e.g. [IRDiRC Orphan Drug Development Guidebook \(ODDG\)](#))
- + A more coherent strategy should be agreed at European level to unite relevant actors in repurposing of medicines for rare diseases, building on the work of the [STAMP expert group](#), IRDiRC and others
- + Regulators should ensure real-time publication of information concerning the status quo of available medicinal products and products in development for rare diseases, to expedite the decision-making process for therapy approvals



To improve access:

- + National HTA bodies should ensure transparency of the decision-making process and criteria regarding orphan medicinal products
- + Post-marketing HTA decisions and reports for orphan medicinal products should not take place on a country-by-country basis, but at a pan-European (and sometimes, ultimately, global) level
- + In the case of advanced therapies for the rarest diseases (those affecting fewer than 1 /100,000), an EU-Fund should be established to co-finance the generation of post marketing authorisation evidence across EU Member States during the years initially following approval, in order to reduce uncertainties
- + A dedicated body to facilitate EU collaboration in HTA should be established, via an EU Regulation (binding for all), but failing this on a voluntary basis as soon as possible, involving as many countries as are willing to collaborate to further the interests of their citizens and share data and assessments on the HTA of orphan medicinal products
- + Actions must be agreed, at European level, to optimise the ecosystem concerning pricing of orphan medicinal products, in order to bring medicines to those who need them and reduce bureaucracy and delay in launching products at national level, whilst ensuring profitability of the rare disease market for companies:
 - Companies should be encouraged to adopt a more open and transparent approach to publicising development costs – without jeopardising core business models - in order to support pricing decisions, enabling a reasonable return on investment which supports profit-making for the private sector but does not debar patients from actually accessing therapies at national level
 - A continuous and value-based approach to pricing should be implemented, involving all stakeholders and sitting within a continuum of dialogue and evidence-generation initiated as early as possible in the developmental pipeline: a robust data ecosystem should support a move towards performance-based pricing in which products which do not show long-term benefits may be removed from market but those performing strongly may warrant their launch prices (recognising that repurposed therapies which have already recovered significant R&D costs may require different consideration)
 - In conjunction with the activities recommended for MoCA above, the EMA and payers should utilise early dialogue/conversations to provide recommendations on - initially - the basic price range which could be considered acceptable for certain types of orphan medicines
 - A workable system should be developed at European level to economically regulate the relationship between public buyers and companies, via a European Table on pricing and negotiations: this is particularly urgent in order to ensure access to advanced therapies such as gene therapies
 - A European-level pilot should support more global discussions here, which are in fact essential to improve access to medicines and therapies for very rare conditions for all patients who need them, leaving nobody behind: the momentum created by the COVID-19 crisis should be leveraged, as an example of feasibility and collective strength of cross-country negotiations and collaborations for the greater good of citizens

“

“I would like to see greater transparency in drug prices and availability. The EU should follow the pricing policies and practices of pharmaceutical companies, including ethically.”

Rare Disease Patient





ADVANCED THERAPY MEDICINAL PRODUCTS

The advanced therapy medicinal products hold promise for the treatment of a variety of rare diseases: specific EU-level actions must be taken to support the availability and accessibility of gene, cell and tissues therapies, which will become more numerous in future:

- + Cross-country collaborations should be put in place, to streamline access to advanced therapies for rare diseases, avoiding the requirement for patients to fund significant costs upfront (or else private funding collections/crowdfunding should be encouraged for the therapies entering the markets)
- + As above, proposals for a shared European fund for advanced therapies' reimbursement should be developed, to support the practicalities of patients receiving care in a diffe-

rent country (ensuring hospitals receive funds from agencies in different Member States in a reliable and timely fashion, for instance)

- + In the case of advanced therapies for the rarest diseases (those affecting fewer than 1 /100,000), an EU-Fund should be established to co-finance the generation of post marketing authorisation evidence across EU Member States during the years initially following approval, in order to reduce uncertainties
- + The precise role European Reference Networks (ERNs) can play in facilitating access to advanced therapies should be explored - and where relevant, enacted - ranging from supporting more experts and informed decision-making concerning which patients would benefit from which therapies, to actually providing advanced therapies in a limited number of centres across Europe, and collecting monitoring data

“

“A cure would be fantastic, but failing that, reasonably priced (or government funded) medication is needed”

Rare Disease Patient

A CONTINUUM OF EVIDENCE GENERATION

Data sources capable of supporting the launch and post-marketing assessment of orphan medicinal products must become more interoperable and federated through a continuum of evidence generation

- + The myriad ways in which the potential for real world data to inform research and development and post-marketing surveillance must be clarified, and new coherent coherent strategies implemented
- + Post-marketing surveillance for orphan therapies should be organised at the European level, through quality-assured shared data registration platforms/disease registries

- + Efficacy as well as safety data should be collected from patients on compassionate use programmes and pooled at the European (and where possible global) level, and be made available to companies to incorporate to evidence datasets, where appropriate
- + The role and capacity of ERNs in generating, collecting and analysing real world data to enhance the availability and affordability of orphan medicinal products should be further defined and adequately supported

(Additional recommendations pertaining to the potential for data to improve the accessibility and affordability of therapies can be found in the chapters dedicated to 'Basic, Clinical, Social and Translational Research for Rare Diseases' and 'Data Collection and Utilisation')



Solutions to improve the development, accessibility and availability of medical devices for rare diseases should be proposed and examined at the European level, and where appropriate, implemented

- + The benefits of a European process for the conditional approval of devices intended for use in rare diseases should be established, accompanied by plans for a robust and shared data-collection and data-submission system.
- + The advantages of European legislation incentivising the development of medical devices intended for rare diseases should be weighed, including the relative advantages of a centralised review for devices intended for orphan use.
- + Notwithstanding the benefits created by the [Medical Device Regulation 2017/745 \(MDR\)](#), the remaining lack of transparency around the clinical evaluation assessments performed by notified bodies should be addressed, and greater cross-talk should be encouraged – at least with respect to rare diseases - between the national bodies in charge of assessing pharmaceuticals, on the one hand, and devices on the other
- + The current silo between post-approval data for orphan medicinal products and medical devices used by people living with a rare disease should be addressed, to develop harmonised data collection plans of benefit to regulators, notified bodies and HTA professionals
- + Entrepreneurial efforts to develop medical devices for people with rare diseases should receive research and development (R&D) and regulatory support, especially if patient-led
- + Particular focus should be placed on the development of devices to collect and convey data from the home environment, which should be positioned within the broader telemedicine strategy
- + The potential for ERNs - and patients - to influence the design and creation of medical devices for rare diseases should be ascertained, along with their suitability for post-launch data collection on effectiveness



WHAT DO PEOPLE LIVING WITH A RARE DISEASE THINK?

The following figures come from several Rare Barometer surveys carried out in Europe between 2016 and 2021. In these surveys, people living with a rare disease told us their views on a range of issues, such as participation in research, access to treatments, social care needs, the impact of COVID-19 on their lives and their hopes for 2030. Find out more: eurordis.org/voices

MORE TREATMENTS AND THERAPIES ARE NEEDED FOR PEOPLE WITH RARE DISEASES

$\frac{2}{3}$ of people living with a rare disease reported having received treatments directly linked to their rare disease. Those treatments are mainly not disease-modifying but symptomatic: only 6% have experienced a curative treatment while 42% had symptomatic treatments, 25% have had a treatment that aimed to slow down or stop their disease and 4% received a treatment that aimed to prevent the disease.

$\frac{1}{3}$ have, on the other hand, reported having never received a treatment directly linked to their rare disease. The main reason being that, according to them, there are no existing treatments.

TREATMENTS NEED TO BE MORE AVAILABLE, ACCESSIBLE AND AFFORDABLE FOR PEOPLE WITH RARE DISEASES

22% of people with rare diseases could not get, in 2019, the treatments they needed because it was not available where they live.

14% of people with rare diseases could not get, in 2019, the treatments they needed because the waiting list was too long.

12% of people with rare diseases could not get, in 2019, the treatments they needed because they could not pay for it.

11% of people with rare diseases could not get, in 2019, the treatments they needed because they did not get the financial support to travel and receive the treatment in another country.

PEOPLE WITH RARE DISEASES ARE LOOKING FOR EQUITY, IN TERMS OF RESEARCH ALLOCATION, AMONG THE DIFFERENT RARE DISEASES

+ The majority of respondents think that research priorities should not be set depending on the existing knowledge of a disease, or the availability of already existing treatments, or the severity of the disease nor on its prevalence - respondents think all rare diseases require investments in research.

+ However, we can consider that this reflects the willingness of reaching equity. Given that some diseases are clearly disadvantaged compared to other (disregarded diseases), we might still be willing to prioritize research in those areas in order to ensure equity.





WHAT DO THE RARE 2030 YOUNG CITIZENS RECOMMEND?

ON COLLABORATION...

Young Citizens recognise that greater availability and accessibility of orphan medicinal products across Europe is highly dependent on stakeholders' willingness to collaborate more and in a more effective way. Therefore they recommend:

- + **Developing a more cohesive and more transparent reimbursement process** throughout Europe, a common baseline that respects the subsidiarity principle while guaranteeing a minimal threshold for availability and accessibility of orphan medicinal products and medical devices
- + **Establishing a European-wide reward for treatment and care abroad**, which would in turn foster cross-border healthcare
- + **Improving cross-country collaboration**, specifically on collaborative efforts to share best practices on topics such as reimbursement and access to care
- + **Fostering joint-HTA assessment**, as some collaborative efforts have proven to be highly beneficial to the rare disease community. However these projects often only concern a relatively small number of countries. Further initiatives would allow more cost-effectiveness and greater availability of innovation and the possibility to safest new technologies throughout Europe



HOW TO MONITOR SUCCESS

- ✓ Monitor the number of new marketing authorisations by the European Medicines Agency
- ✓ Analysing marketing authorisation data on new therapies to ascertain inequalities in disease area coverage
- ✓ Monitor accessibility and availability of new treatments on both national and European levels
- ✓ Regularly monitor the experience and expectations of people living with a rare disease and their carers following the development of a federated and transparent data ecosystem capturing a continuum of evidence on how new treatments are addressing unmet needs
- ✓ Monitor the number of new clinical trials for rare disease treatments registered in the EU Clinical Trial Database and WHO International Clinical Trials Registry Platform (ICTRP)



SUPPORTING THE SUSTAINABLE DEVELOPMENT GOALS BY 2030

3 GOOD HEALTH AND WELL-BEING



IMPROVING HEALTH AND WELL-BEING

By supporting the research and development of accessible and affordable treatments a large percentage of Europe's citizens will directly directly experience better health and well-being



REFERENCES

- + IRDiRC vision
<https://irdirc.org/about-us/vision-goals/>

- + EUCERD Recommendations on the CAVOMP Information Flow
http://www.eucerd.eu/?post_type=document&p=1446

- + Breaking the Access Deadlock
<https://www.eurordis.org/accesspaper>

- + IRDiRC Recognized Resources
<https://irdirc.org/research/irdirc-recognized-resources/>

- + Commission Communication laying down the provisions for implementation of the criteria for designation of a medicinal product as an orphan medicinal product and definitions of the concepts 'similar medicinal product' and 'clinical superiority'
<https://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2000:103:0005:0008:en:PDF>

- + Council Recommendation on an action in the field of rare diseases
<https://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF>

- + Mechanism of Coordinated Access to Orphan Medicinal Products (MoCA)
<https://www.eurordis.org/content/moca>

- + IRDiRC Orphan Drug Development Guidebook (ODDG)
<https://irdirc.org/orphan-drug-development-guidebook-materials/>

- + STAMP expert group
https://ec.europa.eu/health/documents/pharmaceutical-committee/stamp_en-#:~:text=The%20STAMP%20expert%20group%20is,and%20policies%20in%20this%20field

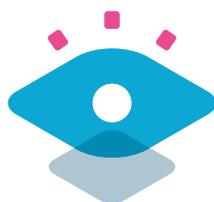
- + Medical Device Regulation 2017/745 (MDR)
<https://eur-lex.europa.eu/legal-content/EN/TXT/PDF/?uri=CELEX:32017R0745>

- + Rare Barometer Survey: Rare disease patients' experience of treatments (report not available yet).
<https://www.eurordis.org/voices>



THE FUTURE OF RARE DISEASES STARTS TODAY

RARE2030.EU/RECOMMENDATIONS



Rare2030

Foresight in Rare Disease Policy