

A better future by design: call for a new European policy framework for rare diseases

Over 230 experts set out recommendations for the next decade of rare disease policies

- Two-year foresight study in rare disease policy supported by EU institutions concludes with call for a new European policy framework for rare diseases
- Eight recommendations set out the roadmap for rare diseases by 2030, across diagnosis, access to treatments, data, research and integrated care
- Recognising progress that has been made, calls to “not lose momentum” in order to meet the unmet needs of people living with a rare disease

23 February 2021, Brussels - To mark Rare Disease Day 2021, 1000 people participate in closing conference of Rare 2030 Foresight Study with conclusions pointing to a refreshed EU policy framework for rare diseases

EURORDIS-Rare Diseases Europe, together with the Rare 2030 consortium of over 230 experts from across the field, call for a new European policy framework for rare diseases at an online conference today concluding the two-year Rare 2030 Foresight Study, a project initiated and funded by European institutions.

The Rare 2030 Plenary Conference, “[The future of rare diseases starts today](#)”, held in the lead up to the fourteenth edition of [Rare Disease Day](#), brings together over 1000 participants to present the long-awaited [recommendations](#) from the Rare 2030 Foresight Study. Prominent EU policy makers, including Commissioner for Health and Food Safety Stella Kyriakides and French Health Minister Olivier Véran are speaking at the conference.

A new European policy framework for rare diseases

While there has been great progress for people living with a rare disease in Europe since the adoption of the Council Recommendation on European Action in the field of Rare Diseases in 2009, the reality is that for 30 million people in Europe unmet needs remain and current policies are no longer fit for purpose.

For people living with a rare disease in Europe:

- It still takes on average four years to receive a diagnosisⁱ
- Only 6% have access to a treatment for their diseaseⁱⁱ

- 52% of patients and carers say their disease has a severe or very severe impact on daily lifeⁱⁱⁱ

A new European policy framework for rare diseases is needed to bridge progress from the last decade and to innovate based on patient needs. Crucially, this framework would:

- Bring together a refreshed concerted strategy across research, digital, healthcare and social welfare complementing existing legislations;
- Guide the implementation of national plans for rare diseases with the same measurable objectives;
- Encourage continued investment in the field of rare diseases at both the European and national levels to ensure we do not lose momentum.

The COVID-19 pandemic has highlighted the vulnerability of our health systems and the rare disease community. It has shown while we cannot plan for the future, we can prepare for it. Lessons must be learned to establish and redress the impact on already vulnerable rare disease populations, whilst capitalising on the positive momentum the crisis has created in terms of rapidly streamlining procedures, research collaborations and regulatory activity.

Yann Le Cam, Chief Executive Officer, EURORDIS-Rare Diseases Europe, said

"It is time to reset Europe's rare disease focus for the next decade: we need a policy framework in Europe adapted to today's realities, to embed excellence and bring policies in line with new technologies, values and infrastructures.

Europe's efforts since 2009 have shown how much progress can be made when national plans and strategies for rare diseases are coordinated across countries and integrated at EU level. We cannot lose momentum now: the Rare 2030 recommendations set the direction we need to go."

Measurable goals for rare diseases

The [Rare 2030 Recommendations](#) offer a roadmap for a new rare disease framework by establishing measurable goals for the first time. In line with the UN Sustainable Development Goals, such goals set a common direction for all EU Member States to make tangible change for all their citizens living with a rare disease.

If a new framework for rare diseases is implemented by EU institutions and Member State governments, the Rare 2030 recommendations hope that by 2030:

- All people affected by a known rare disease will be diagnosed within six months of coming to medical attention;
- 1000 new therapies will be available, and will be 3 to 5 times more affordable;
- The level of psychological, social and economic vulnerability of people with a rare disease and families will be reduced by one third.

The report published today, "[Recommendations from the Rare 2030 Foresight Study: The future of rare diseases starts today](#)" sets out eight overarching recommendations across diagnosis, treatment, data, research and integrated care, each accompanied by measurable goals to monitor progress made across Europe.

Robert Madelin, Chairman of FIPRA International; Director of DG Sante 2004-2010 and Rare 2030 Research Advisor, said:

"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.

The real shifts in policy put forward can unlock society-wide public good. If we can deliver on this vision, it will be to the benefit not only of the millions of people living with a rare disease, their families, employers and communities. It will also bring in its wake crucial improvements in healthcare beyond rare diseases."

Rare 2030 Foresight Study: a better future by design

Over 230 of Europe's most dedicated opinion leaders in the field of rare diseases and beyond have come together in the participative and iterative four stage Foresight Study to decide on the optimum future for people living with a rare disease, concluding today with the rare disease community's recommendations and goals.

The [Rare 2030 Plenary Conference](#) will see over 1000 participants endorse the recommendations, with the participation and support from some of Europe's leading politicians, including Stella Kyriakides (European Commissioner for Health and Food Safety), Olivier Véran (Minister of Solidarity and Health, France), Jan Blatný (Minister of Health, Czech Republic) and several Members of the European Parliament.

Rebecca Skarberg, European patient advocate and member of the Rare 2030 Panel of Experts said:

"Innovation that is needs-led, but also a joint responsibility, will lead to a future that leaves no one living with a rare disease behind. Our quality of life is defined by more than just genetic misspellings and symptoms. It's also about community, independence, empowerment and fulfilling your potential.

Rare 2030 has provided the recommendations, based on our preferred future to allow us to live our best life by 2030."

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More details about the Rare 2030 Recommendations

Eight recommendations: a roadmap for a refreshed policy framework

1. **A European policy framework** guiding the implementation of consistent national plans and strategies, monitored and assessed by a multistakeholder body on a regular basis.
2. **Earlier, faster and more accurate diagnosis** of rare diseases through better and more consistent use of harmonised standards and programmes across Europe, new technologies and innovative approaches driven by patient-needs.
3. **A highly specialised healthcare ecosystem**, with political, financial and technical support at European and national levels, that leaves no person living with a rare disease in uncertainty regarding their diagnosis, care or treatment.
4. **Guarantee the integration of people living with a rare disease in societies and economies** by implementing European and national actions that recognise their social rights.
5. A culture **encouraging meaningful participation, engagement and leadership of people living with a rare disease** in both the public and private sectors.
6. **Rare disease research maintained as a priority** - across basic, clinical, translational and social research.

7. **Data used to its maximum** to improve the health and well-being of people living with a rare disease.
8. **Improve the availability, accessibility and affordability of rare disease treatments**, by attracting investments, fostering innovation and collaboration across countries, to address inequalities.

Read the recommendations in full [here](#).

About the Rare 2030 Foresight Study

Rare 2030 is a [foresight study](#) that gathered the input of over 230 patients, practitioners and key opinion leaders to propose policy recommendations that will lead us to improved policy and a better future for people living with a rare disease in Europe. This was a two-year project is co-funded by the European Union Pilot Projects and Preparatory Actions Programme (2014- 2020).

It followed a four stage process using foresight:

- 1) Establishing the existing [knowledge base](#)
- 2) Identifying [trends](#) likely to impact rare diseases over the next decade
- 3) Preparing four potential [scenarios](#) based on the trends that could play out by 2030, and deciding on the preferred scenario “Investment for social justice”, prioritising patient needs-led innovation and collaboration
- 4) Develop policy [recommendations](#) based on the preferred scenario.

The study was led by [EURORDIS-Rare Diseases Europe](#) with nine partners, and an expert panel of over 230 people who have contributed to the development of the recommendations.

About the Rare 2030 Plenary Conference: The future of rare diseases starts today

The Rare 2030 Plenary Conference “[The future of rare diseases starts today](#)” marks the end of this two-year foresight study and will be the occasion to present the Rare 2030 policy recommendations for a new policy framework in presence of high-level speakers, such as Member of Parliament Frédérique Ries, and European Commissioner for Health and Food Safety Stella Kyriakides.

About EURORDIS-Rare Diseases Europe

[EURORDIS-Rare Diseases Europe](#) is a unique, non-profit alliance of over 930 rare disease patient organisations from 73 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe. By connecting patients, families and patient groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services.

About rare diseases

The European Union considers a disease as rare when it affects less than 1 in 2,000 citizens. Over 6,000 different rare diseases have been identified to date affecting an estimated 30 million people in Europe and 300 million worldwide. 72% of rare diseases are genetic whilst others are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative. 70% of those genetic rare diseases start in childhood.

Due to the low prevalence of each disease, medical expertise is rare, knowledge is scarce, care offerings inadequate and research limited. Despite their great overall number, rare disease patients are the orphans of health systems, often denied diagnosis, treatment and the benefits of research.

Contact

Jenny Steele, Campaign Manager, EURORDIS-Rare Diseases Europe:

- jenny.steele@eurordis.org
- +44 79727 080 307

ⁱ Rare Barometer Survey: [Improve our experience of healthcare!](#) January 2021

ⁱⁱ Rare Barometer Survey: Access to treatments for people living with a rare disease. May 2019. (Not published online, please contact stanislav.ostapenko@eurordis.org for more information.)

ⁱⁱⁱ Rare Barometer Survey: [Juggling care and daily life](#). May 2017