Project Update

May 2020

What is Rare 2030?

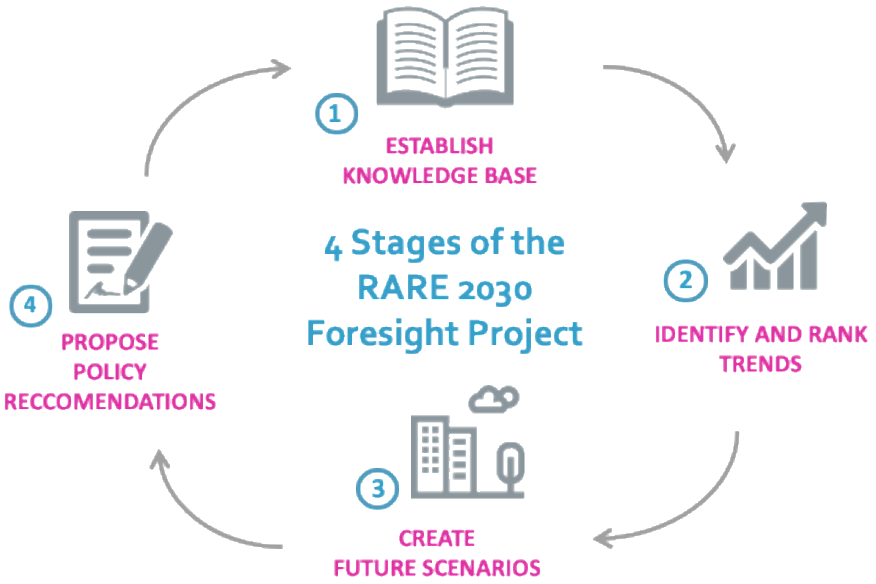
Rare 2030 is a foresight study that gathers the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations that will lead us to improved policy and a better future for people living with a rare disease in Europe. This a two year project that will end in a presentation to parliament at the end of 2020 with recommendations on the most critical areas needing sound policy.

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. Rare 2030 will guide a reflection on rare disease policy in Europe through the next ten years and beyond.

How Does it Work?

The Rare 2030 Foresight Project includes 4 major stages detailed below. The European Conference on Rare Diseases and Orphan Products (ECRD 2020) marks the occasion to present the proposed future scenarios and begin discussion around:

1. Which scenarios are most preferred by the rare disease community?
2. Which scenarios are most likely to happen?
3. How do we prepare to achieve the scenarios we want and avoid those we don't? What are the strategies and policies needed to do this?



1. KNOWLEDGE BASE

To stimulate debate, a background document was prepared for each Sub-Group of the project's Panel of Experts, known as a Knowledge-Base Summary.

These are non-exhaustive summaries of relevant policies, initiatives, resources and actors involved in each broad area. They should be viewed as *dynamic* documents - the table below will always link to the latest versions of each, and indicate when a new version is available. Where possible, the documents incorporate data from the *Resource on the State of the Art of Rare Disease Activities in Europe*, obtained from EU Member States to elucidate national approaches to a range of topics. This data is still being updated by some MS, and thus these sections of the Knowledge-Base Summaries will be updated in the coming weeks and months. Each document ends with the results of a dedicated Literature review and links to both a select and extended bibliography (reflecting work performed by Rare2030 Partner INSERM). The documents are outputs of Rare2030 WP4: their creation was overseen by UNEW, with the support of the Rare2030 partners

- Political & strategic frameworks relevant to rare diseases
- Data Collection and Utilisation
- Availability and accessibility of Orphan Medical Products (OMPs) and medical devices
- Basic, Clinical, Translational and Social Research for Rare Diseases
- Diagnostics
- Integrated, Social and Holistic Care for People with Rare Diseases
- Rare Disease Patient Partnerships
- Access to Healthcare

2. TRENDS

The Panel of Experts of the Rare 2030 project (nearly 200 members across all stakeholder groups) have identified 12 Trends (described fully here: http://download2.eurordis.org.s3-eu-west-1.amazonaws.com/rare2030/Rare2030%20Final%20ALL%20TRENDS.pdf) that will influence rare disease policy in the next 10 years.

3. SCENARIOS

Considering these trends we noticed 2 clusters that emerge around **our society's values in solidarity** and **how they may be applied to drive innovation**. By combining different progressions of these key trends we were able to create four possible scenarios for 2030 along these two axes.

Axis 1 - Societal Attitude toward Solidarity (horizontal axis)

Who is responsible for the health and well-being of people living with rare diseases? Government? Employers? People living with rare diseases themselves? Their families? People living with rare diseases may not only be physically disadvantaged but also socially not because of the individual's inability to fit in with their surrounding environment, but because of society's inability to include them.

- Individual Responsibility - Out of necessity, people living with rare diseases have often been required to take matters into their own hands? Is this ideal? In what circumstances is it "okay"? Individual could also mean individual countries, individual institutions or individual disease associations. Generally speaking in scenarios on this end of the axis collaboration is not fostered.
- Collective Accountability - Equity means giving everyone a chance to achieve the greatest possible health and well-being. This is only possible if there is a collective responsibility to grant people living with rare diseases the same chance to achieve their greatest possible health and well-being. This also implies collaboration across countries and sectors of society.

Axis 2 - Type of Driver of Innovation (vertical axis)

Innovation does not only mean new treatments. Innovation also describes new, better, more effective ways of solving problems - in this case, related to health and well-being of people living with rare diseases or the systems that make them possible. Innovation can include policies, systems, technologies, ideas, services, and products (e.g. surgical innovations, epigenetics, CRISPR-mediated DNA modification, cooperation, patient autonomy...) that provide solutions (e.g. to improve quality, reduce harm, improve access, increase efficiency, eliminate waste, and lower costs...) to existing problems for people living with rare diseases.

- Market-led Innovation We usually think about innovation in the life sciences as being technology-driven where innovation originates in scientific discoveries. When new technologies make it possible to move from the scientific domain to technology implementation, inventors and corporate R&D groups, companies compete to develop commercial applications. Across university research laboratories and throughout the biotech and pharma industries, scientific breakthroughs have been the launching point for major product developments in the traditional bench-to-bedside trajectory. In this approach the focus is more on supply, on curative solutions and on financial return on investments.
- Patient Needs-led Innovation
  Over the past decade, however, a focus on needs-based innovation has emerged as an alternative strategy for innovation, particularly in the domain of biomedical technology (medical devices and diagnostics). Innovators are beginning to focus on developing a deep understanding of needs as the starting point of the invention process. In contrast to the market-led approach, this approach first creates the innovation, then seeks out its market. In this approach the focus is on demand and where possible, preventative solutions. Although the return on investment may be financial it is first and foremost directly responding to the needs of the population it is designed to serve.

PATIENT-NEEDS-LED INNOVATION

Deep understanding of needs as the starting point of the innovation process.

SCENARIO 3

SCENARIO 1

INNOVATION

INDIVIDUAL RESPONSIBILITY

Individualistic mentality: my country, my organization, my research first

SOCIETAL ATTITUDE

TOWARD SOLIDARITY

COLLECTIVE ACCOUNTBILITY

Solidarity among citizens, countries and diseases as policy priority

TYPE OF

SCENARIO 4

SCENARIO 2

MARKET LED INNOVATION

The market-led approach first creates the technology innovation, then seeks out its market.

4. Rare 2030 as the back bone of the 10th European Conference for Rare Diseases

A broad range of experts and project partners have described how several trends could evolve under the four different Scenarios in the video below. The Programme Committee and speakers were briefed on the Foresight Study and asked to integrate the following questions for their audience to consider:

1. Which scenarios is most preferred by the rare disease community?
2. Which scenario is most likely to happen?
3. How do we prepare to achieve the scenarios we want and avoid those we don't? What are the strategies and policies needed to do this?

Rare 2030 Scenarios - VIDEO

SCENARIO – ***Fast But Not Fair***

If we continue as we are, we'll end up in the ***Fast But Not Fair*** world where public and private 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 and the innovations that are proposed are not sustainable as they focus more on developing rewardable products than addressing long term patient needs. Patient organizations and health care professionals working across multiple countries and rare diseases help identify and advocate to improve gaps and disparities in healthcare, treatments and research by collecting the patient reported experience.

With new IT solutions to gather large amounts of health related data there is great potential to advance care and treatment. However, major bottlenecks exist due to a lack of a clear and simple rules and

incentives to share information, especially between publicly funded structures and the private healthcare industry.

Health remains the responsibility of each country in Europe but certain disease areas and some healthcare related services benefit from coordinated public efforts at the European level. Driven by budget constraints, services are prioritized and access to new technologies is slow and limited even if it remains free at the point of access. However, “top-up” payments are more and more common for those breakthrough innovations for those who can afford to pay.

The process for assessing new innovations is more centralized at EU level but lacks transparency and true patient needs are only partially considered. As a result, developers ask high prices for innovative products with small impact on the patients and payers use the process complexity as a tool to block or slow-down the entrance of new products into the market.

The challenges in driving fast but fair innovation are likely to create frustration as well as an increased distrust in science, health care, and the systems that govern them.

SCENARIO - ***It’s Up To You To Get What You Need***

If distrust in supranational efforts to manage health and other societal needs continues, we may find ourselves in the ***It’s Up To You To Get What You Need*** world with distinct national two-tiered health care systems in which basic care is provided by the government and a second tier of care exists for those who can pay for additional, better quality or faster access.

In this scenario, patients may get the holistic care they need but it will highly depend on the country in which they live.

A lack of coordination between health care providers across countries means that many people with complex rare diseases will continue to struggle for diagnosis. And the few multistakeholder networks that do exist across Europe remain significantly underfunded. As a result, their role is limited to an advisory one with a very small impact on health outcomes for people living with a rare disease.

Given that no country, has sufficient numbers of patients to conduct adequate research for rare diseases, great advancements in innovation are hampered in this future due to a lack of investments in multinational research efforts, data collection platforms and data sharing infrastructures.

The innovation that does reach the market for rare diseases become extremely expensive and only accessible for the richest of countries and patients and ultimately interest in investment in developing new technologies decreases due to a diminishing market.

Most people would agree that everyone has a right to healthcare and a right to the highest attainable standard of health. But there may be disagreement about who should be responsible for achieving and paying for it.

Some may believe in personal responsibility, free markets and governance that is limited to providing people the freedom necessary to purse their own goals emphasizing the empowerment of individuals to solve their problems.

SCENARIO - ***Technology Alone Will Save You***

We may find ourselves in such a scenario, ***Technology Alone Will Save You***, where private companies have a greater role in managing the health of people living with rare diseases. Many innovations such as health applications are available for people living with rare diseases to manage their own health. These

innovations can help them in their everyday life but leave them largely responsible for their health and making sure their right to health is respected. As a result, many risk being left behind.

Health care systems are private, insurance based, market-led and profit driven. Voluntary intergovernmental coordination exists but not enough to regulate health, health related data and research.

Several private companies have created fast and accurate diagnostic options but only for those who can pay out of their own pocket. Thanks to the investments of health data companies, artificial intelligence and other cutting-edge science has led to breakthroughs for some of the most complicated and rarest diseases but most diseases remain without treatment options. Again, only the richest of patients can have access to these innovations and often rely on themselves or the generosity of others through crowdfunding to get treatment for their diseases.

Genomics has also developed dramatically and is regularly used by doctors and individuals. When commercially exploitable, the information is gathered in a collective way to find information that is helpful to patients.

Innovators in the field of rare diseases may buy personal data and use artificial intelligence algorithms to combine the prevalence of specific genetic conditions with the willingness and ability of those people to pay. Private investments in research and development for rare disease are therefore led by economic models factoring genetic and income data of the target population. People living with rare diseases are not only extremely burdened with the need to find their own solutions to faster and better diagnosis and treatments but may take significant informational risks in doing so.

Patient organization support has largely been replaced by technologies that allow patients to manage their own needs and those that have access are quite empowered sometimes even creating breakthrough technologies themselves. But what is good for the market may not necessarily be good for the health of most people living with rare diseases and systems fostering such health inequality affects not only those living with a rare disease but the health of everybody, especially in the context of global epidemics.

Others believe it is the duty of governments to achieve equity in health and alleviate social inequalities through innovation and systems of care.

SCENARIO - ***Investments for Social Justice***

If we prioritize societal responsibility, equity and the regulatory 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 equally shared across Europe to ensure the health and well-being of all European citizens – including those living with a rare disease. The European Union's increased legislative power in areas of health and social welfare reduces the risk that patients in some European countries are left behind.

Not only the curative, but also the preventative, rehabilitative and palliative needs of people living with rare diseases are better understood. As such, healthcare systems are led by these holistic patient needs and are driven by better outcomes based on a growing evidence-base achieved through comprehensive health data collection that goes beyond consumer and profit driven companies. Data is shared smoothly across boarders through systems that are interoperable and infrastructures that are connected.

Multistakeholder initiatives prioritize investments in rare disease research that responds to patient needs thus focusing on as many diseases as possible and not just focusing on low hanging fruit. The resulting innovations are evaluated at the European level and with greater transparency, accountability, cost-effectiveness and considering the patient experience. Given the limitation in government funding and regulations required of increased collaboration, cutting edge innovations may develop much more slowly

but despite a lowered competition, needs-led innovation continues in the long run. Existing medicines, non-pharmaceutical treatments and assistive technologies are affordable and equally available no matter where you live in Europe.

The healthcare model for rare diseases has become a model for other health services and health threats where the system is resilient enough to tackle both acute and long-term patient needs. Centres of expertise dedicated to care for rare disease are well-identified, well connected to local primary care as well as to networks within and outside the country., To ensure sufficient funds for these networks, health care decisions are not only driven by cost savings and governments are able to increase the proportion spent on healthcare.

To achieve this scenario people living with rare diseases, their families and all citizens are mobilized to be actively involved in policy decisions in a highly competitive innovation market. The field of rare diseases has played a pioneering role in promoting such a deep change in European health and social policy.

5. NEXT STEPS

General consensus from the conference was a preference for Scenario 1 with a need to balance benefits of Scenario 2 (and to some extent the other scenarios)

Through the remaining portion of 2020 and into 2021 the project partners will be organizing several workshops to get detailed feedback from the following groups on their perspectives. A culmination of policy recommendations from this vast group of opinion leaders and decision makers will be reflected in the projects final recommendations presented at the Parliament on the occasion of Rare Disease Week 2021. Please mark your calendar!

- Panel of Experts Survey and Workshop/EURORDIS CNA and CEF 7 Nov 2019 Brussels (https://www.rare2030.eu/live/)
- European Conference on Rare Diseases (ECRD) 15-16 May 2020 ONLINE
- Young Citizen Conference 8 July 2020
- Regional Workshops aligned with upcoming EU presidencies June-Nov 2020
    - Croatia (Romania)
    - Germany
    - Spain (and Portugal)
    - Slovenia
    - France
    - Czech Republic (and Slovakia)
    - Sweden
- European Policy Backcasting Workshop (with European Reference Networks and other European level KOLs) Sept 2020
- Policy Conference @Parliament February 2021