



## Rare 2030 Panel of Experts Conference

“Validation of Trends to Build Future Scenarios  
in Rare Disease Policy”

Report

7 November 2019

Hotel Pullman

Brussels



## Objectives

Stakeholders comprising the Rare 2030 [Panel of Experts](#) have convened in a series of 18 teleconferences throughout the first year of the project to identify over 100 future-facing trends that will influence people living with rare diseases and the policies that support them. These [trends](#) were consolidated into 12 overarching trends (as below) and ranked in terms of importance and uncertainty via a survey and interactive debate. This activity complemented an expert panel debate, in which representatives of the [project's Research Advisory Board](#) provided their insights related to the future of rare disease policy. Together these presentations served as a refresher for the interactive portion of the workshop (afternoon) where participants continued to elaborate and validate the trends presented, ultimately leading to the next stage of the project which involves the proposal of future scenarios of rare disease policy and final recommendations.



Lucia Monaco, Chair IRDIRC (project partner), Raquel Castro Director of Social Policy, Durhane Wong, Chair of RDI Council (Members of Panel of Experts)

*"Europe has built a strong ecosystem for rare diseases... and this project is one opportunity for continued multi-stakeholder dialogue to prepare for the future. I am, however, concerned that we have lost the platform for exchange giving an impetus to the European Parliament, the Council of Ministers and the Commission to really design ambitious plans for how we get there together and design policies where we are not only dependent on the accidental progress in one or another field but actually get together to design what kind of future we want."*



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Terkel Anderson, President of EURORDIS-Rare Diseases Europe

Rare 2030 Research Advisor

Participants were reminded of the importance and strategic impact of the project by European Member of Parliament, Frederique Ries and Yann le Cam, CEO of EURORDIS.



*"The adoption of Rare 2030 by the European Parliament is one our proudest achievements."*

Frédérique Ries  
Member of European Parliament

## Attending Participants

The large majority of the Panel of Experts (151 attendees) as well as observers from the European Commission and Parliament joined the productive gathering. The [participants](#) include representatives from a very wide range of stakeholder groups, including European Reference Network representatives.



*"You have shaped this study very well, you have asked us to look 10 years forward and see where we want to be. For my part I would like to see a network of Centres of Excellence globally."*

Ruediger Krech, Director for Health Systems and Innovation, World Health Organisation

Rare 2030 Research Advisor

# Structure of the Meeting and Recorded Presentations

The [Agenda](#) and corresponding videos describes key speakers in the morning session

9.00-9.10	Welcome: Frederique Ries	Watch video <a href="#">here</a>
09.10 - 09.30	Review of the Rare2030: Preparing the Future of Rare Disease Policy <ul style="list-style-type: none"><li>• Yann Le Cam, EURORDIS-Rare Diseases Europe</li></ul>	Watch video <a href="#">here</a>
09.30 – 10.30	Rare 2030 Research Advisory Board Panel Discussion: Global insights Moderated by Andrea Ricci, ISINNOVA <ul style="list-style-type: none"><li>• Robert Madelin, FIRPA, former Director DG CNECT and DG SANTE – (10' min)</li><li>• Didier Schmidt, ESA, former advisor on Foresight to the EC President – (10')</li><li>• Rüdiger Krech, WHO Health Systems and Innovation(video) (10')</li><li>• Terkel Andersen, EURORDIS-Rare Diseases Europe Board of Directors (10')</li></ul>	What video <a href="#">here</a>
10.30 -10.50	Coffee break	
10.50 – 11.15	Presentation of Methods and Trends <ul style="list-style-type: none"><li>• Victoria Hedley, University of Newcastle Institute of Genomic Medicine</li><li>• Giovanna Giuffrè, ISINNOVA</li></ul>	What video <a href="#">here</a>
11.15-13:00	Interactive Session Validating the Trends - Rare 2030 Project Partners	What video <a href="#">here</a>

# Workshops Outcomes

Following several months of [virtual consultation with the Panel of Experts](#) 12 emerging trends important to the future landscape of rare disease policy were identified:

Overarching Trends in RDs	Specificities for RDs
1. <a href="#">Rise of pan-European multi-stakeholder networks to advance diagnostics, treatment and care for rare diseases</a>	Europe is now firmly in the age of the European Reference Network (ERNs) - networks of centres of expertise and healthcare providers organised across EU borders whose future depends on continued support. Multi-stakeholder collaboration is also becoming increasingly popular in the research and innovation sectors (e.g. European Joint Programme for Rare Diseases (EJP RD)). Stakeholders in the rare disease field are increasingly collaborating with actors from complimentary fields including social sciences, health policy, regulatory science, eHealth, big data, -omics approaches, bioinformatics, nanotechnology, etc.
2. <a href="#">Strains on the health care budget and the emergence of new care delivery models</a>	As healthcare budgets continue to strain and rare diseases “compete” with more increasingly prevalent non-communicable diseases, health care delivery models for people living with rare diseases become more person-centred and holistic to maximize impact
3. <a href="#">Greater variation in access to treatments and care resulting in more inequality across Europe for people with rare diseases</a>	High market price of orphan medicinal products allows for return on investment and continued R&D in the sector but results in heterogeneous availability and accessibility across member states
4. <a href="#">Demographic change of RD patients introducing new challenges</a>	As more people with rare diseases are living longer the RD population is faced with new challenges such as reproductive choices, transition into adolescent/adult care, comorbidities of ageing and age related disease and a better understanding of the natural history of many rare diseases
5. <a href="#">Threats to solidarity equity, and the prioritization of rare diseases</a>	On the one hand increased threats to solidarity and (such as inequalities in access to care and treatments between and within countries in Europe) are anticipated. At the same increased efforts for solidarity and equity on the global scale are underway such as efforts for integration of people living with rare diseases and related disabilities in society
6. <a href="#">Increasingly empowered rare disease patient and the patient advocacy evolution</a>	Dynamics in patient advocacy and in the role of the patient in health care, research and innovation are changing due to a number of social, technological, political and economic drivers leading to a new era in patient partnership.
7. <a href="#">Rise in innovation-oriented, multi-stakeholder, needs-led (patient-led) research</a>	The lack of disease-modifying treatments and devices for the vast majority of rare diseases suggests continued gaps in design, execution, delivery, and ultimately the outputs of rare disease research. A globalization of efforts; a rise in needs-led objectives and co-creation are a few of the trends filling these gaps.

<p><a href="#"><u>8. Facilitation of knowledge exchange and local care delivery through digital health</u></a></p>	A greater potential use of virtual care and eHealth technology to facilitate knowledge exchange and deliver care locally is being recognised though implementation remains heterogeneous across countries. Challenges and opportunities exist.
<p><a href="#"><u>9. Increased potential for large sets of standardised and interoperable data</u></a></p>	The potential to help advance understanding of RD and accelerate research (with coded and structured data) continues to be recognised as do the technical, legal, ethical, social obstacles in data collection and sharing (e.g. data donation, data portability, dynamic e-Consent control; threats: GDPR over interpretation) that require resolution.
<p><a href="#"><u>10. Rise in the use of AI for diagnostics, treatment and care, opening-up the potential of 'big data'</u></a></p>	A rise in the potential of AI for diagnostics, treatment and care, (via eHealth, codification of eHealth records, portable data, etc.) opening-up the use of 'big data' which nevertheless remains limited in the field of rare diseases and still requires significant regulatory attention and a clearer ELSI (ethical, legal, social issues) framework.
<p><a href="#"><u>11. New technologies and advanced therapeutics</u></a></p>	A better understanding of the mechanisms behind developmental, functional and degenerative rare diseases, new technologies such as gene editing and advanced therapeutics including precision medicine introduce breakthrough opportunities to improve the lives of people living with rare diseases but also potential undesirable consequences
<p><a href="#"><u>12. Application of Whole Genome Sequencing from the research to the clinical sphere</u></a></p>	With a great majority of rare diseases being genetic, advances in the technology around Next Generation Sequencing (NGS) offer significant promise for unravelling the epidemiology of rare disease, accelerating accurate diagnosis and better targeting treatments. These advances introduce a need for an updated ELSI (ethical, legal, social issues) framework.

Prior to the Conference, conference attendees were asked to rank these trends according to their importance and uncertainty having reviewed the [full Rare 2030 emerging trends](#). Along with afternoon breakout sessions where attendees elaborated on their ranking of trends, participants also began to explore the interrelation of these trends and potential indicators to measure their evolution through time. The analysis of this collection of insights will be summarized in Deliverable 5.1 of the project and disseminated to all workshop participants.

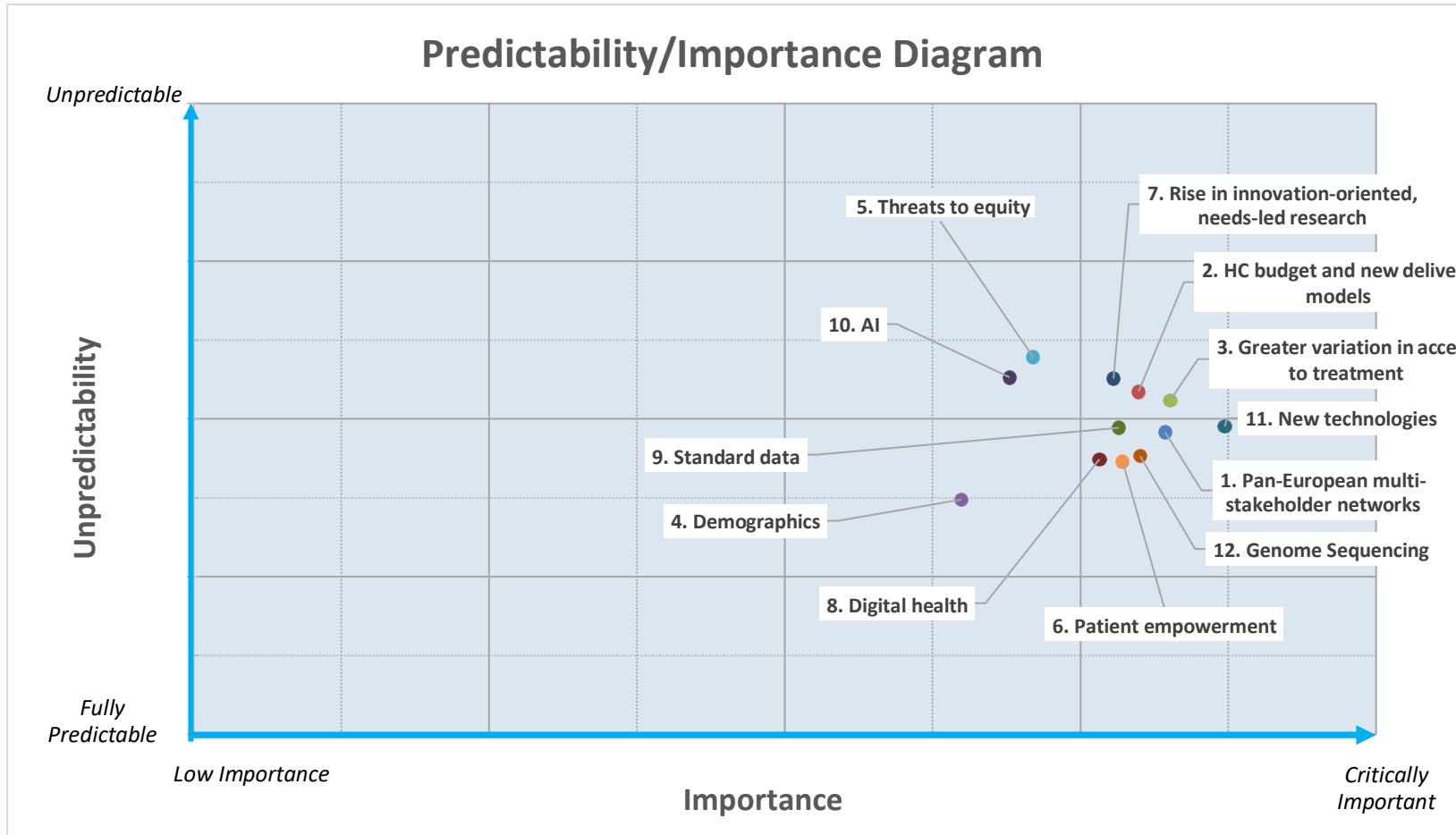
*“How will we define rare diseases... will we be more individualized in the way we see PLWRDs or will we be able to address them given the groupings we have today?”*

Robert Madelin  
 Chairman, FIRPA International  
 Rare 2030 Research Advisor

Preliminary results include the following ranking:

How important and predictable do you consider the evolution of these trends to be at 2030/2040?

*Graphic representation of the results*



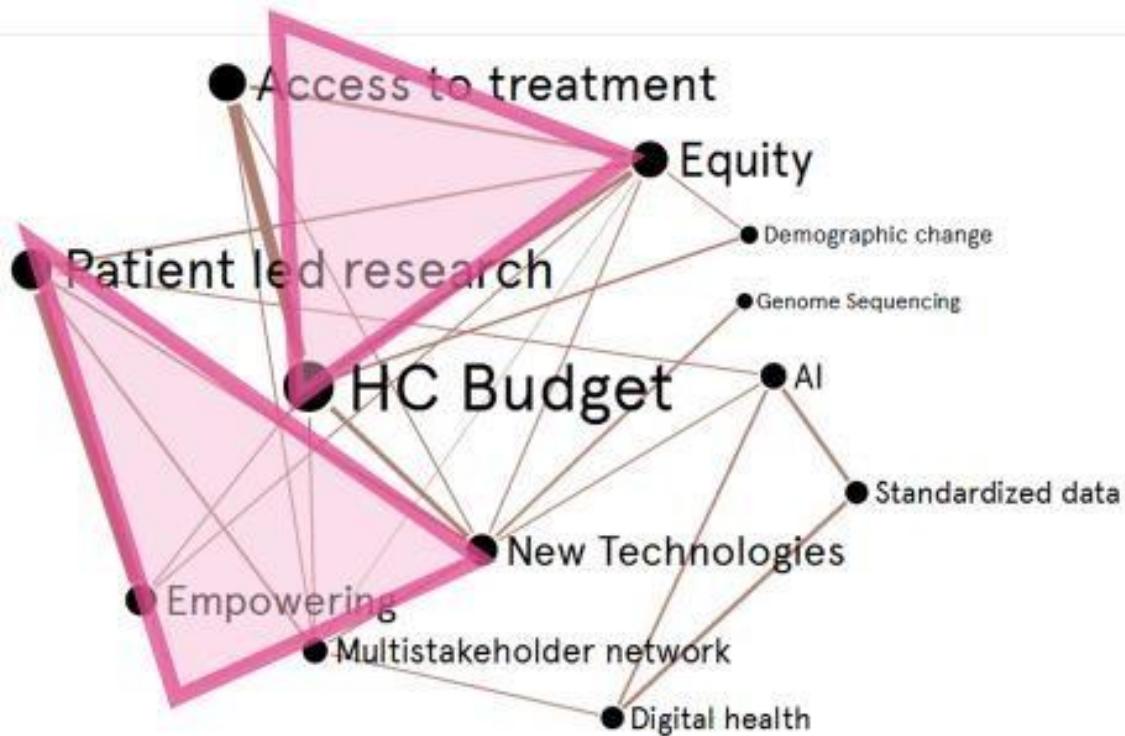
The following trends are identified as most unpredictable and important and are clustered into the following two groups based on analysis of interrelationships:

## CLUSTER OF THE TOP SIX TRENDS BY IMPORTANCE AND UNCERTAINTY



- > 3. Greater variation in access to treatments and care resulting in more inequality across Europe for people with rare diseases
  - > 2. Strains on the health care budget and the emergence of new care delivery models
  - > 5. Threats to solidarity equity, and the prioritization of rare diseases
- 
- > 11. New technologies and advanced therapeutics
  - > 1. Rise of pan-European multi-stakeholder networks to advance diagnostics, treatment and care for rare diseases
  - > 7. Rise in innovation-oriented, multi-stakeholder, needs-led (patient-led) research

## CLUSTERS OF TRENDS PER CONNECTIONS



*"Technology can be a good servant but also a bad master....In 2040 will we remain "human"?"*

Milan Macek,  
Board Member of the European Society of  
Human Genetics (ESHG)  
Rare 2030 Research Advisor

## Summary and Next Steps

The most uncertain and important trends will be used to develop potential scenarios of what life could be like for people living with rare diseases in the coming decades. Translated into narratives and animations, they will be presented at the [European Conference for Rare Diseases and Orphan Drugs](#) online May 14-15<sup>th</sup> 2020. This event will serve an opportunity for Conference participants and the wider rare disease community at large to consider their preferred scenarios and use the platform of the conference and its [six themes](#) to begin discussing in detail the policies needed to get to the preferred future scenarios.

*"One policy framework in 2008/2009 does not do the job. Fortunately, one of the key recommendations [of the European Auditors report on the CBHC directive] is that the Commission needs to review, adopt and potentially replace the framework on RDs before 2023. The environment is evolving and we need policies to address the needs of the future. The reflections in this project can serve as groundwork for such a framework."*

Yann le Cam, CEO, EURORDIS-Rare Diseases Europe



## Additional key events to translate scenarios into policy recommendations

### KEY EVENTS

- Panel of Experts Workshop/EURORDIS CNA and CEF  
7 Nov 2019 Brussels
- European Conference on Rare Diseases (ECRD) 15-16 May 2020 Stockholm
- Regional Backcasting Workshops aligned with upcoming EU presidencies May-Nov 2020
  - 2020 – Croatia, Germany
  - 2021 – Portugal, Slovenia
  - 2022 – France, Czech Republic
  - 2023 – Sweden, Spain
- European Backcasting Workshop Sept 2020
- Young Citizen Conference June 2020
- Policy Conference @Parliament December 2020



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**We look forward to the active participation of the Panel of Experts, Research Advisory Board, project partners and all interested key opinion leaders and decision makers at these events.**



For the latest updates and more information:

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