Research Advisory Board

Milestone 1.1

March 31 2019

WP Participants:
EURORDIS

V1	18 February	EURORDIS
V2	20 February	All partners
Final version	22 Feburary	All partners

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.

Contents

Purpose of the Research Advisory Board....3
List of Candidates and Bios....3
Formal Invitation....7
Terms of Reference....11
Responses to Date (31 March 2019)....17

Purpose of the Research Advisory Board

The Rare2030 project will utilise the support and expertise of an approximately 10-member Research Advisory Board (Rare 2030 RAB) to act as a sounding board to the Executive Management Committee and the Management Board during the project's duration (January 2019 – December 2020) assessing the scientific value of the project achievements (are we using the right methods?), their innovative contents (do we consider related sectors such as IT, artificial intelligence, social innovation?), widely and promptly outcomes beyond the RD community (how does this fit with "mainstream" health policy?).

List of Candidates and Bios

Ruxandra Draghia-Akli
Merck (formerly DG for Research and Innovation, European Commission)

Dr Ruxandra Draghia-Akli is currently Vice President Public Health and Scientific Affairs at Merck Global Vaccines.

Prior to this position she served as Director of the Health Directorate at the Research and Innovation DG of the European Commission, where she initiated the International Consortium for Rare Disease Research (IRDiRC) along with her counterpart at the NIH (US) Francis Collins. She also initiated or provided support for legislative, regulatory and policy issues in the area of health research and innovation, and established, in consultation with stakeholders, research priorities for the Health Research Programme of the EC.

Previously, Dr Draghia-Akli served as Vice-President of Research at VGX Pharmaceuticals (now Inovio) and VGX Animal Health. Her research activities focused on molecular biology, gene therapy and vaccination. She is a global leader in the field of nucleic acid delivery for therapeutic and vaccination applications. She is an inventor on more than a hundred patents and patent applications.

Dr Draghia-Akli received an MD from Carol Davilla Medical School and a PhD in human genetics from the Romanian Academy of Medical Sciences. She also completed a doctoral fellowship at the University of René Descartes in Paris, supported by the rare disease association "Vaincre les Maladies Lysosomales", and a post-doctoral training at Baylor College of Medicine (BCM), Houston, Texas, USA, and served as faculty at BCM. In 2012, she became an honorary member of the Romanian Academy of Medical Sciences.

Rüdiger Krech, *Director,* Universal Health Coverage and Health Systems, WHO

Dr Rüdiger Krech is currently the Director, Health Systems and Universal Health Coverage at the World Health Organization (WHO) covering WHO's work in the areas of integrated people-centered health services, safe and effective medicines, health workforce, financing and health governance. He also facilitates coordination and coherence between the health systems work of the Organisation and other areas of public health. Prior to that he was Directors of the Department of Ethics, Equity, Trade and Human Rights also at the WHO from 2009 to 2012. In this capacity, he was also responsible for WHO's work on Social Determinants of Health and Health in All Policies.

Before joining WHO, he was in charge of German International Cooperation's (GiZ) work on social protection from 2003-2009 both in its Headquarters in Eschborn near Frankfurt and in India. He has also held various management positions at the World Health Organization (WHO) Regional Office for Europe in Copenhagen in the fields of health systems, health policies, health promotion and ageing between 1992-2003. Dr Krech has studied educational sciences, medicine and public health and holds a doctoral degree in public health.

Robert Madelin
FIPRA (formerly Director General DG SANTE and DG CONNECT, European Commission)

Robert Madelin is the International Chairman of FIPRA (Foresight International Regulatory and Policy Advisers).

From 2004-2016, Mr. Madelin held a series of senior leadership positions at the European Commission: as Senior Adviser for Innovation, as Director General for Communications Networks, Content and Technology (DG CONNECT) and as Director General for Health and Consumer Policy (DG SANTE). Prior to that, Mr. Madelin was for 20 years a negotiator in international trade and investment, first for the UK, and then for the EU. Robert served notably in the Cabinet of European Commission Vice-President Leon Brittan. He studied at Magdalen College, Oxford and at the French Civil Service College (ENA). His other current major engagement is as Visiting Research Fellow at the University of Oxford's Department of Politics and International Relations. He is also an Honorary Doctor of the University of Edinburgh, an alumnus Policy Fellow of Cambridge University's Centre for Science and Policy and an Honorary Fellow of the Royal College of Physicians of London. Robert is the author of 'Opportunity Now: Europe's mission to innovate' (2016).

Milan Macek
Orphanet, Eurogentest, RD Connect

Professor Milan Macek Jr. MD, DSc is the chairman of the Department of Biology and Medical Genetics at the Charles University in Prague - the largest academic medical and molecular genetics institution in the Czech Republic. He was a past President of the European Society of Human Genetics (ESHG), currently a board member of the European Society for Human Reproduction and Embryology and of the European Cystic Fibrosis Society (ECFS). His institute contributes to dissemination of knowledge in genetics gathered within various international European projects, such as CF Network, EuroGentest, EuroCareCF or Techgene, to Central and Eastern Europe.

Prof. Macek did his postdocs at the Institute of Human Genetics in Berlin and at the McKusick-Nathans Centre for Genetic Medicine, Johns Hopkins University in Baltimore. During that time, he was also a fellow at Harvard School of Medicine in Boston. Prof. Macek is national coordinator of Orphanet, active member of Eurogentest, has been the chief advisor

of the Czech EU Council Presidency under which the "EU Council recommendation on an action in the field of rare diseases was adopted in June 2009. He also serves at the EUCERD committee on rare diseases and is involved in the rare disease-focused initiatives EURenOmics and RD-Action.

Cécile Wendling, Group Head of Foresight, AXA

Dr Cécile Wendling is head of foresight at Axa Group, where she conducts forward-looking analyses for the group on data, digital and cyber issues, among others. She is also associate researcher at the Centre de sociologie des organisations, a joint programme of the Sciences Po university, Paris and the CNRS (National Centre for Scientific Research). Her areas of research focus mainly on the sociology of risks, crisis management, foresight tools and methods.

Natasha Azzopardi-Muscat, President EUPHA

Natasha Azzopardi-Muscat, MD, MSc Public Health, MSc Health Services Management, PhD, FFPH, DLSHTM. Ms Azzopardi-Muscat is the President of the European Public Health Association (EUPHA).

She currently works as a consultant in public health medicine at the Directorate for Health Information and Research in Malta, having previously held various senior positions in the Ministry of Health in Malta, including that of Chief Medical Officer.

Since 1999 she has also been a resident academic at the University of Malta where she lectures on health systems and health policy. Her main research interests are European Union health policy and small state health systems. She has authored several publications on these topics.

Didier Schmitt, former Bureau of European Policy Advisers to the EC President, advisor on Foresight

Didier Schmitt currently works on space policy at the European External Action Service, the diplomatic service and foreign and defence ministry of the European Union. At the European Commission his first focus was on space policy, having worked in the Space Policy Unit at the European Commission from 2009–12. From 2012-2014 he was scientific adviser and foresight coordinator in the Bureau of European Policy Advisers to the President of the European Commission. He co-authored the first inter-institutional report of the European Union on future trends: "Global trends to 2030: Can the EU meet the challenges ahead."

Prior to that, at the European Space Agency he managed human and robotic exploration preparation programmes, including the use of the International Space Station (1997–2009). In his academic career, he was associate professor at the Toulouse medical school and the International Space University (1992–1997). His educational background is a PhD in biosciences in addition to being a certified medical doctor. Currently working in the European Union

diplomatic service, he is a regular opinion writer in mainstream French newspapers on future issues in science, technology and policy.

Philine Warnke
Fraunhofer Institute

Dr. Philine Warnke has been coordinating the Business Unit Futures Thinking and Dialogs in the Competence Center Foresight at Fraunhofer ISI since 2014.

After finishing her studies of mechanical engineering at the University of Essen, Philine Warnke completed her PhD within the interdisciplinary DFG postgraduate program "technology and society" at the University of Darmstadt in an STS (social science technology studies) framework. Since then, she has been active for more than a decade as a researcher in innovation studies, with a focus on socio-technical change and Foresight processes.

As project and team leader at Fraunhofer ISI, the "Institute for Prospective Studies" of the European Commission (JRC-IPTS) in Sevilla, Spain, and the Austrian Institute of Technology AIT in Vienna, Austria, she designed and implemented a number of Foresight processes in support to decision makers in policy, society and industry in Europe and beyond. Through many contributions to conferences, seminars, guidebooks and journals, she contributed to advancing and sharing insights on Foresight theory and practice.

Simon Kos
CMO Microsoft

Dr. Simon Kos joined Microsoft in 2010. He brings insight and experience from over 19 years working in the healthcare and Health IT industries. As the doctor on Microsoft's Worldwide Health team, he is responsible for clinical strategy and industry engagement. He raises awareness of the Microsoft brand, technologies and partner community, and works to find clinical relevance for Microsoft products. Prior to coming to Microsoft, Dr. Kos worked with global health IT companies Cerner and InterSystems as they implemented some of the largest e-Health initiatives in the Asia-Pacific region. His responsibilities included product design and localization, clinical engagement, industry thought leadership and team management. Before his career in health IT Dr. Kos practiced clinical medicine Australia for several years with a focus on critical care. Dr. Kos has an MBBS and BSc(Med) from UNSW, and an MBA with a major in change management from AGSM. He is passionate about improving healthcare through technology.

Kate Bushby,
Emeritus Prof of
Neuromuscular
Genetics Consultant
Clinical Geneticist

Professor Kate Bushby is a clinical academic with joint appointments between Newcastle University and the NHS. She is a member of the Neuromuscular Research Group within the Institute of Genetic Medicine and plays a leading role in the National Commissioning Group (NCG) for rare neuromuscular diseases. Professor Bushby has a long-standing interest in the molecular genetics of the limb-girdle muscular dystrophies and related disorders and is interested in the best possible development and implementation of care guidelines as well as clinical trials. Her team has developed an extensive programme of research in NMD from basic molecular pathology to clinical studies. In November 2014, Kate Bushby along with Volker Straub and Hanns Lochmüller launched the John Walton Muscular Dystrophy Research Centre. The Centre will focus on three key research areas: translational research, innovative clinical trials and international networking.

Prof Bushby has been actively involved in many European projects including being a founding co-ordinator of the TREAT-NMD Network of Excellence whose objective is to ensure that the most promising new therapies reach patients affected by neuromuscular diseases as quickly as possible.

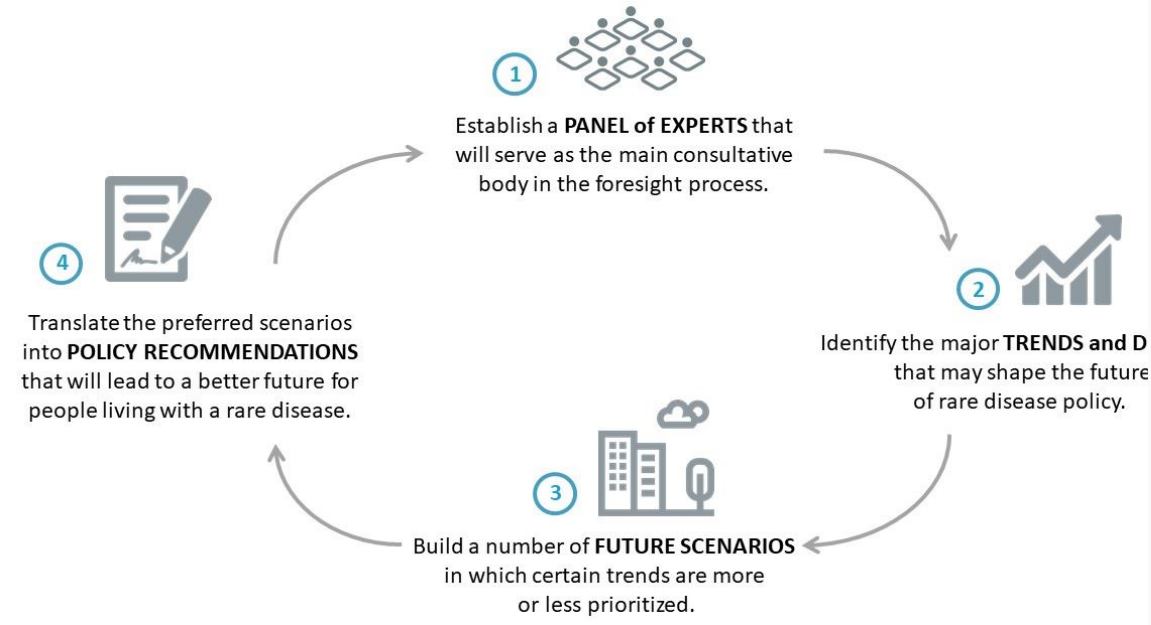
Kate has played a leading role in the European and national rare disease policy area, acting as vice chair on the European Union Committee of Experts on Rare Diseases (EUCERD) from 2010 to 2013, mandated to assist the European Commission in the implementation of rare disease activities in all member states, and the led the EUCERD Joint Action on Rare Diseases. She also acted in the capacity of expert on the Commission Expert Group on Rare Diseases until 2017 and was one of the key experts spearheading the development of European Reference Networks.

Formal Invitation

Dear XXX,

On behalf of EURORDIS-Rare Diseases Europe, I am pleased to invite you to participate as a member of **Research Advisory Board (Rare2030 RAB) in the Rare2030 project** - a participatory foresight study to reflect on the long-term future of people living with a rare disease in Europe. Your enthusiasm for the cause of rare diseases and your genuine commitment to making concrete changes in the lives of children living with rare diseases on a global scale have been instrumental to the success of XXX and would add great success and value of this forward-looking reflection. EURORDIS has been privileged with the responsibility of coordinating this project and, along with the following partners, to **provide policy recommendations on rare diseases through 2030 and beyond.**

The project is co-funded by the European Commission and will span two years (January 2019 - December 2020) with four key steps:



Your involvement

The Rare2030 RAB is a high-level advisory group that will act as a sounding board to the Executive Management Committee and the Management Board throughout the duration of the project.

In accepting this role, we look to your support in assessing the scientific value of the project achievements (are we using the right methods?), their innovative contents (do we consider related sectors, e.g. IT, artificial intelligence, social innovation?) and widely disseminating outcomes beyond the rare disease community (how does this fit within "mainstream" health policy?).

Specifically, as a member of the Rare2030 RAB you will be requested to:

- Participate in a 30-minute interview in the next three months (with 30 minutes preparation) to identify major trends that will shape the future of rare diseases
- Attend a mid-project Executive Management Committee meeting at the end of 2019
- Suggest members for the Rare2030 Panel of Experts
- Review two key deliverables of the project
- Review key impact indicators

You have been invited alongside a board of 11 other experts from the following institutions and backgrounds:

- Experts in Foresight methods

- Former Director Generals of DG RTD, DG SANTE and DG CONNECT
- Former Members of the European Commission Expert Group on Rare Diseases
- Private companies and foundations investing in rare diseases

The Terms of Reference further detailing the role and responsibilities of the Research Advisory Board will follow in a separate email. As a Rare2030 RAB member, your attendance will be supported for all Rare2030 project meetings you are able to attend.

We would greatly value your participation and would very much appreciate it if you could kindly let us know by **25 March** if you accept this invitation and guidelines.

Should you respond positively to this invitation, the next step will be to schedule a 30-minute interview in the coming months.

If you have any questions, please contact Anna Kole, our Rare 2030 Lead.
anna.kole@eurordis.org
+33 (0) 6 14 93 36 00

With my best regards,

Yann Le Cam
Chief Executive Officer
EURORDIS-Rare Diseases Europe

Terms of Reference

Rare2030 Research Advisory Board

(Rare2030 - RAB)

TERMS OF REFERENCE

About Rare2030

Since the adoption of the Council Recommendation on European Action in the field of Rare Diseases in 2009, we have seen tremendous progress in policies that improve the lives of people living with a rare disease in Europe. The European Union Committee of Experts on Rare Diseases (EUCERD) (2010-2013) and the Commission Expert Group on Rare Diseases (2014-2016) have subsequently assisted the European Commission in preparing and implementing community activities in the rare disease field through a number of recommendations.

In 2018, as a result of a Pilot Project adopted by the European Parliament, the European Commission (DG SANTE) issued a call for a 'Foresight Study' dedicated to planning the next ten years of rare disease policy. A consortium was established, led by EURORDIS, and commenced operations on 1st January 2019. The project will run until 31st December 2020. The project has been funded by the European Commission with the support of a consortium of 8 partners:

EURORDIS RARE DISEASES EUROPE — Non-profit alliance of rare disease patient organisations that work together to improve the lives of 30 million people living with a rare disease in Europe

orphanet Inserm — Information portal for rare diseases and orphan drugs

Newcastle University — John Walton Muscular Dystrophy Research Center: translational research to bring diagnosis, care and therapy to people with neuromuscular disease

FONDAZIONE Telethon — Non-profit organisation fostering research that leads to cures for rare genetic diseases

isinnova — Research institute working in the field of policy design, analysis and impact assessment with a particular expertise in forward looking methods

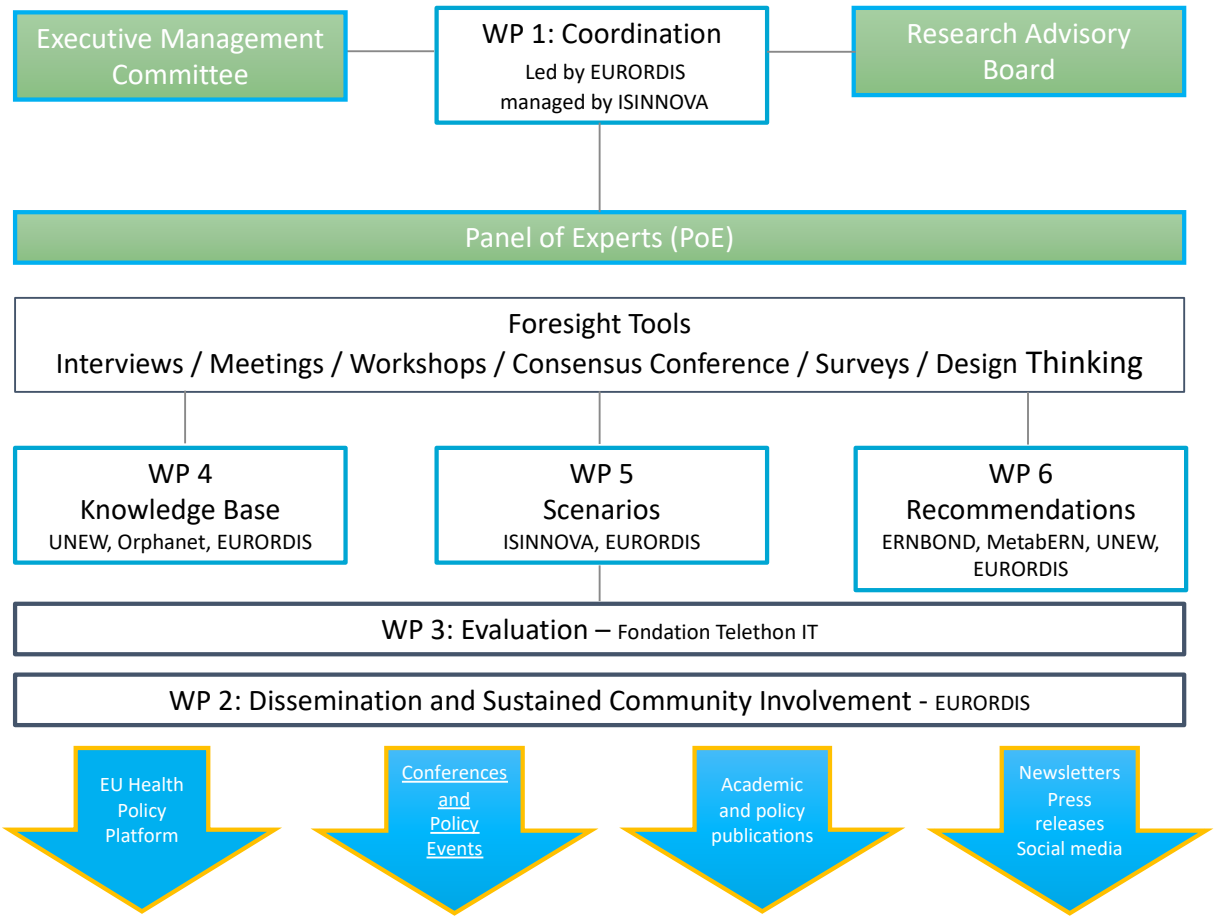
UNIVERSITÀ DEGLI STUDI DI UDINE — European Reference Network for Hereditary Metabolic Disorders (University of Udine)

European Reference Network | MetabERN

European Reference Network on Rare Bone Disorders (Istituto Ortopedico Rizzoli)

European Reference Network | Bone disorders (ERN BOND)

Imperial College London — Centre for Health Economics and Policy Innovation: assessing impacts of public policies on chronic diseases and other areas of health



The goal of the project is to employ innovative research-based methods of a foresight study to guide and support future policy decisions in the field of rare diseases. Trends and drivers of change for people with rare diseases -both 'traditional' i.e. well-known determinants such as organisation of care and 'wild card' factors- will be identified and ranked, to develop policy scenarios for the years leading up to 2030.

The partners will then use these scenarios to prepare recommendations to guide future policy by using the participatory foresight approach and additional innovative consensus-building methods, encouraging broad and sustainable uptake by patients, all relevant stakeholders, (in particular policy makers) and society at large. Each project partner plays a key role in the 4 stages of the foresight study. A panel of experts comprised of up to 250 members (from an exhaustive range of experts both in and peripheral to the field of rare diseases) serves as the primary consultative body in the Horizon Scanning and Scenario Building stages of the project.

The Rare2030 Research Advisory Board (Rare 2030 RAB)

The Rare2030 project will utilise the support and expertise of an approximately 10-member Research Advisory Board (Rare 2030 RAB) to act as a sounding board to the Executive Management Committee and the Management Board during the project's duration (January 2019 – December 2020) assessing the scientific value of the project achievements (are we using the right methods?), their innovative contents (do we consider related sectors such as IT, artificial intelligence, social innovation?), widely and promptly outcomes beyond the RD community (how does this fit with "mainstream" health policy?).

Objectives and responsibilities of the Rare2030 RAB

The Rare 2030 RAB is a high-level advisory group whose main contribution to the project is to share strategic input with the Executive Management Committee and Management Board of Rare2030 by:

- Monitoring the progress of the workplan implementation following initial validation of the evaluation plan, assess the value of the project achievements in terms of health policy and their innovative contents with respect to the European and international state-of-the-art, and suggest, as appropriate, re-orientations of specific tasks and methodologies;
- Contributing to the establishment of a network of specialists (Rare 2030 Panel of Experts) with the twofold objective of (i) ensuring a relevant level of debate on the main issues developed within Rare 2030, and (ii) ensuring that the advances achieved by Rare 2030 are widely and promptly disseminated within the greater health care and health policy context.
- Providing any additional strategic feedback that will best guide the methods and outcomes of the project.

Specifically, members of the Rare 2030 RAB will be requested to:

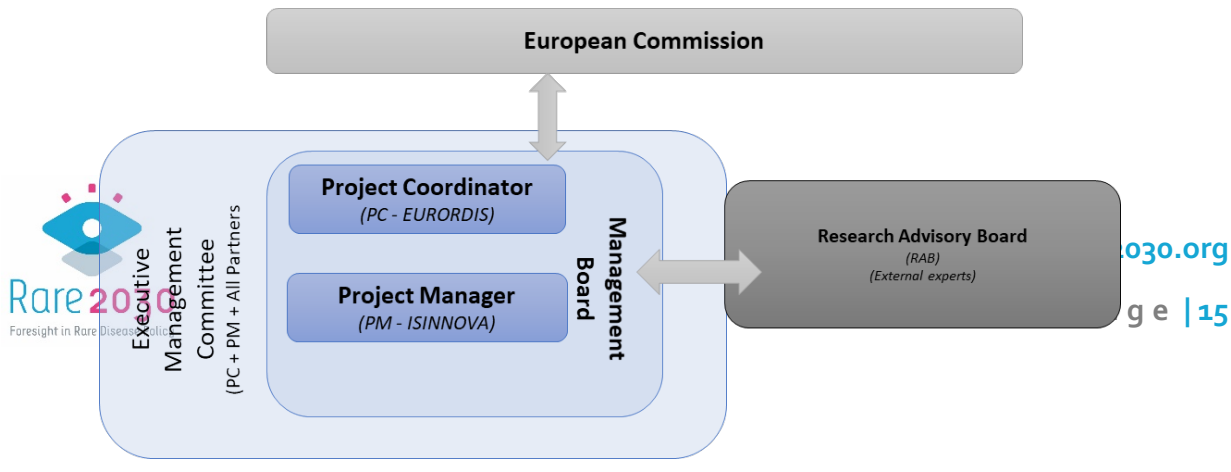
1. Participate in a 30-minute interview (with 30 minutes preparation) to identify major trends that will shape the future of rare diseases
2. Attend a mid-project Executive Management Committee meeting

3. Suggest members for the Rare2030 Panel of Experts
4. Review two key deliverables in the first two key stages of the project
    - During the Horizon Scanning stage, a list of key trends and drivers of change in the field of rare diseases will be identified via a thorough literature review, 10 interviews with key opinion leaders in the field, teleconferences with experts and webinars and workshops with patients and patient advocates. These trends and drivers will be prioritized and ranked to serve as the basis for the building of potential future scenarios in the care and treatment of people living with a rare disease. The RAB's review of these drivers will ensure that no important drivers or trends have been missed for consideration and that sound methods were used to arrive at a validated final list.
    - During the Scenario Building stage, the validated list of drivers and trends in the field of rare diseases will be used to create potential future scenarios of policy. Once again, the RAB will review scenarios to ensure that all trends and drivers were considered and that scenarios are consolidated and plausible.
5. Review key impact indicators

Rare 2030 RAB members will be welcome to attend additional events or review additional materials upon request. Communication will be maintained between the Management Board and the Rare 2030 RAB members by a members' only distribution list or individual telephone calls. The Rare 2030 RAB will be responsive to specific requests for advice, clarification and information from the Executive Management Committee and the Management Board of the project.

Structure and membership of the Rare 2030 RAB

Membership of the Rare 2030 RAB is voluntary and lasts for the entire duration of the project (2019-2020). It consists of approximately 10 members, balanced, to the extent possible, by background, location and gender, with long-standing or widely recognised excellence and expertise in their field: experts in the field of future studies, health-related policy or fields of research or cutting-edge industry that may have an impact on the future of people living with rare diseases. Members participate in their personal capacity. The Rare 2030 RAB is chaired by the project coordinator and will work closely with Executive Management Committee.



Outputs of the Rare2030 RAB

The Rare 2030 RAB's role will primarily be an advisory one. Any outputs will be part of the contractual periodic reports due to the EC. Nevertheless, Rare2030 RAB members may consider a number of benefits from their accepted role:

1. Contribution to a better future for people living with rare diseases and a strategic role in setting policy recommendations that lead to such a better future.
2. A visible role in joining the project partners, panel of experts and public in these efforts through the project website, social and traditional media channels and printed materials.
3. An opportunity to join publications of project process and outcomes either in peer reviewed journals or related gray literature.

Any questions?

Contact Anna Kole, Rare2030 Project Lead @ EURORDIS – Rare Diseases Europe

anna.kole@eurordis.org

+33 6 14 93 36 00

Responses to Date (31 March 2019)

Research Advisory Board

High level opinion leaders in foresight, innovative industries and health policy

Ruediger Krech
WHO

YES

Ruxandra Draghia-Akli
Merck
(foremerly DG RTD)

Robert Madeline
FIPRA
(foremerly DG SANTE, CONNECT)

YES

Milan Macek
Orphanet, Eurogentest, RD Connect

Cécile Wendling, AXA Group Public Affairs and Corporate Responsibility

YES

Didier Schmitt, former advisor to EC on Foresight

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Simon Kos
CMO Microsoft

YES

YES

Natacha Assopardi-Muscat
President EPHA

Kate Bushby
Former leader JA on Rare Diseases

YES

Philine Wanke
Fraunhofer Institute

EURORDIS
RARE DISEASES EUROPE

1