INDUSTRY, INNOVATION AND INFRASTRUCTURE

GOAL LEADERS:

Ruxandra Draghia-Akli, The Janssen Pharmaceutical Companies of Johnson and Johnson

Simone Boselli, EURORDIS

Hans-Georg Eichler, Association of Austrian Social Security Bodies

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With the support of:

Maurizio Scarpa, MetabERN

Anton Ussi, EATRIS

Goal description

Track E will address the challenge of building a cohesive rare disease health data ecosystem echoing the resilient infrastructure mentioned in SDG 9.

"Within the next 10 years, outcomes, actionable data should be routinely used to accelerate diagnosis, to guide care and management, to support better coordination of care, drug development, HTA and regulatory decision-making and inform health policy."

This vision for a seamless European rare disease health data ecosystem, as described in the Rare2030 Recommendations, envisages a cohesive data landscape where epidemiological, healthcare, research, quality of life and treatment-related data are shared and linked at a European and global level where possible. Sharing of data for different purposes is optimised across infrastructures and countries, relying upon commonly adopted codification systems (Orphanet nomenclature), harmonised standards and interoperability requirements. Under this vision, national data ecosystems are linked seamlessly to the European ecosystem via Findable, Accessible, Interoperable and Reusable (FAIR) data approaches.

Such an ecosystem, centred on robust European Reference Networks (ERNs) and by extension their clinical research capacities, the European Platform on Rare Disease Registration, and other key infrastructures would be well anchored within the European Health Data Space (EHDS), benefitting from this new framework for data sharing, while also contributing to build this space in a way that acknowledges the specificities of rare disease data and is responsive to the needs and expectations of the rare disease community.

So, how can we transform this vision into reality? Is the rare disease community ready to tap into the opportunity that the EHDS represents to address the challenges faced by people living with a rare disease? The answer: not yet.

Using the health data journey as a compass to structure the discussion, these two sessions will help to disentangle the myriad of initiatives and facilitate an honest discussion among stakeholders to identify what data are being collected; when and for what purposes each

stakeholder needs to use the data and what would be acceptable conditions to share the health data sets that each is collecting and curating.
How can we make sure that data collected and used upstream (e.g. research/diagnosis/care) can be re-used downstream (e.g. to improve the quality of care/therapeutic development/regulatory process/policy and HTA decision-making) and vice-versa, and how can this be orchestrated? What are the immediate measures to set the foundations of a cohesive European health data ecosystem for rare diseases that can benefit from and contribute to the larger EHDS?

Track F will address mainly the second part of SDG 9, hence innovation, Europe's attractiveness for investment and medicine development, as well as sustainability. The two main objectives are as follows:

- How can we keep Europe attractive for development of treatments and interventions for people living with a rare disease?
- Can Europe be attractive and sustainable at the same time?

To approach these questions, we will look at what is needed to fix framework issues if we continue to operate within the current ecosystem for treatment and interventions development and access for patients. What choices could we make if we would like to address the needs of the entire society based on solidarity and equity, as called upon by the Rare 2030 recommendations?

When looking at the medicine lifecycle from development to access, points to be tackled can be easily identified. The first is the so-called 'valley of death' which exists between the basic research and early stage of development, and the late stage of clinical trials. Indeed, this 'valley of death' is not only problematic in the field of rare diseases, but specific issues and ways of de-risking product development in rare diseases are acute and need to be urgently addressed. A major obstacle is linked to the challenges of clinical trials in small patient populations, on a methodological, operational and/or a financial level. This brings into perspective the importance of the Directive on Patients' Rights in Cross-Border Healthcare (CBHC), the directive for clinical trials and the various funding mechanisms (angel and private investor funds, public, private, and/or public-private partnerships and funding sources). By no means least is the fragmentation of the EU market: products are granted centralized European Marketing Authorisation, but pricing and reimbursement decisions taken at a national or regional level lead to inequalities in access and availability. The current revision of the EU Orphan Medicinal Product (OMP) regulation and of the general pharmaceutical framework is paving the way for some of these issues to be solved by the legislators, but it cannot fix everything. Innovative solutions must also be put forward on the R&D and access fronts in order to unlock the potential of currently disregarded diseases, to ensure the continuity of evidence generation and to allow the defragmentation of the marketplace, i.e. new models of collaboration and funding.

On top of these improvements, we need to reflect on what kind of society we want to live in and how people living with a rare disease want to see the society evolve while keeping rare disease at the top of the policy agenda. Building on the Rare 2030 recommendations and on

the principles of solidarity, equity and social justice, what political choices are needed to combine the EU's attractiveness for companies - ensuring that Europe is a good place for investment and innovation - and the resilience of the healthcare systems for orphan drugs expenditure, reduction of inequalities between countries and within rare diseases? How do we integrate effectively into our society the ground-breaking innovation that tackle the genetic root causes of the great majority of rare diseases?

The central question is: how can we align values across the development chain? From the decision maker's point of view, what criteria should preside within the distribution of resources for healthcare budgets; how do payers become buyers? On the developer's side, where do we stand with regards to ESG (Environmental, Social and Governmental) approaches in view of fulfilling the SDGs? What are the different options to work upstream on assets, fair price, value-based pricing, procurement, sustainable bonds and so on? What would it take to switch from profit-only-based approaches to approaches which bring societal and environmental benefits? Is B-Corp (or Benefit corporations) the way to go for companies? Should there be this type of indicator attached to this value-based decision making on pricing and reimbursement? These are the questions that we would like to discuss with you all!

Track E - The health data challenge: Building a Cohesive Rare Disease Health Data Ecosystem

Session 1: Thursday 30th June 2022, 14:00 – 15:30 CET

Building a seamless health data ecosystem

This session will provide a state of the art of the needs of rare diseases in the European Health Data Space, where are we now, where are we going and where do we want to go? How do we connect data to the wider eco-system to allow those who need it to be able to find it and access it when required? This session will explore the specific challenges and opportunities for the rare disease community. How can we orchestrate exchange and what key immediate actions are needed to build a seamless health data ecosystem?
For this, we will get a clear view of the roadmap that is designed in the EHDS (definition, scope, boundaries and timelines) and reflect as well on how it relates to the Members States level. We will then hear from the current pilot that is put in place for rare diseases and see which opportunities it brings for the whole community. Finally, we will get inspired by the COVID situation which seems to show us that the issues are not technical and that when there is willingness and budget, nothing is impossible - as per the subtitle of our conference, mission …possible.

Chaired by
Mark Hanauer, Co-Director and Director of Innovation Strategy, Orphanet

Speakers: To be confirmed

Session 2 : Thursday 30th June 2022, 16:00 – 17:30 CET

How to make the best use of ERNs & improve the efficiency of the whole system

This session will cover how we generate data, the amount of data generated, for what purposes and for whom. Some key questions are: How can we make better use of data to advance research on rare diseases? How to better enable real-world data (RWD) for legitimate research and decision making? How does GDPR affect research on rare diseases?

In this session, we will try to find answers to these questions while also debunking some myths related to data sharing. Different practical examples and initiatives on health data and real-world evidence will be presented. We'll identify the points of discussion, map actions needed and prioritise actions to build from.

The speakers are clinicians/researchers who are working hand-on with data. They will share their experience on how to generate data of good quality, how to organise the data collection better in the context of diagnosis, epidemiology, natural history and more. We also hear what conditions are needed to use or re-use the data, what types of data handling are needed, how the machines use the data (machine learning, AI, natural languages processes, algorithms, keywords). And finally what does that mean for healthcare professionals, how do we offer tutoring and training for the medical professionals and prepare for the digital doctors of 2030?

Chaired by:
Maurizio Scarpa, Coordinator of the European Reference Network for Hereditary Metabolic Diseases, MetabERN **.**

Speakers:
Prof. Ronald Cornet, Associate Professor, Department of Medical Informatics at Amsterdam Public Health Research Institute
Dr Sergi Beltran, Head of Bioinformatics Unit and Data Analysis Team at Centro Nacional de Análisis Genómico
Dr Francesco Cremonesi, Technical Lead, Datawizard
Prof. Ronan Fleming, Associate Professor, National University of Ireland, Galway, & Assistant Professor, Leiden University

Track F: Building resilient infrastructure, promoting inclusive and sustainable industry and fostering innovation for people living with a rare disease

Session 1: Thursday 30th June 2022, 14:00 – 15:30 CET

Making Europe attractive for therapies development for PLWRD- addressing the valley of death

This session will focus on addressing the 'technical efficiency' issues that exist in Europe, focusing specifically on the ongoing revision of the incentives framework, the implementation of the new regulation on HTA cooperation in Europe, and the possibility for more equitable access to authorised therapies provided by new models.
This session will be formatted as a series of three short moderated conversations between speakers to break down the opportunities and challenges laying ahead of us, preceded by an opening introduction by the Chair.

Chaired by:
Ruxandra Draghia Akli, Global Head, Global Public Health R&D at The Janssen Pharmaceutical Companies of Johnson & Johnson
Simone Boselli, Director of Public Affairs, EURORDIS

Speakers:
Avril Daly, Chief Executive Officer, Retina International
Others TBC

Session 2: Thursday 30th June 2022, 16:00 – 17:30 CET

Can Europe be attractive and sustainable at the same time?

This session will focus on addressing the 'allocative efficiency' or on how to distribute resources to rare diseases therapies within budgets. Furthermore, this session will look at the maturing concepts that are gaining momentum to ensure sustainability and affordability in Europe through ethical approaches (ESG indicators, benefit corporation, social pacts, etc.)
This session will begin with a keynote speech introducing the topic by the chair (Hans Georg Eichler) followed by a series of short presentation to highlight the concepts and progresses made in integrating new ways to address sustainability and accessibility of therapies for rare diseases

Chaired by
Hans-Georg Eichler, Consultant Physician, Association of Austrian Social Insurance Institutions

Virginie Hivert, Therapeutic Development Director, EURORDIS

Speakers:
Giacomo Chiesi, Head of Global Rare Diseases at the Chiesi Group
Yann Le Cam, CEO, EURORDIS
Momir Radulovic, Executive Director, Agency for Medicinal Products and Medical Devices of the Republic of Slovenia
Kasha Witkos, Senior Vice President and Head of International, Alnylam
Sarah Garner, Acting Program Manager - Access to Medicines and Health Products, World Health Organisation