GOOD HEALTH AND WELL-BEING

GOAL LEADERS:

Michela Gabaldo, Fondazione Telethon

Holm Graeßner, ERN RND and University of Tübingen

Victoria Hedley, Rare 2030 and University of Newcastle-upon-Tyne

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With the support of:

Matt Bolz-Johnson, EURORDIS

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Goal description

The 3rd Sustainable Development Goal to *ensure healthy lives and promote well-being for all people living with a rare disease at all* ages is closely linked to the concept of Universal Health Coverage (UHC). Achieving UHC is about having timely access to diagnosis as well as access to effective, responsive, safe and affordable care and treatments to ensure better health outcomes.

The rare disease community envisions a future where, with increased solidarity, countries will overcome healthcare fragmentation and inequalities in access, through interconnected and comprehensive health & social care systems that answer to the evolving needs faced by the 30 million people living with a rare disease in Europe.

While the Rare2030 Foresight Study provides the overall direction to achieve such a future in the next decade, the rare disease community still needs to define the specific organisational and institutional arrangements that will move us from the aspirational scenario depicted in these recommendations to transforming care delivery and achieving better health outcomes. This goal will on the hand, identify specific measures to strengthen health systems arrangements to address some of the wellknown access challenges faced by people living with a rare disease. On the other hand, it will address emerging challenges that call for a deeper transformation of our current health systems propose innovative solutions to tackle these.

The sessions under Track A will focus on well-known "access" challenges and explore immediate measures, low hanging fruits, to increase access to effective diagnostic tools, to highly specialised healthcare services and to allied treatments and interventions for all people living with a rare disease across Europe. Whereas Track B will be devoted to imagining new solutions for emerging challenges and to envision pan-European arrangements, under a future European Health Union, for the delivery of highly specialised healthcare for ultra-rare diseases, as well as the delivery of advanced therapy medicinal products (ATMPs).

Each track will feature a workshop showcasing good practices and evidence, followed by a policy formulation session workshop. The workshops and sessions will address jointly upstream (screening and diagnosis) and downstream (healthcare services and treatments) aspects to formulate policy options that consider as much as possible the interdependencies of upstream and downstream associated health services.

Other key components of UHC such as financial protection, well-being promotion and social inclusion will be addressed by the sessions under Goal 2 and Goal 3.

Track A: Strengthening national health systems to improve access to effective diagnostic testing technologies, care and treatments.

Session 1: Tuesday 28 June 2022, 14:00 – 15:30 CET

Good practice and evidence

This session will present evidence and good practice along the continuum of care to inspire short-term policy action at EU and/or national level. Participants will learn what are the main personal and external factors affecting patients' access to diagnosis, according to the findings of the latest Rare Barometer Voices survey on diagnosis. They will also discover how well-designed care pathways, that optimise access to rare disease Expert Centers, may improve patients' experience with care.

Several case studies will showcase how rare disease Expert Centres can arrange care continuity and coordination with social services and will present innovative approaches to improve access to allied therapies for people living with a rare disease.

Chaired by
Elizabeth Vroom, Chair World Duchenne Organization and EURORDIS Board member

Speakers
Sandra Courbier, Social Research Director - Rare Barometer Programme Lead, EURORDIS
Sanne Bouwman, Marketing and communication advisor, ParkinsonNet
Julie Vallortigara, Research Fellow, Department of Clinical and Movement Neurosciences, University College London
Vinciane Quoidbach, European Brain Council
Johanna Blom, Physiotherapist, Neurology Department of the University Hospital of Skåne in Malmö, Sweden
Monika Benson, Executive Director, Dystonia Europe
Eduard Pellicer, Social Worker - Chronic Complex Patients, Hospital Sant Joan de Déu, Barcelona

Session 2: Tuesday 28 June 2022, 16:00 – 17:30 CET

Policy formulation session

This session will build on the experiences and insights shared in Session 1 to propose concrete policy recommendations to improve access to i) effective diagnostic technologies and services, ii) highly specialised healthcare and iii) allied health and therapy services.

Speakers will identify specific measures to remove the barriers to access diagnostic technologies for rare diseases and strengthen health systems capacities to treat the increasing number of people that are being diagnosed. The panel will also discuss policy and organisational measures to optimise the design of care pathways to accelerate access to expert care as well as ways to ensure continuous and timely access to allied health and therapy services.

Chaired by
Holm Graessner, Head Research Management Unit, COO Centre for Rare Diseases Tübingen & Coördinator, ERN-RND.
Elizabeth Vroom, Chair World Duchenne Organization and EURORDIS Board member

Speakers
Laurence Faivre, Professor at the University of Bourgogne, Hospital Practitioner
Hortensia Gimeno, Occupational Therapist, National Health Service of United Kingdom
Holm Graessner, Head Research Management Unit, COO Centre for Rare Diseases Tübingen & Coördinator, ERN-RND.
Eileen Treacy, Clinical Lead, National Rare Diseases Office

Track B: Inventing innovative health systems arrangements to address new and emerging challenges.

Session 1: Tuesday 28 June 2022, 14:00 – 15:30 CET

Good practices and evidence

This session will showcase a range of good practices in the broad area of 'innovation across the care pathway', starting from accelerated applications of genomic technologies, expansion of newborn screening panels for actionable diseases, and integration of diagnostic approaches for undiagnosed cases, all of which should serve to facilitate an accurate and timely diagnosis and offer the opportunity for all PLWRD to be fast-tracked on an effective pathway.

Larger populations of accurately diagnosed individuals must be accompanied by equally innovative approaches to 'open the door' to curative and transformative therapies; therefore, this session will also consider innovation in health systems to deliver and access highly specialised healthcare services for very rare diseases, intended to speed up the rate at which innovative therapies become accessible to the rare disease community.

Chaired by
Victoria Hedley, Policy Officer, RARE 2030 Foresight Study & University of NewcastleuponTyne.

Speakers
Michela Gabaldo, Head, Translational Project Management & Regulatory Affairs at Fondazione Telethon.
Mark Briggs, Assistant Director of Innovation, Cardiff and Vale University Health Board, United Kingdom
Marie-Christine Ouillade, Board of Directors, AFM-Téléthon & SMA Europe
Tudor Groza, Phenomics Team Lead, European Bioinformatics Institute
Olaf Riess, Head of the Institute of Medical Genetics and Applied Genomics

Session 2: Tuesday 28 June 2022, 16:00 – 17:30 CET

Policy formulation session

'Healthy lives' and greater wellbeing for people living with rare diseases is only possible through a coordinated policy approach which unlocks the potential of innovation in multiple domains, including digital systems, genomic technologies, health systems infrastructure, manufacturing processes, workforce development, knowledge building, and sharing data. Besides spanning multiple disciplines, policies to support innovation in rare disease health systems must often also span many geographical borders.

Building on the good practices and insights shared in Session 1, a panel of experts will discuss policy opportunities to scale-up innovation to ultimately increase life expectancy and improve quality of life for the 30 million Europeans affected by a rare disease or rare cancer (as opposed to the few).

Recognising the need for concerted action for rare disease, the session will discuss some of the core building blocks for a new strategic policy framework for rare diseases, identifying where innovation can be better leveraged to allow EU citizens to benefit from innovative and curative therapies. The pros and cons -and feasibility- of different approaches will be discussed, by considering underdeveloped tools, programmes and approaches which already exist, whilst contemplating what radical future policies might look like.

The session will explore if the existing health legal and institutional framework would allow the flexibility to keep pace of innovation developments as well as fleshing out greater informal or formal collaboration across the European Union, centralising planning and organisations of services, taking a whole-system approach to accelerate adoption and access of innovation into frontline services, access to innovation to all.

Chaired by

Michela Gabaldo, Head, Translational Project Management & Regulatory Affairs at Fondazione Telethon.

Speakers

Nick Meade, Joint Interim Chief Executive & Policy Director, Genetic Alliance UK

Eva Schoeters, Director, RaDiOrg- Rare Disease Organisation Belgium

Giancarlo La Marca, Deputy Head of the Newborn Screening, Clinical Chemistry and Pharmacology Laboratory, Meyer Children's Hospital, Italy

Wendy van Zelst-Stams, Clinical Geneticist, Radboud University Medical Centre, Netherlands

Victoria Hedley, Rare Disease Policy Manager, Newcastle University