# THEME 3

# SHARE, CARE, CURE: TRANSFORMING CARE FOR RARE DISEASES BY 2030

**THEME LEADERS:**

**Prof. Alberto Pereira**, Coordinator of the European Reference Network on Rare Endocrine Conditions (Endo-ERN) & Leiden University Medical Centre, The Netherlands.

**Dr. Birute Tumiene**, Clinical Geneticist, Coordinator for Competence Centres, Vilnius University Hospital Santaros Clinics, Lithuania.

**Matt Bolz-Johnson**, ERN and Healthcare Advisor, EURORDIS

**THEME SUPPORT :**

**Ines Hernando**, ERN and Healthcare Director, EURORDIS

## THEME DESCRIPTION:

Fast forward 20 years, the very fabric of our national health and welfare systems will be unrecognisable, transformed by the disruptive innovation and technologies of our modern age. Even today, many of our everyday services have already been radically transformed - private hire car companies now don't own any cars (Uber), retailers without shops (Amazon) and mobile-only banks (N26) are now common place. Our healthcare and welfare systems will not be immune to these changes. The rule book for healthcare will be re-written, and the once familiar building blocks of our traditional hospitals will be transformed into a new suite of health and social care competencies, on-the-go and on-demand experts and virtual clinics.

The digital transformation of healthcare has the potential to bring great benefits to the rare disease community, but it will also create new challenges. Our thirst for knowledge and unwavering scientific advancements will conscribe the diagnostic odyssey to our history books, with most rare diseases being diagnosed at birth or within the first year of the first symptom – all culminating to improve health outcomes, and resulting in a shift in the population needs and burden of care of an ageing rare disease community, facing different challenges of living longer with increased multiple co-morbidities. This survivorship effect will translate into a shift in our demand for different health and social services and service competencies. Adoption of technology, smart sciences and increasing automated health will force an evolution in the role of both health professionals and patients. The line between health care, social care and research will become increasingly blurred, as will our expectations of how care should be delivered.

Theme 3: Share, Care, Rare 2030: transforming care for rare diseases will explore the rare disease population needs in 2030-40 and explore both the opportunities and challenges of the care provision of the future. The seeds of our future are already visible today: the five sessions will put a spotlight on the emerging trends in best practice, promising technologies and cutting-edge thinking; showcase the forward-looking services and their potential to be scaled-up; and transform the way in which our healthcare will be delivered.

**SESSION 0301:** Thursday 14 May 2020, 14:45 – 16:15

## "Live longer, healthier lives": Rare Disease Population Needs 2030 (and beyond)

*Session 1: RD Population Needs 2030 (and beyond)* will present the emerging policy trends and map the future population needs of the rare disease population, as well as scenario planning on the changing demographics, health inequalities modelling, horizon scanning on the availability of evidence-based services and treatments, and pinpointing the shift in the burden on care for an ageing population and the effect of increased survival. These emerging trends will shape healthcare, hospital systems and the integration of health and social care, for the next decade and beyond.

This session will present a high-level narrative on the four 'dimensions' of our future healthcare and hospital systems, specifically:

- Healthcare provision under mature ERNs (structure)
- Organisation of care under healthcare digital pathways (processes)
- Medical advancements and technology (innovation)
- Changes in role, profile and competency of medical teams and patient community (human resources)

**Chairs: Prof. Kate Bushby**, Institute of Genetic Medicine & Emeritus Professor, Newcastle University, UK

**Speakers:**

**Prof. Alberto Pereira**, ENDO-ERN Coordinator; Head of the Division of Endocrinology, Chair Centre for Endocrine Tumors Leiden (CETL), Leiden University Medical Centre, The Netherlands

**Dr. Birute Tumiene**, Institute of Biomedical Sciences, Faculty of Medicine, Vilnius University, Lithuania

**Dr. Dalia Aminoff**, Head of Patient Organisation, , AIMAR Onlus, Italy

**Dr. Enrique Terol,** Senior Policy Officer, Seconded National Expert, European Commission, Belgium

**Victoria Hedley,** Newcastle University John Walton Muscular Dystrophy Research Centre, UK

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**SESSION 0302:** Thursday 14th May 2020, 16:45 – 18:15

## ERN & CoE Accreditation as Quality Improvement Framework

The EUCERD Recommendations published in 2011 on the organisation of highly specialised healthcare were and remain ambitious. Even today, they continue to be relevant and far-reaching, with many countries still only beginning the process of implementing them. Session 2 will explore these key recommendations and conduct a deep dive into national recognition of expert centres and how European Reference Networks are developing.

Identification of experts in each and every Member State (MS) is the first step towards securing an accurate diagnosis and accessing appropriate care. National processes for endorsing rare disease expert centres continue to be developed across EU MS. Endorsement and accreditation is a dynamic quality improvement process that incrementally raises the quality thresholds services need to meet to be

approved. The future trend will be two-fold - universal coverage of national accreditation of highly specialised healthcare and rare diseases centres, and the incremental step-wise maturing of the accreditation process to come to a final result that will be measured on treatment outcome.

What will ERNs look like in 10-20 years' time? Session 2 will present the opportunities, benefits and challenges foreseen in a maturity ERN System. ERNs won't mature in isolation, but need to be fully integrated into national health systems, see recent Statement of the ERN Board of Member States on Integration of the ERNs to the healthcare systems of Member States.

The need for a more robust and universal care coordination across EU-ERN-wide care pathways, that are supported by shared care arrangements between hospitals as well as between health and social care. Future sustainability of many hospital systems on creating a fine balance between centralisation of supra-specialist care and shared care arrangements for local access, where the expertise travels, not the patient.

**Chair: Dr. Birute Tumiene**, Institute of Biomedical Sciences, Faculty of Medicine, Vilnius University, Lithuania

**Speakers:**

**Prof. Alberto Pereira**, ENDO-ERN Coordinator; Head of the Division of Endocrinology, Chair Centre for Endocrine Tumors Leiden (CETL), Leiden University Medical Centre, The Netherlands

**Anke Widenmann-Grolig**, KEKS & EAT, Germany

**Prof. Till Voigtländer**, Austrian Representative on ERN Board of Member States & Medical University of Vienna, Austria

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**SESSION 0303:** Friday 15th May 2020, 11: 30 – 13:00

## Clinic of the Future & Digital Care Pathways

Session 3: Clinic of the Future & Digital Care Pathways will draw together the building blocks of the 'clinic of the future' and its clinical model, where research is fully embedded in daily clinical care; and its interface with other services along 'digital care pathways'. What will care look like under the clinic of the future? How will it feel to progress along the future 'digital care pathways'?

The healthcare we receive will be connected as 'networks of networks', beyond the traditional hospital building, to wider EU and global infrastructures. Centres of Expertise will act as comprise crucial hubs: they provide quality standards and connections with all stakeholders in the national network, and they are connected to EU-wide networks (for the further development of standards and implementation of all the activities, where ERNs provide economies of speed, scale and scope).

EU Green Corridors will connect hospitals across Europe, through digital healthcare pathways that are rooted in the latest evidence-based practice and support timely access, giving the 'green light' for people with a diagnosed rare disease to access the services when they need it. These healthcare pathways will provide transparency of care quality standards and centralize care only when necessary, but also make it close to home whenever possible.

Will we have witnessed the full impact of the genetic revolution on screening, surveillance, diagnosis and personalised treatment and hold real-world experience of gene therapy and genomic editing in the next

10-20 years? The two worlds of healthcare and research will be fully integrated in our clinic of the future, as we see happening today in a few countries, enabling undiagnosed rare disease patients to be fast track to research setting for an accurate diagnosis and advancing research with real-world evidence. Despite all our hopes and the pace of scientific development, there will always be some RD without specific treatment or for which the symptomatic treatments do not minimise all the complex impairments generated by the disease. For this reason, evidence-based clinical guidelines must be ensured for every disease, as well as coordinated care between health and social care.

**Chair: Prof. Alberto Pereira**, ENDO-ERN Coordinator; Head of the Division of Endocrinology, Chair Centre for Endocrine Tumors Leiden (CETL), Leiden University Medical Centre, The Netherlands

**Speakers:**

**Prof. Dr. Daniel Hommes**, Leiden University Medical Centre, The Netherlands

**Dr. Sofia Douzgou**, ESHG Representative, Manchester Centre for Genomic Medicine, UK

**Prof. Dr. Helge Hebestreit**, Universitäts-klinikum Würzburg, Germany

**Dr. Valter Fonseca,** Director of the Department for Quality in Health, Ministry of Health, Portugal

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**SESSION 0304:** Friday 15th May 2020, 14:00 – 15:30

## Addressing Health Workforce Challenges and training the New Generation of Rare Disease Experts

Advancement in healthcare innovation and technology will lead the way in changing the competency profiles and skill mix of the healthcare workforce. The accessibility of medical information online and private online healthcare provision has already changed the doctor-patient relationship, with patients being more informed than ever, while the volume of new research and changing knowledge that healthcare professionals need to digest seems exponential. How far are we from seeing the role of doctors and surgeons being made redundant, where automated systems and artificial intelligence will have replaced them in traditional healthcare? Many may argue that the family doctor has already been made redundant thanks to this online medical revolution.

This has been the longstanding modus operandi for those living with a rare disease because they are experts in their condition. A single family doctor has never been their primary source for information. With increased health literacy, wearable technology, and direct-to-consumer genetic testing, more people than ever are taking their health into their own hands. However, what are the risks of over-relying on technology and losing the relationship with a healthcare professional? With the changing role of patients there is sure to be a change ahead for healthcare professionals. Are generalists obsolete or do they just need to be given the tools to evolve? What are the skills that our next generation of healthcare professionals need to adjust to this new environment and complement innovation?

For those with rare diseases there cannot be any doubt that greater inter-connectivity and patient power has been beneficial. However there is potentially a fly in the ointment - there's a difference between a patient who's done a rudimentary google search about some transient symptoms and an expert patient who is one of a handful with a specific condition in the country. How will doctors be able to distinguish between the two when they come to them holding out printed information to be read and understood in a 10-minute appointment? Is the term "expert patient" being devalued?

Will the breaking down of our traditional healthcare systems see a similar overhaul of the medical training system underpinning them? Not just moving from classroom-based education to knowledge-sharing online communities, but performing a review of what is being taught and how much emphasis is being placed on it. For example, should physical examinations still include the search for late-onset stigmata of diseases that should be caught earlier in their natural history with standard diagnostic tests? What other signs could replace these that would be more informative?

Our next generation of experts are today's medical students and doctors in training. Given the changing patient and doctor roles, what skills do future doctors require in order to deliver evidence-based and compassionate care? What will the world look like when they graduate in 10 years' time and beyond? Is current medical education moving fast enough that what a medical student has learned during their studies is irrelevant by the time they graduate? What skills and knowledge will they need to learn to work in collaboration with innovative health solutions?

People with rare diseases often share their stories about disbelieving and unsympathetic doctors. Perhaps the changing role of doctors needs to be dominated more by what has been coined as 'soft skills' such as communication skills, interpersonal skills and leadership skills, leaving the pattern matching to the machines. After all, technology is only ever as good as the information you put into it. Obtaining a thorough and accurate history, while making the patient feel listened to and at ease, is an art that needs a lot of practice.

Focusing on staffing the medical community of the future, where are the gaps in the workforce and is the ever-continuing supra-specialisation creating an imbalance in our medical workforce? What is the appropriate workforce skill-mix in an individual nation and across Europe? We need to plan today in order to have the workforce in place that will address the needs of the patient population in ten years' time. Many countries are facing a brain-drain - how can we preserve our expertise, knowledge and experience? How can we get better at sharing knowledge and expertise and finding new ways of collaborating to provide cross-border healthcare?

Potentially the changing role of the patient and healthcare innovation could work favourably to make up for challenges caused by gaps in the workforce. However, as experienced by many rare diseases, innovation doesn't always mean change and can be hindered by evolutionary lag in training and system updates. As we look to 2030 we discuss how best to ready the workforce so that the full potential of innovation can be realised.

**Chairs: Dr. Lucy McKay**, CEO, Medics 4 Rare Diseases, UK

**Speakers:**

**Prof. James Buchan,** Queen Margaret University Edinburgh, UK

**June Rogers,** Paediatric Continence Specialist, Bladder & Bowel, UK

**Dr. Svetlana Lagercrantz,** Chair of the ERN GENTURIS Task Force on Education & Training, Sweden