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 for the rare disease community, but 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 aging 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 and so 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, and showcase the forward-looking services and their potential to be scaled-up and transform the way our healthcare will be delivered.
SESSION 0301: Friday 15th May 2020, 14:00 – 15:30

“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, pinpointing the shift in the burden on care for an aging 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 provide under mature ERNs (structure)
- Organisation of care under healthcare digital pathways – (processes)
- Medical advancements and technology (innovation)
- Change in role, profile and competency of medical teams and patient community (human resources)

Sessions 2-5 will conduct a deep-dive exercise to explore the opportunities and challenges of our forecasting in each of these dimensions.

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

Speakers:

Dr. Lennart Christiansson, Representative from Sweden and Chair, ERN Board of Member States

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

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. Enrique Terol, Seconded National Expert, Policy Officer, Directorate-General for Health and Food Safety, European Commission

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

Dr. Dalia Aminoff, President, AIMAR Onlus, Italy

SESSION 0302: Friday 15th May 2020, 16:30 – 18:00

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 endorsement of centres of expertise 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 process 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 finally be based on benchmarking on treatment outcome, like in the Swedish model.

What will ERNs look like in 10-20 years time? Session 2 will present the opportunities, benefits and challenges foreseen in a mature ERN System. ERNs will not sit in splendid isolation, but will need to be fully integrated into national health systems. Future sustainability of many hospital systems need to strike a balance between centralisation of supra-specialist care and shared care arrangements for local access, where the expertise travels, not the patient. What are the opportunities and value foreseen of ERNs integrating and complimenting with national health systems and what are the expectations from a national perspective?

Chair: Dr. Lennart Christiansson, Representative from Sweden and Chair, ERN Board of Member States

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

SESSION 03: Saturday 16th May 2020, 09:00 – 10:30

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, give 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 centralizing care only when necessary, but also 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:
Dr. Sofia Douzgou, ESHG Representative, Manchester Centre for Genomic Medicine, UK

SESSION 0304: Saturday 16th May 2020, 11:00 – 12:30

Driving Innovation and Codification of Knowledge

Systems are inclined to self-perversive. The speed of technological development outpaces organisations’ ability to adapt. Innovation and technological advancement, whether incremental or radical, is the one constant in our everyday lives. Healthcare systems’ ability to adapt to this ‘force of nature’ of our modern age is currently the rate-limiting factor to unlocking its potential to tackle our burden of needs. Harnessing innovation in front-line services is set to be one of the new service competencies of our future care services, instead of shying away from the unknown. Innovation in its broadest sense – innovative technology, workforce and knowledge - will all shape both the environment and the services we receive. Affordable technologies hold the promise to successfully target health inequalities through digital applications and devices, and tele-medicine aiding timely access to expertise and advice.

Session 4: Driving Innovation will explore the different models of innovation and test the use-ability, applicability and effectiveness of emerging products and interfaces in the health ecosystem. The seeds of our future are already here, waiting only to be scaled up. This session will draw a future vision of care delivery through existing examples of promising innovation.

Ask someone in the 1970s about innovation, and they would say: yes, it is at hand. Innovation is not something novel. Some very “innovative” things of the 21st century (including AI) were created several decades ago. What is novel is the rate of R&I development and the challenges - some novel, some old - that innovation creates. Hence, the main task here would be not “how to create innovation“, but “how to create a fertile ecosystem for innovation adoption“.

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

Speakers:
Prof. Olaf Riess, Head of the Institute of Medical Genetics and Applies Genomics, University Hospital Tübingen, Germany
Addressing Health Workforce Challenges and training the New Generation of Rare Disease Experts

Advancement in healthcare innovation and technology will lead the way in rebalancing the competency profiles and skill mix of the healthcare workforce. How far off are we, to seeing the role of the doctor and surgeon being made redundant, where automated systems, artificial intelligence, robotic surgery has replaced them in traditional healthcare? What will be lost and what will remain? Are we already over-reliant on technology and innovation? What are the roles and skills we need our next generation of healthcare professionals and experts to develop?

With the changing role of experts, there will be also a changing role of patients, increase health literacy to navigate complex healthcare systems, to be informed and take an active role and responsibility in their own decision-making.

Will the breaking down of our traditional healthcare systems see a similar overhaul of the underpinning medical training system, moving from classroom based education to knowledge sharing online communities?

Our next generation of experts are today’s medical students and doctors in training. What skills do they need to deliver evidence-based and compassionate care? What will the world look like when they graduate in 10 years’ time and beyond? Is there enough understanding of the future health needs to change the curricula now? What areas of knowledge and soft skills should be included in today’s medical curriculum? Are there gaps in key professionals for our future healthcare systems or is the ever-continuing supra-specialisation creating an imbalance of our medical workforce? What is the appropriate workforce skill-mix? Can health systems protect clinicians time for many of the tasks around data analysis? How do we preserve our expertise, knowledge and experience from the brain-drain many of our countries are facing? How do we attract new experts? Several more questions: it is just not possible to foresee many of the innovations and skills that await us in the future; hence, the aim is how to educate open-minded, innovation and novel challenges-ready medical staff?

Chair: Dr. Lucy McKay, CEO, Medics 4 Rare Diseases

Speakers:

June Rogers, Bladder & Bowel UK

Prof. James Buchan, Queen Margaret University Edinburgh, UK