

THEME 1

THE FUTURE OF DIAGNOSIS: NEW HOPES, PROMISES AND CHALLENGES

THEME LEADERS:

Prof. Christine Patch, Clinical Lead for Genetic Counselling, Genomics England, UK

Virginie Bros-Facer, Scientific Director, EURORDIS

THEME SUPPORT :

Gulcin Gumus, Research and Policy Project Manager, EURORDIS

THEME DESCRIPTION:

Recent scientific and technological developments have meant that the diagnosis of rare diseases has improved considerably over the last few years.

In this theme we will closely examine the current landscape and also debate future trends and scenarios. We will present the current state of play in several national Newborn Screening (NBS) programmes, and discuss challenges to expanding NBS across Europe, highlighting impacts for patients and families. This theme will also explore how new technologies can be applied to accelerate and improve access to diagnosis, taking into account the implications, opportunities and challenges that are associated with Next Generation Sequencing and Artificial Intelligence by showcasing several platforms. The diagnostic odyssey is still very much a reality for a vast number of rare disease patients despite these recent technological advances. Relevant tools and services will be discussed to understand how to better support the undiagnosed rare disease community.

Genetic counselling represents a critical milestone in the search for a diagnosis and is integral to Genetic Health Services. A dedicated session will present how partnerships and innovative ways of working can benefit all involved and improve care delivery.

SESSION 0101: Thursday 14th May 2020, 14:45 - 16:15

Rare 2030: How can we achieve faster and more accurate diagnosis?

We anticipate that the future will hold a shortened diagnostic odyssey. A number of advances in technology - such as whole genome sequencing as a first line practice (bringing it into the clinic) - present new opportunities to achieve this.

The future of diagnostics will include new trends: Big data and AI capabilities; New innovation such as WGS in the clinic; Patient engagement in the diagnostic process; Wearable technologies; Data platforms integrating many data sources (genetic, phenotypic etc.)

And old drivers of change: Continued raising of awareness; Networking of health care professionals for more efficient diagnosis (e.g. CPMS type system).

Chair: Prof. Milan Macek, Department of Molecular Genetics and National Cystic Fibrosis Centre, Motol University Hospital and Charles University, Prague

Speakers:

Anne-Sophie Chalandon, Head of European Rare Diseases Public Affairs and Patient Advocacy, Sanofi Genzyme, France

Dr. Lucy McKay, Chief Executive Office, Medics 4 Rare Diseases, UK

Dr. Lucy Raymond, Addenbrooke's Hospital & University of Cambridge, UK

Julian Isla, Data and Artificial Intelligence Resource Manager, Microsoft; Dravet Syndrome European Federation (DSEF); Founder, Fondation 29, Spain

SESSION 0102: Thursday 14th May 2020, 16:45 – 18:15

Newborn Screening: Now and in the Future

The session will compare differing national approaches and explore the limits and potential of current and future approaches to newborn screening, exploring technical, societal, ethical and scientific considerations.

Chair: Jayne Spink, CEO, Genetic Alliance UK

Speakers:

Dr. Richard Scott, Clinical Lead for Rare Diseases, Genomics England

Nick Meade, Director of Policy, Genetic Alliance UK

Sara Hunt, Chief Executive Officer, Alex TLC

Prof. Martina Cornel, Amsterdam University Medical Centre, The Netherlands

Simona Bellagambi, UNIAMO (Italian Federation for Rare Diseases), Italy

SESSION 0103: Friday 15th May 2020, 11:30 – 13:00

Diagnosing Undiagnosed Rare Disease Patients: Tools and Resources to strengthen the voice of the undiagnosed Rare Disease Community

Progress in the application of genomic and other technologies (including web-based), has increased the diagnostic rate of patients with rare disorders to 50%. This is a great success but still leaves unanswered questions for the other 50% of the rare disease community. This session will focus on providing updates on existing initiatives of interest to the undiagnosed rare disease community, including patients, families and healthcare professionals.

The first part focuses on the views and voices of the undiagnosed community, their expectations of and outlooks on rare patients and their families. The overall aims of the session are to empower the community with tools and resources to strengthen their voices alongside policy makers and researchers, and to support them in getting closer to finding a diagnosis.

Chair: Dr. Holm Graessner, Solve-RD and ERN-RND, Institute of Medical Genetics and Applied Genomics, University of Tübingen, Germany

Speakers:

Prof. Christine Patch, Clinical Lead for Genetic Counselling, Genomics England

Dr. Holm Graessner, Solve-RD and ERN-RND, Institute of Medical Genetics and Applied Genomics, University of Tübingen, Germany

Lauren Roberts, Director of Support, Genetic Alliance UK, SWAN UK

Prof. Olaf Riess, Head of the Institute of Medical Genetics and Applied Genomics, University of Tübingen, Germany

Stephanie Broley, Senior Genetic Counsellor and Program Coordinator of the Undiagnosed Diseases Program WA, Australia

Dr. Pablo Botas, Head of Science, Fondation 29, Spain

Prof. Peter Krawitz, Institute for Genomic Statistics and Bioinformatics, University Hospital Bonn, Germany

Vanessa Lemarié, Lead Rare Disease Initiative, Business Development Life Sciences at Ada Health, Germany

SESSION 0104: Friday 15th May 2020, 14:00 – 15:30

What's Next After the Search for a Diagnosis? The Future of Specialised Health Services

This session will explore what happens after the completion of genetic analysis from the perspective of patients and health professionals. The session aims to explore what is important to patients regarding their potential diagnosis (or lack thereof) and future care pathways; to discuss innovative ways of working

with health professionals and patients to develop interventions which support these care pathways; and to appreciate the developing role of networks in the delivery of new approaches to aspects of care for rare diseases

Chair: Prof. Christine Patch, Clinical Lead for Genetic Counselling, Genomics England

Speakers :

Dr. Alessia Costa, King's College London, UK

Prof. Glenn Robert, King's College London, UK

Dr. Holm Graessner, Solve-RD and ERN-RND, Institute of Medical Genetics and Applied Genomics, University of Tübingen, Germany

Dr. Vera Frankova, Univerzita Karlova

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

