THEME 1

STRUCTURING THE RESEARCH & DIAGNOSIS LANDSCAPE

THEME LEADERS:

Daria Julkowska, Scientific Coordinator, E-Rare, France

Lauren Roberts, Director of Support, Genetic Alliance UK, National Coordinator, Swan UK, UK

EURORDIS SUPPORT:

Virginie Bros-Facer, Scientific Director, EURORDIS-Rare Diseases Europe

THEME DESCRIPTION

In the last few years, the research and diagnosis landscape has changed significantly in the field of rare diseases. Integration of new technologies in healthcare, and increased connection between research and care has opened up new possibilities for faster diagnosis and treatment. Acknowledging the patient as a key actor in their own health and putting them at the centre is strongly contributing to these tangible benefits. Encouraged by collaborative achievements of rare diseases stakeholders, the IRDiRC has published new, more ambitious goals and Europe is at the point of launching an integrative joint programming rare diseases initiative. But are we yet close to a fully collaborative and effective ecosystem that can provide all rare disease patients a diagnosis within a year?

The "STRUCTURING THE RESEARCH AND DIAGNOSTIC LANDSCAPE" theme will explore how we can exploit current achievements in genomics, how to prepare for new developments on the horizon and how to ensure no patients are left behind.

Starting by exploring how recent advances in research have transformed diagnostic pathways, we will also examine the potential challenges associated with new technology enabling self-diagnosis and consider how we support those patients for who, despite these all innovations, their condition is likely to remain undiagnosed.

Assuming cooperation between patients, clinicians, researchers and sponsors to be the bedrock upon which successful research occurs, the next sessions will investigate what is required to aid this collaboration. Sessions two and three will explore recent, innovative schemes of co-design and funding, how to carry out research that profits all stakeholders and provide examples of how to attract investment. In session four we will challenge the idea of whether it is enough for a patient to simply be 'an expert by experience' and consider what skills and experience is required for them to truly be respected, equal partners.

In the closing session we expect lively debate as we invite ethicists, researchers and patients to scrutinize the impact of recent developments in gene editing – are we heading towards a world without rare diseases?

SESSION 0101

Friday 11 May 2018 | 14:00-15:30

TRANSFORMATIONS IN DIAGNOSTICS: HOW RESEARCH AND EUROPEAN REFERENCE NETWORKS ARE RE-SHAPING THE DIAGNOSIS LANDSCAPE

Session Chair: Olaf Riess, Member of ERN-RND, Medical Director and Head of the Department of Medical Genetics, University of Tübingen, Germany

Introduction state of the art in diagnostics and presentation of future trends in scientific breakthrough
Olaf Riess, Member of ERN-RND, Medical Director and Head of the Department of Medical Genetics, University of Tübingen, Germany

Transformational diagnostic pathways: conversation between families and clinicians/researchers:

- A family diagnosed with an ultra-rare disease
  Isabelle Bros, Solhand, France
  Olaf Riess, Member of ERN-RND, Medical Director and Head of the Department of Medical Genetics, University of Tübingen, Germany
- A family with no diagnosis
  Louise James, SWAN UK, UK
  Alessandra Renieri, Professor, Department of Medical Biotechnologies, University of Siena, Italy

SWAN EUROPE: Keeping patients at the heart of diagnostic advancements
Lauren Roberts, Director of Support, Genetic Alliance UK, National Coordinator, Swan UK, UK

Interview with Centogene
Peter Bauer, Chief Scientific Officer of Centogene, Germany
Olaf Riess, Member of ERN-RND, Medical Director and Head of the Department of Medical Genetics, University of Tübingen, Germany
Virginie Bros-Facer, Scientific Director, EURORDIS-Rare Diseases Europe

SESSION 0102

Friday 11 May 2018 | 16:30–18:00

RESEARCH: FROM AN IDEA TO THE REAL WORLD

Session Chair: Diego Ardigo, Chair Therapies Scientific Committee of IRDiRC; Project Lead, Chiesi, Italy

Overview of the major bottlenecks in translating research
Diego Ardigo, Chair Therapies Scientific Committee of IRDiRC; Project Lead, Chiesi, Italy

How do you get research done on your conditions?
Daniel Lewi, Co-founder and Chief Executive, CATS Foundation, UK

How to develop and adapt a co-design model for rare disease research?
Alison Metcalfe, Associate Dean for Research and Professor of Health Care Research, Kings College London, UK

How to make exploitable research?
Lucia Monaco, Chief Scientific Officer, Fondazione Telethon, Italy

Crack It challenges from the industry perspective
Jon Timmis, CEO and Co-founder, SimOnics, UK

SESSION 0103

Saturday 12 May 2018 | 09:00-10:30

INNOVATIVE FUNDING PARTNERSHIPS: CHALLENGES AND OPPORTUNITIES

Session Chair: Daria Julkowska, Scientific Coordinator, E-Rare, France

Innovative funding partnerships: challenges and opportunities
Daria Julkowska, Scientific Coordinator, E-Rare, France

Case studies:

Patient associations joining forces to fund rare disease research
Speaker to be named

How can a non-profit advance research into a specific rare disease?
Majid Jafar, Co-founder, Loulou Foundation, UK

Research perspective
Heather Etchevers, Research Scientist, Inserm, France

MegaFund
Dimitrios Athanasiou, Head of Parents Project, Muscular Dystrophy Association Hellas, Greece

Panel Discussion

SESSION 0104

Saturday 12 May 2018 | 11:00-12:30

PATIENT INVOLVEMENT: IS IT ENOUGH TO BE AN 'EXPERT BY EXPERIENCE'?

Session Chair: Orion Buske, Chief Executive Officer, Gene42 Inc, Canada

What does it mean to be a patient expert?
Orion Buske, Chief Executive Officer, Gene42 Inc, Canada

Why and how patients can be trained in research/science to become stronger partners?
Virginie Bros-Facer, Scientific Director, EURORDIS-Rare Diseases Europe

How can the patient organisations support best the patient expert for a meaningful engagement?
Mathieu Boudes, Public/Private Partnership Coordinator, European Patients' Forum (EPF)

Interview: What does it mean for you to be a patient expert?
Chris Sotirelis, Patient advocate and volunteer, EURORDIS-Rare Diseases Europe, former patient representative, UK Thalassameia Society (UKTS), UK
Mathieu Boudes, Public/Private Partnership Coordinator, European Patients' Forum (EPF)

SESSION 0105

Saturday 12 May 2018 | 14:30-16:00

GENOME EDITING DEBATE: ARE WE HEADING TOWARDS A WORLD WITHOUT RARE DISEASES?

Debate Session

Moderator: Vivienne Parry, Head of Engagement, Genomics England, UK

Chair of position 1:
Heidi Howard, Senior Researcher, Uppsala University, Sweden

Chair of position 2:
Simon Woods, Policy, Ethics & Life Sciences Deputy-Director, Newcastle University, UK

THEME 2

BREAKTHROUGH MEDICINES ON THE HORIZON: REGULATORS, HEALTH TECHNOLOGY ASSESSOR (HTA) EXPERTS AND PATIENTS WORKING

THEME LEADERS:

Wim Goettsch, Executive Board Chair, EUNetHTA, Netherlands

Jordi Linares Garcia, Head of Scientific and Regulatory Management, EMA, EU

EURORDIS SUPPORT:

François Houÿez, Treatment Information and Access Director, Health Policy Advisor, EURORDIS-Rare Diseases Europe

Matteo Scarabelli, Patient Engagement Manager – HTA, EURORDIS-Rare Diseases Europe

THEME DESCRIPTION

Over the past two years, regulators and health technology assessors have engaged in an unprecedented exchange of information: an agreement to create a one-stop-shop for parallel European Medicines Agency/ health technology assessor's scientific advice and the sharing of early reports from regulators during the evaluation phase of pharmaceuticals so that health technology assessors can start before marketing authorisation. The European Medicines Agency and health technology assessors work together to scan the horizon and to see which medicines are likely to fit their respective procedures. This is preparing for future European cooperation on health technology assessors, as a permanent scientific secretariat to host European health technology assessor's activities is needed.

Theme 2 will cover important initiatives such as Priority Medicines at the European Medicines Agency (PRIME); the current cooperation on health technology assessors (EUnetHTA joint action 3) – the European Medicines Agency -EUnetHTA three-year work plan which was announced in November 2017; plans for the future of health technology assessors, and will describe where we are in the development of orphan medicinal products in 2018.

Lastly, it will explain the new roles of patients and their representatives when working with regulators, health technology assessors and/ or industry.

SESSION 0201

Friday 11 May 2018 | 14:00-15:30

BREAKTHROUGH PRODUCTS / PRIORITY MEDICINES –SYNERGIES BETWEEN REGULATORS AND HEALTH TECHNOLOGY ASSESSORS

Session Chair: Russell Wheeler, Patient Advocate at Leber's Hereditary Optic Neuropathy Society, UK

PRIME: where are we in May 2018: products, diseases, interactions with Health and Technology Assessment bodies and submission of MA for products benefiting from PRIME
Zahra Hanaizi, Scientific Officer, PRIME coordinator, European Medicines Agency, London

Can PRIME attract innovation towards unmet needs / disruptive medicines?
Speaker to be named

PRIME Scientific Advice with Health Technology Assessors: experience and feedback
Speaker to be named

Experience of the Committee for Orphan Medicinal Products
Violeta Stoyanova-Beninska, Senior clinical assessor Agency Medicines Evaluation Board, Member COMP and Expert CNS WP at European Medicines Agency, Netherlands

SESSION 0202

Friday 11 May 2018 | 16:30–18:00

CURRENT EU COOPERATION ON HEATH TECHNOLOGY ASSESSORS: EUNETHTA

Session Chair: Dimitrios Anathasiou, Board Member of United Parents Projects Muscular Dystrophy; Duchenne Muscular Dystrophy (DMD) Patient Advocate; EMA Patient Expert in DMD, Greece

Early Dialogues 2.0: Early Dialogue Working Party and what's new in Early Dialogues
François Meyer, Advisor to the President, International Affairs, Haute Autorité de Sante (HAS), France

Joint Health Technology Assessors for pharmaceuticals
Wim Goettsch, ZIN, and Director of the EUnetHTA Directorate

Preparing the contribution of patients in regulatory / Health Technology Assessors procedures
Matteo Scarabelli, Patient Engagement Manager – HTA, EURORDIS-Rare Diseases Europe, France

Health Technology Assessors and significant benefit, and how it relates to Relative Effectiveness Assessment
Speaker to be named

SESSION 0203

Saturday 12 May 2018 | 09:00-10:30

PREPARING THE CONTRIBUTION OF PATIENTS IN REGULATORY / HEATH TECHNOLOGY ASSESSOR PROCEDURES

Session Chair: to be named

The Community Advisory Boards Programme
Rob Camp, Patient Engagement Senior Manager – CABs, EURORDIS-Rare Diseases Europe

Patients invited to the oral explanations for the marketing authorisation opinion: report
Nathalie Bere, Patient Engagement, European Medicines Agency, UK

First EMA public hearing, EMA Network of Young People
Nathalie Bere, Patient Engagement, European Medicines Agency, UK

Possibility to submit topics for joint HTA (EUnetHTA/ medical devices)
Sabine Ettinger, Researcher & Scientific Project Manager at Ludwig Boltzmann Institute for Health Technology Assessment, Austria

SESSION 0204

Saturday 12 May 2018 | 11:00-12:30

OMPS (ORPHAN MEDICINE PRODUCTS) IN THE PIPE: WHAT CAN WE SEE COMING?

Session Chair: Kerry Leeson-Beevers, National Development Manager, Alstrom Syndrome UK

Characteristics of the 1800+ designated products
Violeta Stoyanova-Beninska, Senior clinical assessor Agency Medicines Evaluation Board, Member COMP and Expert CNS WP at European Medicines Agency, Netherlands

Abandoned OMPs
Viviana Giannuzzi, Senior Researcher, Gianni Benzi Pharamacological Research Foundation, Italy

Drug repurposing
Diego Ardigo, Chair Therapies Scientific Committee of IRDiRC; Project Lead, Chiesi

Horizon scanning at EMA
Kristina Larsson, Head of Orphan Drugs, European Medicines Agency, UK

SESSION 0205

Saturday 12 May 2018 | 14:30-16:00

THE FUTURE OF HEALTH TECHNOLOGY ASSESSOR COOPERATION

The Commission will present the Regulation Proposal at the session starting at 13:30 with all conference participants

Flora Giorgio, Policy Officer, DG Sante, Unit B4, European Commission, Brussels

Session Chair: Cees Smit, Patient Advocate, Patients Network for Medical Research and Health, EGAN, Netherlands and François Houÿez, Treatment Information and Access Director, Health Policy Advisor, EURORDIS-Rare Diseases Europe

European Commission Legislative Proposal
Speaker to be named

Views of a Member of the European Parliament
Speaker to be named

What patients can expect
François Houÿez, Treatment Information and Access Director, Health Policy Advisor, EURORDIS-Rare Diseases Europe

What decision makers can expect
Speaker to be named

What industry can expect
Ansgar Hebborn, Head of Global Market Access Policy, Roche Pharmaceuticals, Switzerland

What an HTA agency can expect
Speaker to be named

THEME 3
THE DIGITAL PATIENT

THEME LEADERS:

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

Justina Januševičienė, Executive for the development of health care technologies and innovations, Lithuanian University of Health Sciences, Lithuania & Former Director, Healthcare resources and innovation management department, Ministry of Health, Lithuania

EURORDIS SUPPORT:

Elisa Ferrer, Patient Engagement Manager, EURORDIS-Rare Diseases Europe

Virginie Hivert, Therapeutic Development Director, EURORDIS-Rare Diseases Europe

THEME DESCRIPTION

While other industries are fully immersed in the digital era, the health industry is struggling to undergo a real digital transformation. The foundations of health science date back centuries and the transition to the digital world is complex. The obstacles to create digital assets and relationships in the field of health range from unbalanced physician-patient relationships to clinical institutions focused on transactions and non-continuous care. Patients with rare diseases are suffering from this situation even more than other chronic patients: the complexity of their conditions, the low number of patients and the scarcity of effective treatments are big problems but also are great opportunities for a new medicine based on the P4 pillars (predictive, preventive, personalised and participatory). We will explore how technology can help patients with rare diseases, how the regulatory world is evolving, the initiatives in Europe to embrace this digital transformation and real examples from patient organisations already starting this journey. New technology will create fabulous opportunities but also new risks, as information will be more accessible to hackers and medical systems will be more exposed to cyberattacks. Information and awareness are elements crucial to understand in order to mitigate the risks while we are evolving into a new era of medicine.

SESSION 0301

Friday 11 May 2018 | 14:00-15:30

EVERYTHING IS TECHNICALLY POSSIBLE

Digital technologies are revolutionizing society and offering innovative solutions to improve patients' lives and to advance medical research at an unprecedented pace. In this session, we will explore what technology can offer to patients and what challenges lay ahead.

Session Chair: To be named

Technology panel discussion:

- What can technology offer & what are the challenges?
- Looking to the future
- New solutions applicable to patients' daily life
- Disruptive technology – Block chain in health care
- Technology bringing value to society

Panellists:

- **Ivo Ramos**, Atos Health Sector, Research and Innovation, France
- **Vytautas Kašėta**, Blockchain consulting services, Lithuania

SESSION 0302

Friday 11 May 2018 | 16:30–18:00

SOCIETAL, LEGAL AND ETHICAL FRAMEWORK

Are patients willing to share their health data for the sake of advancing research and accelerating diagnosis? Is it safe? Who owns the data? We will explore the answers to these questions with legal experts, cyber security specialists and patient advocates.

Session Chair: Petra Wilson, Director at Health Connect Partners, UK

Role Play: Overview on the General Data Protection Regulation
Petra Wilson, Director at Health Connect Partners, UK
Šarūnas Narbutas, president of the the Lithuanian Cancer Patient Coalition (POLA), Lithuania

Panel Discussion: The real life of data

Introductory presentation
Marc Hanauer, Chief Technology Officer, Orphanet, France

Panellists:

- **Marius Pareščius**, CEO International Security Cluster, Lithuania
- **Sandra Courbier**, Rare Barometer Senior Manager, EURORDIS-Rare Diseases Europe
- **Orion Buske**, Chief Executive Officer, Gene42 Inc., Canada

SESSION 0303

Saturday 12 May 2018 | 09:00-10:30

DIGITAL STRATEGY IN EUROPE – BREAKING DOWN THE BARRIERS

Digital technologies are transforming cross-border health care and offering new hope to patients living with rare diseases. This session will show how EU policies are supporting the implementation of digital health solutions and the use of health data for research and innovation.

Session Chair: Justina Januševičienė, Executive for the development of health care technologies and innovations, Lithuanian University of Health Sciences, Lithuania

Panel Discussion:

- Challenge the European Commission from the European Reference Networks and healthcare professionals point of view – Feedback on their discussions on how to interact with industry and on the European Reference Networks roadmap in between
- European Joint Programme
- Digital Health Society
- Exchanges of national experiences – data sharing between countries
- Future policy-shaping

Panellists:

- **Tapani Piha**, Head of Unit, Cross-border healthcare and e-Health, DG SANTE, Luxembourg
- **Brian O'Connor**, European Connected Health Alliance, UK
- **Henrique Martins**, CEO, Shared Services of the Ministry of Health, Portugal
- **Zoi Kolitisi**, eHealth strategist, eGov senior policy advisor, affiliated member of the Information Security Laboratory of the Aristotelian University of Thessaloniki, Greece

SESSION 0304

Saturday 12 May 2018 | 11:00-12:30

EUROPEAN REFERENCE NETWORKS AS A FUTURE MODEL OF HEALTHCARE

The European Reference Networks (ERNs) are transforming diagnosis and care for patients living with a rare disease. We will explore how online consultations and patient data sharing is currently happening in the ERN framework and how the digital infrastructure is supporting this transformation.

Session Co-Chairs:
Kate Bushby, EURO-NMD Coordinator, John Walton Muscular Dystrophy Research Centre, UK
Ana Rath, Director, Orphanet, France

How virtual health care is happening in the ERN framework

- **Rima Nabbout**, European Reference Network on Rare Epilepsies (EpiCARE), Hôpital Necker-Enfants Malades, France
- **Sofia Douzgou**, European Reference Network on Rare Congenital Malformations and Rare Intellectual Disability (ITHACA), Manchester Centre for Genomic Medicine, University of Manchester, United Kingdom

Results of RD-Action WP5 on Steering, maintaining and promoting the adoption of OrphaCodes across member states
Stefanie Weber, Director Deutsches Institut für Medizinische Dokumentation und Information, Germany

Interoperability (national vs European)

- **Elisa Salamanca**, Operations Director, French national database on rare diseases, France
- **Ana Rath**, Director, Orphanet, France

Debate on CPMS system: theory vs real life
Moderator: Victoria Hedley, RD-ACTION Thematic Coordinator for Rare Diseases at Newcastle University Institute of Genetic Medicine, UK

Panellists:

- **Tapani Piha**, Head of Unit, Cross-Border Healthcare & eHealth, DG Sante, Luxembourg
- **Marie Claude Boiteux**, President and Co-Founder of Cutis Laxa Internationale, ERN Skin, France
- **Russel Wheeler**, Rare eye diseases ERN patient representative, Leber's Hereditary Optic Neuropathy Society UK
- **Rima Nabbout**, European Reference Network on Rare Epilepsies (EpiCARE), Hôpital Necker-Enfants Malades, France
- **Sofia Douzgou**, European Reference Network on Rare Congenital Malformations and Rare Intellectual Disability (ITHACA), Manchester Centre for Genomic Medicine, University of Manchester, United Kingdom

SESSION 0305

Saturday 12 May 2018 | 14:30-16:00

PATIENTS AND THE DIGITAL REVOLUTION

Digital revolution is happening and patients are taking the lead. This session will focus on how patient-led technological solutions are helping diagnosis, treatment and care and paving the way for patient-centric medicines development.

Conversational interfaces to identify patient-relevant outcome measures (PROMs): development of a Duchenne muscular dystrophy data platform
Speaker to be named

Deep learning project for symptoms identification
Julian Isla, Foundation 29, Spain

Development of a mobile app in the context of the ERN on multisystemic vascular diseases (VASCERN)
Claudia Croccione, Managing Director, Hereditary hemorrhagic teleangiectasia, Italy

Case studies of remote patient monitoring: use of wearables
Elin Haf Davies, Founder of Aparito, UK

EMA qualification process of new methodologies for medicines development
Kristina Larsson, Head of Office for Orphan Medicines, European Medicines Agency (EMA), UK

THEME 4

QUALITY OF LIFE: MAKING WHAT MATTERS, MATTER

THEME LEADERS:

Lene Jensen, Chief Executive Officer, Rare Diseases Denmark, Denmark

Ursula Holtgrewe, Head of Work & Equal Opportunities, Zentrum für Soziale Innovation, Austria

EURORDIS SUPPORT:

Raquel Castro, Social Policy Senior Manager, EURORDIS-Rare Diseases Europe

THEME DESCRIPTION

Rare diseases pose serious health, social and everyday challenges, which are often highly debilitating, and significantly affect the autonomy and the fundamental human rights of people living with a rare disease and their carers. However, people living with rare diseases and their carers should be recognised and esteemed as persons, not as diagnoses. They should have the possibility of living a life with fulfilling personal relationships, of being able to contribute meaningfully to the lives of others and to society. Freedom to decide on their own lives, autonomy, security and dignity are important factors of what we call "quality of life".

All rare disease stakeholders are working to improve the quality of life of all rare people. Nevertheless, health and social systems was well as the different spheres of access to care, treatment and support to inclusion and participation in society do not always successfully address their complex needs in ways that create actual improvements. How can we continue to build win-win collaborative strategies to advance this mission?

This theme revisits the concept of quality of life and explores the ways in which it can contribute to decision making and to shaping the provision of treatments and care. Discussions will also unveil the invisible burden of rare diseases and explore case studies of innovative services that bridge the existing gaps to effectively and sustainably achieving integrated care.

Lastly, the theme will venture into thinking of how care may look like in 30 years and how all stakeholders can prepare to develop the next best practices, building on the advances and challenges of tomorrow rather than those of today.

SESSION 0401

Friday 11 May 2018 | 14:00-15:30

QUALITY OF LIFE – WHAT REALLY MATTERS TO PATIENTS & HOW TO MEASURE IT

A lot is said and researched on Quality of Life - but what does Quality of Life really mean for patients and carers? What really matters? How can we set meaningful and measurable Quality of Life indicators?

Session Chair: Avril Daly, Vice-President, EURORDIS-Rare Diseases Europe, CEO, Retina International, Ireland

Quality of life, what matters to people living with a rare disease and their carers?
Avril Daly, Vice-President, EURORDIS-Rare Diseases Europe, CEO, Retina International, Ireland

Overview of traditional quality of life assessment methodologies
Jakob Bjørner, Chief Science Officer, Optum Patients Insights, Denmark

The role of European Reference Networks in developing Quality of Life indicators
Sofia Douzgou, European Reference Network for Rare Congenital Malformations and Intellectual Disability (ITHACA), Central Manchester University Hospitals, NHS Foundation Trust, United Kingdom

Debate Session: What really matters?

SESSION 0402

Friday 11 May 2018 | 16:30–18:00

HOW CAN QUALITY OF LIFE CONTRIBUTE TO DECISION MAKING?

How can Quality of Life systematically inform decision making on the provision and reimbursement of treatments, health care and social services? How can we bridge the gaps between what counts for decision making and what really matters to patients and carers?

Session Chair: Anna Bucsics, Project Advisor, MoCA (Mechanism of coordinated Access to Orphan Medicinal Products) Austria

Debate Session:

- **Pauline Evers**, Dutch Federation of Cancer Patient Organisations, Patient Representative at Committee for Orphan Medicinal Products (COMP), European Medicines Agency's (EMA), Netherlands
- **Virginie Hivert**, Therapeutic Development Director, EURORDIS; Vice-Chair of Therapies Scientific Committee, International Rare Diseases Research Consortium (IRDiRC)
- **Karl-Johan Myrén**, Global Head of Health Economics, Outcomes & Value, Sobi, Sweden

SESSION 0403

Saturday 12 May 2018 | 09:00-10:30

DISABILITY: UNVEILING THE INVISIBLE DOUBLE-BURDEN OF RARE DISEASES

Rare diseases = disability? How disabling are rare diseases? How can rare diseases be visible on the disability agenda? How can the disability generated by the time and care burden of rare diseases be taken into account?

Session Chair: Lene Jensen, Chief Executive Officer, Rare Diseases Denmark, Denmark

Patients and carers perspectives: results of European-wide survey on the social impact of rare diseases
Raquel Castro, Social Policy Senior Manager, EURORDIS-Rare Diseases Europe, France

Key findings of the Orphanet Disability project
Ana Rath, Director, Orphanet, France

Debate Session: How to integrate rare diseases into the disability agenda? How to consider the time and care burden aspects?

Panellists:

- **Ana Lucia Arellano**, First Vice-Chair of International Disability Alliance, and President of the Latin American Network of Non-Governmental Organizations of Persons with Disabilities and their Families, Ecuador

SESSION 0404

Saturday 12 May 2018 | 11:00-12:30

INTEGRATED CARE: BRINGING TOGETHER HEALTH & SOCIAL CARE, TWO SIDES OF THE SAME PATIENT

People living with a rare disease have full lives and multidisciplinary needs. Multidisciplinary and integrated health and social care is key for their Quality of Life. But, for patients and carers, finding one's way in through the care systems takes skills, coordination and maybe a bit of luck. How can integrated care for rare disease become a reality across Europe? How can European Reference Networks support the bridging of health and social care?

Session Chair: Ester Sarquella Casellas, Connected Health and Care Business Development Director for Southern Europe, Tunstall Healthcare, United Kingdom

Case Studies - Bridging the gap between health and social care for rare diseases:

Case management at NoRo Centre in Romania (INNOVCare project)
Dorica Dan, President, Romanian Prader Willi Association, Romania

Experience of Centre of Expertise
Anja Diem, Manager of outpatient clinic, EB-Haus, Austria

Patient testimonial of successful experience
Beata Ferencz, Mother of a child with Williams Syndrome, Project Manager, Rare Diseases Sweden, Sweden

Debate Session: Innovative practices to achieve integrated care; key success factors and main hurdles

SESSION 0405

Saturday 12 May 2018 | 14:30-16:00

FROM BEST PRACTICES TO NEXT PRACTICES: BUILDING A COLLABORATIVE VISION

The first sessions focused on the challenges and best practices of today. How about tomorrow? What will care look like 30 years from today? What are the game changers for the future and how should we start getting prepared? What will be the future solutions on future problems 360°?

Session Chair: Peter O'Donnell, Brussels Correspondent, APM Health Europe, Belgium

Key messages from all sessions
Ursula Holtgrewe, Head of Work & Equal Opportunities, Zentrum für Soziale Innovation, Austria

Game changers of the future

Vision from young patient advocates

- **Synne Lerhol**, Secretary General, The Norwegian Association for Youth with Disabilities, Norway
- **Courtney Coleman**, Patient Involvement and Engagement, European Lung Foundation (ePAG), United Kingdom

Closing Speech
Anders Olauson, Agrenska, Honorary President, European Patients' Forum, Chair at RareResourceNet, Sweden

THEME 5

ECONOMICAL PERSPECTIVES IN RARE DISEASES

THEME LEADERS:

Michael Schlander, Professor of Health Economics, University of Heidelberg, Germany

Ruediger Gatermann, Director, Healthcare Policy and External Affairs Europe, CSL Behring, Germany

EURORDIS SUPPORT:

Simone Boselli, Public Affairs Director, EURORDIS-Rare Diseases Europe

THEME DESCRIPTION

Rare diseases pose serious health, The theme will aim to look at economical aspects in rare diseases from different stakeholder perspectives, evaluate existing collaborative approaches and discuss options to further develop an environment conducive to innovation and to faster access to patients care and cure.

The sessions in this theme will explore our ambitions to refine a shared understanding on how to improve access to rare disease therapies and how to ensure a sustainable orphan drug business model for all stakeholders involved.

We will share findings on economic and financial impact of rare disease on healthcare systems and societies, including testimonials/ case studies from patients.

The theme will look both into the impact of the current policies on access to rare disease therapies as well as into innovative concepts and collaborative approaches which are being experimented throughout Europe both in view of value recognition, rewarding and funding.

A look into the future will complete the theme to explore consensual ideas on what is needed to further develop the rare diseases ecosystem and how to ensure sustainable access to rare disease care in 2030.

SESSION 0501

Friday 11 May 2018 | 14:00-15:30

ECONOMIC IMPACT OF RARE DISEASES ON PATIENTS, FAMILIES AND SOCIETY

Session Chair: To be named

Societal dimension of rare diseases
Speaker to be Named

Facts and figures from studies (cost of illness/burden of disease)
Speaker to be Named

Patient testimonials / case studies
Speaker to be Named

SESSION 0502

Friday 11 May 2018 | 16:30-18:00

DYNAMICS OF THERAPY DEVELOPMENT FOR RARE DISEASES

Session Chair: Emmanuel Chantelot, Executive Director, Head of Government Relations and Policy Europe, Celgene, Belgium

Economics of Orphan Medicine Products development

Rare Diseases business model

Rare Diseases ecosystem

Role of incentives

Speakers:
Tim Wilsdon, Vice President Charles River Associate, UK

SESSION 0503

Saturday 12 May 2018 | 9:00-10:30

A PARADIGM SHIFT IN VALUE FRAMEWORKS FOR ACCESS

Session Chair: Prof Michael Schlander, Professor of Health Economics, University of Heidelberg, Germany

New value frameworks/concepts

New value frameworks: merging valuation paradigms with empirical evidence on "social preferences"

The wish to share healthcare resources

European Social Preferences Measurement (ESPM) Project – first presentation of findings

Speakers:

- **Prof Jeff Richardson**, Monash University, Melbourne, Australia
- **Prof Michael Schlander**, DKFZ & University of Heidelberg, Germany
- **Sheela Upadhyaya**, NICE HST Programme, London, United Kingdom

SESSION 0504

Saturday 12 May 2018 | 11:00-12:30

NEW APPROACHES TO PRICING AND FUNDING AND IMPLICATIONS FOR ACCESS

Session Chair: to be named

New approaches to pricing / Innovative pricing models
Speaker to be named

New approaches to funding / Innovative payment models
Speaker to be named

Managed Entry Agreements (MEA), payment based on outcomes, how to deal with uncertainties and differences with EUnetHTA
Speaker to be named

EURORDIS Breaking the Access Deadlock paper
Speaker to be named

SESSION 0505

Saturday 12 May 2018 | 14:30-16:00

A LOOK INTO THE FUTURE – HOW TO ENSURE SUSTAINABILITY ACCESS TO RARE DISEASES CARE IN 2030

Session Chair: to be named

Value assessment continuum, evidence generation (Real World Evidence), role of European Reference Networks and patient registries
Speaker to be named

Foresight 2030 study
Speaker to be named

THEME 6

GLOBAL RARE EQUITY: ARE WE THERE YET?

THEME LEADERS:

Durhane Wong-Rieger, President & CEO Canadian Organization for Rare Disorders, Chair of Rare Diseases International, Founder of the Asia Pacific RD Alliance (APARDO), Canada

Professor Hugh Dawkins, Director, Office of Population Health Genomics, Health Department of Western Australia, Australia

EURORDIS SUPPORT:

Paloma Tejada, Director, Rare Diseases International, EURORDIS-Rare Diseases Europe

Clara Hervas, Public Affairs Junior Manager, EURORDIS-Rare Diseases Europe

THEME DESCRIPTION

It's time to commit to global equity for rare diseases. When rare diseases are neglected anywhere, people living with a rare disease are harmed everywhere.

People with rare diseases are connected globally by their genes and their challenges; they should also be connected by their hope and opportunities. Our vision is a world where all people living with rare diseases receive equitable treatment and support and all advances in rare diseases benefit all those affected, regardless of where they live.

This theme is set up as five inter-related workshop sessions that explore how to achieve global equity for rare diseases from top-down and from bottom-up levels, from policy and research to products and practical solutions.

Each session will be facilitated by an animateur with several "thought leaders" who will set the stage for total audience participation.

SESSION 0601

Friday 11 May 2018 | 14:00-15:30

HOW CAN WE LEVERAGE GLOBAL POLICIES AND GLOBAL AGENCIES TO EXPLICITLY SUPPORT RARE DISEASES RECOGNISING DIVERSITY AND ENSURING EQUITY?

Moderator: Mark Krueger, President, MK&A, USA

Overview presentation
Yann Le Cam, CEO and Co-Founder, EURORDIS-Rare Diseases Europe

Panel Discussion

- **Angela Chaves Restrepo**, CEO, Federación Colombiana de Enfermedades Raras, Colombia

SESSION 0602

Friday 11 May 2018 | 16:30-18:00

WHAT GLOBAL OPPORTUNITIES DO WE UNLOCK WHEN ALL PEOPLE LIVING WITH A RARE DISEASE HAVE ACCESS TO A TIMELY ACCURATE DIAGNOSIS AND OPTIMISED CARE

Moderator: Jeff Sturchio, President & CEO, Rabin Martin, USA

Overview presentation
Speaker to be named

Panel Discussion

- **Matthew Harold**, International Public Affairs, Rare Diseases, Pfizer, UK

SESSION 0603

Saturday 12 May 2018 | 9:00-10:30

IRDIRC NEXT HORIZON 2027: RESEARCH FROM VISION TO THE REAL WORLD

Moderator: Paul Lasko, Scientific Director of the Institute of Genetics, Canadian Institutes of Health Research - Institute of Genetics (CIHR-IG), Canada

Overview presentation
Christopher Austin, Director of NIH/NCATS, USA

Panel Discussion

- **Makoto Suematsu**, President, Agency for Medicial Research and Development (AMED), Japan
- **Kym Boycott**, Senior Scientist, Children's Hospital of Eastern Ontario Research Institute; Care4Rare, Canada

SESSION 0604

Saturday 12 May 2018 | 11:00-12:30

BUILDING THE RARE DISEASE KNOWLEDGE AND INFORMATION ECO-SYSTEM THROUGH BETTER CONNECTIONS

Moderator: Professor Hugh Dawkins, Director, Office of Population Health Genomics, Health Department of Western Australia, Australia

Overview presentation
Marshall Summar, Director, Rare Disease Institute, Children's National Medical Centre - NIH RD Clinical Research, USA

Panel Discussion
Panellists to be named

- **Gareth Baynam**, Clinical Geneticist, Genetic Services Western Australia (GSWA), Australia
- **Christina Waters**, CEO and Founder, Rare Science, United States
- **Dr Mike Brudno**, Scientific Director, SickKids, Associate Professor, Department of Computer Science, University of Toronto, Canada

SESSION 0605

Saturday 12 May 2018 | 14:30-16:00

WHAT ARE OUR KEY ENABLERS TO BRING A VISION FOR EQUITY AND OPTIMISED CARE GLOBALLY TO PEOPLE LIVING WITH A RARE DISEASE LOCALLY?

Moderator: Durhane Wong-Rieger, President & CEO Canadian, Organization for Rare Disorders, Chair of Rare Diseases International, Founder of the Asia Pacific RD Alliance (APARDO), Canada

Overview presentation
Matt Bolz-Johnson, Healthcare and Research Director, EURORDIS-Rare Diseases Europe

Panel Discussion

- **Ritu Jain**, President, DEBRA Singapore and Member of the Executive Committee of DEBRA International
- **Shikha Mittoo**, Assistant Professor, Department of Medicine, Mount Sinai Hospital, Canada
- **Clarisa Marchetti**, Scientific Committee Member, Federación Argentina de Enfermedades Poco Frecuentes, General Coordinator of the Course Integral Management in Rare Diseases, Universidad Isalud, Argentina