9th European Conference
on Rare Diseases and Orphan Products

10-12 May 2018 Vienna

Messe Wien Exhibition & Congress Center

Rare Diseases 360°

Collaborative strategies
to leave no one behind

PROGRAMME

Organised by: EURORDIS RARE DISEASES EUROPE

Co-organised by: DIA, orphanet

With co-funding from: AFMTELETHON INNOVER POUR GUERIR; Co-funded by the Health Programme of the European Union

#ECRDVienna

MOTTO OF **THE EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS (ECRD)**

- The European Conference on Rare Diseases & Orphan Products is the unique forum across all rare diseases, across all European countries, bringing together all stakeholders - patients' representatives, academics, health care professionals, researchers, healthcare industry, payers, regulators and policy makers.
- It is a biennial event, providing the state-of-the-art of the rare disease environment, monitoring and benchmarking initiatives. It covers research, development of new treatments, health care, social care, information, public health and support at European, national and regional levels.
- It is synergistic with national and regional conferences, enhancing efforts of all stakeholders. There is no competition with them, but efforts are complementary, fully respecting initiatives of all.

MESSAGE FROM **THE CO-CHAIRS**

Vinciane Pirard
Co-Chair, EFPIA-EuropaBio Joint Task Force on orphan drugs and rare diseases; Public affairs, Sanofi-Genzyme, Belgium

Rainer Riedl
President, Pro Rare Austria, Austria

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

It is with great pleasure that we welcome you on behalf of the organisers, EURORDIS-Rare Diseases Europe and co-organisers Orphanet and DIA at the 9th edition of ECRD in Vienna.

The ECRD meeting is a special moment for the rare disease community as it fosters collaboration between stakeholders from various backgrounds with a unique interest in advancing diagnosis, treatment and care for patients living with a rare disease.

The very first ECRD was held in Copenhagen 15 years ago, in 2001, and gathered more than 300 attendees. This ground breaking initiative had such a positive impact that there has been an ECRD held in different EU countries every 2 years since then. The rare disease community, including all rare disease patient organizations and their partners, can be proud of its many achievements over the last 20 years. The landscape has changed drastically during this time and has gone from near ignorance of rare diseases to its recognition as a public health priority in Europe. Outputs from past meetings have informed national and local policy initiatives. This amazing community has also managed during all these years to keep a spirit of collaboration across very different diseases, countries and stakeholders in Europe and elsewhere, inspiring many to follow this path.

The future holds many opportunities to accelerate the momentum we have built over the last decades; advancements in science and medicine, transformative treatments in the pipeline, game-changing digital technologies, innovation in the organisation of health and social care and increased empowerment of patients all hold promises for a more inclusive, holistic approach to leave no one behind.

At ECRD 2018 we will look to the future and facilitate effective policy discussions and collaborative strategies to make these promising opportunities a reality. The sessions have been organized in six themes reflecting the 360° collaborative approach needed to improve the lives of rare disease patients and families, which, we believe, each attendee will find relevant.

We hope you will enjoy contributing to the different discussions that will set out next steps to shape better research, services and policies that will improve patients' access to the best possible medicines, healthcare and social care. More than in any other field, the expertise on rare diseases is scarce and ambitious cross-border initiatives like the European Reference Networks (ERN) and IRDiRC are great ways to respond to today's changing environment.

We invite you to take a glance at the full programme and make the most of this meeting by listening to the inspiring visions developed during the plenary sessions, getting lost amongst the high quality posters that will be on display and participating in the debate during the different sessions.

Our thanks go also to our host city for 2018, Vienna, which is a wonderful city in which you'll find ample opportunities to relax and network in an atmosphere steeped in history.

We sincerely hope you enjoy this year's meeting,

Sincerely,

ECRD 2018 Programme Committee Co-Chairs

COMMITTEES

PROGRAMME **COMMITTEE**

Matt Bolz-Johnson
ERN Healthcare Advisor, EURORDIS, Germany
Valentina Bottarelli
Public Affairs Director & Head of European and International Advocacy, EURORDIS, Belgium
Kate Bushby
Professor of Neuromuscular Genetics, Newcastle University, United Kingdom
Emmanuel Chantelot
EUCOPE representative & Executive Director, Head of Government Relations and Policy Europe, Celgene, Belgium
Miriam Dalmas
Consultant in Public Health Medicine, Ministry for Health, Malta
Yann Le Cam
Chief Executive Officer and Co-Founder, EURORDIS-Rare Diseases Europe
François Houÿez
Treatment Information and Access Director, Health Policy Advisor, EURORDIS, France
Julian Isla
Data and Artificial Intelligence Resource Manager, Microsoft and Dravet Syndrome European Federation (DSEF), Spain
Lene Jensen
Chief Executive Officer, Rare Diseases Denmark, Denmark
Daria Julkowska
Executive Programme Manager, E-Rare, France
Jordi Llinares Garcia
Head of Orphan Medicines at the European Medicines Agency, EU
Luca Pani
Professor of Clinical Psychiatry, University of Miami, USA
Anne Pariser
Deputy Director of the Office of Rare Diseases Research (ORDR), NCATS, NIH, USA
Ana Rath
Director, Orphanet, France
Olaf Riess
Member of ERN-RND, Medical Director and Head of the Department of Medical Genetics, University of Tübingen, Germany
Violeta Stoyanova Beninska
Senior Clinical Assessor, Medicines Evaluation Board (Netherlands), Member COMP and Expert CNS WP at EMA, Netherlands
Till Voigtländer
Chair ERN Board of Member States, Clinical Institute of Neurology, Medical University of Vienna, Austria

OUTREACH **COMMITTEE**

Alba Ancochea
Chief Executive Officer, FEDER, Spain
Simona Bellagambi
Volunteer and project collaborator at UNIAMO FIMR onlus, Italy
Mariana Campos
Membership and Public Engagement Manager, Genetic Alliance UK
Anja Helm
Senior Manager of Relations with Patient Organisations, EURORDIS
Ulrike Holzer
Vice Chairwoman, Pro Rare Austria
Lene Jensen
Chief Executive Officer, Rare Diseases Denmark
Olivier Menzel
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Executive Director, Russian Association for Rare Diseases
Martina Michalova
Associate Producer and Office Manager, Czech Association
Gábor Pogany
Executive Vice President, Hungarian Federation of People with Rare and Congenital Diseases

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PROGRAMME OVERVIEW

Thursday, 10 May 2018

09:30 - 16:00 ePAG representatives meeting
09:30 - 13:00 RDI General Assembly (members only)
14:00 - 16:00 RDI Annual meeting (open to members and non-members of RDI)
16:30 - 18:30 EURORDIS General Assembly (members only)
19:00 - 21:00 **Patient Groups Welcome Reception**

Friday, 11 May 2018

09:00 - 09:45 Opening Session
09:45 - 10:15 **Coffee Break**
10:15 - 12:30 Plenary Session
12:30 - 13:30 **Lunch**
13:00 - 14:00 Shire symposium: Transforming lives for those living with rare diseases in Austria
13:00 - 14:00 Poster Session
13:00 - 13:30 Moderated Poster Walk: Theme TBC, Lucia Monaco, Téléthon Italia
13:30 - 14:00 Moderated Poster Walk: ERNs, Matt Bolz-Johnson, EURORDIS-Rare Diseases Europe
14:00 - 15:30 Choose from 6 parallel sessions
15.30 - 16.30 **Coffee break** and poster session
15:30 - 16:00 Moderated Poster Walk: Rare Disease Patient Groups Innovations, Danijela Szili, Rett Syndrome Europe
16:00 - 16:30 Moderated Poster Walk: ERNs, Matt Bolz-Johnson, EURORDIS-Rare Diseases Europe
16:30 - 18.00 Choose from 6 parallel sessions
18:00 - 19:00 **Reception (for all participants)**

Saturday, 12 May 2018

09:00 - 10:30 Choose from 6 parallel sessions
10:30 - 11:00 **Coffee Break** and Poster Session
11:00 - 12:30 Choose from 6 parallel sessions
12:30 - 13:30 **Lunch**
13:30 - 14:30 "Soap box" Plenary Session featuring Flora Giorgio, European Commission
14:30 - 16:00 Choose from 6 parallel sessions
16:15 - 17:00 Closing Plenary Session
17.00 - 17.30 **Farewell coffee break**

GENERAL **INFORMATION**

Information on Interpretation & Language for each Session

Pre-conference satellite meetings

All pre-conference workshops will be conducted in English only.

Opening & Plenary Sessions

The Opening & Plenary Session, taking place on the morning of Friday, 11 May will be simultaneously interpreted from English into 2 languages: French and German.

Other Sessions

All other sessions will be conducted in English only.

Registration Opening Times

The registration desks are located on the ground floor of the conference venue. A separate Speakers registration desk will be set-up and open during the following times:

Thursday, 10 May	**12.00 – 18.00**
Friday, 11 May	**08.00 – 18.00**
Saturday, 12 May	**08.30 – 14.00**

On-site Speaker Room (Speaker Preview)

The Speakers Room is located on the ground floor of the conference venue opposite the registration desks, in the Media Lounge. On-site welcome staff will direct you.

ACKNOWLEDGMENTS AND CREDITS

CREDITS, SUPPORTS AND LEGAL INFORMATION

We wish to thank the following institutions for their active collaboration

CONFERENCE ORGANISER

The 9th European Conference on Rare Diseases and Orphan Products is organised by EURORDIS-Rare Diseases Europe

CO-ORGANISED BY

DIA and Orphanet

With co-funding from

- AFM-Téléthon
- Health Programme of the European Union

In partnership with

- Austrian Government
- EFPIA (European Federation of Pharmaceutical Industries and Associations)
- ESHG (European Society of Human Genetics)
- EUCOPE (The European Confederation of Pharmaceutical Entrepreneurs)
- EuropaBio (European Association of Bioindustries)
- NIH/NCATS (National Institutes of Health USA)
- Pro Rare Austria
- FDA (Food and Drug Administration – OOPD – Office of Orphan Products Development)
- HOPE (The European Hospital and Healthcare Federation)

Associate Partners

- BlackSwan Foundation
- Childhood Cancer International
- CORD (Canadian Organisation for Rare Disorders)
- CORD (Chinese Organisation for Rare Disorders)
- EAHP (European Association of Hospital Pharmacists)
- ECPC (European Cancer Patient Coalition)
- EORTC (European Organisation for Research and Treatment of Cancer)
- EPF (European Patients' Forum)
- E-Rare
- GÖG (Gesundheit Österreich)
- IFSW (International Federation of Social Workers)
- IRDiRC (International Rare Diseases Research Consortium)
- ISPOR (International Society for Pharmacoeconomics and Outcomes Research)
- Medical University of Innsbruck
- NORD (National Organisation for Rare Disorders)
- PHARMIG (Association of the Austrian Pharmaceutical Industry)
- Rare Diseases International
- Russian Patients' Union
- Social Platform
- SWAN Europe
- ZSI (Zentrum für Soziale Innovation)

Continuing Education

The Commission for Professional Development (CPD) of the Swiss Association of Pharmaceutical Professionals (SwAPP) and the Swiss Society of Pharmaceutical Medicine (SGPM) has approved this conference. The conference will be honoured with 14 credits for pharmaceutical medicine.

SwAPP
Swiss Association of Pharmaceutical Professionals

DIA is an authorised training organisation accredited under the number 11 99 53383 75 to the Préfet of Ile-de-France.

CONTACTS

Conference Venue

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Conference Organiser

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Tel. +33 1 56 53 81 37
contact.orphanet@inserm.fr

NETWORKING EVENTS

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PATIENT GROUPS **WELCOME RECEPTION**

Thursday, 10 May 2018 | 19:00-21:00 | First Floor
Messe Wien Congress Center

A welcome reception will be held for registered patients and patients' advocates on Thursday, 10 May 2018 from 19.00 to 21.00 in the 1st Floor Foyer. Drinks and snacks will be served.

NETWORKING **RECEPTION & POSTER AWARD PRESENTATION**

Friday 11 May 2018 | 18:00 – 19:00
Strauss Foyer, Ground Floor

All registered participants are invited to attend this informal networking reception and poster awards presentation taking place from 18.00 to 19.00 in the Strauss Foyer at the ECRD 2018 conference venue. Austrian Folklore music by Die Tanzgeiger is generously provided by ProRare Austria.

FAREWELL **COFFEE**

Saturday 12 May 2018 | 17:00 – 17:30
Strauss Foyer, Ground Floor

All registered participants are invited to attend this informal farewell coffee break taking place from 17.00 to 17.30 in the Strauss Foyer. This will be the occasion to say goodbye to fellow participants before leaving Vienna or enjoying an extended stay in the city.

PROGRAMME AT A GLANCE

Thursday 10 May 2018						
09:30 - 16:00	ePAG representatives meeting					
09:30-13:00	RDI General Assembly (members only)					
14:00-16:00	RDI Annual meeting (open to members and non-members of RDI)					
16:30-18:30	EURORDIS General Assembly (members only)					
19:00-21:00	Patient Groups Welcome Reception					
Friday 11 May 2018						
09:00-09:45	Opening Session					
09:45-10:15	Coffee Break					
10:15-12:30	Plenary Session					
12:30-13:30	Lunch					
13:00-14:00	Poster Sessions & Shire Satellite symposium					
Themes	**1/ Structuring the Research and Diagnostic Landscape**	**2/ Breakthrough medicines on the horizon**	**3/ The Digital Patient**	**4/ Quality of Life: Making what matters, matter**	**5/ Economical Perspectives in Rare Diseases**	**6/ Global Rare Equity: Are we there yet?**
14:00-15:30	Session 0101 Transformations in diagnostics: how research and European Reference Networks are re-shaping the diagnosis landscape	Session 0201 Breakthrough products and priority medicines	Session 0301 Everything is technically possible	Session 0401 Quality of life - what really matters to patients & how to measure it	Session 0501 Economic impact of rare diseases on patients, families and society	Session 0601 How can we leverage global policies and global agencies to explicitly support rare diseases recognising diversity and ensuring equity?
15:30-16:30	Poster Sessions					
16:30-18:00	Session 0102 Research: from an idea to the real world	Session 0202 Current EU cooperation on Health Technology Assessment: EUnetHTA	Session 0302 Societal, legal and ethical framework	Session 0402 How can quality of life contribute to decision making?	Session 0502 Economic dynamics of therapy development for rare diseases	Session 0602 What global opportunities do we unlock when all people living with a rare disease have access to a timely accurate diagnosis and optimised care?
18:00-19:00	Reception (for all participants)					
Saturday, 12 May 2018						
Themes	**1/ Structuring the Research and Diagnostic Landscape**	**2/ Breakthrough medicines on the horizon**	**3/ The Digital Patient**	**4/ Quality of Life: Making what matters, matter**	**5/ Economical Perspectives in Rare Diseases**	**6/ Global Rare Equity: Are we there yet?**
09:00-10:30	Session 0103 Innovative funding partnerships: challenges and opportunities	Session 0203 The Future of Health Technology Assessment Cooperation	Session 0303 EC Digital Single Market Strategy	Session 0403 Disability: unveiling the invisible double-burden of rare diseases	Session 0503 A paradigm shift in value frameworks for access	Session 0603 IRDiRC next horizon 2027: Research from vision to the real world
10:30-11:00	Coffee Break & Poster Sessions					
11:00-12:30	Session 0104 Patient involvement: Is it enough to be an 'expert by experience?'	Session 0204 Orphan medicinal products in the pipeline: what can we see coming and disappearing?	Session 0304 European Reference Networks as a future model of healthcare	Session 0404 Integrated care: bringing together health & social care, two sides of the same patient	Session 0504 Enhancing patient access to care: new approaches to pricing and funding	Session 0604 Building the Rare Disease knowledge and information eco-system through better connections
12:30-13:30	Lunch					
13:30-14:30	**"Soap box" Plenary Session featuring Flora Giorgio**, Head of Sector Health Technology Assessment, DG SANTE B4, European Commission.					
14:30-16:00	Session 0105 Genome editing debate: Are we heading towards a world without rare diseases?	Session 0205 Preparing the contribution of patients in regulatory / Health Technology Assessment procedures	Session 0305 Patients and the digital revolution	Session 0405 From best practices to next practices: building a collaborative vision	Session 0505 A look into the future - how to ensure sustainability access to rare diseases care in 2030?	Session 0605 What are our key enablers to bring a vision for equity and optimised care globally to people living with a rare disease locally?
16:15-17:00	Closing Plenary Session					
17:00-17:30	Farewell Coffee					

OPENING SESSION AGENDA

Friday, 11 May 2018 - 09:00 – 09:45

The Opening and Plenary Sessions will be simultaneously interpreted from English into 2 languages:

French
German

WELCOME ADDRESS AND OPENING REMARKS

09.00 – 09.20

- **Terkel Andersen**, President, EURORDIS-Rare Diseases Europe
- **Magdalena Daccord**, Associate Director, Head Of Operations EMEA - DIA
- **Ana Rath**, Director, Orphanet
- **Rainer Riedl**, President, Pro Rare Austria

KEYNOTE ADDRESSES

09.20 – 09.45

- **Beate Hartinger-Klein**, Austrian Federal Minister of Health and Women's Affairs
- Video message: **Vytenis Andriukaitis**, European Commissioner for Health and Food Safety
- **Martin Seychell**, Deputy Director-General, Directorate-General for Health and Food Safety, European Commission

COFFEE BREAK

09.45 – 10.15

PLENARY SESSION AGENDA

Friday, 11 May 2018 - 10:15 – 12:30

WELCOME & INTRODUCTION

10:15 – 10:30

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

KEYNOTE SPEAKER

10:30 – 10:45

Dr Rüdiger Krech, Director, Universal Health Coverage and Health Systems Office of the Assistant Director-General, World Health Organization

PATIENT TESTIMONY

10:45 – 10:55

Maria Weigl, living with MPS IVA, Morbus Morquio

VIDEO PRESENTATION

10:55 – 11:00

Daniela Bas, Director, Division for Social Policy and Development Department of Economic and Social Affairs, United Nations

RARE DISEASE PATIENTS' NEEDS AND GOALS

11:00 – 11:30

Lene Jensen, Chief Executive Officer, Rare Diseases Denmark

RARE DISEASE POLICY PRIORITIES FOR THE FUTURE

PANEL DISCUSSION

11:30 – 12:30

Moderator: **Yann Le Cam**, Chief Executive Officer and Co-Founder, EURORDIS-Rare Diseases Europe

Panellists:

- **Holm Graessner,** Coordinator Solve-RD and ERN-RND
- **Lene Jensen**, Rare Diseases Denmark
- **Dr Rüdiger Krech, Director**, Universal Health Coverage and Health Systems Office of the Assistant Director-General, World Health Organization
- **Nathalie Moll**, Director General, EFPIA
- **Martin Seychell**, Deputy Director-General, Directorate-General for Health and Food Safety, European Commission
- **Dr. Christa Wirthumer-Hoche**, Head of Austrian Medicines and Medical Devices Agency

CLOSING PLENARY SESSION AGENDA

Saturday, 12 May 2018 - 16:15 – 17:00

TAKE HOME REMARKS

16:15 – 17:00

Moderator: **Vinciane Pirard**, Co-Chair, EFPIA-EuropaBio Joint Task Force on orphan drugs and rare diseases ; Public Affairs, Sanofi-Genzyme, Belgium

- **Theme 1: Lauren Roberts**, Director of Support, Genetic Alliance UK, National Coordinator, Swan UK, UK
- **Theme 2: Violeta Stoyanova-Beninska,** Committee for Orphan Medical Products (COMP) Member, Chair of National Scientific and Regulatory Advice, Netherlands
- **Theme 3: Julian Isla**, Data and Artificial Intelligence Resource Manager, Microsoft and Dravet Syndrome European Federation (DSEF), Spain
- **Theme 4: Ursula Holtgrewe**, Head of Work & Equal Opportunities, Zentrum für Soziale Innovation, Austria
- **Theme 5: Michael Schlander**, Professor of Health Economics, University of Heidelberg, Germany
- **Theme 6: Durhane Wong-Rieger**, President & Chief Executive Officer, Canadian Organization for Rare Disorders, Chair of Rare Diseases International, Founder of the Asia Pacific RD Alliance (APARDO), Canada

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

ADDITIONAL SUPPORT:
Mathieu Boudes, Public/Private Partnership Coordinator, European Patients' Forum (EPF)

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 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

Are we about to enter a world where all rare diseases will be diagnosed within a year? How are recent scientific breakthroughs impacting on diagnostic pathways and what trends can we expect in the near future? Join us as we explore what these trends will offer to patients and their families and how we can ensure they are kept at the centre of the debate.

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, 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

How can patient groups encourage research on their condition? How can researchers ensure that any research undertaken is exploitable and can be translated beyond the lab 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, Chief Executive Officer and Co-founder, SimOnics, UK

SESSION 0103

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

INNOVATIVE FUNDING PARTNERSHIPS: CHALLENGES AND OPPORTUNITIES

Can non-profits sucessfully advance research into specific areas? Are megafunds the answers to the challange of funding rare disease reasearch? Join us to answer these questions as we also explore the challenges and opportunities of innovative funding partnerships, how non-profuts can work together and what researchers want from patient groups.

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
Sean Kelly, Chief Executive, Action for A-T, UK

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'?

Join us as we explore what it takes to make an 'expert' patient an equal partner. Hear how and why patients can be trained in research skills, how patient organisations can best support their members to engage meaningfully in research and share your views to help us build a cloud of words defining just what an expert patient is!

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

Developing tools to empower patient experts
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 patient organisations best support 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?

Are you worried advancements in genomics mean we are heading towards a world without rare diseases? Or do you think advancements should be celebrated? Join us for lively debate as we explore the ethics of genome editing.

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 ASSESSORS (HTA) AND PATIENTS WORKING TOGETHER

THEME LEADERS:

Wim Goettsch, Special Advisor HTA for the Dutch National Health Care Organisation, Netherlands

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

Violeta Stoyanova-Beninska, COMP Member, Chair of National Scientific and Regulatory Advice, Netherlands

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

EURORDIS SUPPORT:

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

For rare diseases more than for others the concept of priority medicines is a relevant tool to stimulate development and timely registration of innovative breakthrough medicines. An overview of the experience from the COMP and the PRIME and HTA dialogue will set the scene to conclude how to move forward.

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

Can PRIME attract innovation towards unmet needs / disruptive medicines?
Steven Hall, Pfizer Global Research & Development, UK

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

What has changed since the existence of the current cooperation on health technology assessors (EUnetHTA)? What to expect from the EMA/EUnetHTA three-year work plan, announced in November 2017?

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
Speaker to be named

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

Analysis of HTA and reimbursement procedures in EUnetHTA partner countries
Peter O'Neill, Scientific Adviser, National Institute for Health and Care Excellence (NICE), UK

SESSION 0203

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

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

Patients are increasingly present and involved in the EMA regulatory process. But are they ready to contribute to the HTA assessments? How can they be prepared to step in?

Session Chair: to be named

The Community Advisory Boards (CAB) 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

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

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?

If you wonder why there is a big difference between the number of orphan designated products and orphan medicinal products on the market, you might find some answers after attending this session.

Session Chair: to be named

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

SESSION 0205

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

THE FUTURE OF HEALTH TECHNOLOGY ASSESSOR COOPERATION

What is the future of HTA cooperation in Europe? The Commission will present the Regulation Proposal at the session starting at 13:30 with all conference participants, for 20-30 minutes

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
Flora Giorgio, Head of Sector Health Technology Assessment, DG SANTE B4, European Commission

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
Mirjana Huic, Croatian Agency for Quality and Accreditation in Health Care and Social Welfare and Head of Department for Development, Research and HTA, Croatia

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 Senior 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: Elena Bonfiglioli, Director of Health Industry Business, Microsoft

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
- **David Martin Lindstrom**, Head of Device & Data Security at ElevenPaths, Telefónica, Spain

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**, Chief Executive Officer, 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
- **Brian O'Connor**, European Connected Health Alliance, UK
- **Henrique Martins**, Chief Executive Officer, 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:
Victoria Hedley, RD-ACTION Thematic 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
Elizabeth Vroom, Duchenne Parent Project, The Netherlands

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 Crocione, 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)

THEME 4

QUALITY OF LIFE: MAKING WHAT MATTERS, MATTER

THEME LEADERS:

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

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

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 as 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 achieve integrated care.

Lastly, the theme will venture into thinking about what 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, Board of Directors, EURORDIS-Rare Diseases Europe, Chief Executive Officer, Retina International, Ireland

Quality of life, what matters to people living with a rare disease and their carers?
Avril Daly, Vice-President, Board of Directors, EURORDIS-Rare Diseases Europe, Chief Executive Officer, 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**, Head of Patient Access at Wilson Therapeutics, Sweden
- **Ri De Ridder,** Director-General of Healthcare, National Institute for Health and Disability Insurance (RIZIV-INAMI), Belgium

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:

- **Gunta Anca**, General Secretary, European Disability Forum, Belgium
- **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 diseases 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:

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

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

EURORDIS SUPPORT:

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

THEME DESCRIPTION

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 diseases 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

The session aims to examine the patient burden in rare diseases from different angles. We will address the health, psycho-social and economic impact of rare diseases on patients, caregivers and the wider health care system. Results from recent cost of illness studies will be presented as well as experience from the perspective of clinicians and patients. The inclusion of the societal dimension is essential to measure the impact across all meaningful parameters. A better understanding of the full burden of a disease would help to assess the real value of a therapy and to implement a holistic policy approach to address persistent gaps in care and cure.

Session Chair: Sandra Nestler-Parr, Managing Director, Rare Access, UK

Speakers:

- **Jamie O'Hara,** University of Chester, UK
- **Mondher Toumi,** Aix-Marseille University, France
- **Mariangela Pellegrini,** ERN BloodNet Programme Manager, France
- **Lise Murphy,** EURORDIS-Rare Diseases Europe

SESSION 0502

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

DYNAMICS OF THERAPY DEVELOPMENT FOR RARE DISEASES

Developing a new rare disease therapy is a fascinating yet complex and costly challenge. This session will deep dive into the dynamics of R&D for rare diseases therapies, underline the specificities of business models focusing on rare diseases, the role of incentives in the rare disease ecosystem.

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

Speakers

- **Tim Wilsdon**, Vice President Charles River Associate, UK
- **Chris Sotirelis,** EURORDIS-Rare Diseases Europe
- **Maurizio Scarpa,** MetabERN coordinator, Germany
- **Anant Murthy,** Vie President, Market Access & Pricing, Alnylam Pharmaceuticals, Switzerland

SESSION 0503

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

A PARADIGM SHIFT IN VALUE FRAMEWORKS FOR ACCESS

The session will focus on the reasons why the conventional health economic paradigm often fails to capture the full social value of interventions for rare and very diseases. Elements of an extended or alternative evaluation paradigm will be discussed. Presentations will build on new empirical research, providing evidence for the will of citizens to share scarce health care resources and for the implications of changing the cost perspective – from incremental cost per case (and length and quality of life gained per case) to incremental cost per member of a National Health Scheme ("NHS", or mandatory health insurance plan) caused by adding a health care programme to the "basket" offered by a national health service.

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

Speakers:

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

SESSION 0504

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

NEW APPROACHES TO PRICING AND FUNDING AND IMPLICATIONS FOR ACCESS

People with rare diseases across Europe still experience difficulties and inequalities in access to adequate therapies for their conditions. This session will explore new approaches to funding and innovative payment models, including collaborative approaches, payment based on outcomes, how to deal with uncertainties and other types of cooperation mechanisms at European level.

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

Speakers:

- **Diane Kleinermans,** Advisor to the Ministry of Health, Belgium
- **Alexander Natz,** Secretary General, EUCOPE
- **Allen King,** Pipeline Lead – Rare Disease Patient Health and Impact (PHI), Pfizer, USA
- **Brian O'Mahony,** President, European Haemophilia Consortium (EHC), Ireland

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

This final session will wrap up the Theme 5 with a look to the future. If science continues to deliver and progress at this pace, what will need to be in place by 2030 to ensure that people with rare diseases have access to the treatment they need? How will healthcare providers be able to provide them? What should the R&D framework look like?

Session Chair: Avril Daly, Vice-President, Board of Directors, EURORDIS-Rare Diseases Europe, Chief Exectutive Officer, Retina International, Ireland

Speakers:

- **Miriam Dalmas**, Health Ministry, Malta, ERN Board of Member States representative, Malta
- **Martin de Graff**, ZIN, Netherlands
- **Agnès Jaulent**, EspeRare, Translational Project Leader, Switzerland

THEME 6

GLOBAL RARE EQUITY: ARE WE THERE YET?

THEME LEADERS:

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

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

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)

How can we leverage global policies and agency frameworks to explicitly support rare diseases recognising diversity and ensuring equity? In this session, key individuals drawn from influential global entities, inside and outside of the rare diseases space, will set the stage for a vibrant discussion on how to translate this from a challenge into a timely opportunity to transform policy, and informed by engaging the entire audience.

Moderator: Jeff Sturchio, President & Chief Executive Officer, Rabin Martin, USA

Overview presentation
Yann Le Cam, Chief Executive Officer and Co-Founder, EURORDIS-Rare Diseases Europe

Panel Discussion

- **Angela Chaves Restrepo**, Chief Executive Officer, Federación Colombiana de Enfermedades Raras, Colombia
- **Rüdiger Krech**, Director of the Department of Ethics and Social Determinants of Health, World Health Organisation (WHO)
- **Emmanuel Akpakwu**, Project Lead, Value in Healthcare, Global Health and Healthcare Industries
- **Matthew Harold**, International Public Affairs, Rare Diseases, Pfizer, UK

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 ?

What global opportunities do we unlock when all people living with a rare disease have access to timely accurate diagnosis and optimised care? This panel will explore some of the advances and innovations in rare disease diagnosis and care and, importantly, the impact for patients, families; transformation for health systems; and more broadly the innovation for all sectors of society.

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

Overview presentation
Moeen Alsyed, Global Commission on Ending Diagnostic Odyssey, Saudi Arabia

Panel Discussion

- **Moeen Alsyed**, Global Commission on Ending Diagnostic Odyssey, Saudi Arabia
- **Ross Selby**, Head of Global Patient Access, Takeda Oncology, UK
- **Olivia Romero-Lux**, World Federation of Hemophilia, Canada
- **Laura Arbour,** Department of Medical Genetics, University of British Columbia, Canada

SESSION 0603

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

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

What to do when you exceed your targets ahead of your deadline? Post more audacious targets with more ambitious timelines and push for real world action! Join bold actors of the International Rare Disease Research Consortium as they promote IRDiRC's 2027 targets and challenge you to become a part of the planning and activity to get there.

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
- **Sonja van Weely,** E-Rare, Netherlands
- **Prof. Getnet Tadele**, Addis Ababa University, Ethipoia

SESSION 0604

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

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

How are we building the rare disease knowledge and information eco-system; what components, such as different knowledge (data) platforms and resources (information) banks could be aligned to this vision through better connections (fibre optic vs copper wire)? What are innovative, emerging and revolutionary technologies converging for real time connectivity? How have multi-stakeholder networks and collaborations been effective in redefining the new rare disease knowledge and information eco-system?

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

Overview presentation
Speaker to be named

Panel Discussion

- **Christina Waters**, Chief Executive Officer and Founder, Rare Science, United States
- **Dr Mike Brudno**, Scientific Director, SickKids, Associate Professor, Department of Computer Science, University of Toronto, Canada
- **Olivier Menzel,** Chairman, BLACKSWAN Foundation, Switzerland
- **Arndt Rolfs**, Chief Executive Officer, Centogene

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?

How can we create a powerful shared vision for equity and optimised care globally to assure all persons living with a rare disease have access to the best care possible in their local environment? How can we take advantage of learning from impactful programmes and initiatives in different locales to collectively address the challenge of global equity in diagnosis, care, treatment and support?

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

Overview presentation
Matt Bolz-Johnson, ERN & Healthcare Advisor, EURORDIS-Rare Diseases Europe

Panel Discussion

- **Ritu Jain**, President, DEBRA Singapore and Member of the Executive Committee of DEBRA International Singapore
- **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
- **Professor Hugh Dawkins**, Director, Office of Population Health Genomics, Health Department of Western Australia, Australia
- **Ramaiah Muthyala**, Indian Organization for Rare Diseases

POSTERS

POSTER SESSIONS AND MODERATED POSTER WALKS

FRIDAY

13:00 - 14:00 Poster Session

13:00 - 13:30 Moderated Poster Walk: Theme TBC, Lucia Monaco, Telethon Italia

13:30 - 14:00 Moderated Poster Walk: ERNs, Matt Bolz-Johnson, EURORDIS-Rare Diseases Europe

15.30 - 16.30 Poster Session

15:30 - 16:00 Moderated Poster Walk: Rare Disease Patient Groups Innovations, Danijela Szili, Rett Syndrome Europe

16:00 - 16:30 Moderated Poster Walk: ERNs, Matt Bolz-Johnson, EURORDIS-Rare Diseases Europe

SATURDAY

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Silvia Sestini, Lia Millucci, Giulia Bernardini, Daniela Braconi, Marco Bardelli, Barbara Marzocchi, Ottavia Spiga, Maurizio Orlandini, Bruno Frediani and Annalisa Santucci

P 4 | Developing the roadmap for collaboration between patients and researchers about genome ELSI on clinical research and policy
Shun EMOTO, Go YOSHIZAWA, Kei KANO, Kunihiro NISHIMURA, and Yukiko NISHIMURA

P 5 | An integrated interactive ecosystem for alkaptonuria: a tool for physicians and researchers
Silvia Sestini, Vittoria Cicaloni, Andrea Zatkova, Lia Millucci, Giulia Bernardini, Andrea Bernini, Neri Niccolai, Alfonso Trezza, Daniela Braconi, Ottavia Spiga, Annalisa Santucci.

P 8 | BH4-response prediction in PKU patients in Georgia
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P 9 | Diagnosis of genetic diseases in developing countries: is it possible to work by the guidelines? DYT1 dystonia in a Romanian patient. Case study
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P 11 | RD-Connect: an integrated infrastructure for data sharing and analysis in rare disease research
S. Beltran, D. Piscia, S. Laurie, J. Protasio, A. Cañada, J.M. Fernández, R. Kaliyaperumal, S. Lair, P. Sernadela, M. Girdea, R. Thompson, H. Lochmüller, D. Badowska, V. Straub, M. Roos, P.A.C. 't Hoen, A. Valencia, L. Monaco, CM. Wang, D. Taruscio, S. Gainotti, Y. Kodra, C. Carta, P. Torreri, D. Salgado, C. Béroud, I. Gut and the RD-Connect Consortium

P 13 | Undiagnosed: Genetic conditions and the impact of genome sequencing
Emily Muir, Farhana Ali, Nick Meade, Lauren Roberts

P 14 | Progress in Rare Diseases Research 2010–2017: An IRDiRC Perspective
Jagut M, Jonker AH, Lau LPL, Cutillo CM, Rath A, Dawkins HJS, Austin CP on behalf of IRDiRC

P 15 | PROFILE: Immunoprofile-directed stratification of patients with the autoimmune disorder thrombotic thrombocytopenia purpura
Nuno Graça, Johana Hrdinova, Nick Geukens, Wim Maes and Karen Vanhoorelbeke

P 17 | Initiative for the harmonization of the quality assessment for analyses performed by the Belgian Centers for Human Genetics in the context of Rare Diseases
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P 18 | OKIDS Activities Q3-2013 to Q4-2017
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P 19 | Developing Integrated Care in the Context of Rare Chromosomal Conditions: 22q11 Deletion Syndrome - A parent/clinician action research collaboration
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P 20 | Recommendations For Improving Quality Of Rare Diease Registries
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P 25 | Depression and anxiety in patients with pulmonary hypertension: Looking beyond disease status
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P 27 | Training and empowering patients on scientific research: the example of Fondazione Telethon
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P 28 | Target 5000 – Gateway to Vision for Irish Retinal Degeneration Patients
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Vivienne Beckett; Jane Lewthwaite; Sindisiwe Mnkandla

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Zsuzsanna Bojtor Pogány, Krisztina Scholtz, Anita Pataki, Anna Pogány, Beáta Boncz

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Gábor Pogány, Eszter Fogarassy, Zsuzsanna Bojtor

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Barbara Glinsner, Irina Vana, Ursula Holtgrewe

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T. Harris (PKD Charity Chief Executive); M. Kokocinska, L. Kerecuk, D. Milford, S. Hulton, C. O'Brien, M. Muorah, S. Stephens, S. Parkes (Birmingham Women's & Children's NHS Foundation Trust); M. Dillon (UK Renal Registry), L. Charles, C. Cotter (NIHR WM CRN)

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Chloe Tuck, Helen Lycett, Eva Raebel, Matt Johnson and Tom Kenny

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Ritchie Head; Christina Olsen; Dimitrios Athanasiou, Eric Hoffman

P 249 | Pitt-Hopkins syndrome
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Srinivasa Rambhatla, Alexander Moffat, Priyesh Chauhan, Aniket Sonsale, Sharon Parkes, Maria Kokocinska, Larissa Kerecuk, Tarekegn Geberhiwot

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Georgia Hollier-Hann, James Wordsworth, Stephen Ralston, David Cork
Name of person presenting the poster on-site: Stephen Ralston

P 256 | Assessing Criteria for NICE recommendation with the HST Programme
Caroline Upton, James Wordsworth, David Cork, Alistair Curry, Georgia Hollier-Hann, Stephen Ralston

P 259 | Survey on rare disease patients' experiences of participation and possibilities of influencing in social and health care service structures
Airos M., Franck K., Heikkinen R., Kiikala-Siuko M., Saari K. and Vataja P.

P 260 | Assessing the role of Material and Technology among the families of children and young people with Metabolic Rare Diseases
Dr Suja Somanadhan: Assistant Professor of Children's Nursing, UCD, Dublin, Ireland, Prof Philip Larkin: Professor of Clinical Nursing (Palliative Care), UCD, Dublin, Ireland

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P 263 | Rare diseases magazine - information platform for all stakeholders in rare diseases in Russia
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N.-K. Walleczek, K. Arbeiter, P. R. Espina, T. Hörtenhuber, A. Kreissl, S. Greber-Platzer

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Nutini Michele, Mingarelli Rita, Ciampa Serena, Radio Francesca Clementina, Dallapiccola Bruno

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Kari Hagen, Lene S Gloslie, Elisabeth F Bækken, Rasmus S Dinessen, Bjørn-Magne Stuestø

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P 276 | Efficacy of an orphan drug: The patient perspective
Cornelia Dechant, MD; Francesca Granata, PhD; Jasmin Barman-Aksözen, PhD

P 277 | Things Left Unsaid - Important topics not discussed during systemic sclerosis consultations: impact on patients and caregivers
CP Denton, B Laird, L Moros, JL Luna Flores

P 279 | The extended newborn's screening has finally become a compulsory national public service in Italy: none of them will be left behind anymore
Manuela Vaccarotto, Giuliana Valerio

P 280 | Challenges in physician-patient communication limit patient understanding and support in SSc-ILD
CP Denton, B Laird, L Moros, JL Luna Flores

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Bobinec A, Ivankov AM, Kero M, Sansović I, Barišić I

P 285 | The UN Sustainable Development Goal 4 - How can Ågrenska and the Rare Disease Community cooperate and try to contribute?
AnnCatrin Röjvik, Gunilla Jaeger, Ågrenska

P 286 | Easy access to reliable information about rare diseases
The Swedish Information Centre for Rare Diseases

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P 291 | Cooperation in the Field of Rare Diseases: A Social Science Perspective
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P 296 | Overview of Rare Renal Diseases at a UK Paediatric Renal Centre through the National Registry of Rare Kidney Diseases (RaDaR)
S. Parkes, M. Kokocinska, L. Kerecuk, D. Milford, S. Hulton, M. Muorah, C. O'Brien, S. Stephens, (Birmingham Women's and Children's Hospital); M. Dillon (UK Renal Registry); L. Charles, C. Cotter (NIHR WM CRN)

P 299 | National Plan of Action for People with Rare Diseases. Further developments
Dr. Miriam Schlangen, Katharina Heuing, M.Sc.

P 301 | Lessons learned from IDeAI - 33 recommendations from the IDeAI-Net about design and analysis of small population clinical trials
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Katharina Niedermayr (MD), Gerhard Pölzl (MD), Sabine Scholl-Bürgi (MD), Christine Fauth (MD), Edda Haberlandt (MD), Ursula Albrecht (MD), Wolfgang Sperl (MD), Johannes A. Mayr (PhD), Daniela Karall (MD)

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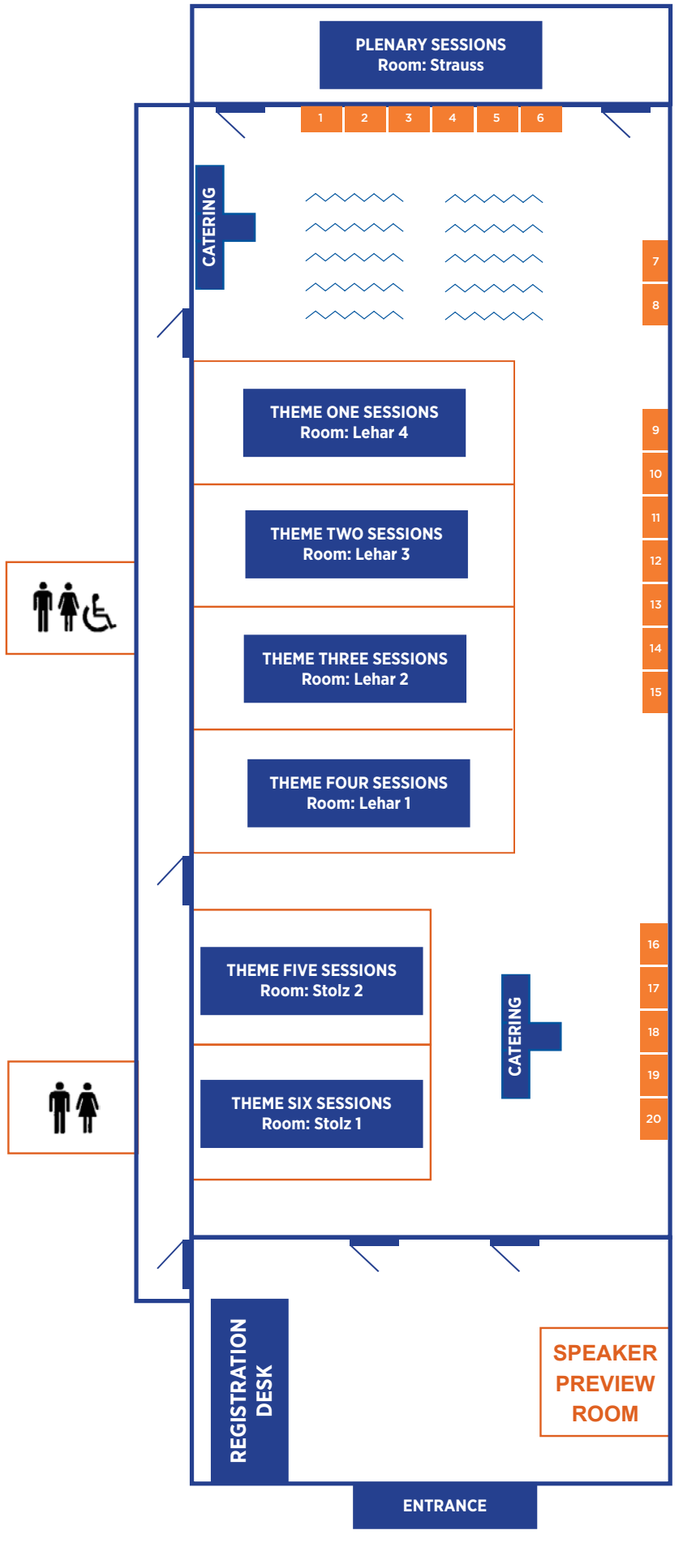
Hanisch L., Hanisch M., Timme M., Benz K , Danesh G.,Jackowski J.

EXHIBITION HALL FLOOR PLAN



1 Table-top exhibits

Poster boards

Doors

MESSE WIEN CONGRESS CENTER (GROUND FLOOR)

EXHIBITING ORGANISATIONS

VISION-DMD | SPACE 1

VISION-DMD is a US-EU collaborative Horizon 2020 project building on patient driven and academic-led research. The aim is to bring an innovative affordable therapy to clinic that delivers a significant improvement on current therapeutic approaches and standard of care for all Duchenne Muscular Dystrophy patients. The project also advances innovate biomarkers and MRI techniques for assessing DMD.

RD-CONNECT | SPACE 2

RD-Connect is an EU-funded global platform that facilitates research on rare diseases by connecting databases, patient registries, biobanks and clinical bioinformatics data into a central resource and analysis tool for researchers and clinicians worldwide.

EUROPEAN REFERENCE NETWORKS | SPACE 3

The ERN exhibition space will showcase some of the activities developed by the European Reference Networks (ERNs) that deal with rare and low prevalence complex medical conditions. Visitors will be able to get a better understanding of what these Networks have achieved so far and what are some of the projects underway.

ORPHANET-INSERM, FRANCE | SPACE 4

Orphanet (www.orpha.net) is a unique resource, gathering and generating knowledge on rare diseases so as to improve the diagnosis, care and treatment of patients with rare diseases. Orphanet maintains the Orphanet rare disease nomenclature (ORPHA number) and provides access to data via www.orphadata.org. Orphanet is coordinated by INSERM US14.

RD-ACTION | SPACE 5

RD-ACTION ('Data and Policies for Rare Diseases') is the European Joint Action for Rare Diseases, and can be viewed as a successor to both the EUCERD and the Orphanet Joint Actions. Uniting 34 beneficiaries and over 30 collaborating partners from 40 countries, RD-ACTION elaborates and implements policies pertaining to rare diseases across Europe.

OPENAPP | SPACE 6

OpenApp is one of the leading health informatic companies focussed on delivering professional services for disease registries. We develop tailored patient-centric solutions primarily focused on patient, clinical and research organisations. We also have extensive experience in health data analytics including geospatial analysis.
Our platforms make a real difference for patients, clinical care, research and health managers. Our clients include DG Sante with the Clinical Patient Management System, the European Cystic Fibrosis Foundation, CATs Foundation, Alpha 1 and some of the leading pharmaceuticals developing drugs for rare diseases.
Come and talk to us about patient registries, portals and how we help with observational/Phase IV drug trials.

CENTOGENE I SPACE 7

CENTOGENE is dedicated to the highest quality genetic and biochemical diagnostic testing. Our molecular understanding of rare diseases allows for a personalized and focused therapy and improved control of rare and congenital diseases. As one of the world's leading companies in molecular diagnostics of rare hereditary diseases, we support clinicians with expert genetic counseling services. We deliver solutions for rare disease diagnostics on a laboratory scale. High quality reporting is our key essential for building a partnership of trust.

PFIZER | SPACE 8

Pfizer Rare Disease is committed to transforming the lives of the rare disease community through life-changing innovations, trusted partnerships, and relentless passion. For thirty years, Pfizer Rare Disease has been a part of the journey with the rare disease community. We partner with patients, advocacy groups, health care professionals, scientists, and payers from discovery to delivery of medicines, relentlessly committed to providing access to medicines and sponsoring support programs in more than 80 countries.
With the backing of Pfizer's R&D, manufacturing, medical expertise, and educational resources, Pfizer Rare Disease has an established global footprint. We have a dedicated research unit with more than 70 passionate scientists working together to identify potential treatments for patients. We have a portfolio of multiple medicines within a number of disease areas, including hematology, neuroscience, and inherited metabolic disorders. To date, Pfizer Rare Disease has treated 76,000 people with its medications globally.
We have been here, we are here, and we will be here for the rare disease community. We believe it's time to seize the moment; and we are here to listen, to learn, and make a difference.

EURORDIS-RARE DISEASES EUROPE | SPACE 9 & 10

EURORDIS-Rare Diseases Europe is a non-governmental patient-driven alliance of patient organisations representing 798 rare disease patient organisations in 69 countries. EURORDIS represents the voice of an estimated 30 million people living with a rare disease in Europe. Please come to stand 9 & 10 to meet members of the EURORDIS team and find out more about key initiatives including Rare Diseases International, Rare Disease Day and the Rare Barometer survey initiative.

RARE BAROMETER | SPACE 11

At the heart of the EURORDIS Rare Barometer Programme is the idea that the advocacy work of EURORDIS and its members should continue to be increasingly based on patient perspectives.
Rare Barometer consists of surveys aiming to collect qualitative & quantitative data on the experience and expectations of rare disease patients and their families to facilitate and streamline the inclusion of patient perspectives into EURORDIS advocacy work.
As part of the Rare Barometer Programme, Rare Barometer Voices, an online panel of more than 7000 people living with a rare disease who are willing to participate in EURORDIS' surveys, is the tool used to carry out quantitative surveys.
The results of Rare Barometer studies and surveys are communicated to:
Patient organisations, so that they can use them to raise awareness among policy makers and the general public
European and International-level policy makers and other influential figures so that they are made aware of actions that need to be taken for the rare disease community in Europe and internationally
The general public including health care providers, to raise awareness about rare diseases.

EXHIBITING COMPANIES

DIA | SPACE 12

DIA is a global association that mobilizes life science professionals from across all areas of expertise to engage with patients, peers, and thought leaders in a neutral environment on the issues of today and the possibilities for tomorrow.

NORD | SPACE 13

The National Organization for Rare Disorders (NORD) is leading the fight to improve the lives of rare disease patients and families in the United States. NORD, along with its members, is dedicated to the identification, treatment, and cure of rare disorders. We do this by supporting patients and organizations, accelerating research, providing education, disseminating information, and driving public policy.
Learn more by visiting www.rarediseases.org

PPTA EUROPE | SPACE 14

The Plasma Protein Therapeutics Association (PPTA) represents the private sector manufacturers of plasma-derived and recombinant analog therapies and the collectors of source plasma used for fractionation. These therapies are used by small patient populations worldwide to treat a variety of rare diseases and serious medical conditions.

NEM.IO FOUNDATION | SPACE 15

NEM's blockchain technology delivers a world-class platform for management of almost any kind of asset: supply chain, notarizations, claims, records, etc. Whether you're building the next mobile app or bringing blockchain into your business infrastructure, NEM gives you a platform to deploy the best of blockchain technology for your business.

SAREPTA THERAPEUTICS | SPACE 16

Sarepta Therapeutics is a commercial-stage biopharmaceutical company focused on the discovery and development of precision genetic medicines to treat rare neuromuscular diseases. The Company is primarily focused on rapidly advancing the development of its potentially disease-modifying Duchenne muscular dystrophy (DMD) drug candidates and is proud to support the ECRD 2018. For more information about Sarepta, please visit http://www.sarepta.com

OPEN HEALTH | SPACE 17

OPEN Health is a family of specialist healthcare communications and market access businesses with extensive experience and expertise in rare disease. In a complex world of evidence-based medicine and market access challenges we remove the boundaries between data and analytics, scientific expertise, creativity and digital know-how to provide bespoke solutions that meet the challenges of rare disease.

BMORE | SPACE 18

Our global work with Ablynx, Kyowa Kirin, Shire and Ultragenyx has taught us that the life of a rare disease patient can often be full of complex messages and confusion.
It can be very difficult for a patient and their families to identify and diagnose their condition with HCPs and then find the most effective course of treatment for a 'normal way of life'
Both Pharma companies and digital solutions can sometimes over complicate things in terms of terminology and techie phrases.
Bmore group has 4 divisions: Bmore educated, Bmore creative, Bmore digital, Bmore connected.
What do we do? We simplify complexity.
We use knowledge, understanding of science, global healthcare, creativity and innovation to deliver multi-channel campaigns for better health for life, from trial to treatment.
bmore.agency - www.bmore.co.uk

SUPPORTING PARTNERS

We would like to thank our supporters for their financial contributions in the context of this important event for the rare disease community

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